SIXTH EDITION

Conditions in Occupational Therapy

Effect on Occupational Performance

Ben J. Atchison
Diane Powers Dirette

Wolters Kluwer

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Contributors

Debbie Amini, EdD, OTR/L, FAOTA
Director of Professional Development
American Occupational Therapy Association
Bethesda, Maryland

Ben J. Atchison, PhD, OTR/L, FAOTA
Professor and Chair Emeritus
Department of Occupational Therapy
Western Michigan University
Kalamazoo, Michigan

Molly Bathje, PhD, MS, OTR/L
Assistant Professor
Department of Occupational Therapy
Rush University
Chicago, Illinois

Mary Frances Baxter, PhD, OT, FAOTA
Professor and Associate Director
School of Occupational Therapy
University of Indianapolis
Indianapolis, Indiana

Shirley Blanchard, PhD, OTR/L, ABDA, FAOTA, FHDR
Professor
Department of Occupational Therapy
Creighton University
Omaha, Nebraska

Angie K. Boisselle, PhD, OTR
Utilization Management
Therapy Manager
Cook Children's Health System
Fort Worth, Texas

Lori E. Breeden, EdD, OTR/L
Associate Professor
School of Occupational Therapy
University of Indianapolis
Indianapolis, Indiana

Cara L. Brown, OTReg (MB), PhD
Assistant Professor
University of Manitoba
Winnipeg, Manitoba, Canada

Susan M. Cahill, PhD, OTR/L, FAOTA

Founding Program Director
Lewis University
Romeoville, Illinois

Susan D. Charnley, DrOT, OTR/L, CHT
Assistant Professor and Program Director
Lewis University
Romeoville, Illinois

Joan Ziegler Delahunt, OTD, MS, OTR/L
Associate Professor
Department of Occupational Therapy
Rockhurst University
Kansas City, Missouri

Diane Powers Dirette, PhD, OTL, FAOTA
Professor
Interdisciplinary Health Sciences PhD Program
Western Michigan University
Kalamazoo, Michigan

Rosanne DiZazzo-Miller, PhD, DrOT, OTRL, CDP
Associate Professor of Occupational Therapy Director
Division of Health Sciences Mentoring Program
Wayne State University
Detroit, Michigan

Kathryn Ellsworth, MA, CCC-SLP
Speech-Language PathologistKalamazoo Speech Associates
Kalamazoo, Michigan

Yael Goverover, PhD, OTR/L
Professor and Director Post-Professional Programs
New York University
New York, New York

Cynthia A. Grapczynski, EdD, OTR, FAOTA
Professor and Chair (Retired)
Occupational Science and Therapy Department
Grand Valley State University
Allendale, Michigan

Holly Grieves, OTD, OTR/L
Faculty Clinical Specialist II
Department of Occupational Therapy
Western Michigan University
Kalamazoo, Michigan

Sharon A. Gutman, PhD, OTR/L, FAOTA
Professor Occupational Therapy Doctorate Program
Rutgers University
Newark, New Jersey

Midge Hobbs, OTD, OTR/L IMPACT
Practice Curriculum Director and Assistant Professor
MGH Institute of Health Professions
Boston, Massachusetts

Nancy Hock, PhD, OTR/L, CHT, FMOTA
Master Faculty Clinical Specialist and Site Coordinator of Grand Rapids Campus
Department of Occupational Therapy
Western Michigan University
Kalamazoo, Michigan

Catherine R. Hoyt, PhD, OTD, OTR/L
Instructor Program in Occupational Therapy
Washington University
St. Louis, Missouri

Debra Latour, PP-OTD, MEd, OTR/L
Assistant Professor of Occupational Therapy
Western New England University
Springfield, Massachusetts

Sheila M. Longpré, PhD, MOT, OTR/L
Assistant Professor Occupational Therapy Program
Eastern Michigan University
Ypsilanti, Michigan

Allison Chamberlain Miller, MS, OTR/L
Visiting Clinical Professor
Department of Occupational Therapy
Indiana University South Bend
South Bend, Indiana

Brandon G. Morkut, MS, OTR/L, CAPS
Occupational Therapist
Van Buren Intermediate School District
Lawrence, Michigan

Shelley Mulligan, PhD, OTR/L, FAOTA
Associate Professor
Department of Occupational Therapy
University of New Hampshire
Durham, New Hampshire

Linda M. Olson, PhD, OTRL, FAOTA
Chairperson and Program Director
Department of Occupational Therapy
Rush University
Chicago, Illinois

Rebecca Ozelie, DHS, OTR/L, BCPR
Associate Professor and Academic Fieldwork Coordinator
Department of Occupational Therapy

Rush University
Chicago, Illinois

Katie M. Polo, DHS, OTR, CLT-LANA
Associate Professor
School of Occupational Therapy
University of Indianapolis
Indianapolis, Indiana

Pat Precin, PhD, PsyaD, NCPsyA, LP, OTR/L, FAOTA
Assistant Professor
Rehabilitation and Regenerative Medicine Programs in Occupational Therapy
Columbia University
New York, New York

Emily Raphael-Greenfield, EdD, OTRL, FAOTA
Assistant Professor
Programs in Occupational Therapy
Columbia University Medical Center
New York, New York

Gayle Restall, PhD, OTR/L
Proffessor
Department of Occupational Therapy
College of Rehabilitation Sciences
Rady Faculty of Health Sciences
University of Manitoba
Winnipeg, Manitoba, Canada

Mylene Schriner, PhD, OTR/L
Associate Dean College of Health and Human Services
Rockhurst University
Kansas City, Missouri

Jennifer L. Smith, MS, OTR/L
Occupational Therapist
Team Rehabilitation
Bingham Farms, Michigan

Michelle A. Suarez, PhD, OTR/L
Associate Professor
Department of Occupational Therapy
Western Michigan University
Kalamazoo, Michigan

Diane B. Thomson, MS, OTR/L, ATP
Senior Occupational Therapist
Rehabilitation Institute of Michigan
Detroit, Michigan

Wendy Tremaine, PhD, OTR/L
Clinical Assistant Professor
Department of Occupational Therapy

University of Michigan-Flint
Flint, Michigan

Michael J. Urban, MS, OTR/L, MBA, CEAS, CWCS
Senior Lecturer
University of New Haven
West Haven, Connecticut

Amy Wagenfeld, PhD, OTR/L, SCEM, EDAC, FAOTA
Lecturer
Post-Professional OTD Program
Department of Occupational Therapy
Boston University
Boston, Massachusetts

Beth Ann Walker, PhD, MS, OTR/L, FAOTA
Associate Professor
School of Occupational Therapy
University of Indianapolis
Indianapolis, Indiana

Andrea L. Washington, BS, OTR/L
Clinical Occupational Therapy Specialist
Inpatient Rehabilitation
Children's Hospital of Michigan
Detroit, Michigan

Lee Ann Westover, MS, OTR/L
Private Practitioner and Consultant
New York, New York

Jillian Woodworth, DrOT, OTR/L
Clinical Assistant Professor
Occupational Therapy Program
University of Michigan-Flint
Flint, Michigan

Tracy R. Young, MHS, OTRL
Faculty Clinic Specialist
Department of Occupational Therapy
Western Michigan University
Kalamazoo, Michigan

Preface

Welcome to the sixth edition of *Conditions in Occupational Therapy: Effect on Occupational Performance*. Since 1993, when the first edition was published, the purpose has remained consistent: to provide occupational therapists with epidemiological information about common conditions of the people with whom they work and to identify the impact these conditions have on their occupational performance. Each chapter provides detailed information about the etiology, incidence and prevalence, signs and symptoms, diagnosis, course and prognosis, and medical/surgical management of a condition and discusses the impact of the deficits in client factors on areas of occupational performance. The areas of occupational performance that are addressed in this new edition include activities of daily living, instrumental activities of daily living, health management, rest and sleep, education, work, play and leisure, and social participation. Each chapter also includes case illustrations to provide the reader with examples of how a person with whom they work might experience the condition.

In this new edition, we have updated the areas of occupational performance based on the *Occupational Therapy Practice Framework*, fourth edition (OTPF-4). New to OTPF-4 is the inclusion of health management as an area of occupational performance. Health management is the ability to develop and maintain health and wellness routines, organize and manage one's own medical care, maintain personal care devices, and manage medication. In addition to guidance from the OTPF-4, the World Health Organization's International Classification of Functioning (ICF) was used to guide the concepts of health conditions and their relationship to body functions and structures, activities, and participation.

We have reorganized this sixth edition of the textbook into four units: Unit 1, Pediatric Conditions; Unit 2, Mental Conditions; Unit 3, Physical Conditions; and Unit 4, General Medical Conditions. The first three units were in the fifth edition but the General Medical Conditions Unit is new to this edition. The General Medical Conditions Unit includes conditions for which occupational therapists may not get primary diagnosis referrals, but which may be common in people who are treated by occupational therapists, either because they are referred for other conditions or for treatment of the secondary effects from these conditions.

We have added new chapters that provide epidemiological information and impact on occupational performance for five additional conditions. Sickle cell anemia is a new addition in the Pediatric Conditions Unit. Musculoskeletal pain and amputations are new additions in the Physical Conditions Unit. General deconditioning and infectious diseases are new additions in the General Medical Conditions Unit.

The experts who have written the chapters in this textbook provide occupational therapists with the tools they need to formulate comprehensive evaluation and treatment plans for people with these conditions either individually or in combination. The textbook does not prescribe the steps for evaluation and treatment but rather provides all the necessary information to understand the underlying condition and how it can impact a person's ability to participate in their daily occupations. We expect that each chapter is a starting point for discussion and analysis of the condition which then will lead to the development of effective intervention planning.

Diane Powers Dirette
Ben J. Atchison

Contents

CHAPTER
1

Thinking Like an OT

Ben J. Atchison and Diane Powers Dirette

KEY TERMS

Core values
International Classification of Functioning, Disability and Health
Occupational Therapy Practice Framework
Personalized medicine
Person-first language
Philosophical assumptions

It is more important to know what kind of person has the disease than what kind of disease the person has.
—Sir William Osler (Address at Johns Hopkins University, February 1905)

Lindsey is finishing her course work in occupational therapy and is now beginning her first level II fieldwork experience. Throughout her education, she has learned the importance of evidence-based practice to guide her treatment decisions. Her challenge now is to develop her clinical reasoning skills to merge the science she has learned with the art of practice. To achieve this, she must understand the person's diagnosis, analyze the person's unique set of problems based on the person's individual characteristics and determine the impact on occupational performance. The first step of this process is the referrals she receives. Each referral gives her some basic information about the person including the person's diagnosis. Her job is to decide what to do next.

How does a student learn to correlate general information about a diagnosis with the needs of a particular person and to identify the problems that require occupational therapy intervention? How does a staff therapist set priorities for problems and decide which require immediate attention? How much problem identification can be done before the therapist actually sees the individual? How do supervisors know when their student or staff therapist is effectively screening referrals and anticipating the dysfunction that the patient might be experiencing? These first steps are essential to the actual intervention process and are the first steps on ongoing clinical reasoning.

The clinical reasoning procedure used by each health care professional is somewhat different. The main focus of intervention for a speech therapist will differ from that of a psychologist or a nurse. An essential skill of the occupational therapist is to effectively gather and apply information that leads to a plan to help people function in their daily activities. Such data gathering and analysis provide the therapist with the foundation for a treatment plan through a prioritized list of anticipated problems or dysfunctions for an individual.

To comprehend the unique aspects of occupational therapy requires an understanding of the core values, philosophical assumptions, and domain of concern of the profession, as well as the language that is used to communicate information clearly and precisely.

Core Values of Occupational Therapy

The **core values** of occupational therapy were initially and officially published in the document "Core Values and Attitudes of Occupational Therapy Practice" (Kanny, 1993) and have been continuously reaffirmed by way of official documents published by the American Occupational Therapy Association including the Occupational Therapy Code of Ethics (AOTA, 2015) and the most recent revision of Occupational Therapy Practice Framework (4th edition) (AOTA, 2020). In addition, the Accreditation Standards for Occupational Therapy Education (ACOTE) includes the requirement that all levels of educational programs in occupational therapy ensure that students demonstrate knowledge of the AOTA Code of Ethics through application of the core values in everyday practice. There are seven

core values that have been identified including altruism, dignity, equality, freedom, justice, truth, and prudence and are defined as follows:

1. Altruism is the unselfish concern for the welfare of others. This concept is reflected in actions and attitudes of commitment, caring, dedication, responsiveness, and understanding.
2. Dignity emphasizes the importance of valuing the inherent worth and uniqueness of each person. This value is demonstrated by an attitude of empathy and respect for self and others.
3. Equality requires that all individuals be perceived as having the same fundamental human rights and opportunities. This value is demonstrated by an attitude of fairness and impartiality.
4. Freedom allows the individual to exercise choice and to demonstrate independence, initiative, and self-direction.
5. Justice places value on the upholding of such moral and legal principles as fairness, equity, truthfulness, and objectivity.
6. Truth requires that we be faithful to facts and reality. Truthfulness or veracity is demonstrated by being accountable, honest, forthright, accurate, and authentic in our attitudes and actions.
7. Prudence is the ability to govern and discipline oneself through the use of reason. To be prudent is to value judiciousness, discretion, vigilance, moderation, care, and circumspection in the management of one's affairs, to temper extremes, make judgments, and respond on the basis of intelligent reflection and rational thought (Kanny, 1993).

These seven core values are the foundation of the belief system that occupational therapists use as a moral guide when making clinical decisions. A seminal article by Peloquin (2007) suggested that occupational therapists consider five additional core values to further reflect the "ethos" of our profession. She eloquently described a profession's ethos as "an interlacing of sentiment, value, and thought that capture its character, conveys its genius, and manifests its spirit." (p. 475). Simpson and Weir (1989; as cited in Peloquin) defined these five additional core values that include the following.

- Courage: "the act of dealing with anything seen as dangerous, difficult, or painful instead of withdrawing from it."
- Imagination: "the act or power of forming mental images of that which is not actually present; it is the act of creating new images or ideas; it is resourcefulness in dealing with new or unusual experiences."
- Resilience: "the quality of bouncing back after being stretched or challenged … the quick recovery of strength, spirit, and good humor; resilience is buoyancy."
- Integrity: "the quality or state of being complete, unbroken, and entire; it is the quality of being whole; it is being of sound principle."
- Mindfulness: "the state of being thoughtful and aware."

Peloquin provides examples of how these five additional values, plus the current seven core values, are expressed in the earliest occupational therapy literature by the founders of the profession. Readers are encouraged to review her article to further understand how they collectively describe the ethos of occupational therapy.

Philosophical Assumptions

The **philosophical assumptions** of the profession guide occupational therapists in providing client-centered therapy that meets the needs of the client and society. These assumptions express our basic beliefs about the client and the context in which the client functions (Mosey, 1996). These assumptions are as follows:

- Each individual has a right to a meaningful existence: the right to live in surroundings that are safe, supportive, and comfortable, and over which he or she has some control; to make decisions for himself or herself; to be productive; to experience pleasure and joy; to love and be loved.
- Each individual is influenced by the biological and social nature of the species.
- Each individual can only be understood within the context of his or her family, friends, community, and membership in various cultural groups.
- Each individual has the need to participate in a variety of social roles and to have periodic relief from participation.
- Each individual has the right to seek his or her potential through personal choice, within the context of accepted social constraints.

- Each individual is able to reach his or her potential through purposeful interaction with the human and nonhuman environment.
- Occupational therapy is concerned with promoting functional interdependence through interactions directed toward facilitating participation in major social roles (occupational performance); and development of biological, cognitive, psychological, and social components (client factors) fundamental to such roles.
- The extent to which intervention is focused on the context, the areas of occupational performance, or the client factors depends on the needs of the particular individual at any given time.

Personalized Medicine

The core values and philosophical assumptions of the profession of OT lead occupational therapists to a focus on **personalized medicine**. According to Burke, Trinidad, and Press (2014), "personalized medicine is best understood as a comprehensive process to determine the best health care options for a particular patient, deriving from a partnership between patient and clinician. This approach offers the opportunity to weigh personal values and preferences as well as clinical findings" (p. 196). In addition, Topol (2014) defines personalized medicine as the tailoring of medical treatments to the individual characteristics of each patient with a focus on the individual as the source of medical data and as the driver of health care.

The core values, especially dignity, equality, and freedom, are the profession's moral guide to personalized medicine. They guide us to value differences, to treat people equally despite those differences, and to allow individuals to make their own choices based on differing perspectives and preferences.

The philosophical assumptions summarize the OT profession's basic beliefs about focusing on the rights and preferences of individuals relative to their biological and social environments. In addition, the philosophical assumptions help guide occupational therapists to form a partnership with each individual to determine the focus of the intervention. Each of these concepts forms a practice in which personalized medicine is an essential element.

Whereas the primary purpose of this book is to describe the potential impact of a condition on occupational performance, the descriptions should not be considered prescriptive or exhaustive. It is necessary to understand common facts of these conditions, including etiology, basic pathogenesis, commonly observed signs and symptoms, and precautions. However, it is equally important to recognize that the effects of a condition on occupational well-being will also be dependent on contextual factors such as age, developmental stage, health status, and the physical, social, and cultural environment (Dunn, Brown, & McGuigan, 1994). Rather than viewing an individual as a diagnostic entity, as a condition, or as the sum of biological cells, the treatment must be personalized.

Language

Although many language systems and mechanisms are available, we will discuss language from two perspectives. First, is a philosophical discussion of using **person-first language**. Second is the use of the ***Occupational Therapy Practice Framework****: Domain and Process, 4th Edition* (AOTA, 2020) that presents the professional language and the occupational therapy domain of concern.

Person-First Language

In many cases, the literature and the media, both popular and professional, describe a person with a given condition as the condition—the arthritic, the C.P. kid, the schizophrenic, the alcoholic, the burn victim, the mentally disabled. All of these terms label people as members of a large group rather than as a unique individual. The use of person-first language requires that the person be identified first and the disease used as a secondary descriptor. For example, a woman, who is a physicist, is active in her church and has arthritis; the fourth-grade boy, who is a good speller, loves baseball and has cerebral palsy. The condition does not and should not be the primary identity of any person.

Consider the following: a father is introducing his son to his coworkers. Which of the following is the best introduction?

"Hey, everyone, this is my disabled son, John."
"Hey, everyone, this is my son, John, who is disabled and loves soccer and video games."
"Hey, everyone, this is my son, John. He loves soccer and video games."

Of course, the third statement is the best choice. Yet it is common when describing a person who has a disability to emphasize the disability first. The consequence is a labeling process. According to Hansen (1998), "Although such shorthand language is commonplace in clinics and medical records, it negates the individuality of the person. Each of us is a person, with a variety of traits that can be used to describe aspects of our personality, behavior, and function. To use a disease or condition as the adjective preceding the identifying noun negates the multiple dimensions that make the person a unique individual."

The Occupational Therapy Practice Framework

The official language for the profession of occupational therapy was revised in 2020 and presented in a document entitled the *Occupational Therapy Practice Framework: Domain and Process, 4th Edition* (AOTA, 2020). The Practice Framework outlines the language and constructs that describe the occupational therapy profession's domain of concern. The domain defines the area of human activity to which the occupational therapy process is applied. The process facilitates engagement in occupation to support participation in life. The specific aspects of the domain are outlined in the language of the Practice Framework.

The Framework is organized into five aspects—occupations, contexts, performance patterns, performance skills, and client factors (Table 1.1). Occupations are various kinds of life activities in which individuals, groups, or populations engage. Occupations include activities of daily living, instrumental activities of daily living, health management, rest and sleep, education, work, and play, leisure, and social participation. Contexts are a variety of interrelated conditions that are within and surrounding the person. Performance patterns are the habits, routines, roles, and rituals used by the person in the process of engaging in occupations or activities. These patterns may enhance or hinder occupational performance. Performance skills are observable elements of action that have an implicit functional purpose. These skills are separated into the categories of motor skills, process skills, and social interaction skills. Client factors are values, beliefs and spirituality, the body functions, and the body structures that reside within the person. These client factors influence the person's participation in occupations.

TABLE 1.1 Occupational Therapy Practice Domain

Occupations	Contexts	Performance Patterns	Performance Skills	Client Factors
Activities of Daily Living (ADL) Instrumental Activities of Daily Living (IADL) Health Management Rest and Sleep Education Work Play and Leisure Social Participation	Environmental Factors Personal Factors	Habits Routines Roles Rituals	Motor Skills Process Skills Social Interaction Skills	Values, Beliefs and Spirituality Body Functions Body Structures

Each of these client factors has a relationship and influence on the others. The outcome is, of course, the ability to function and engage in occupations. Although at any given time the occupational therapist may focus on an

occupation or client factors, the ultimate concern is whether the individual is able to function in daily life. For example, a therapist may evaluate a person's attention span, but not in isolation. Attention span is evaluated within the realm of the performance patterns and context of the person—the attention span required to work on an assembly line, to drive a car, to learn a card game, or to conduct a business meeting.

Once a therapist knows the diagnosis and age of the person, he or she can use this Practice Framework to examine systematically the deficits that occur in the client factors, as well as how these particular deficits can and do alter the person's ability to complete functional activities relevant to occupations. In other instances, the therapist may focus primarily on the occupation or the context, without paying much attention to the underlying client factors that influence the occupational performance.

International Classification of Functioning, Disability and Health

A traditional medical model that focuses solely on the medical condition is not always sufficient for considering the true impact of the condition on a person's ability to function. **The International Classification of Functioning, Disability and Health** (ICF) was developed by the World Health Organization as a comprehensive classification of function and dysfunction related to physical and psychosocial conditions that is applicable across multiple contexts and cultures (World Health Organization, 2001). The ICF begins with the condition and then frames the condition in terms of the impact on body functions and structures, activities, and participation with the acknowledgement that this impact is influenced by environmental and personal factors (Fig. 1.1).

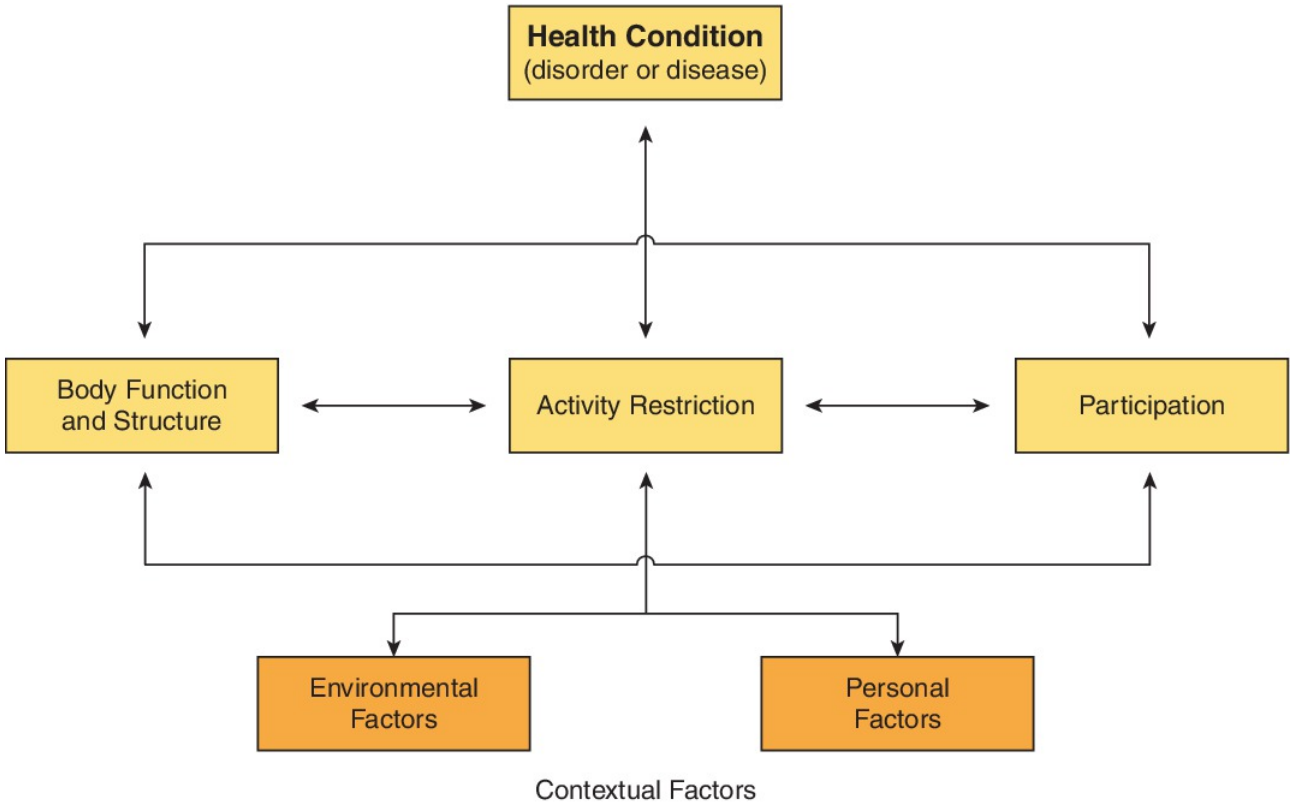

Figure 1.1 International Classification of Function, Disability and Health (ICF) model of health and disability. (Reprinted with permission from Ferkel, R. D. (2016). *Foot and ankle arthroscopy* (2nd ed.). Wolters Kluwer.)

Further, the ICF describes the interaction among these components that influence and modify one another. This model is especially useful for occupational therapists who are concerned not only with the health condition but also with the impact of the condition on occupational performance.

The ICF matches well with the framework of this textbook and is a useful tool for understanding the interaction of the components that are discussed in each chapter. As with the ICF, each chapter begins with a definition and description of the condition. The signs and symptoms outline the impact on body functions and structures and the

sections describing the impact on occupational performance elucidate the impact of the condition on activities and participation. Finally, the case studies illustrate how varied environmental and personal factors interact with the other components to influence a person's ability to function.

Framework of this Textbook

As an instructional tool, this book provides an opportunity to examine each condition closely. The reader is urged to use the information as a springboard for further study of the conditions included here and the many other conditions that occupational therapists encounter in practice. The analysis of the impact on occupational performance for a particular condition is dynamic, and the identification of the most important areas of dysfunction and, therefore, treatment will vary from therapist to therapist. In addition, factors such as secondary health problems, age, gender, family background, and culture contribute greatly to the development of a unique occupational performance profile for each individual served.

The occupational performance approach to the identification of dysfunction described in this book can be used to examine the effects of any condition on a person's daily life. This process will enable the therapist to identify and set a priority for problems in occupational performance, which, in turn, will serve as the foundation for creating an effective intervention plan.

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UNIT
1

Pediatric Conditions

The Pediatric Conditions Unit includes the most common conditions that children have who are treated by occupational therapists as determined by the National Board of Certification in Occupational Therapy. These chapters focus on conditions that are typically diagnosed in childhood, but many of them affect people throughout their life span. Each chapter provides information about the etiology, incidence and prevalence, signs and symptoms, diagnosis, course and prognosis, medical/surgical management, and impact on occupational performance of these conditions. Case illustrations are used to provide examples of lives affected by the condition. The conditions included in this unit are the following:

CHAPTER
2

Cerebral Palsy

Angie K. Boisselle

Key Terms

Ataxia
Athetosis (dyskinetic)
Contracture
Diplegia
Dysarthria
Dystonia
Extrapyramidal
Gastroesophageal reflux
Hemiplegia
Hydrocephalus
Hypertonicity (spasticity)
Hypotonicity
Nystagmus
Primitive reflexes
Quadriplegia
Scoliosis
Strabismus

A couple who had been trying to conceive a child for several years were thrilled when a family friend asked if they would be interested in adopting a baby girl that had just been born to a young unmarried woman in her church. The baby was born 6 weeks early and weighed only 4 lb, but she appeared to be healthy. After initiating the paperwork for a private adoption, they brought the baby home and named her Jill. By the time of Jill's 6 month well baby visit, her parents had become concerned. She appeared to be a bright baby who smiled and cooed and enjoyed reaching for and playing with toys, but her legs seemed stiff and she was not yet rolling over. They spoke with their family doctor about their concerns but he assured them that Jill was developing normally, and they had nothing to be concerned about. By the time of Jill's 9-month well baby visit, her parents' concerns were only growing. Jill was still not sitting up and had not yet learned to roll over or crawl. Her doctor decided to refer Jill to the county early intervention program for a developmental assessment. Jill was assessed by the early intervention team consisting of an occupational therapist, physical therapist, and speech and language pathologist. The occupational therapist noted some mildly increased tone and incoordination in her upper extremities and a 2- to 3-month delay in fine motor and self-help skills. The physical therapist noted that Jill had hypertonicity and retained primitive reflexes in her lower extremities, which was causing significant delay in the acquisition of gross motor skills. The speech and language therapist found Jill's cognitive, language, and social skills to be at age level. The team suggested to the parents that they have a pediatric neurologist assess Jill, as she was demonstrating some of the signs and symptoms of cerebral palsy. Both the occupational and physical therapist recommended that therapy services begin as soon as possible. An Individualized Family Service Plan (IFSP) was developed at a subsequent meeting, and Jill began receiving weekly physical and occupational therapy services.

Jill's parents took her to a pediatric neurologist who conduct a neurological assessment and cranial MRI. She was diagnosed with spastic diplegia, a type of cerebral palsy. Her parents were initially overwhelmed and devastated by the diagnosis. The next year was very difficult as they grieved the loss of so many dreams that they had for Jill and faced so much uncertainty about her future. They waded through an array of possible therapy approaches and medical and surgical interventions that were recommended trying to decide which would be right for Jill and their family. They struggled to find time to work on home exercises that had been prescribed for Jill. The strain became

so great that they even separated for a period but eventually reconciled. By the time Jill turned 3 years old, she was walking with a walker and able to sit in a chair independently, although she needed some assistance with changing positions. She was feeding herself but not yet dressing herself. They enrolled her in a preschool special education classroom where she received therapy services. By kindergarten, Jill was in a regular education classroom with a paraeducator for safety and support. Jill was a happy child, who had many friends and did well academically. Jill most likely will continue to need some type of additional support in order to be an independent adult, but all involved were optimistic about her future.

Description and Definition

Cerebral palsy is a complex, heterogeneous condition that primarily impacts motor function. It can occur at different times during development: congenital cerebral palsy occurs before or during birth, and acquired cerebral palsy occurs after birth. The international consensus definition of cerebral palsy has been prominent over the past decade.

Cerebral palsy is a brain-based, nonprogressive, permanent condition that has a prenatal or perinatal origin. The most prominent features include motor impairment with accompanying disorders of sensory function, cognition, speech, and in some cases, seizures (Rosenbaum, Paneth, Leviton, Goldstein, & Bax, 2007).

Overall, the injury or insult occurs when the brain is still developing. It can occur anytime during the prenatal, perinatal, or postnatal periods. There is some disagreement about the upper age limit for a diagnosis of cerebral palsy during the postnatal period. An upper age limit ranging from 2 to 8 years of age is applied to postneonatally acquired brain injury. Cerebral palsy is not progressive. Once the initial insult to the brain has occurred, there is no further worsening of the child's condition or further damage to the central nervous system. However, secondary conditions and activity limitation may contribute to further functional impairment throughout the life span (Bosanquet, Copeland, Ware, & Boyd, 2013).

Cerebral palsy always involves a disorder in sensorimotor development that is manifested by abnormal muscle tone and stereotypical patterns of movement. The severity of the impairment ranges from mild to severe. The sensorimotor disorder originates specifically in the brain. The muscles themselves and the nerves connecting them with the spinal cord are normal. Although some cardiac or orthopedic problems can result in similar postural and movement abnormalities, they are not classified as cerebral palsy. Some premature babies demonstrate temporary posture and movement abnormalities that look similar to patterns seen in cerebral palsy but resolve typically by 1 year of age. For children with cerebral palsy, these difficulties persist, and it is often a lifelong disability.

Classification of Cerebral Palsy

The manner in which cerebral palsy has been classified has changed over the years. Currently, it is commonly described by several different factors, including the areas of insult within the brain, motor typology related to anatomical location, and functional status. Motor typography is described in the following sections according to neurological locations (Fig. 2.1): pyramidal and extrapyramidal.

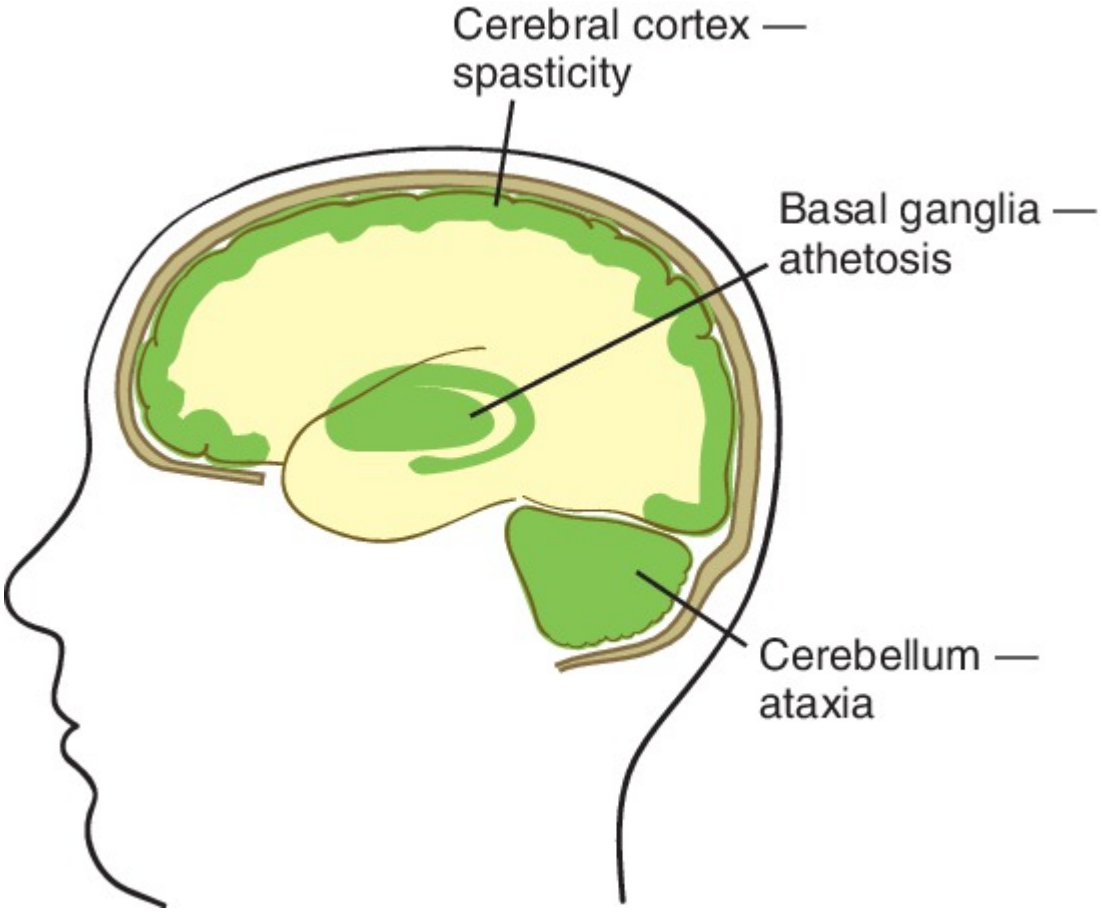


Figure 2.1 Cerebral palsy. Shown are the major parts of the brain involved in each of the three major types of cerebral palsy: spastic, athetoid, and ataxic. (Reprinted with permission from Wilkins, E. M., & Wyche, C. J. (2013). *Clinical practice of the dental hygienist* (11th ed.). Wolters Kluwer Health/Lippincott Williams & Wilkins.)

Pyramidal

Pyramidal cerebral palsy involves the occurrence of spasticity and arises within the pyramidal tracts of the brain, which includes the corticospinal tract that ends in the spinal cord and the corticobulbar tract that ends in the brain stem.

Spasticity is a type of **hypertonicity** that is velocity dependent, meaning the muscles are more resistive to sudden, passive movement. Deep tendon reflexes are present in affected limbs and motor control is affected by the hypertonicity. This type is the most common and accounts for ~80% of cases of cerebral palsy (CDC, 2020). The impact on motor function can range from a mild impairment that does not interfere with functional skills, such as not having isolated finger movement, to a severe impairment, where there is an inability to reach and grasp. **Contractures**, which result in permanent shortening of a muscle or joint and deformities, are common (Fig. 2.2). Spastic cerebral palsy is categorized anatomically according to the area of the body that is affected. Often each area is distinguished by either weakness (–paresis) or paralysis (–plegia). Spastic hemiplegia, spastic diplegia, and spastic quadriplegia are the most common types and described below. More nontraditional types include triplegia, which involves both lower extremities and one upper extremity, and double hemiplegia, which asymmetrically involves all limbs (Glader & Stevenson, 2019).

Figure 2.2 Due to spasticity and limited controlled movement and range of motion in upper extremities and hands, this person is using a mouth stick to activate a computer. Note wrist flexion contracture. (Reprinted with permission from Carter, P. J. (2011). *Lippincott's textbook for nursing assistants: A humanistic approach to caregiving*. (3rd ed.). Wolters Kluwer Health/Lippincott Williams & Wilkins.)

Spastic Hemiplegia

Spastic **hemiplegia** involves one entire side of the body, including the head, neck, and trunk. Usually, the upper extremity is most affected. Early signs include asymmetrical hand use during the first year or dragging one side of the body when crawling or walking. The child learns to walk later than is typical and when walking the child typically hyperextends the knee, and the ankle is in equinovarus or equinovalgus position on the involved side. The child often lacks righting and equilibrium reactions on the involved side and will avoid bearing weight on this side. The shoulder is held in adduction (internal rotation); the elbow is flexed; the forearm is pronated; the wrist is flexed and ulnar deviated; thumb is adducted; and the fingers are flexed (Fig. 2.3). Spasticity increases during physical activities and emotional excitement. Arm and hand use is limited on the involved side, depending on the severity. The child may use more primitive patterns of grasping and lacks precise and coordinated movement. In more severe cases, the child may totally neglect the involved side or use it only as an assist during bilateral activities.

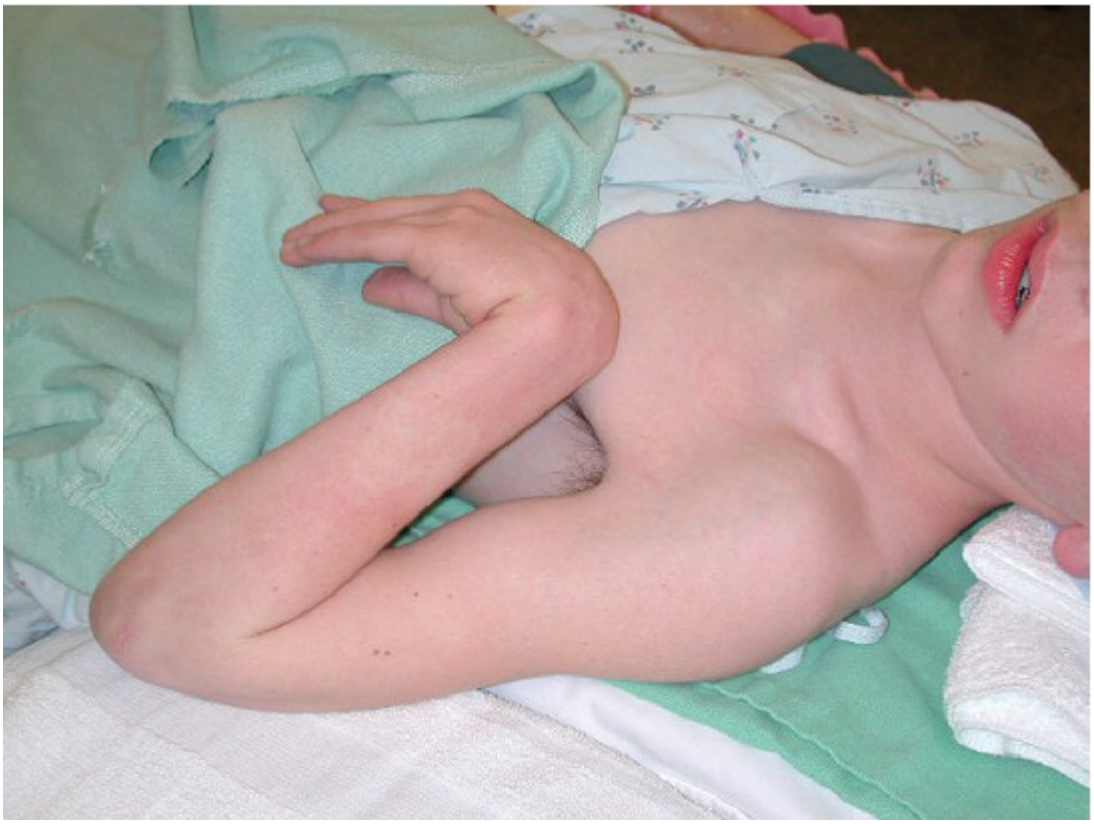


Figure 2.3 Abnormal upper extremity position due to spasticity. Note thumb adduction and flexion in the wrist and elbow. (Courtesy of Ghazi Rayan, MD.)

Spastic Diplegia

Spastic **diplegia** involves both lower extremities, with mild incoordination, tremors, or less severe spasticity in the upper extremities. It is most often attributed to premature birth and low birth weight and is, therefore, on the rise as more infants born prematurely survive as a result of medical advances. The ability to sit independently can be delayed up to 3 years of age or older because of inadequate hip flexion and extensor and adductor hypertonicity in the legs (Bobath, 1980). Frequently the child will rely on the arms for support. The young child will move forward on the floor by pulling along with flexed arms while the legs are stiffly extended. Getting up to a creeping position is difficult because of spasticity in the lower extremities. Similarly, standing posture and gait are affected to varying degrees, depending on severity. Because of a lack of lower extremity equilibrium reactions, excessive trunk and upper extremity compensatory movements are used when walking. Lumbar lordosis, hip flexion and internal rotation (scissoring), plantar flexion of the ankles, and difficulty shifting weight when walking are common (Fig. 2.4). Many of these problems result in contractures and deformities, including dorsal spine kyphosis, lumbar spine lordosis, hip subluxation or dislocation, flexor deformities of hips and knees, and equinovarus or equinovalgus deformity of the feet (Bobath, 1980).

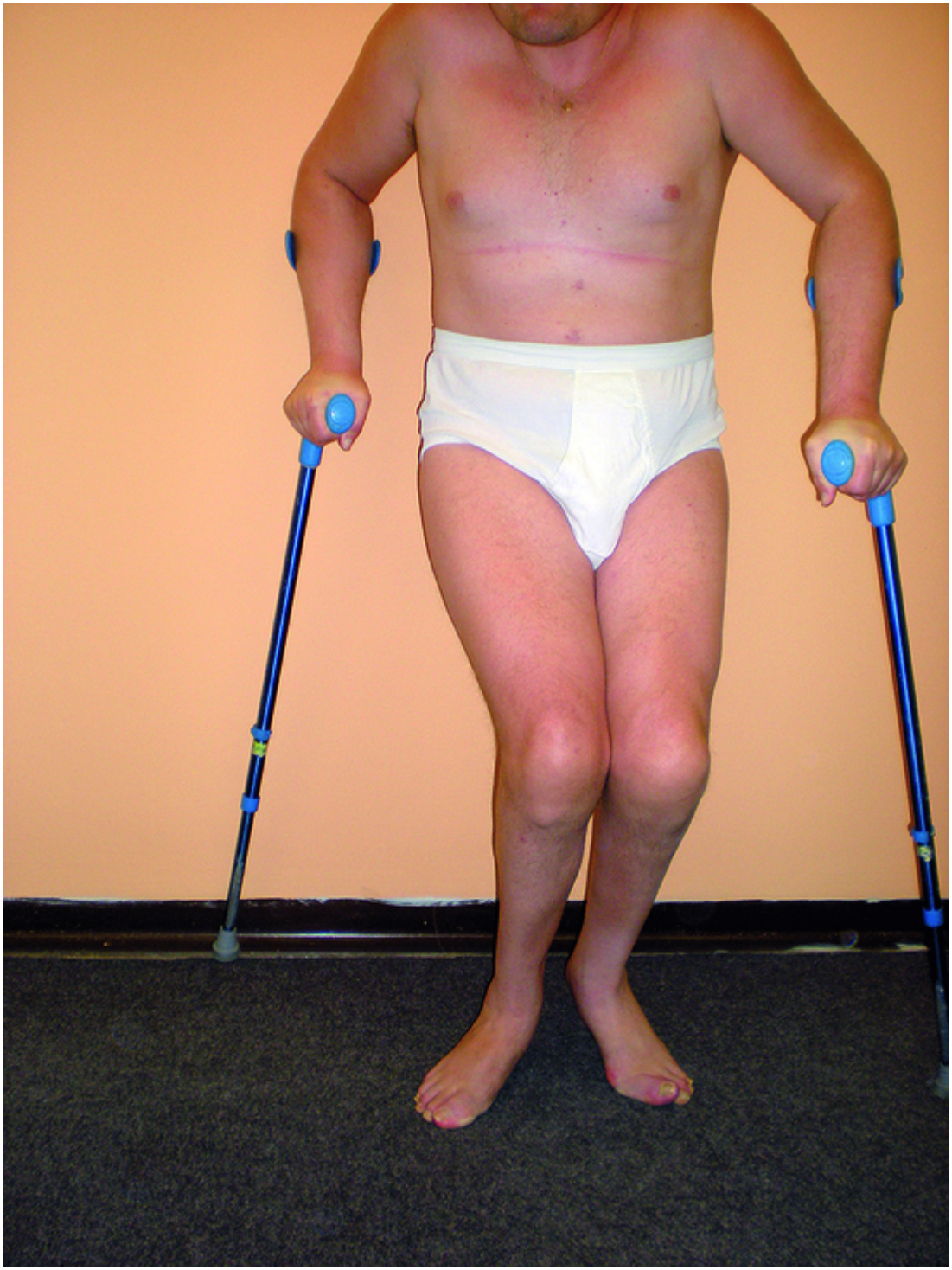


Figure 2.4 Postural effects from cerebral palsy (spastic diplegia). Note crouched posture due to abnormal muscle tone and strength: hip flexion and internal rotation; knee flexion; and equinovalgus positioning of feet. (Reprinted with permission from Liebenson, C. (2014). *Functional training handbook*. Wolters Kluwer.)

Spastic Quadriplegia

Spastic **quadriplegia** impacts all limbs symmetrically. Children with spastic quadriplegia are more likely to experience higher associated conditions and have cognitive impairment, and are less likely to ambulate. The arms typically demonstrate spasticity in the flexor muscles, with spasticity in the extensor muscles in the lower extremities. Because of the influence of the tonic labyrinthine reflex (TLR), shoulder retraction and neck hyperextension are common, particularly in the supine position. This results in difficulty with transitional movements such as rolling or coming up to sitting. In the prone position, there is increased flexor tone, also a result of TLR influence, causing difficulty with head raising and bearing weight on the arms. Independent sitting and standing are

difficult for the child because of hypertonicity, the presence of primitive reflex involvement, and a lack of righting and equilibrium reactions. Individuals are susceptible to contractures and deformities, particularly hip dislocation and scoliosis, and must be closely monitored (Fig. 2.5).

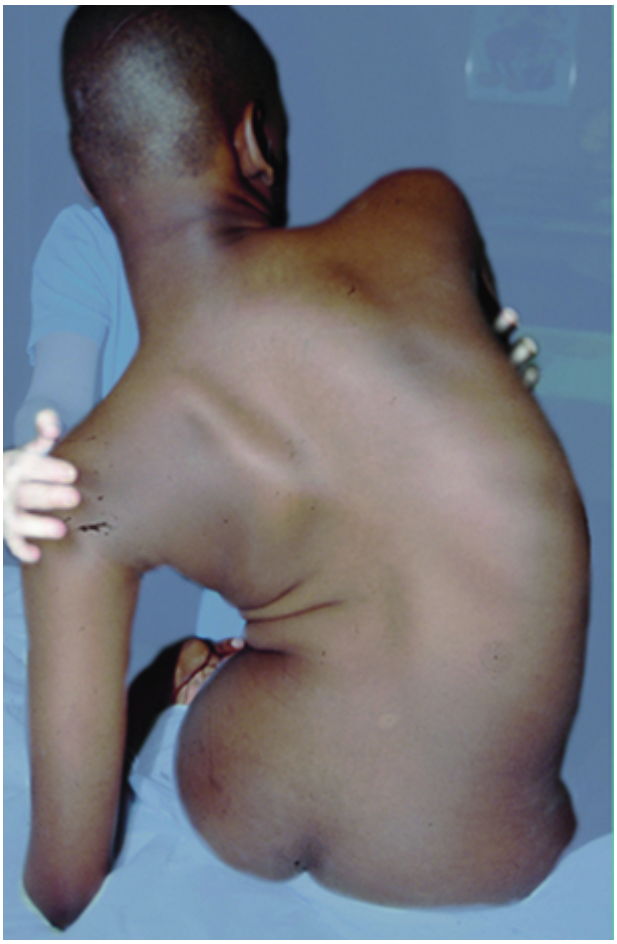


Figure 2.5 Severe scoliosis with pelvic obliquity in person with spastic quadriplegia. Scoliosis with this severity can compromise respiratory function. (Reprinted with permission from Flynn, J. M., & Wiesel, S. W. (2010). *Operative techniques in pediatric orthopaedics*. Wolters Kluwer Health/Lippincott Williams & Wilkins.)

Extrapyramidal

The **extrapyramidal** system is that aspect of the central nervous system that allows for modulation of muscle tone, posture, and movement. Extrapyramidal cerebral palsy occurs outside of the pyramidal tracts of the brain and involves a fundamental movement disorder including dyskinetic and ataxia.

Dyskinetic

Dyskinetic type is characterized by involuntary and uncontrolled movements. These movements are typically slow and writhing. It is noted to be the most common type of **dystonia**, with an incidence of 4%-17% (Fehlings et al., 2018). **Athetosis** is the most common type of dyskinesia, characterized by slow, writhing, involuntary movements of the face and extremities or the proximal parts of the limbs and trunk. Abrupt, jerky, distal movements (choreiform) may also appear. The movements increase with emotional tension and are not present during sleep. Head and trunk control is often affected, as is the oral musculature, resulting in drooling, dysarthria, and eating difficulties.

Ataxia

Ataxia is characterized by unsteadiness and difficulties with balance, particularly when ambulating. It is the least common type of cerebral palsy and results from involvement of the cerebellum or its pathways. It is common for there to be mixed forms where two of the types occur together as a result of diffuse brain damage. The most

common is spastic with athetoid. Persons with this type have signs of athetosis, and postural tone that fluctuates from hypertonicity to hypotonia. Athetoid combined with ataxia is less common. Ataxia is characterized by a wide-based, staggering, and unsteady gait. Children with ataxia often walk quickly to compensate for their lack of stability and control. Controlled movements are clumsy. Intention tremors may be present. The ability to perform refined movements such as handwriting is affected. **Hypotonicity** is often present (Glader & Stevenson, 2019).

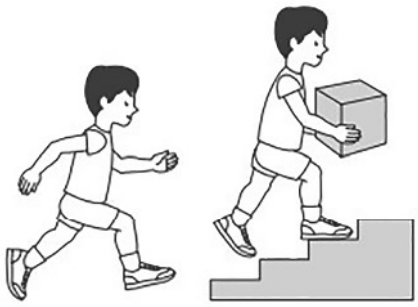
Mixed Types

It is common for there to be mixed forms, in which two of the types occur together as a result of diffuse brain damage. Dystonia with spasticity is the most common coexisting type. Dystonia is thought to originate in the basal ganglia. It is suspected that the impact of dystonia is significantly underdiagnosed because it is fluctuating and may be difficult to identify (Fehlings et al., 2018). The American Academy of Cerebral Palsy and Developmental Medicine (AACPDM) provides a care pathway titled Dystonia in Cerebral Palsy, which offers evidence and guidelines for clinicians (American Academy of Cerebral Palsy and Developmental Medicine, 2017).

Functional Classification

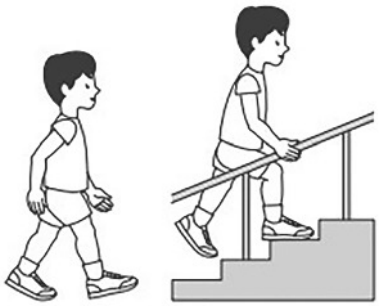
Clinicians who specialize in care of those with cerebral palsy commonly use classification systems to understand current level of motor function and limitation of children and youth with cerebral palsy. The Gross Motor Functional Classification System (GMFCS) classifies gross motor abilities and functional limitations for infants to 12 years of age with cerebral palsy. It consists of five levels ranging from level I, which includes walking without limitation, to level V, which requires transportation in a wheelchair. Each level is related to current performance with ambulation and movement within a variety of physical environments such as home, school, and community (Palisano et al., 1997). The extended, revised version (GMFCS-E & R; Fig. 2.6) was later developed to expand focus through 18 years of age (Palisano, Rosenbaum, Bartlett, & Livingston, 2008). Similarly, the Manual Abilities Classification System (MACS) categorizes how children from ages 4 to 18 manipulate objects during activities of daily living (ADLs). Level I signifies very few limitations in hand-related activities. Conversely, level V signifies very restricted use of hands for activity (Eliasson et al., 2006). Classification systems such as the GMFCS and MACS are widely used in cerebral palsy research and practice to classify typical performance of motor abilities across home, school, and community.

GMFCS E & R between 6th and 12th birthday: Descriptors and illustrations



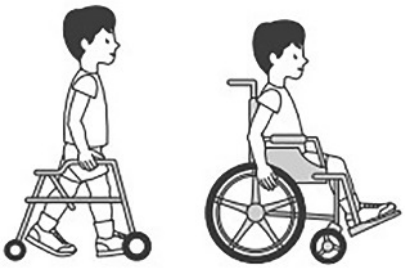
GMFCS Level I

Children walk at home, school, outdoors and in the community. They can climb stairs without the use of a railing. Children perform gross motor skills such as running and jumping, but speed, balance and coordination are limited.



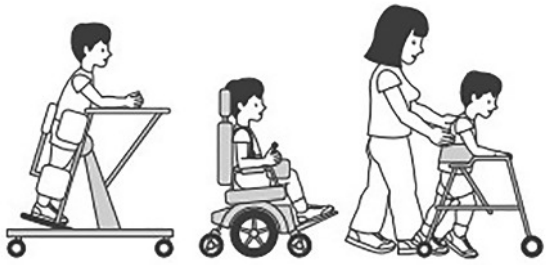
GMFCS Level II

Children walk in most setting and climb stairs holding onto a railing. They may experience difficulty walking long distances and balancing on uneven terrain, inclines, in crowded areas or confined spaces. Children may walk with physical assistance, a hand-held mobility device or used wheeled mobility over long distances. Children have only minimal ability to perform gross motor skills such as running and jumping.



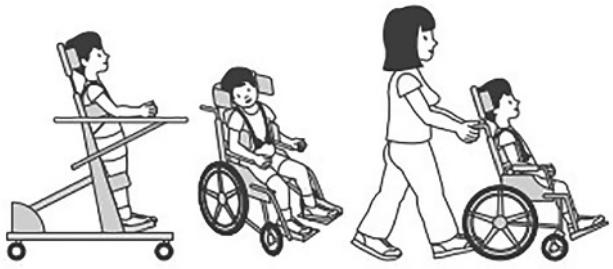
GMFCS Level III

Children walk using a hand-held mobility device in most indoor settings. They may climb stairs holding onto a railing with supervision or assistance. Children use wheeled mobility when traveling long distances and may self-propel for shorter distances.



GMFCS Level IV

Children use methods of mobility that require physical assistance or powered mobility in most settings. They may walk for short distances at home with physical assistance or use powered mobility or a body support walker when positioned. At school, outdoors and in the community children are transported in a manual wheelchair or use powered mobility.



GMFCS Level V

Children are transported in a manual wheelchair in all settings. Children are limited in their ability to maintain antigravity head and trunk postures and control leg and arm movements.

GMCS descriptors: Palisano et al. (1997) Dev Med Child Neurol 39:214-23
CanChild: www.canchild.ca

Figure 2.6 Gross Motor Function Classification System, Expanded and Revised.

(From Palisano, R., Rosenbaum, P., Bartlett, D., Galuppi, B. E., & Russell, D. J. *Gross Motor Function Classification System—Expanded & Revised*. CanChild Centre for Childhood Disability Research. Retrieved from https://www.canchild.ca/en/resources/42-gross-motor-function-classification-system-expanded-revised-gmfcs-e-r, on September 20, 2018. Illustrations Version 2 © Bill Reid, Kate Willoughby, Adrienne Harvey and Kerr Graham, The Royal Children's Hospital Melbourne ERC151050.)

Etiology

Cerebral palsy is not considered an etiological disease but is classified as a phenomenological diagnosis since understanding the signs and symptoms is key to the diagnosis and treatment (Glader & Stevenson, 2019). It is characterized as a developmental disability. Historically, birth asphyxia was considered the major cause of cerebral palsy. Dr. John William Little was known to be the first to identify cerebral palsy in 1860. He suggested that a major cause was a lack of oxygen during the birth process (Little, 1862). In 1897, Sigmund Freud disagreed, suggesting that the disorder might have roots earlier in life. Freud wrote, "Difficult birth, in certain cases, is merely a symptom of deeper effects that influence the development of the fetus" (Freud, 1968). In his monograph entitled "Infantile Cerebral Paralysis," Freud points out that a well-known painting by Spanish painter Jusepe Ribera (1588–1656), which depicts a child with infantile hemiplegia, proves that cerebral paralysis existed long before medical investigators began paying attention to it in the mid-1800s (Freud, 1968). Freud's work as a neurologist is not generally well known and, at the time that his monograph was published in 1897, he was already deep into his work in the area of psychotherapy. However, he was recognized at the time as the prominent authority on the paralyses of children. Today, cerebral paralysis is known as cerebral palsy.

Although Freud made these observations in the late 1800s, it was not until the 1980s that research supported his views (Freeman & Nelson, 1988; Illingsworth, 1985). Only a small percentage of cases of cerebral palsy are a result of birth complications. The Centers for Disease Control and Prevention (CDC, 2020) reports that 85%-90% of cases of those diagnosed with cerebral palsy have congenital cerebral palsy—that is, the injury to the brain occurred in utero. Genome studies indicate that genetic factors contribute to anywhere from 14% to 49% of CP cases. The study of genetic links within CP remains in infancy. It is anticipated that further exploration in this area will result in a more significant genetic link and expand upon the phenotypic definition of the diagnosis of cerebral palsy (Novak et al., 2017; Pham et al., 2020; Stavsky et al., 2017).

There is a large number of risk factors that can result in cerebral palsy, and the interplay between these factors is often complex, making it difficult to identify the specific cause. The presence of risk factors does not always result in a subsequent diagnosis of cerebral palsy. The presence of one risk factor may not result in cerebral palsy unless it is present to an overwhelming degree. Current thought is that often two or more risk factors may interact in such a way as to overwhelm natural defenses, resulting in damage to the developing brain. The strongest risk factors are prematurity of <28 weeks and low birth weight (Oskoui, Coutinho, Dykeman, Jetté, & Pringsheim, 2013). This is largely because premature and low-birth-weight infants are at greater risk for developing complications, especially in the circulatory and pulmonary systems. These complications can lead to brain hypoxia and result in cerebral palsy.

Additional risk factors include intrauterine exposure to infection and disorders of coagulation (National Institute of Neurological Disorders and Stroke, 2020). Maternal infection during the perinatal period is a critical risk factor for cerebral palsy and includes infections such as maternal urinary tract infections (UTI), toxoplasmosis, cytomegalovirus (CMV), and herpes simplex virus (HSV) (Glader & Stevenson, 2019). The infection does not necessarily produce signs of illness in the mother, which can make it difficult to detect. Table 2.1 lists specific risk factors related to both congenital and acquired types of cerebral palsy.

TABLE 2.1 Cerebral Palsy: Contributing Risk Factors and Causes

Parental Influence and Developmental Periods and Events	Risk Factor/Cause
Preconception (parental background)	Biological aging (parent or parents older than 35)
	Multiple gestations
	Biological immaturity (very young parent or parents)
	Environmental toxins
	Genetic background and genetic disorders
	Malnutrition
	Metabolic disorders
	Radiation damage
First trimester of pregnancy	Endocrine: thyroid function, progesterone insufficiency
	Nutrition: malnutrition, vitamin deficiencies, amino acid tolerance
	Toxins: alcohol, drugs, poisons, smoking
	Maternal disease: thyrotoxicosis, genetic disorders
Second trimester of pregnancy	Infection: cytomegalovirus, rubella, toxoplasmosis, HIV, syphilis, chicken pox, subclinical uterine infections
	Placental pathology: vascular occlusion, fetal malnutrition, chronic hypoxia, growth factor deficiencies
Third trimester of pregnancy	Prematurity and low birth weight
	Blood factors: Rh incompatibility, jaundice
	Cytokines: neurological tissue destruction
	Inflammation
	Hypoxia: placental insufficiency, perinatal hypoxia
	Infection: listeria, meningitis, streptococcus group B, septicemia, chorioamnionitis
Intrapartum events	Uterine rupture
	Acute maternal hypotension
	Prolapsed umbilical cord
	Ruptured vasa previa
	Tightened true knot of the umbilical cord

Parental Influence and Developmental Periods and Events	Risk Factor/Cause
Perinatal period and infancy	Endocrine: hypoglycemia, hypothyroidism
	Hypoxia: perinatal hypoxia, respiratory distress syndrome
	Infection: meningitis, encephalitis
	Multiple births: death of a twin or triplet
	Stroke: hemorrhagic or embolic stroke
	Trauma: abuse, accidents

Data from UCP Research and Educational Foundation. Factsheet: Cerebral Palsy: Contributing Factors and Causes. September, 1995; Bosanquet, M., Copeland, L., Ware, R., & Boyd, R. (2013). A systematic review of tests to predict cerebral palsy in young children. *Developmental Medicine and Child Neurology, 55*(5), 418–426. doi:10.1111/dmcn.12140; Glader, L. J., Stevenson, R. D., eds. (2019). *Children and youth with complex cerebral palsy: care and management*. Mac Keith Press; and Stavsky, M., Mor, O., Mastrolia, S. A., Greenbaum, S., Than, N. G., & Erez, O. (2017). Cerebral palsy-trends in epidemiology and recent development in prenatal mechanisms of disease, treatment, and prevention. *Frontiers in Pediatrics, 5*(21), 1–10. doi:10.3389/fped.2017.00021.

There are four types of injuries to the brain that often result in cerebral palsy (National Institute of Neurological Disorders and Stroke, 2020):

- Periventricular leukomalacia (PVL) involves damage to the white matter in the brain adjacent to the lateral ventricles due to ischemia, or restriction in blood supply to the brain tissue. Necrosis occurs, resulting in empty areas or cysts that fill up with fluid. This is most often associated with premature birth before 32 weeks' gestation and decreased birth weight. Shang et al. (2015) conducted a study of 408 children diagnosed with CP and PVL and found that 95% of cases that involved high risk factors such as infection and hypoxia resulted in quadriplegia (CP affecting all four limbs), and the most associated disorders included visual and hearing impairments.
- Hypoxic–ischemic encephalopathy (HIE), commonly known as perinatal asphyxia, occurs when there is a loss of oxygen resulting in damage to brain tissue. It most often occurs in the full-term infant in the perinatal period and can be caused by many factors related to birth and delivery as well as fetal stroke.
- Intraventricular hemorrhage (IVH) involves bleeding into the brain's ventricular system. This most often occurs in infants born more than 10 weeks prematurely due to the blood vessels in the brain not being fully developed at this gestational period. It is rarely present at birth but develops during the first several days of life. There are four types or grades of IVH based on the amount of bleeding that occurs. Grades 1 and 2 involve a smaller amount of bleeding and typically do not result in any long-term developmental problems. Grades 3 and 4 involve more severe bleeding. **Hydrocephalus**, defined as abnormal fluid build-up in the brain, can occur when blood presses on or leaks into the brain tissue resulting in blood clots blocking the flow of cerebral spinal fluid. Grades 3 and 4 bleeds are most often associated with cerebral palsy.

Congenital malformations can occur when the brain does not grow properly or fully develop. They differ from the other three causes of cerebral palsy that are due to lesions or brain injuries. With cerebral dysgenesis, the brain did not grow properly or fully develop. The first 21 weeks of gestation are most critical for brain development, and factors such as maternal infections or trauma can result in a brain malformation. The most common type of brain malformation associated with CP is microencephaly, in which the brain does not grow (Stavsky et al., 2017). The severity of brain malformations can vary greatly with a more severe type resulting in cerebral palsy.

Acquired CP occurs more than 28 days after birth and represents a small percentage of cases. This category includes infection, cerebrovascular accidents (CVA), and injury from external causes such as motor vehicle accidents, and child abuse (CDC, 2020). A closed-head injury that occurs during this period is now classified as traumatic brain injury, even though the resulting impairments are very similar to cerebral palsy.

Incidence and Prevalence

Cerebral palsy is the most common cause of motor limitations and childhood disability globally (McGuire, Tian, Yeargin-Allsopp, Dowling, & Christensen, 2019). It is estimated that the global prevalence is 1 in 500 live births with 17 million from birth to 90 years of age (Gross et al., 2020; National Institute of Neurological Disorders and Stroke, 2020; Novak et al., 2017). Cerebral palsy is more common among males than among females at a ratio of 1:4. Romeo et al. (2016) conducted a critical review on sex differences of the various motor impairment of cerebral palsy and found more male representation of impairment across all types and severity. Cerebral palsy has increased in prevalence in middle- and low-income areas as compared with upper class areas (Novak et al., 2017). The prevalence of CP in non-Hispanic black children is higher than non-Hispanic white and Hispanic children; however, the direct cause is not known. It is suspected that racial disparities may result in lower birth rates and prematurity (Stavsky et al., 2017; Van Naarden Braun et al., 2016).

There has been considerable advancement in obstetric and neonatal care during the past three to four decades. Many hoped these advancements would reduce the incidence of cerebral palsy. Unfortunately, the rate has remained relatively stable over this period. This is probably a result of increased survival rates of very low birth weight and premature infants (Novak et al., 2017). CP is 15 times more prevalent in twins, and even more so in triplets, particularly with the surviving infant following an in utero death of the twin (Stavsky et al., 2017). Another factor may be the use of fertility treatments by older women that have resulted in an increase in the number of multiple births. Multiple births tend to result in infants who are smaller and premature and are at greater risk for health problems. On the average, they are half the weight of singleton infants at birth and arrive 7 weeks earlier.

Signs and Symptoms

The early signs and symptoms common to all types of cerebral palsy are abnormal muscle tone, reflex and postural abnormalities, delayed motor development, and atypical motor performance.

Tone Abnormalities

Tone abnormalities include hypertonicity, hypotonicity, and dystonia, which involves abnormal posturing and repetitive, contorted movements. Muscle tone can be characterized as the degree of resistance when a muscle is stretched. For instance, when there is hypotonicity and the elbow is passively extended, there will be little to no resistance to the movement and hypermobility in the elbow joint. With hypertonicity, there will be increased resistance, and it may be difficult to pull the elbow into full extension if the tone is strong. Dystonia presents as involuntary or irregular muscle contractions triggered by several factors, including physiological changes, startle, emotional state, sleepiness, and difficulty with concentration on cognitive tasks (Fehlings et al., 2018). Most infants with cerebral palsy initially demonstrate hypotonia. Later the infant may develop hypertonicity or dystonia, or continue to demonstrate hypotonia, depending on the type of cerebral palsy.

Reflex Abnormalities

With hypertonicity, reflex abnormalities such as hyperreflexia, clonus, overflow, enhanced stretch reflex, and other signs of upper motor neuron lesions are present. Retained primitive infantile reflexes and a delay in the acquisition of righting and equilibrium reactions occur in conjunction with all types of abnormal tone. When hypotonia is present, there may be areflexia, or an absence of primitive reflexes. **Primitive reflexes**, or infantile reflexes, are those involuntary postural responses to static position and movement that should be present during the first several months of life, and it is of concern when they are not. After 4-6 months, primitive reflexes are diminished as righting and equilibrium reactions emerge.

Atypical Posture

The presence of primitive reflexes and muscle tone abnormalities causes the child to have atypical positions at rest and to demonstrate stereotypical and uncontrollable postural changes during movement. For instance, in an infant lying supine with hypertonicity in the lower extremities, the hips are typically internally rotated and adducted and the ankles plantar flexed. This posture is caused by a combination of hypertonicity in the affected muscles and the presence of the crossed extension reflex (Fig. 2.7). In contrast, when an infant with hypotonicity in the lower

extremities is lying supine, the hips are typically abducted, flexed, and externally rotated because of low muscle tone, weakness in the affected muscles, and the influence of gravity.

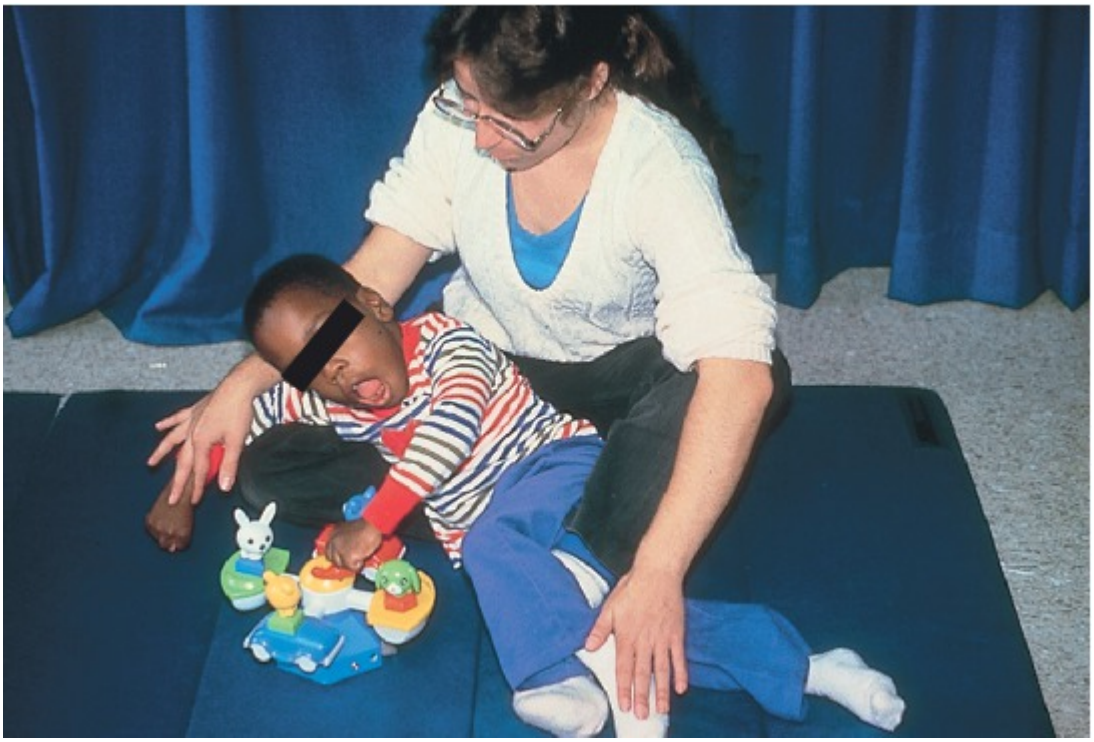


Figure 2.7 Child with spasticity in upper and lower extremities. Note hip adduction and scissoring in his legs, internal rotation at shoulders, fisted hand position, and overflow movements in his mouth. (Reprinted with permission from Hatfield, N. T. (2013). *Introductory maternity and pediatric nursing* (3rd ed.). Wolters Kluwer Health/Lippincott Williams & Wilkins.)

Delayed Motor Development

Cerebral palsy is always accompanied by a delay in the attainment of motor milestones. One of the signs that often alerts the pediatrician to the problem is a delay in the child's ability to sit independently or to crawl. While cerebral palsy is often present at birth, it is often not recognized until the child fails to achieve these early motor milestones.

Atypical Motor Performance

The way in which a child moves when performing skilled motor acts is also affected. Depending on the type of cerebral palsy, the child may demonstrate a variety of motor abnormalities, such as asymmetrical hand use; unusual crawling method or gait; uncoordinated reach; difficulty with speech; or difficulty sucking, chewing, and swallowing.

Associated Disorders

In addition to the motor impairments, there are a number of disorders and difficulties associated with cerebral palsy that can significantly affect functional abilities. In some cases, associated disorders can have a more significant impact on function than the motoric aspects of cerebral palsy. The list of possible associated conditions related to cerebral palsy is vast. The most common ones are highlighted here.

Cognitive Impairment

Of all the associated disorders with cerebral palsy, cognitive impairment has the most significant impact upon functional outcomes impacting 50% of the population of children with cerebral palsy. In about one-third of these instances, the cognitive impairment is mild. The most significant impairments most often occur with mixed types and severe spastic quadriplegia (GMFCS IV-V). Children with dyskinetic types of cerebral palsy have the least occurrence of cognitive impairment as compared to motor impairment (Frazier, 2019). It is important to understand that motor

limitations and dysarthria may mask cognitive abilities. The onus is on the clinician to provide ample time for a response and work collaboratively to determine the need for and methods to access communication devices.

Orthopedic Conditions

Individuals with cerebral palsy often encounter a wide array of orthopedic issues that are exacerbated by compromised muscular integrity related to spasticity, contractures, hypotonia, and dystonia-related torsion.

Hip displacement and dislocation are another primary concern for children with CP; they often result in pain and reduced quality of life. Hip surveillance, which involves regular, ongoing clinical and radiological examinations, is found to be instrumental for monitoring hip abnormalities. Surveillance should begin at 2 years of age or as soon as cerebral palsy is diagnosed. It should occur more frequently for children with higher GMFCS levels as the incidence of hip dislocation of GMFCS is as high as 90% (Romness & Anciano, 2019; Wynter et al., 2015).

Scoliosis, which is defined as a lateral curvature of the spine, is another condition that may occur across several types of cerebral palsy. Severity is found to correlate with the GMFCS levels and often worsens over time (Romness & Anciano, 2019). It contributes to secondary respiratory and gastrointestinal concerns and can significantly impact quality of life. While surveillance is not yet formalized for scoliosis, ongoing assessment can be beneficial. Low bone density or osteoporosis is often evident making fractures a common concern in children with CP. It is estimated to occur in 20% of nonambulators with CP (Fehlings et al., 2018).

Seizure Disorder

Reports of the incidence of seizures in people with cerebral palsy is noted to be ~41%. The percentage rises to ~67% in those with higher GMFCS levels (Day, Wu, Strauss, Shavelle & Reynolds, 2007). The incidence varies across the diagnostic categories. It is most common in spastic quadriplegia and nonambulatory types. Seventy percent of children with CP and epilepsy experience focal (localized) seizures (Mytinger & Goodkin, 2019).

Visual Impairments

Visual and hearing impairments occur at a higher rate with cerebral palsy than in the general population. Those with CP can have a range of visual issues including those with acuity, strabismus, and visual perception. Cortical vision impairment (CVI) is a complex condition that originates in the visual pathways of the brain associated with lesions common in CP such as PVL. It can manifest as deficits of visual field, acuity, nystagmus, strabismus, visual–cognitive function, and contrast issues. **Nystagmus** refers to rapid, involuntary movement of the eyes, and **strabismus**, or "crossed eyes," is a condition in which the eyes do not simultaneously look at an object in exactly the same direction. Due to the mixed presentation, neuro-ophthalmological concerns should be closely followed. It is suspected that CVI occurs in 70% of individuals with cerebral palsy (Fazzi et al., 2012). Visual perceptual impairment is found to be as high as 40%-50% across all types of cerebral palsy and is not specific to one type of cerebral palsy, seizures, or intellectual disability, but rather is more correlated to lower gestational age (Ego et al., 2015). Acuity and oculomotor abnormalities are common in 98%-100% of children with quadriplegia (Fazzi et al., 2012).

Oral Motor and Communication Disorders

Difficulties with oral motor function are commonly present in children with cerebral palsy. They may manifest due to the mechanical difficulties with the act of chewing, swallowing, or speaking. Disturbance in sensation may result in textural aversion to food or inability to manage food within the mouth during the oral phase of eating and swallowing. Dysphagia (difficulty swallowing) may occur due to sensory impairment or as a result of postural and mechanical demands of coordinating all aspects of swallowing, which include the oral, pharyngeal, and esophageal phases (Bickley, Delaney, & Intagliata, 2019). Severe **dysarthria** (difficulty speaking) may affect functional communication, resulting in the need for alternative forms of communication. The Eating and Drinking Ability Classification System (EDACS) (Benfer et al., 2017) and the Communication Function Classification System (CFCS) (Hidecker et al., 2011) offer levels similar to the GMFCS and provide clinicians with a method to classify current functional abilities and limitations for each.

Gastrointestinal

Gastrointestinal difficulties occur frequently in cerebral palsy. **Gastroesophageal reflux** (GERD), which refers to an abnormal, opposite flow of fluid in the esophagus, can create much discomfort and can result in refusal to eat or difficulty transitioning to solid foods. It can also be attributed to additional issues such as anemia, respiratory infections, and weight loss. Pharmacological interventions are often tried first, followed by surgical interventions

such as fundoplication which is the surgical reinforcement of the esophageal sphincter (Beinvogl & Mobassaleh, 2019).

Chronic constipation is also evident in those with cerebral palsy due to spasticity of the rectal sphincter. It can result in pain and discomfort and often impacts appetite. It is important for a multidisciplinary team of clinicians along with the patient and caregiver to monitor signs and symptoms of chronic constipation. Interventions such as dietary changes, prophylactics, and enemas are useful to aid in relief (Beinvogl & Mobassaleh, 2019).

Pulmonary

Individuals with complex motor impairments such as spastic quadriplegia often develop respiratory issues that interfere with respiration. The respiratory muscles may be directly restricted or musculoskeletal changes may develop over time. These individuals are prone to frequent upper respiratory infections, reactive airway disease, and sleep disturbances such as sleep apnea, which can significantly impact their health. Progressive changes in the spine, muscle contractures, and changes in the walls of the lungs can contribute to pulmonary disorders (Welsh & Katwa, 2019). Aspiration pneumonia can develop as a secondary issue of gastrointestinal issues noted above due to frequent reflux, poor swallow function, and muscle weakness needed to clear liquids and food with a productive cough. Ongoing, careful assessment and management of pulmonary function are advisable to monitor risks with respiration and provide pharmaceutical and durable medical equipment as needed.

Diagnosis

The early diagnosis of cerebral palsy was once thought to be difficult. Diagnosis typically occurs between 12 and 24 months; however, research now indicates that accurate diagnosis can occur prior to 6 months adjusted age. Developmental screening and surveillance are crucial to identify concerns in motor abilities early in life. For example, the inability to sit by 9 months, bear weight in the lower extremities, and/or use hands symmetrically should be cause for concern (Maitre et al., 2020; Novak et al., 2017). Following clinical identification, several diagnostic tests are recommended: neonatal brain magnetic resonating imaging (MRI); cranial ultrasound (CUS); genetic testing, which includes phenotyping; developmental testing; and neurological testing (Glader & Stevenson, 2019). Evaluation of gross motor skills is often conducted along with neurological tests such as Hammersmith Infant Neurological Examination (HINE) and Prechtl Qualitative Assessment of General Movements (Kwong, Fitzgerald, Doyle, Cheong, & Spittle, 2018). The combination of the tests noted above can offer up to 98% predictability of cerebral palsy in young children. While early prediction may be arduous, it is necessary to provide early interventions such as hip surveillance and constraint-induced movement therapy (CIMT) (Novak et al., 2017). Because of the high occurrence of associated conditions, children with cerebral palsy should also be screened for cognitive, visual, and hearing impairments, as well as speech and language disorders.

Course and Prognosis

The course of cerebral palsy varies depending on type, severity, and presence of associated problems. With mild motor involvement, the child will continue to make motor gains and compensate for motor difficulties with little to no assistance from caregivers. With more severe forms, little progress may be made in attaining developmental milestones and performing functional tasks. Those with complex cerebral palsy and GMFCS and MACS level V will typically require full-time assistance from a caregiver and need assistive devices such as a custom wheelchair and equipment for activities of daily living (ADLs).

As the child grows older, secondary problems such as contractures and deformities will become more common, especially with spasticity. Adults with cerebral palsy experience musculoskeletal difficulties and loss of function at an earlier age than their nondisabled peers. One study found that walking remains constant in 76% of those who ambulated as a child. Those who used a wheelchair were 34% more likely to quit ambulating by age 25 (Day et al., 2007).

The survival rate for adults with cerebral palsy is good but lower than the general population. An increase in mortality is evident with higher level of GMFCS and typically attributed to secondary conditions. An Australian study of 385 persons found that 75% of mortality cases were attributed to respiratory reasons such as pneumonia, with an average age of 14.6 (Blair, Langdon, McIntyre, Lawrence, & Watson, 2019).

Medical/Surgical Management

Because of the complexity and diversity of difficulties affecting the individual with cerebral palsy, medical management requires a team approach using the skills of many professionals. Depending on the type of cerebral palsy and the presence of associated problems, team members typically include medical doctors from a variety of specialties, occupational therapist(s), physical therapist(s), speech pathologist(s), an educational psychologist, a nurse, and social worker(s). The emphasis of intervention is to help the child gain as much motor control as possible, position the child to maximize independence and minimize the effects of abnormal muscle tone and pain, instruct the parents and caregivers on handling techniques and ways to accomplish various activities of daily living, recommend adaptive equipment and assistive technology to increase the child's ability to perform desired activities, provide methods to improve feeding and speech if difficulties are present, and help parents manage behavioral concerns and family stresses. The primary physician treats the usual childhood disorders and helps with prevention of many health problems. Physicians with various medical specialties may also be involved. The neurologist may assess neurological status, conduct gait analysis, address tone pharmacology needs, and help control seizures, if present. An orthopedist may monitor skeletal changes, prescribe orthotic devices, and address any necessary surgeries. A gastroenterologist may monitor food intake, reflux, and needed surgical intervention. An ophthalmologist would assess and treat any visual difficulties.

Medical management includes both surgical and nonsurgical approaches, with much of the focus on techniques to decrease spasticity and dystonia. Several oral medications such as diazepam (Valium), dantrolene (Dantrium), and baclofen are used to reduce spasticity in severe cases with mixed results (Delgado et al., 2010). Intrathecal baclofen infusion (ITB) administered through a pump implanted in the abdominal wall to the spinal cord fluid has shown to be more effective than oral medications in reducing severe spasticity and dystonia in cerebral palsy. Impact on spasticity of the lower extremities have shown to be positive; however, effect on the upper extremities is inconclusive. Complications from surgery occur in 5 in 100 cases. Further evidence is needed to determine significance for quality of life (Novak et al., 2013; Russman, 2010). Another treatment more widely used in recent years is the injection of botulinum toxin (brand names Botox or Dysport) into muscles. Spasticity is reduced for a period of 3-6 months after injection. Botox or Dysport is injected into specific muscles that, in addition to reducing tone, increases range of motion and reduces deformities as well as provides an opportunity to work on muscle strengthening. A systematic review of interventions found that Botox is effective for decreasing upper extremity spasticity, improving walking and hand function, and decreasing drooling (Novak et al., 2013).

Orthotics and splints may be prescribed for the child with cerebral palsy. Upper extremity splints are worn to maintain range of motion, manage contractures, or facilitate typical grasping patterns during daily activities. A thoracolumbosacral orthosis (TLSO) is a back brace used to support the trunk and slow the progression of spinal deformities. Ankle–foot orthoses (AFOs; Fig. 2.7) and supra malleolar orthotics (SMOs) offer ankle stability, prevent collapse of the arch, and facilitate proper foot alignment. Orthotics and splinting should be used with caution as the evidence is insufficient that it improves function and prevents contractures and deformities. Child and caregiver burden, wearing-schedule compliance, and risk of skin integrity issues related to orthotics or splints should also be considered. Serial (or progressive) casting is shown to be effective for improving range of motion in the lower extremities, but there is insufficient evidence that it is equally effective in the upper extremities. However, CIMT, which involves restraint of the unaffected limb in conjunction with bimanual training (use of both hands together with repetitive activities), is proven to be effective for cerebral palsy intervention (Novak et al., 2013).

Surgical approaches are used to improve the function and appearance of affected areas of the body and to prevent or correct deformities. Common surgical procedures include joint fusions, tendon lengthening to increase range of motion, and tendon transfers to decrease spastic muscle imbalances (Koman & Smith, 2014). Selective dorsal rhizotomy (SDR) is a neurosurgical technique that is used to reduce spasticity and improve function in carefully selected individuals (Ferger, Lunsford, Sauer, Novicoff, & Abel, 2015; Tedroff, Hägglund, & Miller, 2020). The procedure involves dividing the lumbosacral posterior nerve root into four to seven rootlets. Each rootlet is stimulated electrically. The dorsal rootlets causing spasticity are determined through careful neurological examination of those showing pathology. This approach is highly invasive with mixed results. Often, other pharmacological interventions such as baclofen or Botox are still required following the procedure (Tedroff et al., 2020). For children with diplegia, the goal is to improve gait and leg function. For children with spastic quadriplegia who have very limited movement, the goal is to increase their independence by allowing them to sit for longer periods of time, enabling them to use a wheelchair or potty chair and making daily care easier for their caregivers by reducing the spasticity that makes dressing and other daily living tasks more difficult. An essential part of this treatment approach includes intensive postsurgical physical and occupational therapy for a period of several weeks.

Impact on Occupational Performance

It is important for the occupational therapist to be aware of all the client factors that can affect individuals with cerebral palsy, but to not make any assumptions based on the type of cerebral palsy and known associated disorders. Each factor should be directly assessed to determine its impact on occupational performance. Furthermore, each individual is unique and will have their own set of strengths and challenges. Factors such as motivation, cognition, caregiver support, socioeconomic status, and environmental contexts will play a role in participation in occupation. Virtually all of the body function categories can be affected in the individual who has cerebral palsy. Milder forms of cerebral palsy may have limited impact upon occupational performance. Some individuals will require physical assistance, additional training, or assistive technology to participate fully in occupational performance areas, while individuals with severe forms of cerebral palsy will be limited in their performance of all areas of occupation. The body function category that is always affected in individuals with cerebral palsy is neuromusculoskeletal and movement-related function. Overall, the severity of the condition, type of cerebral palsy, and the presence of associated disorders determine how occupational performance is impacted.

Activities of Daily Living and Instrumental ADLs

Participation in activities of daily living (ADLs) and instrumental ADLs varies greatly depending on classification of cerebral palsy and severity of the condition. Many children and adults with cerebral palsy can successfully manage all essential self-care activities. Children and adults with a GMFCS and MACs V will require assistance for nearly all ADLs and IADLs from a caregiver, nurse, or paraeducator while at school. Additionally, consideration of accessibility of everyday items within the lived environment will help foster independence. For example, adaptations can be made to the counter level in the kitchen and bathroom to help promote participation in daily routines and meal preparation. Likewise, environmental controls needed to manage the lights, television, or small appliances can be programmed through communication devices for youth and adults with spastic quadriplegia.

Health Management

As the youth with cerebral palsy transitions into adulthood, health management becomes a primary concern. It is important to establish health and wellness routines such as medication and appointment management, wellness and stress-reduction strategies, and fitness regimes as early as possible. Independence with health care decisions varies greatly depending on functional level and other factors discussed in this chapter. Health care decisions automatically transfer to youth at 18 years of age; therefore, planning for health care needs should be considered in the early to mid-teens (de Gusmao & O'Hare, 2019). It is also necessary to transition from pediatric care to an adult primary care physician and specialists. The youth and caregivers should be prepared for the change in focus from comprehensive chronic care to that specifically targeting acute concerns. Therapies, private duty nursing, and habilitation services may no longer be covered by insurance for the adult with cerebral palsy.

Rest and Sleep

Rest and sleep may be difficult for many with cerebral palsy. This is due to several reasons, including pain, dystonia triggered by movement in the night, seizures, respiratory issues, and/or sleep apnea (Welsh & Katwa, 2019). Disruption of rest and sleep for the child or youth with cerebral palsy may also significantly impact that of the caregiver's due to possible medical needs throughout the night. Goal planning focused on sleep and rest strategies may be beneficial for some with mild sleep disturbances. Activities such as reduction of screen time before bed, sleep schedules, and naps can help facilitate healthy sleep patterns.

Education

Children and adults with cerebral palsy who are participating in education may require minimal to full assistance. Those with lower GMFCS levels may need little to no assistance to participate but may need assistance to transition to classes and need minimal accommodations for assignments. Someone with spastic quadriplegia may require full assistance throughout the day for transfers, assistive technology for communication and learning activities, and school nursing for feeding and bowel/bladder management. Both global and specific mental functions can be affected, particularly if there is an associated learning disability, attention deficit hyperactive disorder, or cognitive impairment.

Work

Many with cerebral palsy are able to lead a full productive work life. Employers may need to provide simple environmental modifications (eg, provision of a stool for a cashier) for those with mild variations of cerebral palsy (GMFCS I-II). Individuals with spastic quadriplegia and limited functional independence can participate in work or volunteer opportunities with an accommodating employer, proper contextual supports, and utilization of complex durable medical equipment such as power wheelchairs with seat controls and communication devices.

Play and Leisure

Cerebral palsy can impact play and leisure opportunities for many children and adults with the condition. Assumptions that participation cannot occur due to motor limitations often creates unnecessary barriers to occupational engagement in activities. Reduced participation may contribute to depression and feelings of isolation. Caregiver stress also may be compounded by the child's limited opportunity to participate in leisure activities (Arakelyan, Maciver, Rush, O'hare, & Forsyth, 2019). According to Rosenbaum and Gorter (2011) in the ground-breaking article, "The 'F-words' in childhood disability: I swear this is how we should think!" the ingredients for participation, not only in leisure activities, but with most occupational performance areas, are simple: ask the child what they want to do, adapt the activities, and don't expect the individual to participate just like everyone else.

Social Participation

In general, people with cerebral palsy need positive engagement with family and peers similar to that of every other individual. It is important to actively involve an individual with cerebral palsy in co-occupations while at home, school, or participating in community activities through a variety of methods. First, many everyday social activities are performed at standing eye level. Effort should be made to address the person in a wheelchair at eye level. Second, it is beneficial to provide ample time for verbal and written response in cases in which dysarthria or delayed motor are responsible. Lastly, the individual should be encouraged to seek opportunities in activities such as social clubs, adaptive sports, online activities, and romantic and age-appropriate sexual relationships (Rosenbaum & Gorter, 2011).

CASE STUDY 1

A.K. is a 2-year-old girl who lives with her parents and older brother. She was born at 37 weeks' gestation at a birth weight of 5 lb 10 oz. Pregnancy and birth were unremarkable. She was healthy at birth but by her well baby visit at 9 months of age, she was not yet rolling, crawling, or sitting independently. Her pediatrician referred her to a pediatric neurologist who diagnosed her with spastic diplegia at 11 months of age. The neurologist referred her to a physical medicine and rehabilitation (PM&R) physician at the local children's hospital as well as the local school district for early intervention services. The PM&R physician signed a referral for her to the orthotics department to fit her with AFOs to help her with standing and walking. He has also recommended botulinum toxin injections and selective posterior rhizotomy for consideration as future treatments. Through the early intervention program, she received a multidisciplinary team assessment, which included physical, occupational, and speech and language assessments. Delays were noted in gross motor, fine motor, and self-help skills. Speech and language, social, and cognitive skills were all determined to be at age level. Weekly occupational and physical therapy home-based services were recommended.

Affected performance skills are in the motor area, which includes posture, mobility, coordination, and strength/effort. She demonstrates spasticity in all lower extremities and her trunk. Her movement patterns reflect significant spasticity in her legs. Her joint range of motion is significantly limited in her hamstrings and hip adductors with mild limitations in her heel cords bilaterally. A.K. can sit independently; however, it is difficult for her to sit on the floor with her legs extended in front of her as a result of hamstring and hip adductor tightness. She requires assistance moving in and out of sitting. In prone, she can push up to hands and knees and can crawl for short distances. She bears weight on her legs in supported standing with knees in a slightly flexed position, hips adducted, and feet plantar flexed and pronated. She has begun ambulating with a walker for short distances.

In her upper extremities, there is mildly increased muscle tone bilaterally as well as some incoordination. A.K. grasps pegs and small blocks and releases objects into a container. She can place a peg in a pegboard but is not yet able to stack objects or complete a shape sorter. Areas of occupation that are affected include activities of daily living (ADLs) and play. Because she is only 2 years old, instrumental activities of daily living, student, work, and

leisure areas of occupational are not yet relevant areas for her. In ADLs, because of her age, she would not yet be expected to be independent. Given this, A.K.'s affected activities of daily living include bathing, dressing, feeding, and functional mobility. A.K. needs assistance with dressing skills such as undressing and removing shoes and socks. She is independent in feeding with some adaptations. She requires assistance with maintaining a stable sitting position in the bathtub and requires assistance getting in and out of the bathtub. She needs assistance with functional mobility, such as getting in and out of chairs and moving from one place to another.

Exploratory play skills are affected by A.K.'s difficulty with moving about her environment to obtain toys she wants to play with. Participation is affected by the need for a stable position in which to free up her upper extremities to manipulate toys. She uses a bench with a pelvis stabilizer and tray for refined fine motor tasks.

CASE STUDY 2

L.N. is a 54-year-old woman with cerebral palsy, spastic quadriplegic type. She has lived alone in an apartment complex for the elderly and disabled for the past 15 years. She supports herself on supplemental security income (SSI) and disability payments from the state. A personal care attendant provided by the Department of Social Services comes in each morning and evening to assist her with activities of daily living, such as meal preparation, bathing, and dressing. L.N. has never been employed but has done volunteer work. She writes articles for a newsletter on her computer and has worked in her church's Sunday school. She has no family support but has many friends. She enjoys learning and taking classes through continuing education.

Spasticity, dystonia, and retained primitive reflexes severely restrict L.N.'s purposeful movement. She has limited range of motion in her left upper extremity and both lower extremities. When reaching with the left arm, she cannot bring it to shoulder height or behind her back. She has a gross grasp in her right upper extremity and can grasp a joystick to operate her electric wheelchair. She cannot write or perform other activities requiring fine motor dexterity. Her left upper extremity is used as an assist for bilateral activities, with no grasping ability present. She can maintain an upright position in sitting, but her weight is shifted to the left (with resulting scoliosis). She can bring her head to an upright position, but neck flexion increases with activities requiring effort. Oral motor muscles are affected, resulting in severe dysarthria, drooling, and difficulty eating. Endurance is a problem, and L.N. becomes easily fatigued.

Communication/interaction skills are also affected. Articulation and modulation when speaking is affected by L.N.'s oral motor control. Limited dexterity and restrictions in movement limit her ability to use gestures and to orient her body in relation to others when engaged in social interactions.

All areas of occupation are affected. In activities of daily living, L.N. needs assistance with bathing, personal hygiene and grooming, toilet hygiene, and dressing. She brushes her teeth and performs light hygiene, such as washing her face, independently. She can transfer herself between her wheelchair and her bed. She needs assistance transferring to the shower seat she uses for bathing. She can transfer on and off the toilet in her apartment with grab bars and the toilet seat at the proper height and position, although it takes her a while to do this. In eating, L.N. can feed herself with adaptations if the food is set up for her, but the process is slow and messy. She drinks from a straw. She takes her own medications if they are set out for her.

In instrumental activities of daily living, L.N. needs assistance in clothing care, cleaning her apartment, household maintenance, and meal preparation. She can use a hand-held portable vacuum cleaner for small cleanups. She has a cat that she cares for. She shops independently but needs assistance getting money out of her wallet at the cash register. All areas of activities of daily living are affected except socialization. L.N. uses a computer for written communication. She uses a speaker phone for telephone communication. If she falls or is in danger at home, she has an emergency alert system that she can activate. Because her speech is difficult to understand, she has an augmented output device for communication but uses it infrequently. She uses a motorized wheelchair for mobility. In the community, L.N. uses public transportation with no difficulty. She has some difficulty transferring herself to and from the toilet when using public restrooms, which sometimes results in incontinence.

In work activities, L.N. has never been employed but has worked as a volunteer for the past several years in the religious education program at her church. She enjoys the interaction with the children that are in the classes.

In leisure activities, L.N. has varied interests. She is an avid reader and enjoys computer games. Social activities include getting together with friends frequently and going out into the community, either alone or with friends. L.N. participates in church retreats as well as community-based trips through an independent living center.

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CHAPTER
3

Autism Spectrum Disorders

Michelle A. Suarez and Kathryn Ellsworth

Key Terms

Auditory processing
Autism spectrum disorder (ASD)
Casein
Echolalia
Food selectivity
Gene
Gluten
Joint attention
Neurons
Restrictive and repetitive behavior
Rote memory
Sensory processing disorder
Theory of mind

Henry was a beautiful firstborn baby boy. His mom and dad delighted when he squeezed their finger with his tiny fist and looked into their eyes. They loved to cuddle him and show him off on walks through the neighborhood while he sat contentedly in his stroller. However, his first year was hard. He was diagnosed with gastroesophageal reflux disease (GERD) and struggled to put on weight. Henry did not sleep for more than a few hours and was hard to soothe. He seemed to be extremely uncomfortable after each feeding. For example, he would cry, arch his back, and squirm while eating. His parents learned how to thicken his formula and hold him upright after each bottle. This seemed to make him more comfortable and the family was relieved that he was crying less.

When Henry was 16 months old, his parents took him to a birthday party for their 1-year-old niece. They noticed that she was saying a few first words and that she delighted in games like peek-a-boo. She was walking, exploring her environment, and extending items to show others while making eye contact and smiling. Henry's parents started to become worried about Henry's limited engagement and interaction. He was not crawling yet and sometimes would sit and look at parts of his toys for extended periods of time. He also rarely looked at them, especially avoiding eye contact, and did not respond to his name. He did not reach to request desired items and rarely babbled. When he produced sounds, he did not use them to seek the attention of or communicate basic ideas with others. As a result of this decreased responsiveness, his parents worried that his hearing was impaired. However, an audiological test indicated normal hearing abilities.

Dressing this little boy became a challenge. Suddenly, at age 18 months, he did not tolerate the sensation of clothes against his skin. He became intensely distressed when his mother tried to brush his hair or clip his nails. Engaging Henry in daily activities became increasingly difficult, for he appeared frustrated, unable to express his needs and wants, and unable to leave familiar, preferred activities without becoming highly agitated. During episodes of tantrums, Henry would hit himself or bang his head repeatedly against a wall while crying. His mother stated that overall, "he does not seem happy."

When Henry was 2 years and 6 months old, his parents brought their concerns to the attention of early intervention specialists. As a result, Henry began receiving occupational therapy, speech-language therapy, and special education services. During his preschool class, these specialists observed the same concerning characteristics his parents had described. Additionally, they noted that he did not appear interested in the other children in his preschool class. Transitioning from room to room consistently distressed him, causing "meltdowns" during which would he fling himself on the floor and cry inconsolably. He used only a few real words or echoed words of others

but did not appear to understand what they meant. Many times, the classroom environment seemed to provide him with far too much sensory input. As a result, he would close his eyes or seek out quiet places away from others. Yet Henry still showed moments of attachment and joy, such as a strong, loving connection toward his parents and grandparents. Additionally, he smiled, laughed, and shared eye contact during specific activities, such as swinging or singing certain songs.

Despite these moments of engagement, the gaps in his development caused his parents and therapists to seek further neurodevelopmental testing. Henry was therefore assessed by a team of specialists including a developmental pediatrician, a psychologist, a speech-language pathologist, and an occupational therapist who confirmed the suspicions of his family and therapists: Henry presented with autism spectrum disorder.

Description and Definitions

Autism spectrum disorder (ASD) is characterized by impairments in social interaction and communication and by the presence of restricted and repetitive behaviors (RRBs) (American Psychiatric Association, 2013b). Individuals with this developmental disorder often have difficulty with skills such as participating in conversation or may misread nonverbal social cues from others. These challenges adversely impact an individual's ability to make friends and interact with peers. In addition, people who have ASD may have unusual responses to sensory information (eg, tactile, auditory, visual stimulus) or experience high sensitivity to changes in their environment and/or become overly dependent on routines.

The *Diagnostic and Statistical Manual of Mental Disorders (DSM)*, by the American Psychiatric Association, provides differential diagnosis criteria for autism. In 2013, the *Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-5)*, was published and provided major changes to the way individuals with autism are diagnosed. In the earlier manual, the *Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition (DSM-IV),* individuals could be diagnosed with one of five main disorders under the autism spectrum umbrella. These disorders included autistic disorder, Asperger disorder (also known as Asperger syndrome), pervasive developmental disorder not otherwise specified (PDD-NOS), childhood disintegrative disorder, and Rett disorder (American Psychiatric Association, 2000).

In a major departure from the presence of multiple categories of autism, the DSM-5 provides for one universal ASD diagnosis. The rational for this change was the inconsistent and inaccurate use of the DSM-IV criteria (American Psychiatric Association, 2013a). Researchers found that across clinics and professionals, there were little reliable data to support the continued distinction of five separate disorders. A summary of the alterations from the DSM-IV to the DSM-5 include (1) elimination of the five subtypes of Pervasive Developmental Disorder (ie, autistic disorder, Asperger's, PDD-NOS, Childhood Disintegrative disorder, Rett disorder); (2) development of the ASD diagnosis with specifiers for severity and presence or absence of intellectual features; (3) changes in diagnostic criteria from inclusion of social reciprocity, communication, and RRB to only two criteria of social communication/interaction and RRB; (4) inclusion of sensory processing symptoms as one sub feature of RRB; and (5) a change in the diagnostic age of onset from "age three" to "early childhood" (Shin Kim et al., 2014). In addition to these changes, the DSM-5 adds a "Social Communication Disorder (SCD)" category for individuals without RRB (American Psychiatric Association, 2013c).

In the DSM-5, anyone with a DSM-IV diagnosis should still meet the criteria for ASD. In addition, the DSM-5 provides new specifiers to more accurately describe the variants and severity of ASD; theoretically making it of more practical use for developing a treatment plan for an individual based on their specific presentation of this condition. For example, an individual who was previously diagnosed with Asperger disorder (a previous variant of autism where intelligence is normal, language skills are intact, but social skills are impaired) might now be diagnosed with ASD *with or without* intellectual impairment or *with or without* language impairment. Children with ASD share the following diagnostic characteristics (American Psychiatric Association, 2013b):

1. Impairment in social communication and social interaction that occurs over several contexts
2. **Restricted and repetitive behavior**, interests, and/or activities

Additional specifiers include ASD:

- "With or without accompanying intellectual impairment"
- "With or without accompanying language impairment"

Finally, the child's ASD is classified based on severity:

- "Level 3: Requiring very substantial support"
- "Level 2: Requiring substantial support"
- "Level 1: Requiring support"

A child with ASD often demonstrates not only delays but also atypical patterns of development. Some children evidence problems from birth, such as appearing aloof or extremely fussy or aversive to eye contact compared with other infants. In other cases, the characteristics of autism noticeably emerge between 12 and 36 months of age. Recognition of symptoms at this age is often due to one of two reasons. During the 12- to 36-month period, in a child developing typically, there is usually a rapid expansion of language skills. Children with ASD, however, may have been developing more slowly overall, and then this language burst fails to occur. Alternatively, some preschool children with ASD may lose language skills that they had demonstrated during earlier months in their development (Al Backer, 2015).

ASD is considered a lifelong disability for which there is no cure. However, intervention to improve skills, particularly in challenging areas that significantly limit participation in meaningful occupation, can have a positive and powerful impact on quality of life for individuals with this disorder.

In addition to changes introduced with the DSM-5 to create one ASD, a new category was introduced. Social (pragmatic) communication disorder is characterized by social communication dysfunction without the presence of RRB (Norbury, 2014). Social communication requires use of social contexts for understanding meaning. For example, it is necessary to understand a speaker's intentions and nonverbal cues as well as societal norms and expectations in order to understand the message being communicated. Pragmatics is similar and involves use of language in context and can include things like taking turns in conversation or shared meaning between a speaker and a listener.

Children with social (pragmatic) communication disorder share the following diagnostic characteristics (American Psychiatric Association, 2013b):

1. Ongoing difficulty with verbal and nonverbal communication that is not explained by decreased cognitive skills.
2. Difficulty with learning to use language including inappropriate responses in communication.
3. Symptoms limit effective communication, relationships, academic skills, and occupational performance.
4. Must be present in early childhood.

The diagnostic process for ASD changed from the DSM-IV to the DSM-5. However, individuals with autism continue to demonstrate difficulties with social communication and restrictive and repetitive behaviors. Diagnostic accuracy allows for treatment planning specific to an individual's unique needs.

Etiology

There is no single, clearly defined cause of autism. This condition was first described in literature in 1943 by Dr. Leo Kanner (1943). His initial hypothesis was that autism was caused by "cold" or unresponsive parents (Bettelheim, 1967). This hypothesis has been proven false and summarily rejected. Researchers now believe that autism develops from a complex combination of biological, genetic, and environmental factors (National Institute of Health [NIH], 2017).

Biology: Abnormalities in Brain Structure and Function

Children with ASD develop, process, and react to the world differently from typically developing children. This diversion from normal development may arise from physical differences in the brains of children with ASD. Additionally, the way in which life experiences are mapped onto the brain may be altered as a result of these physical differences (Courchesne, Campbell, & Solso, 2011). The precise distinctions are not yet entirely clear, since ASD is not caused by a single obvious lesion in the brain. In fact, studies have indicated differences in the way the brain grows and how different areas of the brain communicate may result in the manifestation of ASD (Ha, Sohn, Kim, Sim, & Cheon, 2015). Through the study of postmortem brain tissue and imaging studies with positron emission tomography (PET) scan, magnetic resonance imaging (MRI), and functional magnetic resonance imaging (fMRI), consistencies have emerged in the research of brain abnormalities.

Brain Volume Differences

Scientists have demonstrated differences in the number of neurons, as well as growth trajectories, in the brains of children with autism. There is evidence that these differences emerge prenatally, during the second trimester, and persist throughout the lifespan (Courchesne et al., 2011). The location in the brain where these abnormalities occur may shed light on the underlying pathology of the symptoms of autism.

ASD may begin before a baby is born. During the second trimester of pregnancy, there is a large production of brain cells or **neurons** (Courchesne et al., 2011). These neurons develop and migrate into their appropriate locations to support the healthy foundational structure needed for future functioning. As this process unfolds, unhealthy and excess neurons are pruned so that smooth and efficient connections can be made to support learning and development. A study of post-mortem brain slices of children with autism revealed dense patches of abnormal neurons located in the wrong layers of the brain in the frontal and temporal lobes (Stoner et al., 2014). It is possible that the process of checking and removing bad cells does not work and the brain begins life cluttered with excess neurons (Courchesne et al., 2011).

Additionally, in children with ASD, there is abnormally accelerated brain growth, particularly in the frontal and temporal lobes, during the first few years of life (Ha et al., 2015; Redcay & Courchesne, 2005). This pathological growth causes large deviations in brain volume between the ages of 2 and 4 years (Fig. 3.1). After this period of time, growth falls into a normal or possibly even arrested growth rate (Courchesne, 2004).

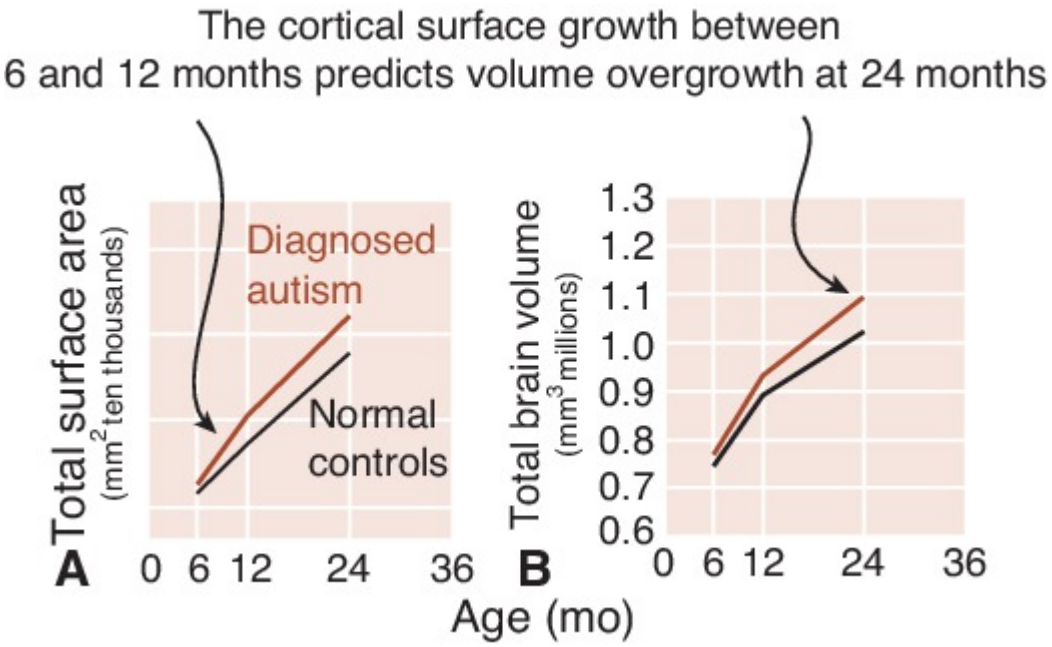


Figure 3.1 A. Toddlers who develop autism showed an accelerated expansion of brain surface area between 6 and 12 months. **B.**Toddlers with autism have greater total brain volume at 24 months. (Reprinted with permission from Higgins, E. S., & George, M. S. (2019). *The neuroscience of clinical psychiatry* (3rd ed.). Wolters Kluwer. Adapted by permission from Nature. Hazlett, H. C., Gu, H., Munsell, B. C., Kim, S. H., Styner, M., Wolff, J. J., … Piven, J. (2017). Early brain development in infants at high risk for autism spectrum disorder. *Nature, 542*, 348–351. Copyright © 2017 Springer Nature.)

It is relevant that abnormal cell development, decreased pruning, and accelerated growth of cells occur in the frontal and temporal lobes of the brains of children with autism (Ha et al., 2015). These areas are responsible for the higher level cognitive, social, emotional, and language functioning that is impacted by ASD. Having excessive neurons with tangled connections may make it confusing and difficult to hone in on the important aspects of the environment that are necessary for learning higher level skills, ultimately leading to autistic behavior (Courchesne, 2004).

It is also interesting to note that the temporal and cortical regions of *typically developing* children grow particularly slowly compared to those with ASD (Courchesne et al., 2011). There is evidence to show that slower growth leads to greater higher-level skill development. It is possible that this slow growth allows typically developing children's brains to be shaped by the complex and nuanced social experiences that they are exposed to over time. Children with ASD may not have this extended brain growth time to allow their brains to be shaped by formative and complex experiences in order to wire correctly.

Eye Contact Differences

Brain structure differences can cause behavior that isolates children with early indicators of autism, serving to focus them increasingly toward their own internal world. For example, eye contact is necessary for learning social skills and perspective taking through reciprocal engagement with other people. Many children with autism struggle with eye contact from very early in development.

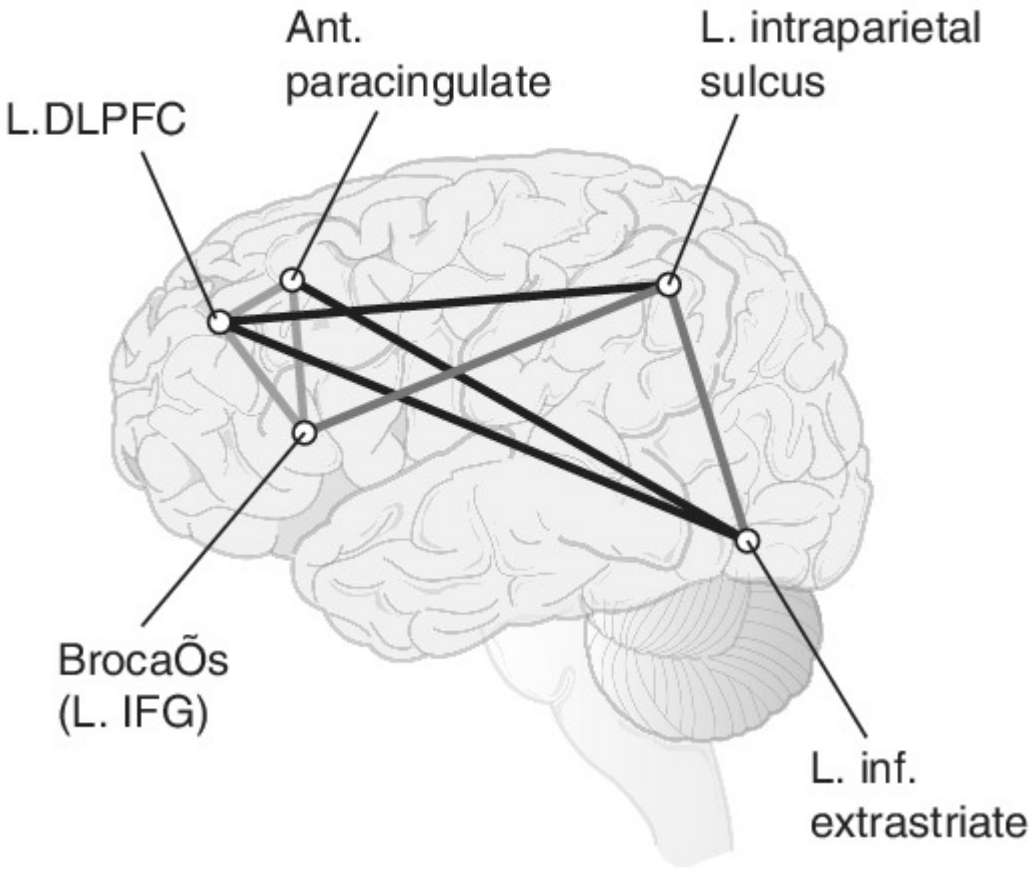
In a fascinating study, Jones and Klin (2013) used sophisticated eye tracking technology to measure eye fixation during the early months and years of life. They found that children who are later diagnosed with ASD have typical interest in people during their first 2 months. After that, eye contact decreases sharply through 6 months of age. At this point, children with autism attend more to objects than people and therefore, their brain is shaped and specialized to attend to this nonsocial world. They do not have the same biologically driven interest in people, closing an important window for learning social communication and interaction skills during the toddler and preschool years. In this way, "autism creates itself" (Klin, 2012) and eye gaze differences persist into adulthood.

This information highlights the importance of early intervention (Courchesne et al., 2011). When children with autism are identified early, intervention can provide intensive and targeted social experiences to influence the connections that are retained in the brain. Neuroplasticity allows for the foundational structures of the brain to be built and re-modeled through flooding the child with positive and developmentally appropriate learning experiences.

Brain Connectivity

Brain connectivity differences have emerged in the research that may account for some of the symptoms of autism (Fig. 3.2). The gray and white matter of the brain serves to connect different areas so that they can communicate and work together. Gray matter is where the cell bodies, dendrites, and axon terminals live and specialized processing occurs (Mackenzie, 2019). White matter is composed of axons that *connect* adjacent gray matter regions, as well as regions across hemispheres and lobes. One hypothesis about the cause of autism relates to differences in white matter, as well as underconnectivity in long-range connections (white matter differences) and overconnectivity in short-range connections (gray matter differences) (Alexander, 2012).

Autistic



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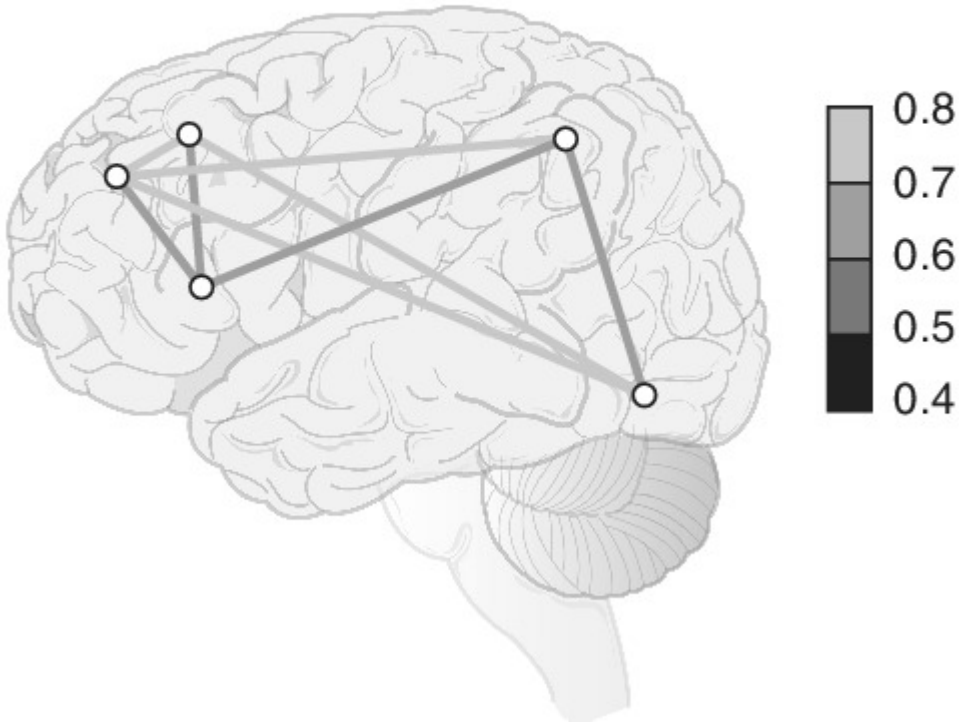

Figure 3.2 Autism: Connectivity in the brain. The behaviors of autism may result from reduced connectivity among the different regions of the brain. This figure illustrates reliably lower functional connectivity for autism participants between pairs of key areas during sentence comprehension tasks. Darker end of scale denotes lower connectivity. (Slide courtesy of Marcel Just, Carnegie Mellon University.)

Several studies have found abnormalities in white matter in key regions of the brain in individuals with autism. In the corpus callosum, the brain structure that bridges the left and right hemispheres, there appears to be less white matter fiber density, suggesting fewer axons to transmit information (Dimond et al., 2019) and atypical development of the microstructure of the white matter in this region (Travers et al., 2015). In addition, Dimond and colleagues (2019) found decreased white matter fiber density in the frontal-occipital fasciculus. This is a white matter tract that connects the occipital and parietal lobes at the back of the brain with the frontal lobe (Conner et al., 2018). This tract is responsible for semantic language processing and goal-oriented behavior. Communication over distances between the two sides of the brain, and the back and front of the brain, may be impacted by these white matter differences.

Underconnectivity, or fewer connections, among long-range regions may be an underlying neural mechanism for autism. Long-range connections provide the wiring of our brain across hemispheres and lobes (Alexander, 2012). These connections allow us to use the different specialized regions of our brain to process and integrate complex information (Park & Friston, 2013). This "cross talk" among different areas of the brain allow us to understand the world and accomplish tasks (Maximo, Cadena, & Kana, 2014). It is clear that these long-range connections are impaired in people with autism (O'Reilly, Lewis, & Elsabbagh, 2017). In fact, Anderson and colleagues (2011) were able to use functional connectivity alone (connection between two brain structures) to predict which children had autism at a ~80% accuracy rate.

The support for the hypothesis of overconnectivity in short-range connections in autism is less clear (O'Reilly et al., 2017). Overconnectivity in short-range connections means that there is a great deal of communication between adjacent areas of the brain. With overconnection, this communication could create confusion or low-level noise in the brain (Maximo et al., 2014). This alteration could reduce efficiency for processing and disrupt the highlighting of important information in the environment and ignoring things that are unimportant. Several studies have identified this overconnectivity in people with autism (Alexander, 2012) but other research has conflicted with these findings (O'Reilly et al., 2017). This area will be continually explored as researchers work to pin down underlying mechanisms in autism.

The brain is made up of about 100 billion neurons that interconnect and communicate information among the various brain regions and from other areas of the body. The behaviors of autism may result from abnormalities in the neural networking between the multiple areas of the brain as it works to process complex information.

Scientists are persistently closing in on the differences in the brain structure and function of this population. In the meantime, the question arises: What has happened in a child's system to cause these deviations in the brain to occur?

Parents of children with ASD, upon hearing their child's diagnosis, typically search for reasons why this condition occurred in their child. The professionals who work with families must act as guides to discuss the most recent and accurate information in the autism field.

Genetics

The differences in the neurobiology of children with autism are most likely accounted for by genetics. **Genes** are composed of DNA and, through heredity, determine the particular characteristics that distinguish one human from another. Genes are encoded instructions—the brain's written guidelines for function. If any abnormalities lie in a brain's genes that are passed on from a parent, or if *de novo* change (spontaneous alteration in a gene) occurs, the brain deviates from a course of typical development (Huguet, Benabou, & Bourgeron, 2016).

ASD is one of the most heritable neuropsychiatric disorders (Jeste, nd). In monozygotic (identical) twins, where both twins develop from one egg, there is a 60%-70% autism concordance rate (Sandin et al., 2012). In dizygotic (fraternal) twins, where twins develop from two separate eggs, there is a 5%-10% autism concordance rate. The recurrence rate for siblings is 10%-20% or 33% if more than one child has already been affected. Clearly, there is no question that autism is genetic. However, it is not necessarily one gene or a single set of genes that causes autism. If it were a single gene, 100% of monozygotic twins would both be affected.

More than 100 genes have been associated with autism (National Institute of Medicine, U.S. National Library of Medicine, 2020). Often, it appears that gene variations may combine with other factors to trigger the condition. A child can have a *de novo* mutation in a gene that was not passed on from their mother or father. Susceptible genes appear to stem from certain biological pathways including those that control gene expression, support the development of synaptic functions, and the serotonin-NAS melatonin pathway that is important for sleep (Huguet et al., 2016). The highly varied triggers for the development of autism are likely one reason for the heterogeneity of the disorder.

Comorbidity with other clearly defined genetic conditions can provide additional insight into the origins of autism. For example, fragile X syndrome is caused by a mutation in *FMR1*, located on the X chromosome. It is the most common single gene disorder that accounts for about 3% of cases of autism (National Fragile X Foundation, 2020). Tuberous sclerosis complex causes the growth of cortical tubers in the brain. This affects the connections between different regions. This disorder affects about 1-6000 people (Jeste, Sahin, Bolton, Ploubidis, & Humphrey, 2008) and ~40% of people with this disorder also have autism (Vignoli et al., 2015). These disorders can provide information about the manifestation of observable characteristics or clusters of symptoms stemming from a known genetic cause.

As we learn more about the genetic origins of autism, in many cases, parents are encouraged to pursue genetic testing (Jeste, nd). The hope is that eventually, information about the genetic origin of the disorder can have treatment value. If we can specify the constellation of symptoms that stem from specific genetic origins, we may be able to individualize a child's treatment for greater efficacy and efficiency. Ultimately, the goal is to provide early, targeted intervention to support independent functioning and quality of life.

Environment

It is clear that genetics influence the risk for autism, but this factor alone does not explain all instances of the disease. Aspects of the environment may combine with genetic predisposition to cause or prevent the development of ASD. Environmental factors include parental age at conception, exposure to toxins, and maternal factors, including nutrition. Finally, prematurity or very low birth weight is also linked. Understanding environmental factors is crucial for improving diagnosis and treatment.

Advancing age of both the father and mother can increase the risk of having a child with autism. Although there is not widespread agreement about the degree of this risk, one study estimated that men who have children over the age of 40 doubled their risk of having a child with autism (King, Fountain, Dakhlallah, & Bearman, 2009). For women, this risk was three-fold. For perspective, in the entire population, children born between 1992 and 2000 had an autism prevalence of 34 per 10 000 births. During this same time period, in children born to parents over 40 years of age, the prevalence rose to 56 per 10 000.

Another factor that can increase the risk of autism is prenatal exposure to toxins. For example, maternal use of the drug valproate, used to treat epilepsy, significantly increases the risk of autism (Christensen et al., 2013). Maternal exposure to insecticides (Brown et al., 2018) and exposure to air pollution from living near a freeway during the third trimester (Volk, Hertz-Picciotto, Delwiche, Lurmann, & McConnell, 2011) also increased the risk. In all studies, exposure to toxins combined with genetic predisposition to increase risk. However, risk needs to be considered in context. For instance, the detriment of uncontrolled seizure activity in women who have epilepsy may outweigh the relatively small increased risk of having a child with ASD.

Evidence that autism begins in utero has increased research into maternal factors that may contribute to the emergence of the disorder. When the mother has a fever in the third trimester of her pregnancy, this can increase the risk of ASD (Brucato et al., 2017). Research suggests that metabolic conditions related to underlying inflammation, including diabetes, obesity, and hypertension, may contribute to neurodevelopmental conditions including autism (Jones & Van de Water, 2019). Maternal antibodies may alter the development of the child's brain development during pregnancy. With obesity increasing, concern about the impact this will have on autism prevalence is important (Krakowiak et al., 2012). In total, these findings encourage doctors and women to closely monitor the mother's health in order to ameliorate autism risk for offspring.

Low birth weight and prematurity increase the risk of autism. A study by Lampi and colleagues (2012) found that children who were born small for gestational age were <2500 g (~5.5 lb) and/or were born at <32 weeks had an increased risk for ASD. It is unclear whether the risk increase is a result of differences in brain development based on these characteristics or the possibility that low birth weight, prematurity, and small size for gestational age share the same neurodevelopmental antecedents. Regardless, babies born small and/or early should be monitored for neurodevelopmental disorder as they age.

To offset ASD risk factors, there are some simple, proactive steps that women can take before and during pregnancy. Merely taking a prenatal vitamin 3 months before conception through the first month of pregnancy can reduce autism risk for genetically vulnerable children (Schmidt et al., 2011). Also, folic acid intake during this same time period appears to have a protective function for babies (Schmidt et al., 2012).

One myth that has been clearly debunked is that vaccines cause autism (DeStefano, Price, & Weintraub, 2013; Hviid, Hansen, Frisch, & Melbye, 2019). The notorious study by Andrew Wakefield that purported this connection has been definitively discredited (Eggertson, 2010). This unsubstantiated link caused substantial harm with the re-emergence of vaccine-preventable disease. Solid scientific support related to the safety of vaccines indicates the value of this preventative step for overall health. The professionals who work with families with young children must act as guides to discuss the most recent and accurate information in the autism field.

The heterogeneity of autism makes it logical that there is no single cause. It is likely that vulnerability in an individual's genetics leads to susceptibility for a factor or factors to trigger brain differences leading to symptoms. Since autism can have serious, lifelong consequences, researchers will continue to develop ways to identify the disorder early and find effective treatment for manifestations of ASD so that individuals can experience productivity and happiness.

Incidence and Prevalence

Incidence, or the measure of new cases per year, is best studied in disorders that have a clear onset. Because the age of onset in autism is usually unclear, accurate incidence rates are difficult to measure. Prevalence of autism is monitored regularly, and rates have increased steadily since the 1960s.

The World Health Organization (2019, November 7) indicates that 1 in 160 children in the world have ASD. However, many well-controlled studies indicate that the prevalence is much higher. In low- to middle-income countries, the prevalence is still unknown. In the United States, autism rates have increased from 1 in 150 in the year 2000 to 1 in 59 in the year 2014 (National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, 2019, September 3). The recent increase from 1 in 69 to 1 in 59 from the years 2012 to 2014 makes the prevalence 16.8 per 1000 children. The reasons for these rate increases are not entirely clear. However, it is likely that more awareness, stress on early identification, and greater availability of services have influenced diagnosis. ASD is ranked one of the most common causes of disability for children under 5 years of age (Baxter et al., 2015).

The likelihood of an autism diagnosis can vary by community and by demographic characteristic (Baio et al., 2018). For example, the CDC studied 11 communities using the Autism and Developmental Disabilities Monitoring Network for prevalence data. Five of the communities reported similar estimates of 1.3%-1.4%. However, the highest prevalence estimate came from a community in New Jersey with 2.9% of children reporting autism. More White children are given an autism diagnosis compared to Black and Hispanic children. However, these disparities were smaller when compared to previous years. Differences in ASD rates by demographic characteristic may be related, at least in part, to socioeconomic status. A study by Durkin and colleagues (2017) found that prevalence of autism increases with socioeconomic status and differences between high and low SES groups were constant over time. This may mean that children with fewer resources do not have access to diagnostic services and consequently do not have access to care. Finally, boys are about five times more likely to be identified with ASD than girls. This difference may be caused by differences in the biological structures of the male and female brain. Autism has been called "extreme maleness" by some researchers (Baron-Cohen, 2009; Baron-Cohen, Knickmeyer, & Belmonte, 2005). This theory relates to the population generality that the male brain is more geared toward "systemizing" and the female brain generally more geared toward empathizing.

The changes from the DSM-IV to the DSM-5 spurred concerns about whether new diagnostic criteria would prevent children who would have previously been given the diagnoses from qualifying for services or make children who already have the diagnosis no longer eligible. Shin Kim and colleagues (2014) evaluated a group of children ages 7-12 years using both DSM-IV and DSM-5 criteria to determine prevalence differences. They found that 99% of children with a previous autistic disorder (n = 114) ended up with a DSM-5 ASD diagnosis, with the remaining 1% (n = 2) receiving the DSM-5 SCD diagnosis. In children with a DSM-IV Asperger disorder diagnosis (n = 34), 91% (n = 31) were given the DSM-5 ASD diagnosis, 6% (n = 2) received the SCD diagnosis, and the remaining 3% (n = 1) had another psychiatric disorder. Finally, in children with a DS-IV PDD-NOS diagnosis, 71% (n = 41) had the DSM-5 ASD diagnosis, 22% (n = 13) received the SCD diagnosis, and 7% (n = 4) got other psychiatric diagnoses. In this study, the estimated prevalence of the DSM-IV autism diagnosis was nearly identical to the combined prevalence of the DSM-5 ASD plus SCD diagnoses. Although this study provides some reassurance that children who have

symptoms of autism will still be given access to care, the impact of the DSM-5 changes will continue to be monitored. Overall, prevalence information can be useful to inform policy and education of health care professionals so that all children on the spectrum have access to services that can improve their quality of life.

Signs and Symptoms

Children with ASD all have difficulty with social communication and interaction as well as RRBs. However, the severity and functional presentation of these symptoms vary widely from one individual to the next. In addition, these core behavior deficits do not encompass the entire picture for these complex human beings. Motor abnormalities, **sensory processing disorders**, feeding disorders, and co-occurring medical disorders (eg, seizures, sleep disturbances, GI problems) are only some of the additional concerns that arise in this population. Every child with autism displays a separate matrix of strengths and challenges with individual differences resulting from personality and experiences (National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention (CDC) 2019, August 27). Core symptoms, described below, are drawn from the DSM-5 diagnostic criteria for ASD.

Core Symptoms

The first of the core symptoms, difficulty in social interaction, includes challenges with both nonverbal and verbal skills. Many children with autism have limited use of eye contact and decreased use of social gestures (eg, pointing, shrugging, reaching arms up to be lifted). Additionally, children on the autism spectrum usually struggle to accurately interpret the body language and facial expressions of others. Children with ASD may not seek out others to share enjoyment, share interest in the same objects with peers, or look for approval or reassurance from parents. Moreover, individuals experience challenges in developing friendships with same-age peers. Those on the spectrum typically have trouble adapting to others across various contexts. For instance, knowing how close or far away to stand from others changes depending on the social situation, as does knowing when to use formal vs casual speech. These nuances are often confusing for individuals with autism (Winner, 2007).

For those with autism, problems with interaction may partially result from limitations in **theory of mind**, which is the ability to understand another person's thoughts, feelings, or intentions. It is how an individual "reads" someone's thoughts, understands another person's perspective on an issue, and predicts another's feelings (Baron-Cohen, 2000; Pedreño, Pousa, Navarro, Pamias, & Obiois, 2017). It is also how a person recognizes how their social behaviors are perceived by others. Without this ability, a person is not able to predict or understand the actions and emotions of others. Consequently, those on the spectrum typically have trouble adapting to others across various contexts (Winner, 2007).

Social communication and language impairments in the autism population include delays in language development as well as abnormalities in language use. Typically developing children progress through several communication milestones in their first few years (National Institute on Deafness and Other Communication Disorders (NIDCD), 2017). During a child's first year of life, an infant gazes at others with interest, begins babbling, and reacts to sounds and voices. By 12 months, gestures and first words emerge, and the child shows recognition of his or her name and some familiar words and phrases. By 2 years of age, the child has built a substantial vocabulary and combines words into two- to three-word phrases. However, children with autism frequently show delays in learning to speak, with some children unable to use words until their school-age years (National Institute of Health, National Institute on Deafness and Other Communication Disorders, 2016). In severe cases of autism, spoken language may not be acquired at all. A child's ability to respond to voices or comprehend verbal language (**auditory processing**) may also be impacted. If a child with autism has challenges with language comprehension, the child may not respond to their name, look toward or get items named, respond to questions, follow directions, or join in an activity into which they have been verbally invited. Furthermore, a child may imitate a speaker rather than responding, or show confusion or emotional dysregulation (Cawn & Osten, 2012).

Social impairments further affect these children's communication because they may not be motivated to communicate with others; are typically challenged to use or understand others' social communication cues such as intonation, gestures, body postures, and facial expressions to interpret meaning; and have difficulty maintaining the "give and take" of conversations (eg, a child may discuss only his or her topic of interest without allowing others to take a turn) (Fig. 3.3).

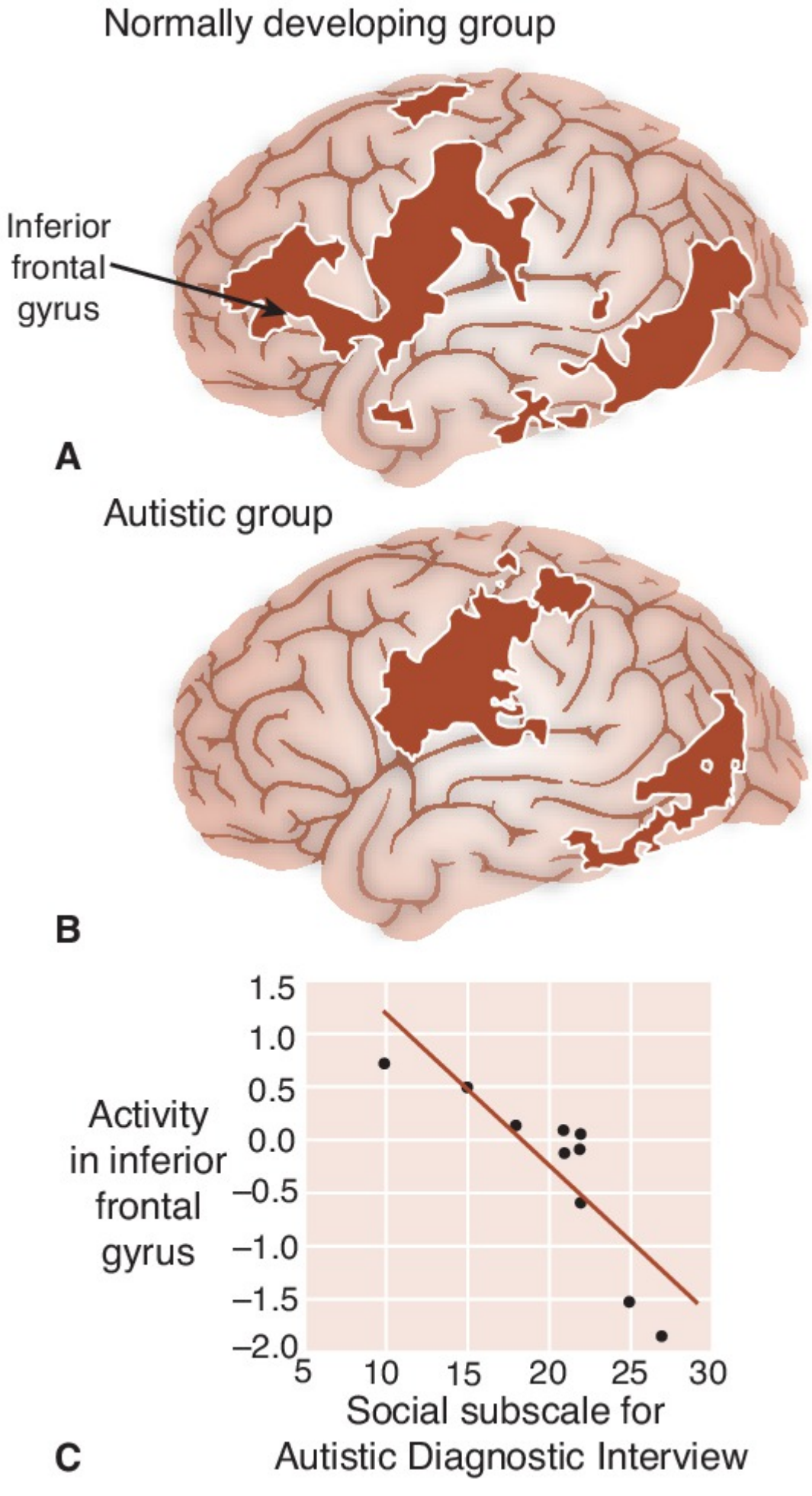
Normally developing group
Inferior frontal gyrus
A
Autistic group
B
Activity in inferior frontal gyrus
1.5
1.0
0.5
0.0
–0.5
–1.0
–1.5
–2.0
5
10
15
20
25
30
Social subscale for Autistic Diagnostic Interview
C

Figure 3.3 Functional MRI studies for normally developing preteens compared with high functioning age-/IQ-matched subjects with autism while imitating emotional facial expressions. The subjects with autism show less activity of the mirror neuronal network, particularly in the frontal cortex. (Adapted by permission from Nature. Dapretto, M., Davies, M. S., Pfeifer, J. H., Scott, A. A., Sigman, M., Bookheimer, S. Y., & Iacoboni, M. (2006). Understanding emotions in others: Mirror neuron dysfunction in children with autism spectrum disorders. *Nature Neuroscience, 9*(1), 28–30. Copyright © 2006 Springer Nature.)

The language of verbal children with autism often appears peculiar because of the presence of echolalia and pronoun reversal. **Echolalia** is the repetition, or echoing back, of another person's speech (Gernsbache, Morson, & Grace, 2016). Though all young children use echolalia as they are learning language, some children with autism retain this characteristic for longer, use it more persistently, or apply it in abnormal ways. Echolalia may be immediate, meaning that it occurs just after another speaker's utterance, or delayed, occurring significantly after hearing a speaker (eg, reciting an entire children's book from memory an hour or day later). Though it may appear unusual in conversation, children with autism frequently use this device in meaningful ways (eg, a child may recite lines from his favorite movie as a self-calming strategy when he feels anxious). For children in this population, as language comprehension increases, immediate echolalia decreases. Echolalia may serve a stepping stone for further language development in some children with autism.

Young, typically developing children initially struggle with correct use of pronouns. This difficulty can persist for longer in some children with autism (Gernsbache et al., 2016). For example, a child with autism may substitute the second-person pronoun "you" for the first-person pronoun "I." However, this behavior appears to resolve in many children with autism if and when their language comprehension ability increases. Although the development of this skill can be protracted, pronoun production problems are often remediated with continued development.

Restrictive and repetitive behaviors comprise the second core feature of autism. Individuals with ASD may exhibit an abnormal or intense preoccupation with routines or patterns. For instance, a child may line up blocks or toy cars for hours without playing with others or using these toys in pretend play. Additionally, the child may scream or cry in reaction to someone joining the child's play or moving the lined-up toys. Obsessions with specific topics are common, such as showing an unusual interest in elevators or numbers and letters (American Psychiatric Association, 2013a). Other common characteristics include rigidity in daily routines; abnormal, stereotyped behaviors such as repetitive hand-flapping or body rocking; and unusual, nonfunctional preoccupation with parts of an object (eg, spinning the wheels of a toy car without showing interest in any other type of play with this toy).

All children diagnosed with ASD demonstrate some combination of these social, social language, and behavioral symptoms. However, the range of severity and functional presentation of these symptoms is vast. The DSM-5 (American Psychiatric Association (APA), 2013a) provides additional specifiers to more accurately capture the impact of these symptoms on individual functioning that are useful for treatment planning and research purposes. First, the presence or absence of intellectual disability, language disorder, associated medical condition, and/or catatonia is captured. Then, severity level for the two core symptoms is classified on a three-point scale from mild to severe. Examples of function within this hierarchy, adapted from the DSM-5, are included in Table 3.1.

TABLE 3.1 Levels of Severity Within the Autism Spectrum

Level 1: Mild to Moderate on Autism Spectrum	Level 2: Severe to Moderate on Autism Spectrum	Level 3: Very Severe on Autism Spectrum
Has use of sentence level language but often has difficulty using language functionally without support	May have some use of language but verbal and nonverbal communication is limited	Often does not have verbal language and/or use of language is very limited and atypical
May have less interest in social engagement than typically developing peers or is sometimes described as odd or atypical	Difficulty with meaningful engagement with others and attempts are often viewed as odd or unusual	Does not seek engagement with others except occasionally to get needs met
Attempts to make friends may fail when social cues are missed	May attempt conversation around a restricted interest like trains, numbers, or electricity and unable to shift to a topic of mutual interest	When attempting to communicate a need, does so with unusual and often ineffectual attempts Frequently ignores or does not respond to even very direct attempts from others to engage socially
Is most comfortable in a well-worn routine	Insistence on sameness and inflexibility is noticeable to others outside of the child's immediate social circle	Extreme inflexibility and resistance to change. May have self-injurious behavior with changes in routine
This child benefits from intervention to engage in meaningful occupations at a greater depth and to improve the richness of social relationships	This child requires substantial intervention to engage in meaningful occupations	Even with intervention, this child's autism very significantly interferes with participation in meaningful occupations

The DSM-5 (APA, 2013a) provides for one umbrella diagnosis of ASD and offers new categorical descriptors to pinpoint the specific presentation of this disorder for an individual. Since the degree of functional impairment varies widely for individuals with autism, this has the potential to allow for more individualized treatment. However, the move from having five diagnostic categories (ie, Asperger syndrome, pervasive developmental disability not otherwise specified, childhood disintegrative disorder, Rett syndrome, and autism disorder) is not without controversy. Detractors are concerned that diagnosis may be more restricted, and that people who are newly diagnosed with autism may not fit the new criteria and may not have access to services and support previously provided (Kulage, Smaldone, & Cohn, 2014). However, recent research provides optimism related to this concern (Shin Kim et al., 2014). The impact of this major change will be followed for years to come, and the evaluation of

the usefulness of this new diagnostic structure will be determined by the successful identification of children in need of services to mediate the functional limitations that are often caused by symptoms of autism.

Co-occurring Conditions

The core symptoms of autism cause many challenges to engagement in everyday life for individuals with ASD and the people who love and support them. These challenges can be compounded by co-occurring conditions that are commonly found in individuals ASD. Sensory processing disorder and possibly related fine and gross motor deficits, food selectivity, sleep disturbances, and GI issues can create additional stressors that profoundly influence the quality of life in this population.

Sensory Processing Disorder

Hypo- and hyperreactivity to sensory information and/or unusual preoccupation with the sensory aspects of the environment are included in the diagnostic criteria in the DSM-5 under RRBs. However, many children with autism have sensory processing problems that go beyond hypo- and hyperreactivity. Also, beginning with the pioneering work of the prominent occupational therapist Jean Ayres, sensory processing disorders are of special interest to occupational therapists and are therefore covered separately here.

In the 1970s, Jean Ayres, an occupational therapist with training in neuroscience and education, developed the theory of sensory integration to explain learning differences in children who struggled with interpreting sensation from their bodies and the environment (Ayres, 1994). Today, estimates of atypical sensory processing in children with ASD are high, with some studies indicating a prevalence as high as 95% of this population (Baranek, David, Poe, Stone, & Watson, 2006; Ben-Sasson et al., 2009).

Sensory processing disorders are organized into several subtypes that manifest in difficulty modulating sensory experiences in order to maintain a calm alert state (ie, under- or hyporesponsivity [SUR], over- or hyperresponsivity [SOR], sensory seeking [SS] or craving [SC]), difficulty with motor skills (ie, dyspraxia and postural instability), and difficulty with discrimination of the quality of sensory stimulus (Miller, 2014). The severity and type of sensory problems in an individual with ASD highly impacts that person's success in daily functioning. Table 3.2 contains the sensory processing disorder subtypes with behavioral examples and the connection between the subtype and children with ASD.

TABLE 3.2 Sensory Processing Disorder in Children With ASD

Sensory Processing Disorder Subtype (Miller, 2014)	Behavioral Example	Implications Specific to ASD
Sensory Modulation Disorder		
SOR Sometimes called "sensory defensiveness." Behavioral response to stimuli that is more intense and longer lasting than expected	Distress with bathing and grooming Difficulty tolerating crowded places Meltdowns with unexpected noises	Strong correlation with symptoms of anxiety in children with ASD (Lane, Reynolds, & Dumenci, 2012) Linked to decreased participation in activities outside the home for children with ASD (Little, Ausderau, Sideris, & Garanek, 2015) Associated with decreased daily living skills (Ismael, Lawson, & Hartwell, 2018)
SUR Take longer to respond to stimuli or do not notice sensory information that is obvious to others	Does not respond to injuries Often unaware of what is going on in the environment Leaves food on face or clothing twisted on body	Is one early predictor of joint attention and social communication skills in young children with ASD (Baranek et al., 2013; Watson, Baranek, Roberts, David, & Perryman, 2010) May be one predictor of social responsiveness (Crasta, Davies, & Gavin, 2019)
SC Insatiable craving for sensory stimuli that interferes with functioning	Touches everything Is always moving Puts nonfood items in mouth	Has been associated with academic underachievement due to difficulty with sustained attention (Ashburner, Ziviani, & Rodger, 2008)
Sensory-Based Motor Disorder		
Dyspraxia Difficulty with motor planning (praxis)	Is delayed in the development of motor skills including learning to walk and/or learning to ride a bike Struggles with multistep activities, like getting dressed or making a sandwich Novel motor activities are especially difficult	Motor coordination deficits are pervasive and some hypothesize that this may be an underlying feature of autism (Fournier, Hass, Naik, Lodha, & Cauraugh, 2010; MacNeil & Mostofsky, 2012; Roley et al., 2015) Have substantial dysfunction in praxis skills when compared to typically developing individuals (MacNeil & Mostofsky, 2012) Somatopraxis (ability to organize own body action using tactile and proprioceptive information) deficits are associated with social participation, possibly due to difficulty with imitation (Roley et al., 2015)

Sensory Processing Disorder Subtype (Miller, 2014)	Behavioral Example	Implications Specific to ASD
Postural disorder and/or poor postural strength and stability	Tires easily Has decreased balance and requires support during static motor tasks	Development of postural stability is delayed and can fail to mature to adult levels in children with ASD (Minshew, Sung, Jones, & Furman, 2004; Travers et al., 2015)
Sensory Discrimination Disorder Difficulty discriminating between the qualities of distinct sensory stimulus can occur in one of more of the senses	Difficulty with stereognosis Struggles to tell the difference between a "b" and a "d" Cannot distinguish between speech sounds	Contradictory evidence (Dickinson & Milne, 2014) for both enhanced sensory discrimination (Bertone, Mottron, Jelenic, & Faubert, 2005; Bonnel et al., 2003) and reduced tactile discrimination (Puts, Woodka, Tommerdahl, Mostofsky, & Edden, 2014)

Fine and Gross Motor Impairment

As described in Table 3.2, children with autism have motor impairments that may be related to their ability to use sensation to plan, guide, and execute movement. Fine and gross motor impairments include problems in skilled movement, hand-eye coordination, speed, praxis and imitation, posture, and balance (Buja et al., 2018; Dawson & Watling, 2000). A particularly debilitating abnormality in individuals with autism includes deficits in motor imitation skills despite intact perceptual and motor capacities, which are most apparent in younger groups of children (Wadsworth et al., 2017; Williams, Whiten, & Singh, 2004). Motor skill deficits may be one underlying factor for differences in language development in children with ASD (Bedford, Pickles, & Lord, 2015).

Food Selectivity

Children with ASD often have an extremely self-restricted diets and disruptive behaviors during mealtime (Schreck, Williams, & Smith, 2004). These children may refuse to eat more than 5-10 very specific foods. For example, a child with ASD might insist on eating a specific brand and shape of macaroni and cheese at every meal. They may insist that food be arranged on the plate a particular way with foods not touching other foods. They may refuse to eat any foods from a particular food group and have meltdowns and/or gag or vomit when encouraged to eat a nonpreferred food. It is common for children with autism and **food selectivity** to refuse to eat any fruits and vegetables (Suarez & Crinion, 2015). This behavior causes worry and stress for caregivers and can make mealtime unpleasant for the entire family (Suarez, Atchison, & Lagerwey, 2014). Also, depending on the degree of food refusal, food selectivity can threaten health and development due to lack of consumption of essential nutrients (Bandini et al., 2010).

Sleep Disturbance

Poor quality sleep is common in the ASD population and up to ~70% have some form of sleep disturbance (Souders et al., 2009). Children with autism may resist going to bed and require extensive time to fall asleep (Liu, Hubbard, Fabes, & Adaom, 2006). They may have long periods of awakenings, awaken many times during the night, or wake up very early in the morning. Causes for sleep disturbance are unknown but may be related to abnormalities in the areas of the brainstem that regulate sleep and in melatonin levels (Malow et al., 2012). Children with ASD who are

poor sleepers have more difficulty with daytime disruptive behavior than children who sleep well (Malow et al., 2012; Wang et al., 2015). In addition, sleep disturbance in children with ASD has implications for the sleep quality of the entire family. This can lead to additional stress on a family already struggling with core symptoms of autism.

Diagnosis

Autism is now diagnosed at a younger age than ever before because characteristics of autism have become more defined and better recognized. Researchers have employed at least two research methods to identify early indicators of ASD (Boyd, Odom, Humphreys, & Sam, 2010). Retrospective studies, using home videos of children later diagnosed with autism, and prospective studies of the younger siblings of children with autism (due to the genetic likelihood of siblings both having autism) have been used to develop an understanding of early behavioral warning signs. Key indicators include delay or disorder in social behaviors like making eye contact, smiling socially, or responding to one's name. Coordination of verbal and nonverbal communication (eg, verbalization with a point or eye gaze, showing objects to others) is often difficult or absent in very young children that are eventually diagnosed with autism. Finally, atypical play, including spinning objects or preoccupations with parts of objects, can be an early indicator. Receiving a diagnosis at an early age can be life changing (Elder, Kreider, Brasher, & Ansell, 2017). The sooner the disorder is recognized, the greater the chances are that the child can make dramatic reductions in symptoms and gains in learning (Fernell, Eriksson, & Gillberg, 2013). In addition, parents are able to receive valuable support and education from professionals and other family members.

A child is usually referred for an assessment because those who interact closely with him (eg, family members, pediatrician, teachers) may observe warning signs either specific to autism or to a development delay in one or more areas. When these concerns become apparent, the child is initially screened, usually by primary care providers or early child care professionals, to look for the "red flags" that may indicate autistic behaviors. Published screening instruments for children with autism include the following:

- The Checklist for Autism in Toddlers (CHAT) (Baron-Cohen, Allen, & Gillberg, 1992)
- The Autism Screening Questionnaire (ASQ) (Berument, Rutter, Lord, Pickles, & Bailey, 1999)
- The Screening Tool for Autism in 2-Year-Olds (Stone, Coonrod, & Ousley, 2000)
- Australian Scale for Asperger Syndrome (Garnett & Attwood, 1998)
- Pervasive Developmental Disorders Screening Test, Stage 1 (PDDST-I) (Siegel, 1998a)
- The Modified Checklist for Autism in Toddlers (M-CHAT) (Robins, Fein, Barton, & Green, 2001).

If the child does not pass the screening, this indicates that enough warning signs are present to warrant a thorough assessment for autism. Since each child on the spectrum displays a unique matrix of strengths and weaknesses, no two children will look the same. Therefore, an assessment with clinicians who are experienced in identifying the characteristics of autism is critical. Additionally, a thorough diagnosis with a team of professionals can gather insight into each child's skills across several areas of development, which, in turn, helps with intervention planning. An autism evaluation team frequently consists of a developmental pediatrician, a psychologist, a speech and language pathologist, and often an occupational therapist. The following elements should be included in every sensitive, comprehensive evaluation of a child with autism:

1. *History*: Though autism is not known to result from complications during pregnancy, it is important to discuss any unusual pre- or perinatal events to rule out other disorders. Because of autism's genetic implications, it is important to determine if other family members have been diagnosed with autism, psychiatric concerns, or developmental disorders. The history portion should also include questioning about autism-specific behaviors in the areas of social, language, behavioral, play, cognitive, and sensory processing abilities (Center for Disease Control, nd).
2. *Medical history*: During this portion of the assessment, parents report when their child's developmental milestones were reached (eg, what age their child said his or her first word, learned to walk), if regression of developmental skills occurred at any point, or if any other medical problems are occurring (eg, psychiatric, sleeping, or eating problems) (Filipek et al., 1999; Hueta & Lord, 2012).
3. *Physical/neurological examination*: Other illnesses such as fragile X syndrome, tuberous sclerosis, or congenital rubella need to be ruled out, since these disorders may look similar to autism (National Fragile X Foundation, nd; Rapin, 1997). The physician also checks for other medical illnesses (eg, GI disorders, ear infections), measures head circumference, gives a general physical examination, examines mental status,

verifies that cranial nerves function normally, and performs a motor examination (Filipek et al., 1999; Hueta & Lord, 2012).

4. *Parent interviewing*: Many diagnostic tools are available to gain parents' insight into their child's autism-specific behaviors. A clinician should also ask parents about their overall impression of their child, since more general questions may reveal further insight beyond the scope of these tools (Hueta & Lord, 2012; National Research Council, 2001). Parent interview tools include the following:

   - *The Autism Diagnostic Interview*: Revised (Lord, Rutter, & LeCouteur, 1994)
   - Functional Emotional Assessment Scale (FEAS) (Greenspan, DeGangi, & Wieder, 1999)
   - The Gilliam Autism Rating Scale (GARS) (Gilliam, 1995)
   - The Pervasive Developmental Disorders Screening Test, Stage 2 (PDDST-II) (Siegel, 1998b)

5. Tests in language development. Examples of tools include the following:

   - Preschool Language Scales (PLS) (Zimmerman, Steiner, & Pond, 2011)
   - Clinical Evaluation of Language Fundamentals (CELF) (Wiig, Semel, & Second, 2013)

6. Tests in cognitive development:

   - Mullen Scales of Early Learning (MSEL) (Mullen, 1995).
   - Wechsler Intelligence Scale for Children (WISC) (Wechsler, 2003)
   - Bayley Scales of Infant and Toddler Development, Third Edition (Bayley-III) (Bayley, 2005).

7. Tests in other developmental areas including sensory processing, motor, and adaptive skills.

   - Sensory Profile 2 (Dunn, 2014)
   - Sensory Processing Measure (Parham, Ecker, Miller Kuhaneck, Henry, & Glennon, 2007)
   - Peabody Developmental Motor Scales, Second Edition (PDMS-2) (Folio & Fewell, 2000)
   - Quick Neurological Screening Test 3R (QNST-3R) (Mutti, Martin, Sterling, & Spalding, 2017)
   - Vineland Adaptive Behavior Scales (Sparrow, Cicchetti, & Balla, 2006)

8. Formal and informal observation of the child interacting with others. This is considered one of the most valuable pieces of the evaluation process, since it typically reveals the qualitative impairments of the child (such as lack of eye contact, limited initiation of interaction, or difficulty transitioning between tasks). A diagnostic instrument should be used that examines autistic behaviors while the clinician observes the child's interests and interactions with others. The Autism Diagnostic Observation Schedule (ADOS 2) (Lord et al., 2012) is considered a gold standard instrument for this purpose.
9. *Audiological testing*: An audiological evaluation is necessary to rule out hearing disorders. Children with autism are often unresponsive to verbal auditory stimuli, and it is important to determine that this behavior is not caused by a hearing impairment. If hearing loss co-occurs with autism, language comprehension may be further impacted than with a diagnosis of autism alone (Beers et al., 2014).
10. *Other testing*: Certain tests may prove beneficial to specific circumstances. For instance, an electroencephalograph (EEG) may be needed if the child is suspected of having seizures (Hartley-McAndrew & Weinstock, 2010). Some children are assessed through university studies, and MRIs are typically included in these autism assessments (Chen et al., 2011). Genetic testing may be appropriate for parents who are considering having another child (Jeste & Geschwind, 2014).

Course and Prognosis

Since the symptom variability within the autism spectrum differs widely among individuals, the course and prognosis depend on an individual's presentation. Many children with autism can have significant functional improvement with treatment. However, a lot of individuals tracked longitudinally retain some characteristics of the diagnosis throughout the span (Levy & Perry, 2011). Several prognostic indicators can partially predict functional outcomes for individuals with this disorder.

Intellectual Disability

Some children with autism also have decreased mental capacity limiting their ability to plan, problem solve, think abstractly, and/or comprehend complex ideas (The Arc, n.d.). Intellectual abilities are measured with assessments that provide an intelligence quotient (IQ) with scores below 75 indicating significant impairment in thinking skills. The prevalence of intellectual disability in children with ASD is likely in the 15%-20% range (Fernell et al., 2013) with estimates ranging as high as 68% (Yeargin-Allsopp et al., 2003). IQ is the most consistent predictor of later adaptive functioning in children with autism (Fernell et al., 2013). For example, the previous DSM (IV) included a diagnosis of Asperger's, which was characterized by social and behavioral challenges *without* presentation of cognitive deficits. Longitudinal studies suggest that prognosis is better for the Asperger disorder with more individuals able to function independently in employment and self-sufficient living (American Psychiatric Association, 2000).

Language

A child's language level is also a strong predictor of functioning over the course of the lifespan. A child's ability to spontaneously, meaningfully, and consistently combine words into phrases or sentences before 5 years of age is a reliable prognostic indicator of cognitive, language, adaptive, and academic achievement measures (Levy & Perry, 2011; National Research Council, 2001). It is important to observe if the child spontaneously constructs sentences rather than echoes others' utterances (echolalia) or uses memorized chunks of language to communicate. In addition, a child's use of joint attention has been found to be a predictor of language outcome. **Joint attention** is the ability to use eye contact and gestures in order to share experiences with others. Children who fail to use early gestural joint attention (eg, failing to point at an object and to turn to his mother to determine if his mother shares his interest) seem to struggle in the development of meaningful language. In Lord and McGee's review of autism research (National Research Council, 2001), findings implied that early joint attention, symbolic play, and receptive language were strong predictors of a child's future outcomes. Another study in this review examined severity of repetitive, stereotyped behaviors and social symptoms and found that the severity later predicted adaptive functioning.

Motor Skills

Early success in hand-eye coordination may predict vocational abilities later in life. Fine motor skills also predicted later leisure pursuits (National Research Council, 2001). Children with autism who displayed a definite hand preference performed significantly better on motor, language, and cognitive tasks (Hauck & Dewey, 2001; Kobylinska et al., 2017). The ability to imitate body movements has been linked to expressive language development, and imitation of actions with objects predicted later levels of play abilities (National Research Council, 2001). The more established behavioral challenges become without intervention, the more these problems persist and worsen.

Medical/Surgical Management

Overall, treatment for autism does not heavily rely on medical intervention, and surgical interventions are not practiced for this disorder. Intensive early intervention is most effective in reducing problem behaviors while increasing language, social, sensory, motor, and cognitive skills (Franz & Dawson, 2019).

A variety of approaches are available, ranging from highly structured to naturalistic. Because no two children with autism present with the same set of symptoms, no one treatment plan is successful for all children with autism. Effective intervention must account for the child's individual strengths and challenges and must consider functional skills to be generalized across a variety of settings in the child's life. Substantial literature supports early intervention for children 0-3 years as the most beneficial time to connect new pathways for more appropriate functioning and behavior, though an individual may continue to make substantial gains following this period of development (National Research Council, 2001). Because it is beyond the scope of this book to discuss these approaches, this chapter focuses on current knowledge of medical interventions meant to accompany intervention techniques.

Pharmacologic Therapies

Use of medication does not cure the core social, language, and repetitive behavioral deficits of autism. However, many children with ASD are prescribed psychotropic medication to address comorbid diagnoses like bipolar, psychoses, and depression and comorbid symptoms like aggression, hyperactivity, and mood dysregulation. A study by Madden and colleagues (2017) found that ~50% of children with autism had visited a mental health professional

compared to 9% of children without ASD. In addition, almost one-half of children with autism were prescribed a psychotropic medication at some time in their history. These medications included those to treat ADHD, antipsychotics, antidepressants, and mood stabilizers.

Research suggests that pharmacologic intervention, used appropriately, may reduce behavioral dysregulation (LeClerc & Easley, 2015). For instance, risperidone, an atypical antipsychotic agent, shows promising results in research for reducing tantrums, irritability, aggression, and self-injurious and repetitive behaviors. However, caution is warranted due to side effects such as weight gain, sedation, and extrapyramidal symptoms with these medications (Madden et al., 2017). Because autism is likely a genetic disorder determined before the child is born, pharmacologic treatments cannot "undo" this disorder. Therefore, these treatments are not intended to replace educational services but rather to supplement them.

Children with autism often have significant difficulty with sleep (Malow et al., 2012). It is common for trial of medication in order to alleviate this issue. These medications include melatonin and α-agonists (eg, clonidine). However, these medications may have some adverse effects, including increased challenging daytime behavior. Close medical supervision is needed when these medications are prescribed, and nonpharmacological strategies (eg, soothing bedtime routine, room darkening) are preferred when possible.

Medical Conditions

Medical intervention is necessary for any co-occurring medical conditions, such as seizures or GI disorders. Children with autism need to be monitored closely for behaviors that may reflect a medical condition, particularly if they are nonverbal and cannot describe any pain or discomfort that they may be experiencing. After a condition is diagnosed, the child should receive a complete medical workup and treatment through a physician who specializes in the child's comorbid diagnoses.

Complementary and Alternative Medicine

Complementary and alternative medicine is defined as a "broad set of health care practices that are not part of that country's own tradition and are not integrated into the dominant health care system" (World Health Organization (WHO), 2019). This type of treatment continues to be popular as a supplement to behavioral and educational services. Since clinicians are often unable to definitively state the reason for a child's autism, families often turn to therapies that are not conventional (Levy & Hyman, 2015).

Those who support complementary and alternative medicine believe the methods target underlying medical difficulties, such as GI and sleep disorders, which are not addressed through behavioral or educational intervention. The goal of many of the complementary and alternative medicine treatments is to aid in the associated problems of autism, rather than cure the disorder. Statistics reveal that over a lifetime, up to 95% of children with autism may be using some form of complementary and alternative medicine (Hofer, Hoffmann, & Bachmann, 2017). However, evidence for the effectiveness of these interventions is generally low and there are indications that many of these practices can be harmful. This is particularly concerning as families report that they rarely consulted with their physician for information about these practices (Levy & Hyman, 2015). Table 3.3 summarizes the results of a systematic review of the effectiveness of complementary and alternative medicine (Levy & Hyman, 2015). Types of treatment are included if they had at least a "low" level or evidence. Treatments with no direct research were excluded from this table.

TABLE 3.3 Summary of Evidence for Complementary and Alternative Medicine

Supplements	Mind-Body Treatment	Biological Treatment
Vitamin C Preliminary evidence of improved sensory, sleep and GI symptoms	Auditory integration Benefit not demonstrated. Low risk	Anti-infectives (eg, antibiotics, antivirals) No proven benefit. Not FDA approved
DMG No benefit. Parents report hyperactivity side effect	Acupuncture No evidence for use in ASD. Risk for injury in uncooperative children	Immunoglobulins No proven benefit. Not FDA approved
Omega-3 No evidence of improvement in children with autism. Some preliminary benefit for other related populations	Equine therapy Potential for benefit in symptom reduction or as a community activity. Risk low if proper riding precautions	Chelation No proven benefit. High level of risk. One death reported
Vitamin B12 No evidence of overall improvement in GI symptoms or food allergies	Massage/Qigong Parent report of benefit in sensory and behavioral outcomes. Low risk involved.	Oxytocin Preliminary indication of possible increase in emotional recognition. Not FDA approved
Melatonin Good physiologic evidence for improved sleep with few side effects	Music Therapy May increase verbalization. Low risk involved	Secretin No benefit. Stress risk with intravenous route. Not FDA approved
	Yoga May improve behavior but more research is needed. Low risk involved	Gluten-free/casein-free diet Benefit not proven. Can cause nutritional deficits without professional consultation
		Hyperbaric oxygen therapy No proven benefit. Not FDA approved
		Transcranial magnetic stimulation May improve social relatedness and anxiety but more research is needed. Not FDA approved. Has the potential for risk

Data from Levy, S. E., & Hyman, S. (2015). Complementary and alternative medicine treatments for children with autism spectrum disorders. *Child and Adolescent Psychiatric Clinics of North America, 24*(1), 117–143. doi: 10.1016/j.chc.2014.09.004

Supplements

Families of children with autism may turn to supplements, like vitamins and minerals, because of the GI and food selectivity issues that many children face (Interactive Autism Network, nd). Proponents of using supplements claim that these supplements improve their child's ability to obtain/absorb nutrients. Frequently used supplements include vitamin C, dimethylglycine (DMG), omega-3, vitamin B12, and melatonin (Levy & Hyman, 2015). Melatonin is one supplement with relatively promising results for improving sleep. More research is needed in this area.

Mind-Body Treatment

The connection between the mind and body has been documented in research. The theory is that changing body systems and structures can change the mind and promote health. There is some limited evidence for the potential benefit of treatments like equine therapy, music therapy, massage, and yoga (Levy & Hyman, 2015). All of these treatments are low risk. However, as with most complementary and alternative medicine treatments, more research is needed.

Biologically Based Treatment

Biomedical treatments include off-label prescription use and other treatments that require a licensed clinician to administer (Levy & Hyman, 2015). These medications have the most potential for negative side effects. Some of these may be appropriate for clinical trial but should be used with caution and under close medical supervision.

- *Secretin*: The human body naturally produces secretin, which is a hormone produced in the small intestine that stimulates secretion by the pancreas and liver. The use of extra secretin through injections became a popular complementary and alternative medicine treatment before it was scientifically analyzed, and studies now show few changes for children with ASDs who have used the extra hormone injection (Levy & Hyman, 2003).
- *Chelation*: Because mercury poisoning produces symptoms similar to those seen in autism, mercury and other heavy metals have been suggested as causes of autism. Though research has not found a link between mercury and autism, those who believe their children have experienced metal poisoning may choose chelation, a process to remove toxins from a child's system (Levy & Hyman, 2003). However, this is a very high-risk procedure without proven benefit and with one death reported (Levy & Hyman, 2015).
- *Antibiotic treatment*: Immune system dysfunctions and antibiotic treatments have been targeted as possible causes of autism, and those who believe in these theories use further antibiotic treatment to alter the course of the symptoms in autism (Levy & Hyman, 2003). However, there has been no proven benefit (Levy & Hyman, 2015).
- *Antifungal treatment*: Yeast overgrowth in the colon is hypothesized to cause many medical disorders, including autism, with a low-sugar diet and the use of probiotic agents (which encourage helpful intestinal bacteria) used as treatments (Levy & Hyman, 2003). Similar to the treatments discussed above, there has been no proven benefit from this approach (Levy & Hyman, 2015).

Gluten-Free, Casein-Free Diet

Parents and professionals who support the theory that **gluten** and **casein** negatively impact a child's development recommend a diet that completely eliminates these products (McCandless, 2003). Parent reports of improvement have been inconsistent, as some parents claim to see significant improvements in their child's behavior and/or developmental skills (eg, improved eye contact), while others report that no change occurs by implementing this diet. Data are currently limited and inconclusive. One review of five available articles researching the effectiveness of gluten- and casein-free diets for behavioral symptom management found no positive effect from this diet (Hurwitz, 2013). In addition, nutritional deficits can result from this diet without professional consultation.

The effectiveness of complementary and alternative medicine stands largely unproven and controversial, though attempts are underway for further studies since some parents report positive results by using these methods. Complementary and alternative medicine treatments are intended to supplement behavioral and educational services rather than to replace them. Clinicians are currently encouraged to use an empathetic stance with families providing complementary and alternative medicine to their children, although no alternative method should be administered without the guidance of an experienced physician.

Impact on Occupational Performance

The presence or absence of intellectual and language impairments, the severity of the autism symptoms, and the influence of coexisting sensory, psychiatric, and motor deficits all have significant implications on the degree of functional impairment present in an individual with an ASD diagnosis. The following describes associated client factors—specifically, body structures and functions—that are commonly observed.

Client Factors: Body Structures and Function

Global Impairment

Autism is considered a global impairment, meaning that it does not reflect damage as a result of one specific lesion in the brain. Since autism likely affects several regions and functioning of the brain, this global impact means that autism impairs multiple areas of a child's development. As a result, all areas of occupation are often impacted by this disorder.

Specific Mental Functions

Each individual with autism presents with different areas of strengths and weaknesses; therefore, individuals with an ASD diagnosis show various combinations of concerns in their mental skills. Concerns in this area can range from very severe, where the individual will need constant support and supervision throughout the lifespan, to very mild, in which the individual can achieve complete financial and interpersonal independence. However, despite the great variability in symptom presentation, several areas of mental function are commonly impaired to a greater or lesser extent depending on the level of ASD severity.

Children with autism often have difficulty regulating their own emotions and often experience features of or possibly comorbid psychiatric disorders (Leyfer et al., 2006; Mattila et al., 2010). The most common issue is specific phobia. Children with autism may experience phobias of typically innocuous things like crowds, bridges, or loud noises. In addition, attention deficits are nearly universal in this population. Unique patterns of attention include disorganization, intense preoccupation for preferred, self-initiated activities for unusual lengths of time, and inability to inhibit distraction (Rapin, 1991; Uzefovsky, Allison, Smith, & Baroon-Cohen, 2016). Features of obsessive-compulsive disorder are also often present in many children with autism. They may need to perform a routine a certain way, like walking through the same side of a double door to get to the lunchroom at school, or not be able to move on until a task is completed, like needing to pick all of the dandelions on the playground before they can play. Finally, ~10% of children with autism also have symptoms of major depression with episodes of things like sadness, hopelessness, crying, and/or flat affect (Leyfer et al., 2006). Difficulty regulating emotion in this population can lead to emotional lability (eg, swinging from laughing to crying without apparent reason), heightened anxiety with temper tantrums, and/or aggressive behaviors such as hitting and biting, self-injury (eg, head banging, hitting self), or self-stimulatory behaviors (body rocking).

A child with autism often shows a scattered pattern of memory functions (Kercood et al., 2014). Overall, "memory performance of individuals with autism becomes increasingly impaired as the complexity of the material increases" (Minshew & Goldstein, 2001, p. 1099). Children with autism also use fewer organizational strategies, relying on stereotyped rules regardless of the task's complexity (Minshew & Goldstein, 2001). Therefore, a child with autism performs more poorly on tasks with higher complexity. However, certain areas of memory remain intact, particularly in the areas of visual and rote memory. **Rote memory** describes the memorization and use of previously heard chunks of language rather than the spontaneous generation of language.

In the area of perceptual functioning, individuals with ASDs demonstrate anomalies in perceptual, sensory, and motor functions (Casartelli, Riva, Villa, & Borgatti, 2018). Integration of perceptual and sensorimotor information allows individuals to respond appropriately with physical and emotional responses. In autism, this integration does not occur in the same efficient way; therefore, this population of individuals has difficulty responding with typical emotional and behavioral responses to the sensory stimuli around them. Between 30% and 100% of individuals with autism demonstrate deviant sensory-perceptual abilities (Dawson & Watling, 2000).

As mentioned above, cognitive impairments or intellectual disabilities are common in, though not universal to, the autism population. The Center for Disease Control (2019) estimates that 31% of children with autism have an IQ <70, qualifying for an intellectual disability. However, 44% have IQ scores that are in the average to above average range. Individuals with cognitive deficits typically demonstrate scattered skills; in other words, they may present with strong skills in some areas with significant concerns in other areas. These children often have difficulty

sequencing a series of items, imitating the actions and words of others meaningfully, generalizing concepts across a variety of situations, demonstrating **theory of mind**, and playing with toys appropriately and symbolically (National Research Council, 2001). Studies examining higher level cognitive functions have revealed that executive functioning skills such as forward planning, cognitive flexibility, and the use of assistive strategies (eg, creating a mnemonic such as a rhyme to assist in remembering information) in learning are impacted for those who have autism (Gordon, 2002).

As previously described, children with autism typically show language delay. Deviations from typical language are noted across all diagnostic categories. Children on the autism spectrum typically have limitations in using language in appropriate contexts and for social purposes. Some children may be highly verbal and articulate though literal or rigid, echolalic, and repetitive; other children may remain nonverbal or use very little speech (National Institute of Health, National Institute on Deafness and Other Communication Disorders, 2016). The goal for these children may be to learn to communicate using gestures or a picture system. Additionally, comprehension of language is affected and exacerbated by a decreased motivation to use language for interacting with others. Often, these children are challenged to understand what topics interest others and have difficulty interpreting nonverbal language such as gestures and facial expressions, as well as abstract language such as jokes or idioms. For those who are verbal, intonation of their voices may sound unnatural as a result of a singsong or monotone pattern. Speaking at unusually loud volumes is another unusual characteristic of children with autism. Though the words may be clear, content may be memorized chunks of language or may focus on topics that are not relevant to a conversation partner. Echolalia is also common for verbal children.

Sensory Functions and Pain

As Dunn describes it, "The experience of being human is imbedded in the sensory events of everyday life. When we observe how people live their lives, we discover they characterize their experiences from a sensory point of view" (2001, p. 608). Sensory processing is particularly challenging in individuals with autism. If a child's sensory system does not interpret stimuli in a typical way, it is easy to understand why this individual may react to the world differently. Sensory processing problems should be addressed through early intervention to reduce these abnormal behaviors (Dunn, 2001; National Research Council, 2001).

Visual perception is usually an area of relative strength and may be used to compensate for challenging areas. For instance, the integration of vestibular, visual, and somatosensory afferent systems is needed to maintain upright postural stability. Molloy, Dietrich, and Bhattacharya (2003) measured the postural stability in children with autism and found that these children relied on visual cues to help them maintain stability. When these visual cues were omitted, children had difficulty maintaining their upright balance and reducing their sway. One area of visual processing that is consistently impaired is integrating details of a figure into a whole (Deruelle, Rondan, Gepner, & Tardit, 2004). For instance, if given a line drawing of a house made up of geometric shapes, these children focus on the shapes rather than seeing the image of a house.

Functions Related to the Digestive System

Children with autism may be at a higher risk for experiencing GI problems such as reflux or gastritis, with persistent gas, diarrhea, and constipation also frequently reported (McElhanon et al., 2014). Additionally, hypotheses exist regarding functioning of the digestive system, such as increased intestinal permeability that allows absorption of morphine-like compounds from gluten and casein. The buildup of these substances theoretically results in the social withdrawal and stereotypical behaviors seen in autism, but as previously described, this theory has not been proven (Levy & Hyman, 2003).

Urinary and Reproductive Functions

In children with autism, the urinary tract is usually typical in its structure and function. However, for those children with cognitive and sensory impairments, toilet training is often complicated. Children with autism often learn to toilet train at a later age than typically developing children; evidence problems such as fear, pain, confusion, frustration, and constipation when learning to train; may not get appropriate cues from their bodies when they need to go; and experience difficulty when a change of routine occurs or when entering an unfamiliar bathroom (Autism Speaks, nd).

Because individuals with autism have difficulty interacting, inappropriate sexual behavior is also a concern. Close relationships with others are challenging for these individuals; therefore, reduced person-oriented behaviors are noted. Additionally, discouraging inappropriate sexual behaviors (eg, public masturbation, stalking) may be difficult in some young adults (Postorino, Siracusano, Giovagnoli, & Mazzone, 2018). The discrepancy between physical and social maturation during adolescence can be particularly challenging for individuals with autism.

Brain Structure

The most significant abnormalities in a child's body structure include the anomalies of the brain that are currently under investigation. Likely caused by genetic abnormalities, these deviations in the brains of individuals with autism are not yet clearly defined, nor is it clear precisely how the differences cause the characteristics of autism. Studies have found brain volume differences (Courchesne et al., 2011), accelerated brain growth (Ha et al., 2015; Redcay & Courchesne, 2005), and brain connectivity abnormalities (Alexander, 2012).

ASDs are pervasive and can cause lifelong impairment. The social and behavioral challenges that accompany autism can create difficulty with participation in all areas of occupation. However, appropriate diagnosis is a first step in developing treatment strategies and environment changes that support functioning for individuals with autism. With support and acceptance, people with autism lead full, productive, and satisfying lives.

Summary of Occupational Performance Areas

Activities of daily living (ADLs) such as dressing and eating can be difficult if the individual is overresponsive to the sensations of clothing or food and/or has difficulty with maintaining the necessary postural stability to support motor planning of these tasks. Instrumental activities of daily living (IADLs), like shopping or driving, are difficult and even dangerous if the individual does not have adequate focused attention and emotional regulation to make in-the-moment safe decisions. Health maintenance can be affected by difficulty with verbal communication. For example, sometimes, pain may manifest as new disruptive behaviors. That is the reason that medical causes of changes behavior must be fully investigated from a medical perspective. Participation in the educational system can be hampered by difficulty with memory functions for learning and socialization skills to develop healthy relationships with peers. Play and leisure activities are sometimes limited by RRBs, making participation outside of the home stressful. Finally, individuals with autism often have difficulty obtaining and maintaining employment due to the complex nature of these tasks and the constellation of symptoms that make professional behaviors challenging. In summary, ASD is a pervasive, lifelong disability that can impact every area of occupational performance. However, intervention can facilitate functional changes to significantly improve the quality of life of the individual with autism and their family.

CASE STUDY 1

At 1 year of age, Jacob was brought to an early intervention clinic because of delays in motor and cognitive skills. An initial evaluation conducted by an occupational therapist, educator, and speech-language pathologist confirmed these concerns. In the area of gross motor skills, Jacob was delayed in learning to walk and demonstrated moderate hypotonia in his trunk. Fine motor concerns included tactile defensiveness with wet or sticky substances and delays in grasping and manipulating objects appropriately. Cognitively, Jacob showed little interest in playing with toys or imitating the words and actions of others. Language impairments were not yet observed since Jacob frequently vocalized, babbled, and expressed his feelings through behaviors such as smiling or crying. At this young age, ASD was not initially suspected. Jacob was clearly a delightful little boy who showed a strong attachment to his parents and was interested in watching children and other adults in his environment.

Jacob was placed in a playgroup at the clinic where his occupational therapist and educator worked directly with him, while a speech-language pathologist monitored his language. As time passed, further concerns became evident. His language skills failed to further develop; as a result, first words did not emerge, nor was he using gestures to indicate what he wanted. At the age of 14 months, he demonstrated limitations in language comprehension and speech production, and his use of eye contact decreased. As the months progressed, he showed frustration more frequently through crying, banging his head, throwing himself on the ground, and arching his back to pull away from a person trying to hold him. His feeding skills were limited since he was not able to bring his hand to his mouth, and he demonstrated extreme sensitivities to many tastes and textures. He did not show an interest in other self-help skills such as learning to bathe or dress himself. While other children his age learned to imitate motor movements in songs, Jacob seemed to content to only listen to the songs. He did not demonstrate typical play skills for his age such as exploring how toys worked, taking toys in or out of a container, or taking turns with others. However, musical or flashing toys captured his attention for long periods of time. While he was engrossed with these toys, he evidenced unusual, repetitive behaviors such as rocking his body back and forth.

Through early intervention by his therapists and parents, Jacob's gross motor skills improved during the following months and he successfully learned to walk at 18 months. He delighted in walking through his home,

through the early intervention center, and outside. His eye contact improved, becoming more spontaneous and consistent. His repertoire of sounds increased, and he vocalized to take a turn in songs or games; in addition, he began signing "more" to request something desirable to happen again. He learned to play with toys in a more functional manner, including stacking rings and blocks and taking toys in and out of containers. Yet other skills continued to be challenging. When he was left to play on his own, he repetitively turned the pages of books or walked aimlessly around a room. He showed hypersensitivities to touching or mouthing certain textures, limited motor or imitation skills, difficulty understanding others, a lack of verbal words, and repetitive behaviors during play.

Currently, at slightly older than 2 years of age, Jacob continues to struggle in several areas of development. However, he has made steady progress in these developmental skills, and his family and team are encouraged that he will continue to make significant gains. More importantly, despite being faced with more challenges than typically developing children, Jacob is a young boy who is often able to share and express joy with others.

CASE STUDY 2

Patrick is a 38-year-old man who was diagnosed with autistic disorder at two-and-a-half years of age. His mother was first concerned he had a hearing impairment, as he did not seem to understand what people said to him, did not respond to a fire truck siren, placed his ear close to the refrigerator, and acted out with negative behaviors. Behavioral and developmental concerns persisted after testing ruled out a hearing impairment. A psychologist specializing in autism assessed Patrick's developmental skills and behaviors and diagnosed Patrick with autistic disorder.

Patrick communicated nonverbally for several years, usually by grabbing a person's hand and leading the person to an item he wanted. He had a very limited diet as a young child, eating only peanut butter, raisins, and yogurt. In time, he increased his repertoire of foods. When Patrick was three, he began attending a local school for children with autism; at the age of eight, he attended a school for children with a variety of special needs. Patrick began using words at the age of nine. He transferred to a local public high school at the age of 18, participating in the school's autism program and working in the school's kitchen to learn to work with others. He graduated high school when he was 26, receiving a diploma of completion for the school's autism program.

Following high school, Patrick moved into a condominium with two other men diagnosed with autism; he currently maintains this living situation. Patrick and his roommates receive 24-hour supervision. He also works 5 days per week at a therapy center that encourages functional skill building. He works alongside eight or nine other adults with autism and receives supervision in his daily tasks, which include shredding legal documents and inserting newspapers into plastic sleeves. To prevent overwhelming stimuli, the room is kept dim and without extraneous sound.

As with all individuals diagnosed with autism, Patrick shows personal strengths and unique challenges, with his own personality shining through. Patrick has achieved several goals in activities of daily living and continues to work toward independence. He dresses himself, with the occasional need for help with fasteners. He toilets independently, though requires assistance with wiping after a bowel movement. He washes himself with verbal prompting from a supervisor and climbs in and out of the tub independently. He applies deodorant and cologne and combs his hair when directed. Once a supervisor has prepared his toothbrush, Patrick brushes his teeth for 2 minutes, needing occasional reminders to not swallow the toothpaste. He carries groceries into the house, takes out the trash, and wipes the table after meals.

Patrick displays unusual and stereotypical behaviors, such as rapidly bouncing or tapping a small ball on a tabletop, spinning lids, and repeatedly blowing up balloons. He eats his food very quickly and needs verbal reminders to eat at a slower pace. In the car, he often bends down to play with the hardware beneath his seat. Recently, Patrick has shown negative behaviors, such as destruction of property or soiling himself. His family and staff believe these behaviors are likely Patrick's way of showing grief from his father's death last year.

Socially, Patrick gets along well with his family, supervisors, and roommates, though he has few verbal interactions with his roommates. His family frequently brings him to community outings such as sports events; Patrick shows interest and enjoyment during these outings. He reciprocates another person's smile easily. He occasionally initiates and maintains eye contact with another person, but not with typical duration or frequency. He often responds to a speaker but does not engage in back-and-forth conversation.

Regarding other developmental skills, Patrick currently understands more language than he uses. He follows familiar one-step directions, though sometimes needs repetition from the speaker to complete the request. He comprehends simple questions that relate to his interests and daily routines. Patrick communicates through single words and phrases. He names familiar people and objects, answers simple questions, and requests desired items and actions. He greets when prompted by another person. In the area of fine motor skills, Patrick does not demonstrate a hand preference. He writes his first name, last name, and the word "love" to sign letters and cards. Patrick shows many strengths in his gross motor skills. He enjoys playing a variety of sports, including swimming and throwing a ball back and forth with a partner. He loves to shoot basketball hoops (dribbling is more difficult for him), which was a favorite activity he had shared with his father. He experiences some difficulty with coordination and motor planning. Cognitively, Patrick has recently learned the days of the week and loves to look at calendars. He can recite the alphabet and recognizes a few familiar written words. He does not yet tell time.

Patrick lives a very active life, filled with family, supportive staff, and opportunities to contribute to his community. His mother and professional team continuously work together to create new goals for Patrick, and he meets these challenges. He is a delightful and admirable individual.

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CHAPTER
4

Intellectual Disability

Tracy R. Young and Michelle A. Suarez

Key Terms

Cortical atrophy
Craniostenosis
Cytomegalovirus
Down syndrome
Fragile X syndrome
Hydrocephaly
Hyperphenylalaninemia
Hypoxia
Intellectual disability (ID)
Spina bifida
Tay-Sachs disease
Toxemia
Tuberous sclerosis

Michael, now 2, was diagnosed by the pediatrician shortly after his birth as having Down syndrome. He exhibits the physical characteristics such as a round face, flattened nose bridge, abnormally small head, low-set ears, short limbs, and abnormally shaped fingers. His motor development is delayed, and his low muscle tone has made learning to walk difficult. Intellectual disability in children with Down syndrome is inevitable but varies in degree of severity. Michael's early intervention program provides services for Michael as well as parent support and education.

Jeffrey is an adorable little boy who just celebrated his third birthday. He was brought to the local school district for a developmental evaluation by his parents who report that the pediatrician is concerned with his language development. He smiled and interacted with examiners but was unable to express his wants and needs or follow single-step directions. An interview with his parents revealed that Jeffrey was unable to dress or undress and was not self-feeding. He was diagnosed with global developmental delay and possible intellectual disability and enrolled in an early childhood special education program where he is working toward language and self-care goals. His family hopes he will eventually attend mainstream kindergarten with special education support.

Kelly, now in second grade, is pleasant and likable. Teachers have been concerned about her inability to write letters or sound out words since kindergarten, and as the curriculum becomes more challenging, she is falling further behind. She has trouble maintaining friendships and often struggles to join peers in play. School staff conducted a multidisciplinary team evaluation and diagnosed Kelly with an intellectual disability. It was determined that Kelly would stay in her present classroom and be educated using an inclusive model. She was provided with special education support and accommodations to match her cognitive ability and learning pace.

Description and Definition

The term **intellectual disability (ID)** has evolved from the previous diagnosis of mental retardation. This terminology change reflects the shift from a view that classifies "mental retardation" as a personal trait residing solely within the individual to a holistic perspective that includes the capabilities of the person within the context of the environment. According to the *Diagnostic and Statistical Manual of Mental Disorders-Fifth Edition (DSM-5)*, published by the American Psychiatric Association (APA), the definition of ID is characterized by significant limitations in both intellectual functioning and adaptive behavior as expressed in conceptual, social, and practical adaptive skills. This disability originates before age 18 (American Psychiatric Association [APA], 2013).

Intellectual Functioning

The first component of this definition, intellectual functioning (or intelligence), is the general mental capability of an individual. This includes the ability to reason, plan, problem solve, think abstractly, comprehend complex ideas, and learn from experience (APA, 2013). While it has its limitations, the accepted measure of intelligence is that which is determined by an intelligence quotient (IQ) score, which involves administration of standardized tests given by a trained professional. In the 2010 publication of the American Association on Intellectual and Developmental Disabilities (AAIDD), a "significant limitation in intellectual functioning" was defined as two standard deviations below the mean, in the context of the standard error and strengths and limitations of the specific instrument (IQ test) used in assessment (American Association on Intellectual and Developmental Disabilities [AAIDD], 2010). The mean of an IQ test is 100, and therefore two standard deviations below the mean would be a cutoff score of ~70. The IQ score is not a complete representation of human functioning and must be considered in the context of adaptive behavior, health, participation, and context. Therefore, clinical judgment must be used to interpret scores when considering diagnosis or service provision.

Adaptive Behavior

Adaptive behavior is the most related domain of concern among occupational therapists. Thus, it is our focus of assessment and intervention for persons with ID. As defined by the AAIDD, "adaptive behavior is the collection of conceptual, social, and practical skills that people have learned so they can function in their everyday lives" (AAIDD, 2010). Significant limitations in adaptive behavior impact a person's daily life and affect the ability to respond to a particular situation or to the environment. Table 4.1 provides specific examples of these three areas, which are published by the AAIDD.

TABLE 4.1 Adaptive Skills

Conceptual Skills	Social Skills	Practical Skills
Receptive and expressive language	Interpersonal	Personal activities of daily living such as eating, dressing, mobility, and toileting
Reading and writing	Social responsibility	
Money concepts	Self-esteem	
Time concepts	Gullibility (likelihood of being tricked or manipulated)	Instrumental activities of daily living such as preparing meals, taking medication, using the telephone, managing money, using transportation, and doing housekeeping activities
Number concepts	Naiveté (wariness)	
	Following rules/obeying laws	
	Social problem solving	Occupational skills
	Avoiding victimization	Maintaining a safe environment

Limitations in adaptive behavior can be determined by using standardized tests referenced to the general population, including people with disabilities and people without disabilities. On these standardized measures, significant limitations in adaptive behavior are operationally defined as performance that is at least two standard deviations below the mean. In contrast to IQ scores for determining intellectual abilities, adaptive behavior is measured with the focus on typical performance and not maximum performance. In other words, the criteria critical to measuring limitations in adaptive behavior is *how a person typically performs*, not performance potential.

Onset Before the Age of 18

The final component of the definition of ID is that it begins early in life, and therefore, a diagnosis of ID is made in childhood. A diagnosis of ID is not considered in adult-onset degenerative diseases such as dementia or those associated with traumatic brain injury. In addition, children under the age of 5 years who have delays in developmental milestones and intellectual functioning are sometimes given a temporary diagnosis of global developmental delay (GDD) that requires reassessment after age 5 (APA, 2013). This diagnosis recognizes that accurate assessment of intellectual functioning is often not possible during this early childhood period, and therefore, the lifelong intellectual functioning prognosis is difficult to determine.

Severity Levels

In the United States, there are two diagnostic and classification systems for the term ID, the DSM-5 and the AAIDD. Although both systems agree that ID is a developmental condition characterized by significant impairment in intellectual functioning and adaptive behavior, they vary by how they classify severity levels.

The DSM-5 uses a classification system to further refine and categorize the diagnosis of ID through severity levels based on an individual's severity of function. The severity levels used by the DSM-5 are mild, moderate, severe, and profound (APA, 2013). Standardized testing along with clinical judgment regarding a person's adaptive functioning across the domains of conceptual, social, and practical domains is used when determining the severity level. Refer to the Signs and Symptoms section of this chapter for additional breakdown on these levels.

The AAIDD (2010) also uses a classification system for severity levels; however, instead of classifying by the severity of function, as done in the DSM-5, the AAIDD evaluates severity based on the intensity of supports needed to improve function. This system considers a person's strengths and abilities, not simply limitations, and is determined through a standardized support need tool. The intensities of support levels are intermittent, limited, extensive, and pervasive. Intermittent support is provided on an "as-needed" basis. Limited support occurs over a limited time span. Extensive support is assistance provided on a daily basis in a life area. Pervasive support refers to the need for support in all life areas across all environments on a daily basis (AAIDD, 2010).

In considering the definition of ID, the AAIDD notes that there are five assumptions that need to be considered in the diagnostic process (AAIDD, 2010):

1. Limitations in present functioning must be considered within the context of community environments typical of the individual's age, peers, and culture.
2. Valid assessment considers cultural and linguistic diversity as well as differences in communication, sensory, motor, and behavioral factors.
3. Within an individual, limitations often coexist with strengths.
4. An important purpose of describing limitations is to develop a profile of needed supports.
5. With appropriate personalized supports over a sustained period, the life functioning of the person with ID generally will improve.

Etiology

There are hundreds of causes of ID, yet despite knowing the many factors that contribute to ID, in a large proportion of cases, the cause remains unknown. Causes of ID may be classified according to their origin (biomedical vs environmental) or when they occurred in the developmental cycle (prenatally, perinatally, or postnatally) (Shapiro & Batshaw, 2011). The ability to determine cause is highly correlated with the level of the ID. The etiology of ID is much less likely to be known with individuals who are mildly intellectually disabled (IQs of 50-70) than with those who are severely affected (IQs of <50) (AAIDD, 2010).

Biomedical Influences

Biomedical factors during the prenatal period that can contribute to ID include genetic aberrations, birth defects that are not genetic in origin, or a combination of factors (Shapiro & Batshaw, 2011). Up to 50% of the individuals diagnosed with ID may have more than one causal factor (AAIDD, 2010).

Genetic Causes

With genetic aberrations, the problem is either with the genes, which are the basic unit of heredity, or with the chromosomes, which carry the genes. Genetic causes can be divided into two types: single gene disorders and chromosomal abnormalities. In single gene disorders, there is a problem with the quality of the genetic material; a specific gene is defective. In chromosomal abnormalities, the problem is with the quantity of material. There is either too much or too little genetic material in a specific chromosome. In many cases of ID, the gene or chromosome that has caused the condition can be identified specifically. In fact, more than 350 inborn errors of metabolism that result from genetic changes have been identified. Many of these metabolic errors lead to ID (Sumar & Lee, 2011). In other cases, the specific genetic aberration has not been identified. Factors such as higher incidences of a condition in specific families or increased recurrence rates among siblings suggest that the defect is genetic (Sumar & Lee, 2011).

Single Gene Disorders

Single gene disorders follow specific patterns of transmission: autosomal dominant, autosomal recessive, or sex linked. Table 4.2 presents the transmission patterns and risk factors associated with each type.

TABLE 4.2 Single Gene Disorders

Type	Autosomal Dominant	Autosomal Recessive	Sex Linked
Transmission pattern	Either parent carries gene or spontaneous transmission	Both parents are carriers	Either parent can transmit gene: mother usually a carrier, father cannot be a carrier but can have the disorder
Risk factors	50% risk of child being affected with each pregnancy	25% risk of child being affected with each pregnancy	If mother has affected gene, 25% risk of having affected son or carrier daughter; if father has affected gene, all his daughters will be carriers and his sons will be normal
Sex distribution	Male and female children equally at risk	Male and female children equally at risk	Primarily male children at risk for having disorder, female children at risk for becoming carriers

The autosomal dominant type is caused by a single altered gene. Either parent may be a carrier, or there may have been a spontaneous mutation of the gene. Dominant inheritance occurs when one parent passes on the defective gene. This occurs even if the other parent passes a healthy gene. Because the defective gene can be passed by either parent, there is a 50% risk of the child being affected in each pregnancy (Ropers, 2008). An example of this type of inherited disorder is **tuberous sclerosis**.

In the autosomal recessive type, both parents are carriers but show no outward signs or symptoms of having the disorder. Inheritance occurs when both parents pass the defective gene to their offspring. Each pregnancy has a 25% risk of the child being affected (Ropers, 2008). Examples of this type of disorder are phenylketonuria (PKU) and **Tay-Sachs disease**.

In X-linked disorders, the affected gene is on the sex chromosomes, specifically the X chromosome, and can occur in either parent. Because males have only one X chromosome, if the father has an affected gene, he will always have the disorder and cannot be a carrier. Because the female has two X chromosomes, she can either be a carrier of the disorder (if only one X chromosome is affected) or have the disease herself (if both X chromosomes

are affected). A carrier mother has a 25% risk of having an affected son. If the father has the affected gene, all his daughters will be carriers, but his sons will not be affected (Ropers, 2008). **Fragile X syndrome** is the result of a mutation at what is known as the fragile site on the X chromosome and is one of the most common causes of ID (Sumar & Lee, 2011). Other examples of X-linked disorders are Duchenne muscular dystrophy, Lesch-Nyhan syndrome, and Hunter syndrome.

Chromosomal Disorders

Chromosomal aberrations include missing or extra chromosomes, either in part, such as a short arm, or the total chromosome, as is found in the trisomal types. Either the autosomes or sex chromosomes can be affected, with the autosomal type resulting in more serious neuromotor impairments (National Human Genome Research, 2019). One of the most common genetic causes of ID is **Down syndrome**, or trisomy 21, which is generally caused by an extra 21st chromosome (Sumar & Lee, 2011). The other most common are trisomies 18 and 13.

Birth Defects

Birth defects that are not considered genetic in origin can also contribute to or cause ID. These could include such things as malformation of parts of the CNS (eg, **cortical atrophy**, **hydrocephaly**, **spina bifida**, **craniostenosis**) (Shapiro & Batshaw, 2011) or congenital cardiac anomalies (Razzhaghi, Oster, & Reefhuis, 2015).

Environmental Influences

Environmental influences related to the etiology of ID are factors that originate external to the person and impact the development or functioning of the brain. These factors can be classified by when they occur in the developmental cycle.

Prenatal Factors

Numerous environmental risk factors may play a role in the prenatal development of ID. They include intrauterine exposure to chemical agents such as nonprescription drugs or alcohol (O'Leary et al., 2013) ingested by the mother during pregnancy. A study by O'Leary et al. (2013) provided the first population-based study with evidence indicating that maternal alcohol use disorder should be classified as the leading known nongenetic cause of ID. Maternal conditions that contribute to ID include rubella, syphilis, hypertension (Huang, Zhu, Qu, & Mu, 2016), diabetes (Huang et al., 2016), **toxemia**, **hyperphenylalaninemia**, a rare form of PKU caused by increased levels of phenylalanine amino acid (Mayo Clinic, n.d.), or to congenital infections such as **cytomegalovirus**, a common herpesvirus infection that has a wide range of symptoms from asymptomatic to fever and to severe signs involving the eyes, brain, or other internal organs (Cheeran, Lockensgard, & Schleiss, 2009).

Low birth weight that results from prematurity or intrauterine growth retardation can also be a contributing factor to ID (Huang et al., 2016). Maternal factors associated with low birth weight include smoking, lack of prenatal care, infections, poor nutrition, toxemia, and placental insufficiency. Exposure to industrial chemicals, or drugs including certain over-the-counter (OTC) prescriptions and illegal substances, also can affect birth weight, particularly during the first trimester of pregnancy.

Perinatal Factors

Two major causative factors of ID in the perinatal period are mechanical injuries at birth and perinatal **hypoxia**, which refers to reduced oxygen supply. Mechanical injuries are caused by difficulties of labor because of malposition, malpresentation, disproportion, or other labor complications that result in tears of the meninges, blood vessels, or other substances of the brain. Factors that cause perinatal hypoxia or anoxia include premature placental separation, massive hemorrhage from placenta previa, umbilical cord wrapped around the baby's neck, and meconium aspiration. Preterm birth has also been a factor with ID (Huang et al., 2016). Very premature infants may also have impaired respiration or an intracranial hemorrhage that can result in brain damage.

If a mother has an active case of herpes simplex II and is shedding the virus at the time of delivery, the baby can acquire the infection in the birth canal, which can cause severe developmental disability. This can be avoided by testing to determine whether the mother has an active case and, if so, delivering by cesarean section. In addition, HIV-1 encephalopathy, acquired perinatally, can lead to developmental disabilities (Mitchell, 2006).

Postnatal Factors

Traumas or infections that result in injury or a lack of oxygen to the brain are a major cause of ID during the postnatal period. Traumas include near-drowning or strangulation, child abuse, and closed head injuries. Early severe psychosocial deprivation (ie, attachment disorder, removal from the family home) can be a significant factor in ID etiology (Shevell & Sherr, 2008). Infections include encephalitis and meningitis. ID that results from meningitis caused by *Haemophilus influenzae* is now preventable, however, with the introduction of the *H influenzae* type B (HiB) vaccine (Baraff, Lee, & Schriger, 1993).

Incidence and Prevalence

ID is one of the most frequently occurring developmental disabilities and is not limited by race or culture. Maulik, Mascarenhas, Mathers, Dua, and Saxena (2011) found that prevalence around the world is around 1% and almost two times higher in low- and middle-income countries. The most recent data in the DSM-5 reports indicate an overall prevalence rate of ~1%, which is lower than previous estimates of 2%-3%. This reduced number is likely due to the fact that IQ is no longer relied on as the specific factor in diagnosis of ID (APA, 2013).

Most professionals recognize that prevalence estimates vary by chronological age (Anderson, Larson, MapleLentz, & Hall-Lande, 2019; APA, 2013; Ropers, 2008), operational definition of ID at the time of the study (Anderson et al., 2019), severity levels of ID (APA, 2013), and sex of the individual (Zablotsky, Black, & Blumberg, 2017). According to the DSM-5, prevalence for severe ID is ~6 per 1000 (APA, 2013) and males are ~1.5 times more likely to be diagnosed with ID than females, which may be related to the sex-linked genetic disorders that result in ID (Zablotsky et al., 2017).

Signs and Symptoms

All individuals with ID will have deficits in intellectual functioning and adaptive behavior, occurring before the age of 18 (APA, 2013). Early symptoms in a young child will vary depending on the severity of the ID. Generally speaking, symptoms might include difficulty with gross or fine motor development, learning to speak, remembering things, poor awareness or use of social rules, trouble understanding of consequences, and solving problems (Center for Disease Control and Prevention, 2019). Individuals with mild ID may not have any symptoms present until they begin elementary school. Once a diagnosis is confirmed, the functional presentation will vary between individuals depending on severity level and possible co-occurring diagnoses.

Core Symptoms

In the DSM-5, one of the core components for the diagnosis of ID is impairment in intellectual functioning. This may initially present as difficulty with reading, writing, mathematics, understanding of time or money, abstract thinking, and memory. To be considered intellectually disabled, a person's performance on a standardized intelligence test must be two standard deviation units or more below the mean. When looking at the levels of ID from the perspective of IQ scores, King, Toth, Hodapp, and Dykens (2009) identified that ~85% of individuals are in the mild range, 10% are in the moderate range, 4.0% are in the severe range, and 2.0% are in the profound range of function. It is important to remember that not all individuals in a particular classification will function at exactly that level.

A second core component for ID is significant limitations in adaptive behavior. As previously stated, adaptive behavior areas include communication, self-care, home living, social/interpersonal skills, leisure, health and safety, self-direction, functional academics, use of community resources, and work (AAIDD, 2010). An individual with ID would therefore have difficulty with skills required to live independently in a safe and responsible manner. Limitations in adaptive behavior is expressed by two standard deviations below the mean on a standardized test in conceptual, social, and practical adaptive skills (AAIDD, 2010; APA, 2013) and should be assessed in all of the individual's performance contexts. Levels of adaptive skill functioning by severity, as adapted from the DSM-5 (APA, 2013), are as follows:

- Mild ID: In children, this is characterized by difficulty learning academic skills and struggles with abstract thinking, executive functioning, and short-term memory. Functional application of previously learned academic skills is noted in adults and highlighted by a concrete approach to problem solving. Immature social and communication interactions can cause difficulty with regulating emotions and behavior, understanding of social risks, and gullibility. When in school, support is required in one or more academic areas to meet age

appropriate expectations. Independence in personal care is possible, but support may be needed for more complex daily living tasks such as health care, legal decisions, and raising a family. Employment is possible in jobs that do not emphasize conceptual skills.

- Moderate ID: This level includes slow development in language and academic skills beginning at the preschool age. Adults typically have an elementary level education. Spoken language is used primarily for communication, but not at the same level as peers. Lack of understanding of social cues can make work settings and friendships with typically developing peers difficult. Daily assistance and supports are needed for all application of academic skills in both personal settings and vocational employment. Significant social and communication support is necessary for success in vocational settings. Individuals can be successful with recreational skills and general self-care needs when provided with an extended period of teaching and occasional reminders.
- Severe ID: Individuals in this level are able to understand simple speech and gestures; however, the development of conceptual skills is limited. Expressive language is highlighted by single words or phrases and may be supplemented by augmentative means. Extensive supports are needed for problem solving. Individuals will require supervision and ongoing supports for all activities of daily living (ADL). Long-term teaching and support can assist with skill acquisition.
- Profound ID: Functional use of objects is possible. There is minimal understanding or use of symbolic communication, but individuals may initiate and respond through gestural and emotional cues. Enjoyment is found in relationships with well-known individuals. Recreational activities are possible and might include water-based activities, taking walks, listening to music, or watching a movie. Dependency for all ADL is common; however, individuals without physical impairment may be able to assist with some routine chores. Co-occurring motor and sensory differences may interfere with functional use of objects, social, recreational, or vocational activities.

Co-occurring Conditions

ID often occurs in tandem with, or as a secondary manifestation of, another diagnosis. The DSM-5 states that mental, neurodevelopmental, medical, and physical conditions frequently co-occur with ID, with some conditions such as cerebral palsy and epilepsy co-occurring up to three to four times more than in the general population (APA, 2013). According to the DSM-5 (APA, 2013), the most common co-occurring mental and neurodevelopmental disorders are:

- Attention-deficit/hyperactivity disorder
- Depressive and bipolar disorders
- Anxiety disorders
- Autism spectrum disorders
- Stereotypic movement disorder (with or without self-injurious behavior)
- Impulse control disorders
- Major neurocognitive disorders

In addition to the associated conditions already mentioned, a high proportion of individuals with ID also have some form of mental illness. Estimates of prevalence of mental illness among people with ID range from 10% to 20% to 40% to 70% (King, Toth, Hodapp, & Dykens, 2009). Some of the common types of mental illness seen in people with ID include personality disorders, affective disorders, psychotic disorders, avoidant disorder, paranoid personality disorder, and severe behavior problems that may include self-injurious behavior (Einfeld, Ellis, & Emerson, 2011). Several misconceptions about people with ID may complicate or prevent appropriate care for their mental illness, including the beliefs that people who are intellectually disabled cannot also be mentally ill, do not experience normal feelings and emotions, and are not affected by changes in their environment. Substance abuse problems, especially with alcohol, may be overlooked. Because of limited communication skills and limitations in abstract thinking caused by the ID, the diagnosis of mental illness and mental health problems can be a very difficult process and is frequently inexact. Good communication with caregivers and significant others in the life of the individual with ID is essential.

Diagnosis

An evaluation must be performed to determine whether a person meets the criteria for being intellectually disabled. Besides ascertaining that the onset of the condition occurred before age 18, there are two main aspects to this process. The first part involves administration of an appropriate standardized intelligence test by a qualified individual. The selection of the specific standardized instrument should be based on factors such as the individual's social, linguistic, and cultural background (AAIDD, 2010). The individual's IQ is interpreted in the context of his or her level of adaptive behavior.

The second aspect of the process is the evaluation of adaptive behavior as it relates to the targeted adaptive life skill areas. Adaptive functioning is an individual's ability to cope with life demands and meet societal expectations for independence depending on age group (AAIDD, 2010). The skills needed for adaptive behavior become more complex and varied as the person ages. For instance, eating and dressing independently are major skills for the young child, but the child does not need to be able to use a phone or manage money. Evidence for deficits and strengths in adaptive function should be gained from one or more independent, reliable sources who are familiar with the individual's abilities in different performance contexts. This information should be used to complete a standardized adaptive behavior scale designed to provide a composite "picture" of the individual's adaptive function. As with the selection of an intelligence test, care should be taken that the adaptive behavior scale chosen is appropriate for the individual's sociocultural background, education, associated handicaps, motivation, and cooperation level (AAIDD, 2010). There are more than 200 adaptive behavior measures and scales. One of the most common scales is the Vineland Adaptive Behavior Scales, Third Edition (VAB-3) (Sparrow, Cicchetti, & Saulnier, 2016), which purports to assess the personal and social skills needed for everyday living. It is an indirect assessment in that the respondent is not the individual in question but someone familiar with the individual's behavior. The VAB-3 measures three domains: communication, daily living skills, and socialization. An Adaptive Behavior Composite is a combination of the scores from the three domains. A second scale frequently used to assess adaptive behavior is the Diagnostic Adaptive Behavior Scale (DABS) (Tassé et al., 2017). This measure was developed by the AAIDD to assess conceptual, social, and practical skills and focuses on the critical "cutoff area" for the purpose of ruling in or ruling out a diagnosis of ID. It should be noted that someone with limited intellectual function who does not have adaptive skill deficits is not considered intellectually disabled (AAIDD, 2010).

Once a diagnosis and classification of ID have been made through testing of intellectual functioning and adaptive behavior, it is important to complete the assessment framework by determining the level and system of supports needed for an individual in order to meet the demands of their environment (AAIDD, 2010). The Supports Intensity Scale (SIS) is a standardized tool designed to measure the type and intensity of supports that a person with ID requires to be successful in their environment (Thompson et al., 2015). The AAIDD is the publisher of SIS products and has a version for adults (SIS-A) and children (SIS-C).

Course and Prognosis

ID is generally considered a lifelong condition, but the course and prognosis will vary depending upon the cause(s) of the disability and access to resources (Sulkes, 2020). In terms of life expectancy, people with mild ID live as long as the general population (Shevell & Sherr, 2008). However, people with more profound ID are less likely to reach old age. This is likely due to more serious neurological deficits and associated disorders.

Most cases of ID are nonprogressive, that is, once the initial insult to the brain occurs, there is no further damage. The emphasis is on managing the medical aspects of the condition and helping individuals to achieve their highest potential. However, certain genetic conditions (eg, muscular dystrophy and Tay-Sachs disease) are progressive, with incremental loss of function and, in some cases, associated early death. Those with Down syndrome experience degenerative changes in the brain, beginning at about age 40 years, that eventually result in progressive dementia similar to Alzheimer's (Carr, 2012). The goal for these individuals is to help them achieve the highest level of independence and maintain it as long as possible.

One significant issue that can impact prognosis in people with ID is the presence of stigma in our society. This group often encounters negative stereotypes and prejudice from society that can lead to isolation and discrimination (Werner, 2015), lower self-esteem, and social exclusion (Robinson, Hill, Fisher, & Graham, 2020). In addition, self-reported stigma from people with ID is strongly associated with symptoms of anxiety and depression (Ali, King, Styrdom, & Hassiotis, 2015). Social inclusion can mediate the impact of stigma and support psychological well-being (Simplican, Leader, Kosciulek, & Leahy, 2015). For example, membership in organizations like the Special Olympics can increase self-esteem, increase quality of life, and reduce stress (Crawford, Burns, & Fernie, 2015).

Another important factor in the improvement of the prognosis for people with ID is access to services that focus on strengths and increase adaptive functioning. With these services, it is possible for individuals with mild ID to gain

adaptive behavior skills through remedial programs to the extent that they no longer meet the diagnostic criteria for being intellectually disabled, although their intellectual function has probably not changed significantly (AAIDD, 2010).

Medical/Surgical Management

There is no surgical or drug treatment for the condition of ID; however, surgery or medications may be needed for some of the conditions that could occur in tandem. In the past, psychotropic medications, and particularly antipsychotics, were often used in an attempt to manage challenging behaviors (Glover, Bernard, Brandord, Holland, & Strydom, 2014). However, due to negative side effects and limited evidence that these medications actually decreased these target behaviors, this practice has declined. Instead, psychotropic medications are now often used to treat diagnosed concomitant mental health problems (Tsiouris, Kim, Brown, Pettinger, & Cohen, 2013). A large-scale study of psychotropic drug use in people with ID by Tsiouris et al. (2013) revealed that 45% were receiving an antipsychotic (eg, risperidone, olanzapine) to treat psychosis and bipolar disorder, 23% were receiving an antidepressant (eg, Prozac, Zoloft) for an affective disorder, and 16% were receiving an antianxiety agent (eg, clonazepam).

In addition to medications for treatment of mental health disorders, ~20% people with ID also have epilepsy, which may require the use of medications to reduce seizure activity (Robertson, Hatton Emerson, & Baines, 2015). Medications include Tegretol for generalized tonic-clonic seizures and tranquilizers like clonazepam for short-term emergency treatment. Unfortunately, diagnosis of epilepsy in people with ID can be complicated by communication and tolerance issues in this population that make standard testing like electroencephalogram magnetic resonance imaging difficult (McCarron, O'Dwyer, Burke, McGlinchey, & McCallion, 2014). Misdiagnosis can increase the risk for serious, sometimes life-threatening consequences of untreated seizure activity. Vigilance and professionals that are trained in recognizing symptoms of seizures in this population can increase appropriate medication dosage and seizure management.

Neuromuscular dysfunction (eg, spasticity) is seen in children with ID who have comorbid cerebral palsy. Several medications are sometimes helpful for increasing active and/or passive range of motion for increased function and prevention of contractures (Smith & Kurian, 2012). For example, botulinum toxin (eg, Botox) is injected directly into the muscle group and will provide a short-term reduction in tone that can be used to strengthen antagonist muscle groups and facilitate the development of functional movement patterns. One longer-lasting medication for tone reduction is intrathecal baclofen. This medication is sometimes delivered by a pump directly into the spinal fluid and can relax spastic muscle groups systemically. All medications have potential side effects and, when given, require close monitoring by the prescribing physician.

Given that there is no surgical or drug treatment for the diagnosis of ID, long-term management of individually designed programs of supports to meet a person's unique needs is required. To maximize an individual's health and wellness, independence, and engagement, best practice is highlighted when an interprofessional team designs the types and intensity of supports needed. Systems of supports chosen are dependent on the individual's level of severity of ID, comorbidities, strengths, and abilities. Examples of systems of supports that might be beneficial include individual therapies, education and vocational training programs, organization-level consultation through community initiatives (Umeda et al., 2017), and local, state, and national programs, such as the Special Olympics (Candan & Dubon, 2019). Team members that might be involved in the design and implementation of medical management and programming include a primary care physician, psychiatrist, psychologist, neurologist, nurse practitioner, special education teacher, behavioral intervention specialist, physical and/or occupational therapist, speech pathologist, social worker, vocational training specialist, and pharmacist (Figs. 4.1 and 4.2).

FIGURE 4.1 Emma just finishing her OT session where she worked on tone management and vocational skills.

FIGURE 4.2 Jason works on carryover of newly learned meal preparation skills.

Impact of Occupational Performance

Virtually all areas of occupational performance and many client factors can be affected by ID, depending on the cause and severity of the ID. As stated previously, the diagnostic criteria for individuals with ID included three categories of adaptive behavior (conceptual, social, and practical skills) (AAIDD, 2010). These areas (communication, language, interpersonal skills, social responsibility, recreation, friendships, daily living skills, work, and travel) fall into all occupational performance areas including (basic) ADL, instrumental ADL, health management, rest and sleep, education, work, play, leisure, and social participation.

Although all of the occupational performance areas and client factors can be influenced by ID, those that are affected will depend on factors such as the presence of additional medical diagnoses and the severity of the

diagnosis. It is imperative that the clinician be informed about the specific diagnosis that accompanies the identification of ID to determine the associated client factors that are involved. For example, occupational therapists often work with persons who have Down syndrome, which is one of the most common genetic causes of ID (Sumar & Lee, 2011).

With this diagnosis, the impact of client factors on occupational performance begins as a baby. Individuals with Down syndrome often have hypotonia, or poor muscle tone (National Down Syndrome Society, n.d.). This reduced muscle tone, which results in oral motor dysfunction such as protruding tongue, often makes feeding babies with Down syndrome difficult. Hypotonia may also affect the muscles of the digestive system, in which case constipation may be a problem. Atlantoaxial instability, a malformation of the upper part of the spine located under the base of the skull, is present in some individuals with Down syndrome. This condition can cause spinal cord compression as well as craniovertebral instability (Brockmeyer, 1999).

Approximately half of children with Down syndrome are diagnosed with a congenital heart disease and associated early onset of pulmonary hypertension, or high blood pressure in the lungs (National Institutes of Health [NIH], 2017). Echocardiography may be indicated to identify any congenital heart disease. If the defects have been identified before the onset of pulmonary hypertension, surgery has provided favorable results. Seizure disorders, though less prevalent than some of the other associated medical conditions, still affect between 5% and 13% of individuals with Down syndrome, a 10-fold greater incidence than in the general population. Congenital hypothyroidism, characterized by a reduced basal metabolism, an enlargement of the thyroid gland, and disturbances in the autonomic nervous system, occurs slightly more frequently in babies with Down syndrome (NIH, 2017).

According to the NIH (2017) up to three-quarters of children with Down syndrome have a hearing loss. A study by Nightengale et al. (2017) evaluated the prevalence of permanent and transient hearing loss and middle ear dysfunction in children with Down syndrome. Results included permanent hearing loss in 24.9% of the children. Most frequent were bilateral (75.4%) and conductive (33.3%) hearing losses. A transient hearing loss, with a high incidence of middle ear pathologies, was discovered in 22%-30%. In addition to hearing disorders, visual problems also may be present early in life. Cataracts occur in ~3% of children with Down syndrome but can be surgically removed. Given that 85% of all ID cases fall in the mild range of cognitive impairment, global and specific mental functions will be most affected with this population.

The following case studies illustrate how ID affects an individual's area of occupational performance in different stages of the life cycle.

CASE STUDY 1

K.R. is a 21-year-old young woman with Down syndrome. Despite the fact that she has reached the legal age of adulthood, she is still in the process of transitioning from the developmental stage of adolescence to young adulthood. She still receives special education services but is also working on developing work and job skills through traditional vocational services. Her "disability" is expected to be lifelong.

K.R.'s condition was identified at birth. She has received continuous support and direct services to facilitate development of her abilities since then. She has always lived at home with her parents and still does so. She has her own room and bathroom at home and generally has exclusive use of the family room for her leisure pursuits. Her parents are professionals who are actively involved in their professions and the community. They are very realistic about her abilities and extremely supportive, allowing her to make most of her life choices, with the exception of the support and guidance needed for health care needs. K.R. has a younger sister who is now away at college. K.R. has always been exposed to and involved in many social and cultural opportunities in the community, both with her family and on her own. Her social circle includes friends with and without disabilities, and she has several close friends as well as many social acquaintances.

K.R. has recently declared her life goals to be getting a job, getting an apartment of her own, and spending leisure time at the local community drop-in/recreational center for individuals with disabilities. Her mother feels that with appropriate supports, all of these goals are attainable.

K.R. is independent in most areas of personal care. As a result of limited fine motor coordination that seems to be complicated by visual-perceptual deficits, she needs assistance with regulating the water temperature for her bath, fastening zippers and buttons, tying her shoes, and keyboarding. She also occasionally needs reminders to straighten her clothing and brush the back of her hair. She has a speech impediment that makes it difficult for individuals who are unfamiliar with her to initially understand what she is saying. K.R. has learned to adapt to this