"This detailed book fulfills a long-time need for individuals with spina bifida and their families."

—TIMOTHY BREI, MD, FAAP

Gillette Children's Healthcare Series

SPINA BIFIDA

Understanding and managing the condition across the lifespan: A practical guide for families

Kelly Cho, MD
Cheryl Tveit, RN, MSN, CNML
Rachelle Lavalier, Parent

Editors
Lily Collison, MA, MSc
Linda E. Krach, MD
Tom F. Novacheck, MD

GILLETTE CHILDREN'S

Praise for *Spina Bifida*

"This book is sure to stand the test of time. It is an invaluable resource for families and individuals who live with the diagnosis as well as for professionals who are involved with the management of patients with spina bifida. It provides a comprehensive overview of the condition, its variations, implications, and, importantly, its effects on the individual as they age. I would have loved to have had this book when our son was born. I highly recommend it as a reference for anyone involved with the condition in any way."

SIOBHAN FLANAGAN, MD, Family Doctor, Ireland, and mother of adult son with spina bifida

"This is a beautifully written, comprehensive guide to myelomeningocele, the most common type of spina bifida, and its care. The content is laid out in logical, easy-to-follow chapters. The graphics are helpful to understanding the content, and the appendices provide excellent supporting information. The patient and family stories are lovely and provide hope for positive outcomes for the condition. All in all, this is a valuable resource for both families in caring for their child with spina bifida and professionals unfamiliar with the condition."

KATHRYN SMITH, RN, MN, MS, DrPH, Co-Director of Spina Bifida Program, Children's Hospital of Los Angeles, Professor of Clinical Pediatrics, Keck School of Medicine of USC

"The information in this book will be a valuable resource to anyone who would like to learn about spina bifida. The summary of key points at the end of each chapter allows interested readers to learn more efficiently, and the many diagrams provide clarity. It is an important resource for families with children with the condition."

JUDY THIBADEAU, RN, MN, Director of Research and Services, Spina Bifida Association

"Drawing on the latest published evidence, this book empowers individuals with spina bifida and provides their families with information they can use both to understand the complexities of this condition and to reinforce skills such as self-management and self-advocacy. Importantly, it uses a lifespan approach, highlighting issues from the prenatal period to adulthood. The inclusion of experiences of individuals and families provides a unique and important perspective. As a rehabilitation physician, I appreciate the emphasis on function and whole-body health."

BRAD DICIANNO, MD, Director, UPMC Adult Spina Bifida Clinic, Physical Medicine and Rehabilitation, Associate Medical Director, Spina Bifida Association

"In my experience, one of the biggest challenges families with a child with spina bifida face is learning the anatomy, physiology, and the vocabulary associated with the condition, the investigations, and treatments. The knowledge required extends far beyond that of most lay people. However, this comprehensive book provides the information needed with ample, clear explanations and illustrations. It will arm families with a strong foundation from the very beginning and throughout life. The sections on life in adulthood are particularly welcome and refreshing; so many shy away from sex, fertility, and aging, despite the clear need for quality information on these important topics."

GILLIAN YAZ, RN, Head of Health, Shine (Spina bifida, Hydrocephalus, Information, Networking, Equality)

"Speaking from the perspective of someone living with spina bifida, I found this book does a phenomenal job of describing all the different ways the condition can affect a person. It also caters to different audiences—people with spina bifida, parents, and those who take care of people with the condition—so every reader can get something out of this book. The pictures do a really good job of showing how some people with spina bifida may function. The suggestions of how to exercise the muscles differently, too, are very helpful for someone who may struggle to find a solid workout routine. I learned some new things about spina bifida by reading this book that I will take with me as I continue my own journey of being an adult with the condition."

GRACIE HADLICH, Adult living with spina bifida

"Spina Bifida *is an excellent resource for individuals with spina bifida, their families, medical professionals, and educators alike. It includes information from embryology to adult transition, and the stories from those living with the condition bring the issues to a personal level."*

CHRISTOPHER J. KEENAN, DO, FAAP, FACOP, FAAPM&R, Pediatric Physical Medicine and Rehabilitation, Children's Hospital of Philadelphia

"This detailed book fulfills a long-time need for individuals with spina bifida and their families. It is full of well-researched information and helpful illustrations, and readers will want to refer back to it often, as people with spina bifida move through the different stages of life. It also incorporates information from the Spina Bifida Association, along with other helpful resources, making it a comprehensive, valuable tool for families seeking to advocate more effectively for their loved ones with spina bifida."

TIMOTHY J. BREI, MD, FAAP, Medical Director, Spina Bifida Association

"In the literary genre of practical medical care, Spina Bifida *is destined to become a classic. The authors provide a thoughtfully organized volume of practical guidance for today's discerning reader, and they draw both from first-hand knowledge and numerous authoritative current sources. The writing style is easy and warm, and the use of graphics along with an international parental perspective bring the tome to life. This handsome resource belongs in the hands of every individual with spina bifida and on the shelf of every professional serving this complex and admirable population."*

JONATHAN CASTILLO, MD, MPH, FAAP, Chief, Division of Developmental Medicine, Children's Nebraska Hospital, Endowed Professor, Department of Pediatrics, University of Nebraska Medical Center, College of Medicine

"Exceptional spina bifida care depends on integrating high-tech medical and surgical advances with practical, everyday self-management and life skills. Spina Bifida *is a highly accessible resource that weaves together these two concepts: medical expertise with practical information for people living with the condition. The photos interspersed throughout the book of Rachelle and her daughter, Maddy, give life to the text, spanning a prenatal diagnosis to a successful transition into adulthood. The glossary demystifies medical terminology, the section on living with spina bifida offers highlights of the lived experience across the lifespan, and the illustrations throughout the text are excellent.This book is a must-read for families and health care providers alike."*

NIENKE DOSA, MD, MPH, Professor of Pediatrics, Medical Director of Spina Bifida Clinic, Upstate Medical University, Syracuse, New York.

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To individuals and families whose lives are affected by these conditions, to professionals who serve our community, and to all clinicians and researchers who push the knowledge base forward, we hope the books in this Healthcare Series serve you very well.

All proceeds from the books in this series at Gillette Children's go to research.

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Series Foreword

You hold in your hands one book in the Gillette Children's Healthcare Series. This series was inspired by multiple factors. It started with Lily Collison writing the first book in the series, *Spastic Diplegia–Bilateral Cerebral Palsy*. Lily has a background in medical science and is the parent of a now adult son who has spastic diplegia. Lily was convincing at the time about the value of such a book, and with the publication of that book in 2020, Gillette Children's became one of the first children's hospital in the world to set up its own publishing arm—Gillette Children's Healthcare Press. *Spastic Diplegia–Bilateral Cerebral Palsy* received very positive reviews from both families and professionals and achieved strong sales. Unsolicited requests came in from diverse organizations across the globe for translation rights, and feedback from families told us there was a demand for books relevant to other conditions.

We listened.

We were convinced of the value of expanding from one book into a series to reflect Gillette Children's strong commitment to worldwide education. In 2021, Lily joined the press as Program Director, and very quickly, Gillette Children's formed teams to write the Healthcare Series. The series includes, in order of publication:

- *Craniosynostosis*
- *Idiopathic Scoliosis*
- *Spastic Hemiplegia—Unilateral Cerebral Palsy*
- *Spastic Quadriplegia—Bilateral Cerebral Palsy*
- *Spastic Diplegia—Bilateral Cerebral Palsy, second edition*
- *Epilepsy*
- *Scoliosis—Congenital, Neuromuscular, Syndromic, and Other Nonidiopathic Types*
- *Spina Bifida*
- *Osteogenesis Imperfecta*

The books address each condition detailing both the medical and human story.

Mac Keith Press, long-time publisher of books on disability and the journal *Developmental Medicine and Child Neurology*, is co-publishing this series with Gillette Children's Healthcare Press.

Families and professionals working well together is key to best management of any condition. The parent is the expert of their child while the professional is the expert of the condition. These books underscore the importance of that family and professional partnership. For each title in the series, medical professionals at Gillette Children's have led the writing, and families contributed the lived experience. These books have been written in the United States with an international lens and citing international research. However, there isn't always strong evidence to create consensus in medicine, so others may take a different view.

We hope you find the book you hold in your hands to be of great value. We collectively strive to optimize outcomes for children, adolescents, and adults living with these childhood-acquired and largely lifelong conditions.

Dr. Tom F. Novacheck

Series Introduction

The Healthcare Series seeks to optimize outcomes for those who live with childhood-acquired physical and/or neurological conditions. The conditions addressed in this series of books are complex and often have many associated challenges. Although the books focus on the biomedical aspects of each condition, we endeavor to address each condition as holistically as possible. Since the majority of people with these conditions have them for life, the life course is addressed including transition and aging issues.

Who are these books for?

These books are written for an international audience. They are primarily written for parents of young children, but also for adolescents and adults who have the condition. They are written for members of multidisciplinary teams and researchers. Finally, they are written for others, including extended family members, teachers, and students taking courses in the fields of medicine, allied health care, and education.

A worldview

The books in the series focus on evidence-based best practice, which we acknowledge is not available everywhere. It is mostly available in high-income countries (at least in urban areas, though even there, not always), but many families live away from centers of good care.

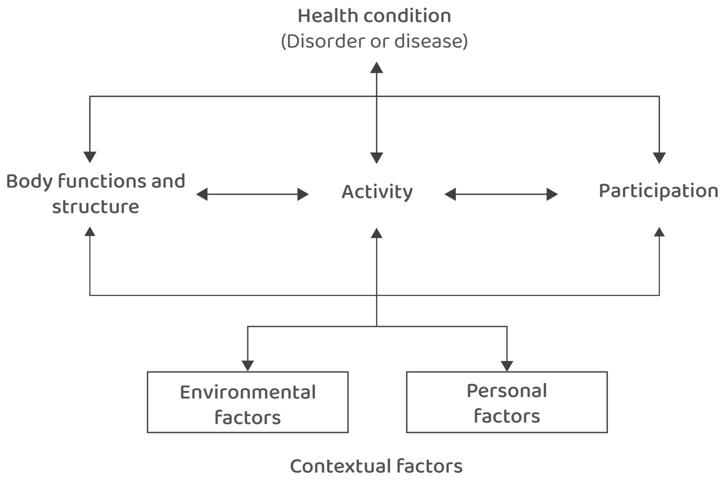
We also acknowledge that the majority of people with disabilities live in low- and middle-income countries. Improving the lives of all those with disabilities across the globe is an important goal. Developing scalable, affordable interventions is a crucial step toward achieving this. Nonetheless, the best interventions will fail if we do not first address the social determinants of health—the economic, social, and environmental conditions in which people live that shape their overall health and well-being.

No family reading these books should ever feel they have failed their child. We all struggle to do our best for our children within the limitations of our various resources and situations. Indeed, the advocacy role these books may play may help families and professionals lobby in unison for best care.

International Classification of Functioning, Disability and Health

The writing of the series of books has been informed by the International Classification of Functioning, Disability and Health (ICF).[1] The framework explains the impact of a health condition at different levels and how those levels are interconnected. It tells us to look at the full picture—to look at the person with a disability in their life situation.

The framework shows that every human being can experience a decrease in health and thereby experience some disability. It is not something that happens only to a minority of people. The ICF thus "mainstreams" disability and recognizes it as a widespread human experience.



International Classification of Functioning, Disability and Health (ICF). Reproduced with kind permission from WHO.

In health care, there has been a shift away from focusing almost exclusively on correcting issues that cause the individual's functional problems to focusing also on the individual's activity and participation. These books embrace maximizing participation for all people living with disability.

The family

For simplicity, throughout the series we refer to "parents" and "children"; we acknowledge, however, that family structures vary. "Parent" is used as a generic term that includes grandparents, relatives, and carers (caregivers) who are raising a child. Throughout the series, we refer to male and female as the biologic sex assigned at birth. We acknowledge that this does not equate to gender identity or sexual orientation, and we respect the individuality of each person. Throughout the series we have included both "person with disability" and "disabled person," recognizing that both terms are used.

Caring for a child with a disability can be challenging and overwhelming. Having a strong social support system in place can make a difference. For the parent, balancing the needs of the child with a disability with the needs of siblings—while also meeting employment demands, nurturing a relationship with a significant other, and caring for aging parents—can sometimes feel like an enormous juggling act. Siblings may feel neglected or overlooked because of the increased attention given to the disabled child. It is crucial for parents to allocate time and resources to ensure that siblings feel valued and included in the family dynamics. Engaging siblings in the care and support of the disabled child can help foster a sense of unity and empathy within the family.

A particular challenge for a child and adolescent who has a disability, and their parent, is balancing school attendance (for both academic and social purposes) with clinical appointments and surgery. Appointments outside of school hours are encouraged. School is important because the cognitive and social abilities developed there help maximize employment opportunities when employment is a realistic goal. Indeed, technology has eliminated barriers and created opportunities that did not exist even 10 years ago.

Parents also need to find a way to prioritize self-care. Neglecting their own well-being can have detrimental effects on their mental and physical health. Think of the safety advice on an airplane: you are told that you must put on your own oxygen mask before putting on your child's. It's the same when caring for a child with a disability; parents need to take care of themselves in order to effectively care for their child and family. Friends, support groups, or mental health professionals can provide an outlet for parents to express their emotions, gain valuable insights, and find solace in knowing that they are not alone in their journey.

For those of you reading this book who have the condition, we hope this book gives you insights into its many nuances and complexities, acknowledges you as the expert in your own care, and provides a road map and framework for you to advocate for your needs.

Last words

This series of books seeks to be an invaluable educational resource. All proceeds from the series at Gillette Children's go to research.

Chapter 1

Neural tube defects and the central nervous system

1.1

Introduction

> The mystery of life isn't a problem to solve,
> but a reality to experience.
> **Frank Herbert**

Neural tube defects are congenital conditions that occur when the neural tube, a structure that forms early in the development of a human embryo, fails to close properly. ("Congenital" means that the condition is present at birth.) Neural tube defects form very early in the pregnancy (within the first month). Congenital conditions can arise due to genetic factors, environmental factors, and other factors, but the exact cause of neural tube defects is not known.

The neural tube eventually develops into the central nervous system, which includes the brain and spinal cord. When the area of the neural tube that becomes the spinal cord does not close properly, the spinal cord, spinal nerves, and surrounding structures do not develop normally. These structures include the skin, muscles, and vertebrae (bones that surround and protect the spinal cord).

Spina bifida is the most common type of neural tube defect and is the focus of this book. “Spina bifida” means split spine (“bifida” means split; “spina” means spine), referring to an opening, or split, that occurs in the spine.[2]

The spinal cord is a cylindrical column of nerves that form the main communication pathway between the brain and the rest of the body. An atypical opening in the spine (called a “lesion”) in an individual with spina bifida allows the spinal cord and nerves to herniate* into a bulging sac. When that happens, the spinal cord and nerves are damaged by several factors, including pressure on the spinal cord and other structures from the uterine wall (during fetal movement or uterine contractions), and exposure to amniotic fluid (especially if the lining around the sac ruptures).[3–5] Figure 1.1.1 shows a developing fetus with spina bifida.

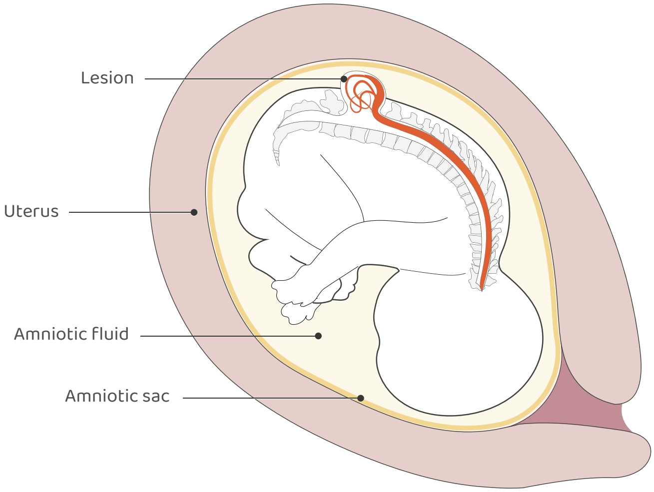


Figure 1.1.1 Fetus with spina bifida.

Spina bifida is considered a neuromuscular condition, which is a condition that affects the nervous system and muscles, and the communication between them. It is a complex, chronic medical condition that requires lifelong management. While the condition is challenging, with appropriate management, individuals with spina bifida can live a full life. Many finish high school, obtain a college degree, are employed, and live independently.[6]

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* Be displaced from their normal position.

Note: There are several types of spina bifida. This book focuses on the most common type, myelomeningocele,[7] often referred to as classic spina bifida. Additional information on other types of spina bifida can be found in **Useful web resources**.

How to read this book

To help you navigate the information in this book, it has been organized so that you can read it from beginning to end or, alternatively, refer to different sections and chapters independently. Because much of the information builds on previous sections and chapters, it is best to first read the book in its entirety to get an overall sense of the condition. After that, you can return to the parts that are relevant to you, knowing that you can ignore other sections or revisit them if and when they become relevant.

This chapter addresses neural tube defects and the anatomy and function of the central nervous system. Chapter 2 presents an overview of spina bifida, including types, the impact on function, and associated conditions. Chapter 3 covers spina bifida from the prenatal stage (before birth) to the newborn stage. Chapter 4 addresses the anatomy and function of the bowel, bladder, reproductive, and musculoskeletal systems, and Chapter 5 presents an overview of the management of spina bifida. Chapters 6 through 15 cover associated conditions and their management. Chapters 16 and 17 address the transition from childhood to adolescence and adulthood, and living as an adult with spina bifida.

Throughout the book, medical information is interspersed with lived experience. Orange boxes highlight the story of Madeline (Maddy) Lavalier, age 17 at the time of writing, and her mother, Rachelle. Both have written about their experience of living with Maddy's spina bifida. Chapter 18 is devoted to vignettes from other individuals with spina bifida and their families from around the globe. Chapter 19 provides information on further reading and research. At the back of the book, you'll find a glossary of key terms.

A companion website with appendices for this book is available at www.GilletteChildrensHealthcarePress.org. A QR code to access **Useful web resources** is included on the next page.

1.2

Prevalence and risk factors

Prevalence

“Prevalence” means “the proportion of persons in a population who have a particular disease or attribute at a specified point in time or over a specified period of time.”[8] Globally, approximately 300,000 babies are born with neural tube defects each year, and about half of those have spina bifida.[5,9,10] In the US, spina bifida occurs in approximately 1 of every 2,900 births.[11]

Prevalence of spina bifida varies by type. Myelomeningocele is the most common type and accounts for 76 percent of all cases in the US.[12] Prevalence also varies geographically and by socioeconomic status. Low-income countries have higher rates of neural tube defects,* particularly where there is no government-mandated program for folic acid fortification.[7,9,13] Race and ethnicity also play a part. For example, in the US, Hispanic Americans have a higher overall prevalence of spina bifida compared to the non-Hispanic white population or the non-Hispanic

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* Registry or surveillance systems that record and report birth defects such as neural tube defects are less available in low-income countries, making accurate estimates of prevalence difficult to obtain.

Black or African American population.[11,14] The reasons for these differences are not completely understood.

Risk factors

The US Centers for Disease Control and Prevention (CDC) defines the term “cause” as “a factor (characteristic, behavior, event, etc.) that directly influences the occurrence of disease.”[15] The term “risk factor” can be defined as “an aspect of personal behavior or lifestyle, an environmental exposure, or an inborn or inherited characteristic that is associated with an increased occurrence of disease or other health-related event or condition.”[15] Causes have a stronger relationship with a condition than do risk factors.

All pregnant women have a risk of having an infant with a neural tube defect, and certain risk factors are associated with the development of the condition. However, no single cause of a neural tube defect has been identified, and spina bifida is *not* caused by anything a pregnant woman did or did not do.[16]

Risk factors for the development of neural tube defects are divided into three categories:

- **Nutritional**
- **Genetic**
- **Environmental**

Having a combination of risk factors (referred to as multifactorial risk factors) most likely leads to an infant developing a neural tube defect.

a) Nutritional

The main nutritional risk factor for developing a neural tube defect is folate deficiency, which describes an inadequate amount of folate in the diet of the pregnant woman. Folate, or vitamin B9, is a naturally occurring compound found in fruits and leafy green vegetables. The synthetic form of folate is known as folic acid and is what is added to vitamins and dietary supplements.[4] Women with folate deficiency are two to eight times more likely to have a child with myelomeningocele (classic spina bifida).[7]

Both folate supplementation and folic acid fortification are methods to help reduce the risk of neural tube defects.

- **Folate supplementation** refers to taking the synthetic form of folate, folic acid, as a supplement to ensure adequate levels for the individual.
- **Folic acid fortification** is the process of adding folic acid to food, such as wheat flour, rice, or cereals, to ensure adequate levels in the population.

Women who could become pregnant are encouraged to take folic acid supplements. Studies have shown that the risk of occurrence of first-time and subsequent neural tube defects can be reduced by 60 to 70 percent when folic acid supplements are taken around the time of conception.[*13,17] Many supplements marketed as prenatal vitamins contain folic acid, but the amount varies. The following folate supplementation is recommended:[†]

- **Women who could become pregnant:** a daily supplement containing 0.4 mg of folic acid[18,19]
- **Women with a previous pregnancy with a neural tube defect, who have a child with spina bifida, or who have spina bifida themselves:** a daily supplement containing 4.0 mg of folic acid for one to three months before pregnancy[18]

Mandated folic acid fortification programs have been implemented in many areas worldwide, and the prevalence of spina bifida is lower in these areas compared to those without such programs.[20–22] For example, the US and Canada implemented mandatory folic acid fortification programs in 1998,[13,23] and both countries have seen positive results.[13,23] In the US, there has been a decrease of up to 32 percent in the occurrence of neural tube defects since the pre-fortification period,[24] and the severity of spina bifida has also decreased.[25] The decrease in overall

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[*] Some studies address the risk of neural tube defects in general, while others address spina bifida specifically.

[†] These are the recommendations in the US. Recommendations in other countries may vary.

spina bifida prevalence in Canada has varied by province with a 50 to 90 percent reduction since the start of fortification.[23]

b) Genetic

Genetics likely play a role in the development of spina bifida, and research is ongoing to identify genes that may cause the defect.[4,11]

Most pregnancies (95 percent) affected by spina bifida occur in women without a family history of spina bifida.[7] Having a family member with spina bifida, however, may increase the risk of having a child with spina bifida, though the risk decreases with the distance of the relationship.[26] Women with spina bifida themselves have an increased risk of having a child with a neural tube defect compared to the general population.[4,7,27] In addition, once a woman has one pregnancy affected by a neural tube defect, the risk of this happening again is about 3 to 8 percent.[11,22]

Certain chromosomal abnormalities or genetic syndromes* may be associated with spina bifida, but these account for a small number of cases.[10,26]

c) Environmental

Environmental risk factors are external physical, chemical, biological, and social influences that may negatively affect health. Those associated with higher rates of pregnancies affected by neural tube defects include:[4,22,26]

- Maternal obesity
- Maternal diabetes, both type 1 and type 2†
- Maternal and paternal exposure to organic solvents and pesticides such as those used in agriculture
- Maternal medications such as those used to manage seizures and epilepsy (Due to the risk of seizures during pregnancy, continuing to take antiseizure medications is typically recommended. Women who take

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* Syndromes are a group of characteristics and findings that consistently occur together and indicate a specific condition. A genetic syndrome is caused by one or more gene mutations or chromosomal abnormalities.

† Type 1 diabetes is an autoimmune condition in which the body destroys cells needed to make insulin. Type 2 diabetes is a condition in which the body either does not use insulin effectively (insulin resistance) or does not produce enough insulin to maintain normal blood sugar levels.

certain antiseizure medications may be advised to take an increased dose of folic acid—5 mg—when considering pregnancy.)

1.3

Diagnosis

Neural tube defects are typically diagnosed in the prenatal period ("pre" means before, "natal" means birth). In countries with screening programs for birth defects, over 90 percent of spina bifida cases are diagnosed prenatally.[10] Neural tube defects may also be diagnosed after birth, in the postnatal period ("post" means after).

Neural tube defects can be classified as closed or open:

- **Closed neural tube defect (occulta):** The lesion is covered by vertebrae and skin, protecting the neural tissues and structures from the external environment.
- **Open neural tube defect (aperta):** The lesion is not covered by vertebrae and lacks skin covering, exposing the neural tissues and structures to the external environment.

In general, open neural tube defects are more likely to be diagnosed prenatally compared to closed neural tube defects.

Prenatal diagnosis

Ultrasound is the most common diagnostic tool for identifying open neural tube defects.[10,13] Other diagnostic tools that may be used are the maternal serum alpha fetoprotein (MSAFP) test and amniocentesis.

a) Ultrasound

Ultrasound uses high-frequency sound waves to create black-and-white images of body tissues that are displayed on a monitor. Prenatal diagnosis of spina bifida with ultrasound has been used since the 1970s and today is considered the gold standard in identifying the condition.[10,13]

Prenatal ultrasound detection of spina bifida typically occurs in the second trimester, between 18- and 22-weeks' gestation, although first trimester ultrasound detection is sometimes possible. Ultrasound also allows visualization of the structures in the brain and other associated neurological conditions that may occur along with a neural tube defect, such as hydrocephalus or Chiari malformation type II (addressed in Chapter 6).[28,29]

A closed neural tube defect is not as reliably detected on ultrasound.[5,10,30,31] As well, factors such as maternal obesity, low levels of amniotic fluid, and positioning of the fetus may contribute to poor quality ultrasound images.[32]

b) MSAFP

An MSAFP test measures the level of alpha fetoprotein (AFP), a type of protein, in the mother's blood. When open neural tube defects occur, AFP, found in cerebrospinal fluid (fluid that surrounds the spinal cord and brain) can leak into the amniotic fluid and then into the mother's blood through the placenta. This can result in higher-than-expected levels of AFP in the mother's blood.[33] This test is performed in the second trimester, between 16- and 18-weeks' gestation. Closed neural tube defects cannot be detected with an MSAFP test.[34]

A positive MSAFP test result does not provide a definitive diagnosis of a neural tube defect, however, and false positive results can occur.[10,33-35] Having a pregnancy with multiple fetuses, or an incorrect estimate of the gestational age, or having some conditions that affect the placenta, may lead to higher-than-expected levels of AFP. Additional tests, such as ultrasound and amniocentesis, are therefore recommended to confirm positive MSAFP test results.[34,36]

c) Amniocentesis

Amniocentesis is a procedure in which a needle is inserted through the mother's abdomen into the amniotic sac to collect a sample of the amniotic fluid that surrounds the fetus. The level of AFP in the amniotic fluid is measured; elevated levels of AFP can indicate the presence of a neural tube defect.[10,36] Amniocentesis is typically done at 16- to 18-weeks' gestation.

Amniocentesis is also used to test for genetic conditions or infections in the fetus. There are risks associated with amniocentesis, which can be discussed with the medical professional.

Postnatal diagnosis

An open neural tube defect may be diagnosed after birth by the presence of an obvious lesion on the back of the infant. Postnatal diagnosis doesn't allow the same amount of preparation time for surgery as prenatal diagnosis does, and decisions may need to be made more rapidly. However, neural tube defects diagnosed postnatally are still highly manageable. Prompt evaluation by a pediatric neurosurgeon is essential.

A closed neural tube defect may be suspected after birth by the presence of a bulge, sacral dimple,[*] patch of hair, or red discoloration of the skin—typically along the spine—along with other signs such as muscle weakness or abnormal reflexes.[22,37,38]

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[*] A small indent in the area just above the buttocks, at the base of the spine.

Chapter 2

An overview of spina bifida

2.1

Introduction

The only person you are destined to become
is the person you decide to be.
Ralph Waldo Emerson.

The initial management of spina bifida focuses on restoring protection for the spinal cord and nerves by surgically closing the lesion either before birth or shortly after. While the protection of the spinal cord and nerves can be restored by closing the lesion, the damage that has already occurred can't be undone.

USEFUL WEB RESOURCES

2.2

Impact of spina bifida on the spine, spinal cord, and brain

The primary impact of spina bifida is on the spine, spinal cord, and brain.

Impact on the spine and spinal cord

Recall that spina bifida is a neural tube defect that results in incomplete closure of the vertebrae. Without the fully formed vertebrae, the contents inside the spinal column (spinal cord, spinal meninges, spinal nerves, CSF) do not remain in place and herniate through the opening.

Figure 2.2.1 shows a cross-section of a single vertebra: one typical and one with incomplete closure (as occurs with spina bifida). Individuals with spina bifida typically have several vertebrae in a row that fail to fully close.

Figure 2.2.2 depicts both a typical spinal cord and one with a lesion. The image of the infant shows what the lesion itself looks like.

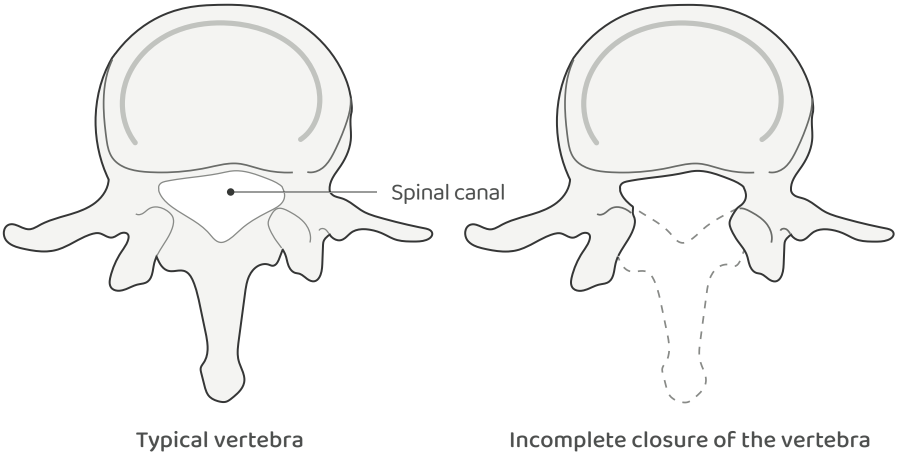

Figure 2.2.1 Left: Typical vertebra. Right: Incomplete closure of a vertebra with outline of the missing bony structures.

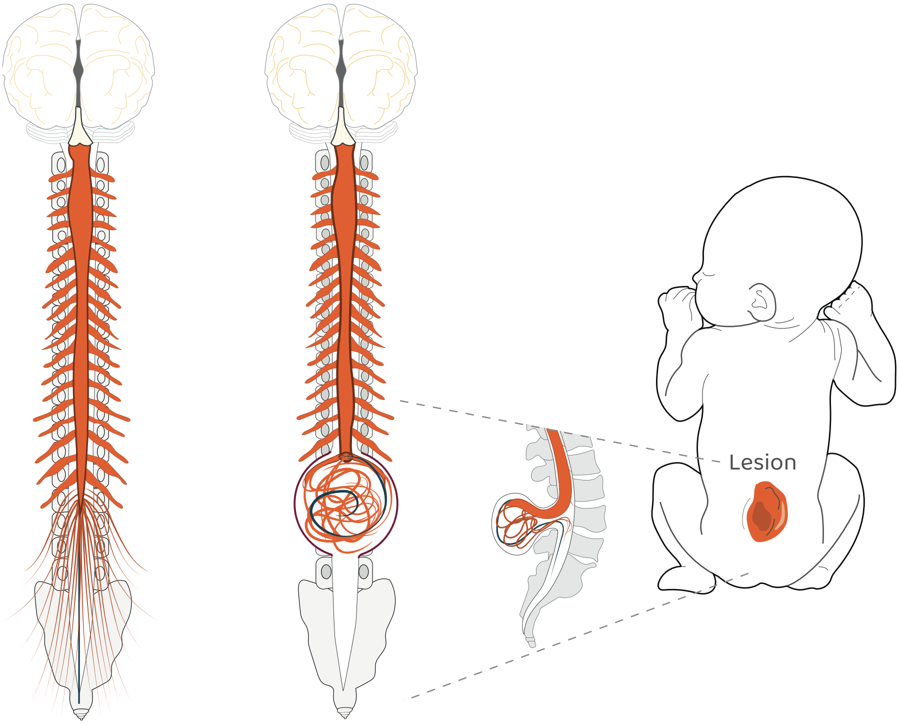

Figure 2.2.2 Left: Typical spinal cord, posterior view. Middle: Spinal cord with lesion, posterior and lateral (side) views. Right: Infant with a lesion on their back.

The central nervous system (CNS) plays a key role in controlling movement, while spinal nerves are responsible for transmitting motor and sensory information. The function of the spinal nerves that branch out from the spinal cord depends on the level of the spinal column they exit. The spinal nerves include:

- Motor and sensory nerves, which impact both motor and sensory function. “Motor function” refers to the ability to move; “sensory function” refers to the ability to receive and interpret information from the environment (e.g., touch).
- Autonomic nerves, which control involuntary (or automatic) functions such as digestion, breathing, heart rate, and bloodflow (circulation). Autonomic nerves may be sympathetic (responsible for automatic fight-or-flight function) or parasympathetic (responsible for automatic rest and digest functions). Sympathetic and parasympathetic nerves work together, helping create balance within the body. An example is bladder function that is impacted both by the sympathetic nerves that arise from vertebrae T10 to L2 and the parasympathetic nerves that arise from vertebrae S2 to S4.

Individuals with spina bifida have impaired motor, sensory, and autonomic function in areas of the body that are *at and below the level of the lesion*. The higher the lesion level, the greater the level of impairment.

Nerves branch out from the spinal cord in pairs and are categorized by the vertebral level from which they originate:

- Cervical nerves are C1 to C8.*
- Thoracic nerves are T1 to T12.
- Lumbar nerves are L1 to L5.
- Sacral nerves are S1 to S5.

Figure 2.2.3 is a simplified depiction of the nerves and their impact on motor, sensory, and autonomic function at each vertebral level. Not shown is that the motor function of each muscle or area of the body (e.g., the legs) is controlled by nerves at several different vertebral levels. This is an important concept to understand as individuals with spina bifida often have muscle function that is different than what the

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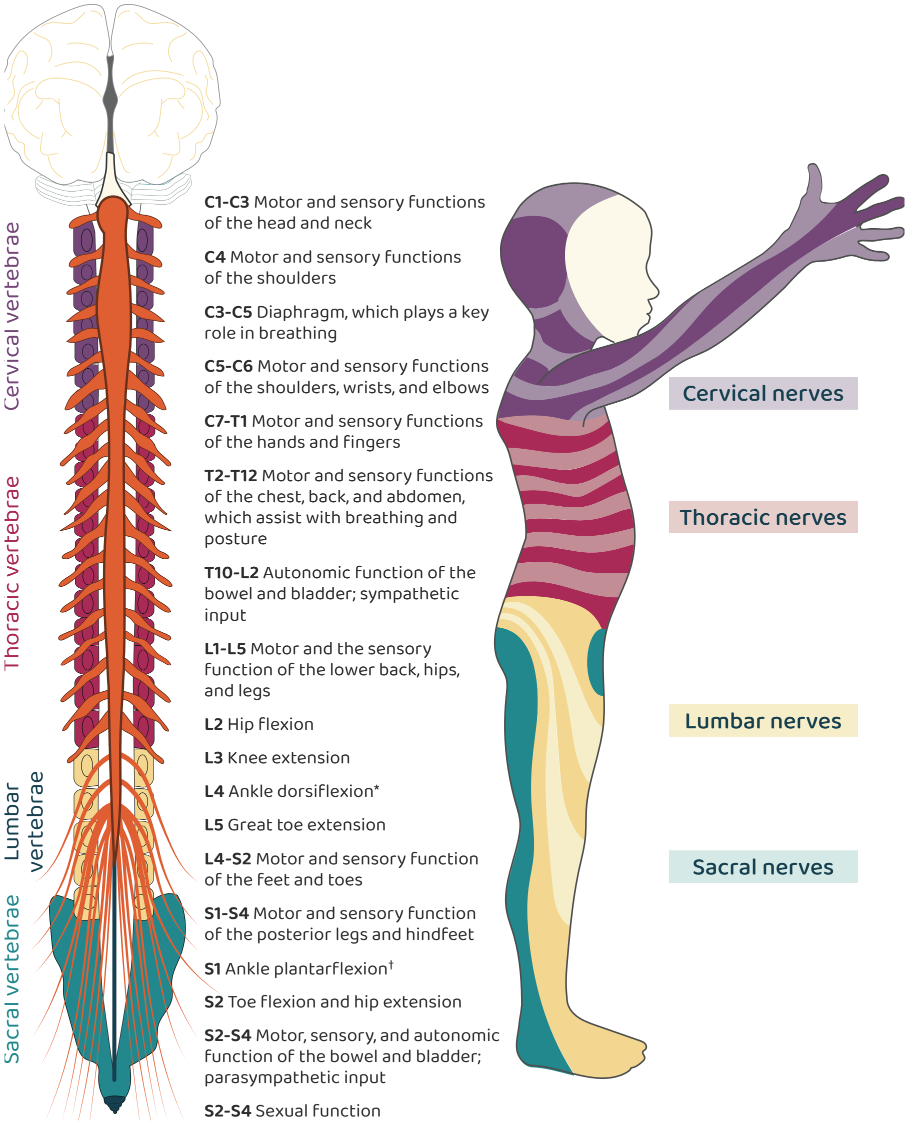
* There are seven cervical vertebrae, but there are eight cervical nerves.

anatomical level of the lesion may predict. It's also important to know that spina bifida can occur in any region of the spinal column, but it most commonly occurs in the lumbar and sacral regions.[47]



C1-C3 Motor and sensory functions of the head and neck

C4 Motor and sensory functions of the shoulders

C3-C5 Diaphragm, which plays a key role in breathing

C5-C6 Motor and sensory functions of the shoulders, wrists, and elbows

C7-T1 Motor and sensory functions of the hands and fingers

T2-T12 Motor and sensory functions of the chest, back, and abdomen, which assist with breathing and posture

T10-L2 Autonomic function of the bowel and bladder; sympathetic input

L1-L5 Motor and the sensory function of the lower back, hips, and legs

L2 Hip flexion

L3 Knee extension

L4 Ankle dorsiflexion*

L5 Great toe extension

L4-S2 Motor and sensory function of the feet and toes

S1-S4 Motor and sensory function of the posterior legs and hindfeet

S1 Ankle plantarflexion†

S2 Toe flexion and hip extension

S2-S4 Motor, sensory, and autonomic function of the bowel and bladder; parasympathetic input

S2-S4 Sexual function

Figure 2.2.3 Spinal nerve function.

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* Pointing the foot upward, bringing the toes closer to the shin.

† Pointing the foot downward, moving the toes away from the shin.

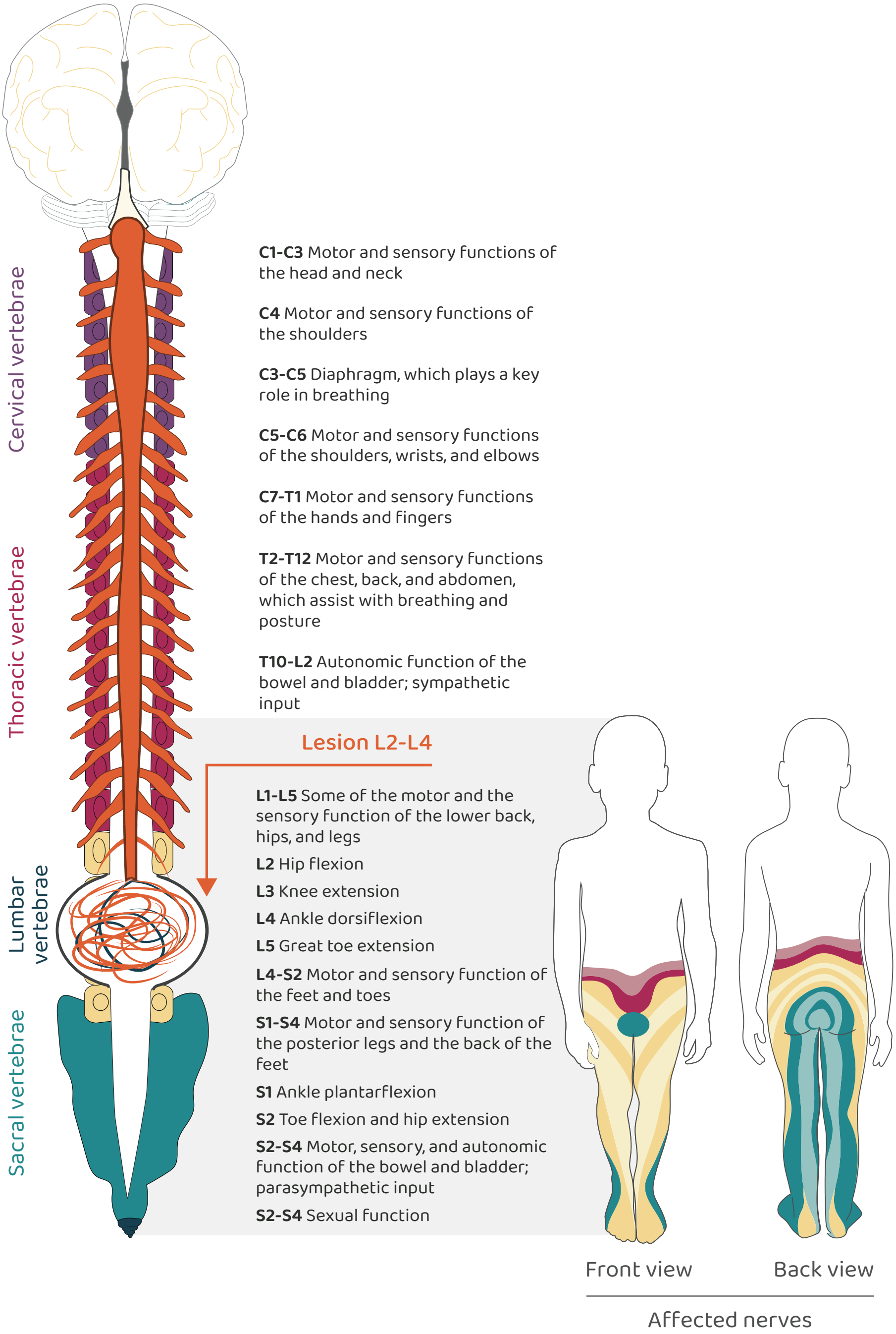
The location of the lesion in the spine is referred to as the anatomical level of the lesion. Figure 2.2.4 shows a spine with a lesion located starting at L2 and extending to L4. In this example, the individual's motor and sensory functions to the lower back, hip and leg muscles, and bowel and bladder, along with sexual function, are likely impacted. Autonomic functions such as the regulation of bloodflow is also impacted; in combination with decreased motor function, this results in decreased circulation to these areas. Consequently, individuals with spina bifida may have colder lower extremities and skin color changes. Function below the highest level of the lesion is disrupted.

It is important to remember, however, that each individual is different, and the impact on two different individuals with a lesion at the exact same level can differ. This occurs because the anatomical level of the lesion may not correlate with the functional level of the lesion (which is based on the level of the spinal cord and nerves with preserved motor function and varies between individuals) and correlates to the functional ability an individual is *likely* to have.

Individuals with spina bifida may also experience other neurological conditions of the spine, including:

- Syringomyelia, an atypical widening of the central canal in the spinal cord, which results in the formation of a fluid-filled area in the spinal cord, known as a "syrinx"
- Tethered cord, a condition in which the spinal cord abnormally attaches to a structure inside the spinal column, restricting its movement

These conditions are addressed in Chapter 6.



C1-C3 Motor and sensory functions of the head and neck

C4 Motor and sensory functions of the shoulders

C3-C5 Diaphragm, which plays a key role in breathing

C5-C6 Motor and sensory functions of the shoulders, wrists, and elbows

C7-T1 Motor and sensory functions of the hands and fingers

T2-T12 Motor and sensory functions of the chest, back, and abdomen, which assist with breathing and posture

T10-L2 Autonomic function of the bowel and bladder; sympathetic input

L1-L5 Some of the motor and the sensory function of the lower back, hips, and legs

L2 Hip flexion

L3 Knee extension

L4 Ankle dorsiflexion

L5 Great toe extension

L4-S2 Motor and sensory function of the feet and toes

S1-S4 Motor and sensory function of the posterior legs and the back of the feet

S1 Ankle plantarflexion

S2 Toe flexion and hip extension

S2-S4 Motor, sensory, and autonomic function of the bowel and bladder; parasympathetic input

S2-S4 Sexual function

Figure 2.2.4 Spinal nerves affected in an individual with a lesion from L2 to L4.

Impact on the brain

In an individual without spina bifida, the CSF flows in a steady, balanced way. The pressure inside the brain and spine stays at a healthy level to keep everything working properly; CSF flows from the ventricles out around the spinal cord and back over the surface of the brain.

In an individual with spina bifida, however, the CSF flow and absorption within and around the brain is disrupted, which results in excess volume in the ventricles. The flow can be disrupted by the inability of the fluid to move normally or, more commonly, by the inefficient absorption of the CSF at the surface of the brain.

Two key conditions that develop in the brain in individuals with spina bifida are **hydrocephalus** and **Chiari malformation type II**.

a) Hydrocephalus

Hydrocephalus occurs when enlarged ventricles in the brain (a condition known as ventriculomegaly) result in increased intracranial pressure (ICP), which is the pressure exerted within the skull by the combined volume of the brain, CSF, and blood. Ventriculomegaly occurs when more CSF is being produced than absorbed. To compensate for this imbalance, the ventricles enlarge to create more space to store the excess volume of CSF. When increased ICP occurs because of the enlarged ventricles, the result is hydrocephalus. However, ventriculomegaly can occur without increased ICP.

Normal ICP allows for optimal brain growth, development, and function. An increase in the amount of any of the contents within the skull (e.g., excess CSF) may result in increased ICP. Prolonged increased ICP can cause brain damage.

Figure 2.2.5 shows a lateral (side) view of an individual without hydrocephalus (normal volume of CSF in the ventricles) and an individual with hydrocephalus (excess volume of CSF in the ventricles).

Management of hydrocephalus involves surgically placing a device known as a shunt in the ventricles of the brain to drain the excess volume of CSF into other areas of the body, most commonly in the abdomen.

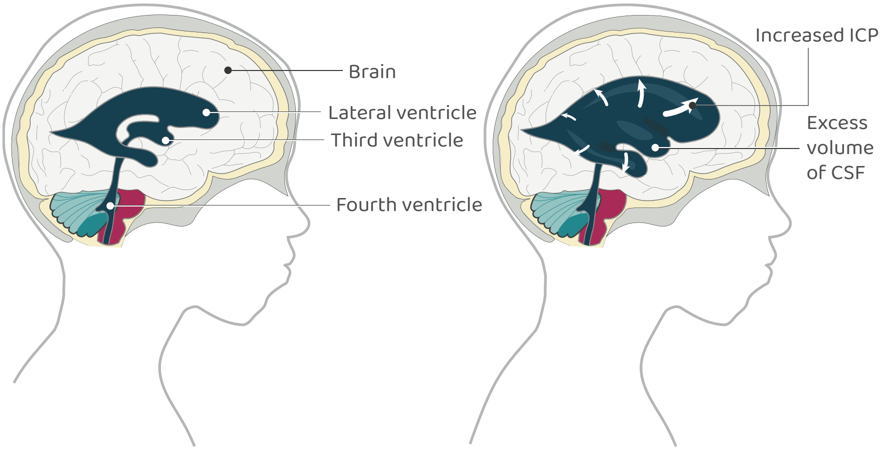


Figure 2.2.5 Left: Individual without hydrocephalus. Right: Individual with hydrocephalus. The arrows indicate increased ICP.

b) Chiari malformation type II

Chiari malformation type II is a condition in which contents of the brain (the cerebellar tonsils) are displaced downward into the foramen magnum (a bony opening at the base of the skull). Since the foramen magnum is made of bone and is not flexible, the displaced contents, as well as structures near the cerebellar tonsils (the brain stem and spinal cord), may become compressed. Chiari malformation type II occurs during the development of the brain and spinal cord in an individual with spina bifida.

Figure 2.2.6 shows a lateral (side) view of an individual without Chiari malformation type II and one with spina bifida and Chiari malformation type II (note the cerebellar tonsils shifting into the foramen magnum).

Hydrocephalus and Chiari malformation type II and their management in spina bifida are addressed in Chapter 6.

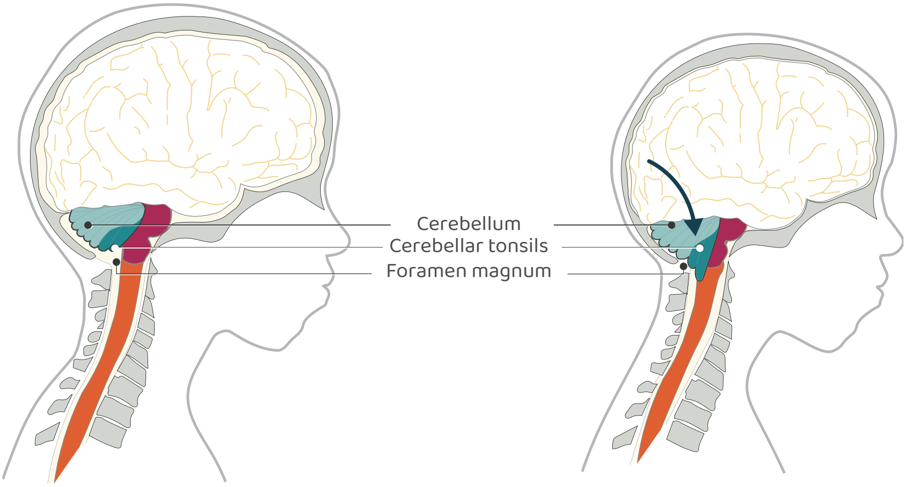


Figure 2.2.6 Left: Individual without Chiari malformation type II. Right Individual with Chiari malformation type II. The arrow indicates the cerebellar tonsils shifting into the foramen magnum.

2.3

Types of spina bifida

There are three main types of spina bifida:

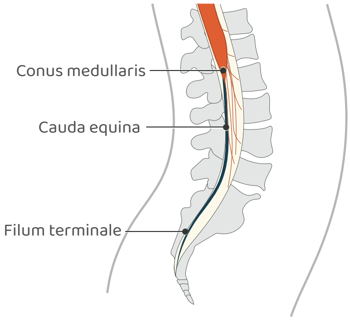
- Spina bifida occulta
- Meningocele
- Myelomeningocele

This book focuses on the last of these, myelomeningocele, which is also termed classic spina bifida.

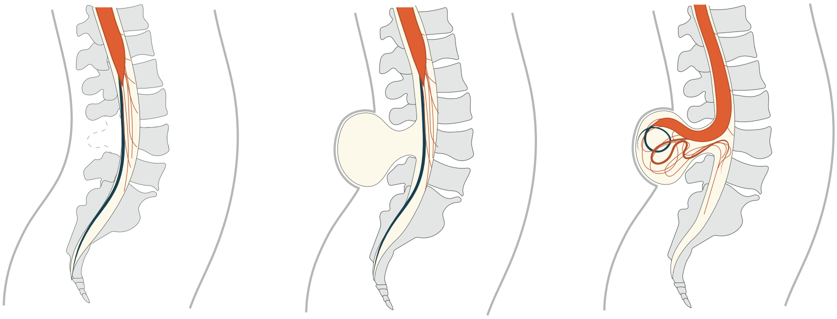
The three types of spina bifida are differentiated by which contents are displaced from their typical position in the spinal column. In general, the more contents displaced, the more severe the **signs** and **symptoms** the individual experiences.

- A **sign** is what can be seen (e.g., a dimple on the skin).
- A **symptom** is what the individual experiences (e.g., headaches).

Figure 2.3.1 shows a typical spinal cord compared to one in each type of spina bifida.



Typical spinal cord



Spina bifida occulta

Meningocele

Myelomeningocele

Figure 2.3.1 A typical spinal cord compared to three types of spina bifida.

Further details about the three types and the differences between them are as follows:

- **Spina bifida occulta** is a closed neural tube defect; the spinal cord and nerves remain in their normal position. A layer of skin is present over the area of the defect and may include a sacral dimple, bulge, patch of hair, or red discoloration in the skin.[38] (See Figure 2.3.2.) Sometimes, muscle weakness or abnormal reflexes are present, as well as bowel or bladder dysfunction. However, this type of spina bifida can occur without any signs or symptoms and is typically discovered in late childhood or adulthood, often found during an examination performed for other reasons. Spina bifida occulta is fairly common, with an estimated prevalence of 12 percent in the typical population.[38]

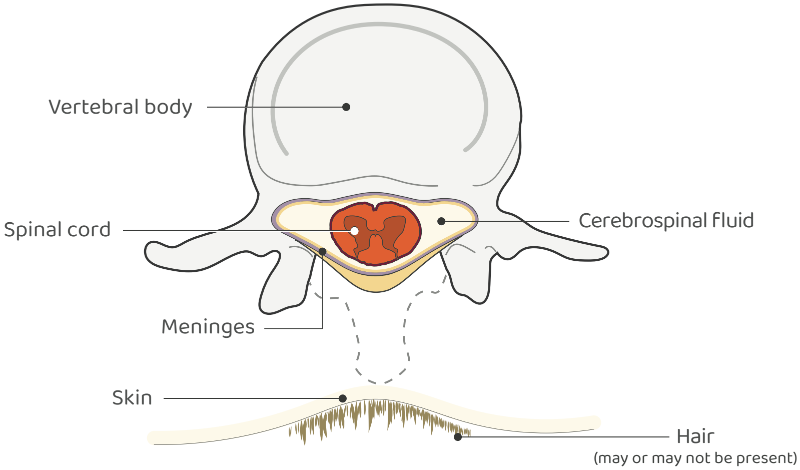


Figure 2.3.2 Cross-section of vertebra with spina bifida occulta.

- **Meningocele** is generally considered an open neural tube defect even if a layer of skin is present over the defect. Meninges are displaced from their normal position, filling with CSF. The spinal cord and nerves usually remain in their normal position. Skin may or may not cover the meninges. (See Figure 2.3.3.) Individuals with a meningocele may experience neurological symptoms including leg weakness, abnormal sensation, or challenges with movement. Bowel and bladder dysfunction may also occur but is less common than in the next type, myelomeningocele. A subtype of meningocele is known as a lipomeningocele, which includes a lipoma (a collection of fat that forms under the skin), as shown in Figure 2.3.4.

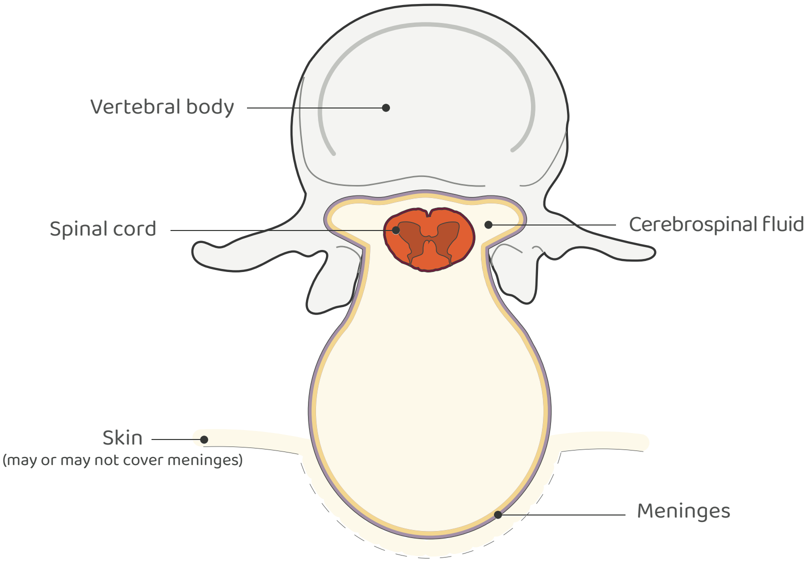


Figure 2.3.3 Cross-section of vertebra with meningocele.

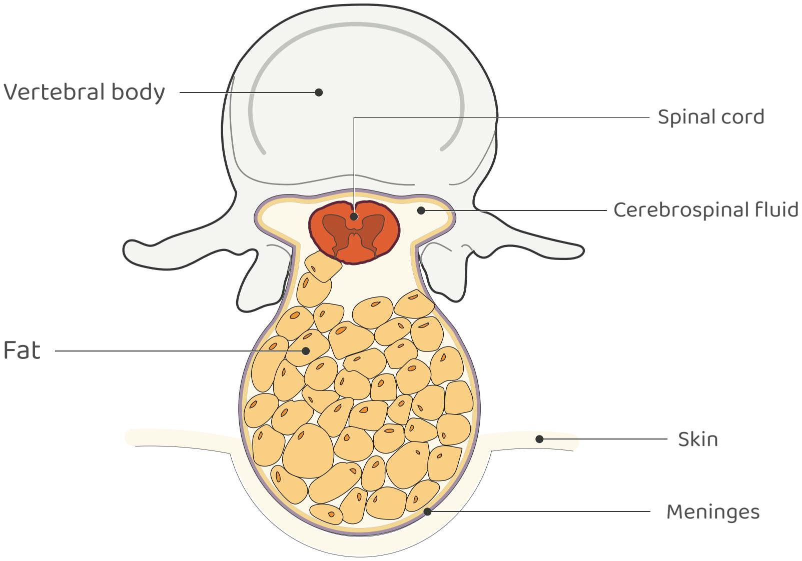


Figure 2.3.4 Cross-section of vertebra with lipomeningocele.

- **Myelomeningocele** is an open neural tube defect. The meninges, along with the spinal cord and nerves, are displaced from their normal position, filling with CSF. Skin is typically absent over the area of the defect. (See Figure 2.3.5.) This is the most common and severe type of spina bifida, and individuals with it may experience muscle weakness, atypical sensation, paralysis, bowel and bladder dysfunction, hydrocephalus, syrinx, tethered cord syndrome, Chiari malformation type II, and neuropsychological conditions.* A subtype of myelomeningocele is known as a lipomyelomeningocele, and includes a lipoma (a collection of fat that forms under the skin), as shown in Figure 2.3.6.

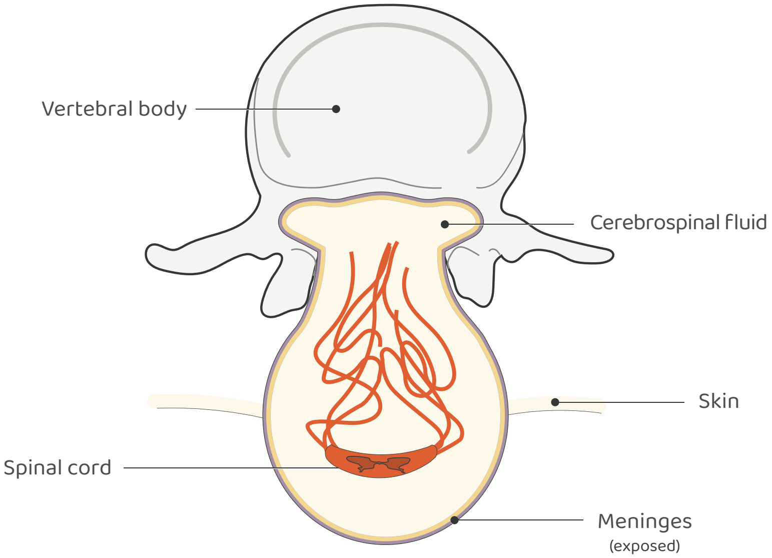


Figure 2.3.5 Cross-section of vertebra with myelomeningocele.

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* Conditions that affect how the brain functions, impacting thinking, memory, behavior, emotions, or learning abilities. See Chapter 15.

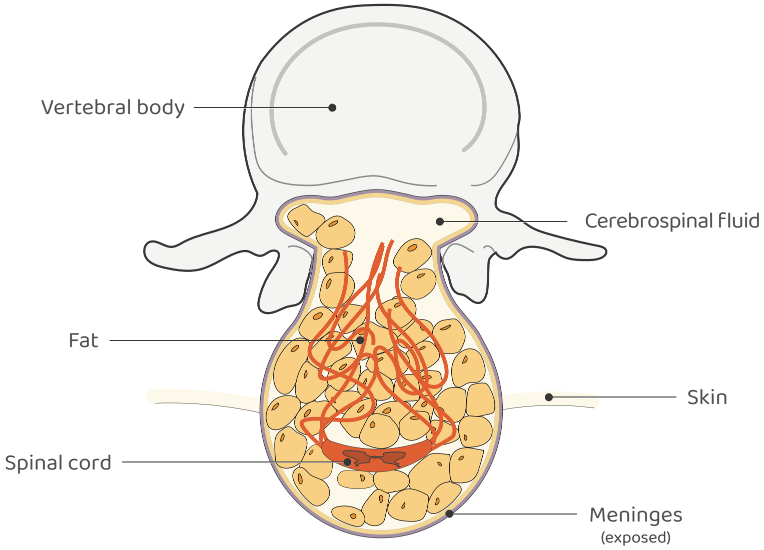

Figure 2.3.6 Cross-section of vertebra with lipomyelomeningocele.

Two conditions of the spinal cord that may occur along with spina bifida include:

- **Split cord malformation:** The spinal cord is split into two parts, divided by a septum, which is either rigid or nonrigid. Split cord malformation is also often accompanied by a tethered cord, a condition in which the spinal cord abnormally attaches to a structure inside the spinal column, restricting its movement. A split cord malformation may occur independently or along with myelomeningocele.
- **Terminal myelocystocele:** The central canal expands and forms a fluid-filled area known as a cyst. As with typical myelomeningocele, the meninges, along with the spinal cord, nerves, and CSF, are displaced from their normal position and herniate through the spinal defect. A syrinx may form in this area and attach to a layer of fat on the skin. “Terminal” refers to the location of the myelocystocele, which occurs at the terminal end of the central canal.

Note: As this book focuses on myelomeningocele (also termed classic spina bifida), the term “spina bifida” is used to refer to this type throughout the remainder of the text.

2.4

Prognosis

"Prognosis" means the "prospect of recovering from injury or disease, or a prediction or forecast of the course and outcome of a medical condition."[48] As it relates to spina bifida, prognosis can provide a prediction of how the condition may impact individuals across the lifespan, including quality of life* and life expectancy. Although spina bifida is a chronic condition and there is no cure, appropriate management of the condition can improve the prognosis and make living a long and fulfilling life possible.

The survival rate of infants born with spina bifida has increased in the last 50 years: today, approximately 90 percent of infants born with spina bifida survive the first year of life, and 83 percent survive to age 18.[49,50] The most common cause of death in the first year is infection; the most common cause in ages 1 to 18 years is complications related to hydrocephalus.[49] The presence of genetic syndromes or multiple birth defects, very premature birth, or low birth weight increase the risk of death in the first month.[49–52]

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* An individual's perception of their position in life (sense of well-being, happiness, and satisfaction) in the context of the culture and value system in which they live, and in relation to their goals, expectations, standards, and concerns.

Life expectancy for those with spina bifida has also increased dramatically over the last decades due to advances in health care.

2.5

Functional ability

Parents of infants newly diagnosed with spina bifida often ask what their child will be able to do. Knowing what to expect can help parents set realistic expectations. “Functional ability” is a measure of an individual’s ability to carry out activities of daily living that are tied to maintaining independence and overall well-being, such as mobility, feeding, and performing personal care tasks. Functional ability is influenced by the level of the lesion. A lower-than-predicted functional ability may occur due to an interruption in the coordination of function between the brain and spinal cord and associated neurological conditions, such as hydrocephalus.

Level of lesion

The *anatomical* level of the lesion (also referred to as the surgical level) is the area of incomplete closure of the neural tube and its contents. It is determined by imaging, either prenatally or postnatally, before repair.

The *functional* level of the lesion, which is related to the anatomical level, is the lowest level of spinal nerves with preserved function. It is more often used to describe individuals with spina bifida than is the anatomical level as it better predicts functional ability.

Individuals with prenatal repair (addressed in Chapter 3) are more likely to have a functional level of the lesion that is two or more levels lower* (further down the spinal cord) than the anatomical level of the lesion.[53] For example, if an individual has a lesion at the mid-lumbar level (anatomical level) and undergoes prenatal repair, they are more likely to have a functional level of the lesion at the lower-lumbar level.

The functional level is determined by physical examination and other tests (not imaging), such as manual muscle testing, which is addressed in Chapter 4. The functional level of the lesion is generally what is referred to when describing an individual's functional ability, and it is used when describing the impact of the level of lesion on associated conditions. It is common to have a different functional level between the right and left sides of the body.

Based on data from almost 12,000 individuals with spina bifida, the US National Spina Bifida Patient Registry reported functional level over several years as follows (see Figure 2.5.1):[12]

- 14 percent thoracic
- 53 percent lumbar (divided into high-, mid-, and low-lumbar)
- 33 percent sacral

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* In the spinal cord, functional levels that are lower result in better functional abilities; therefore, lower is better when referring to functional levels.

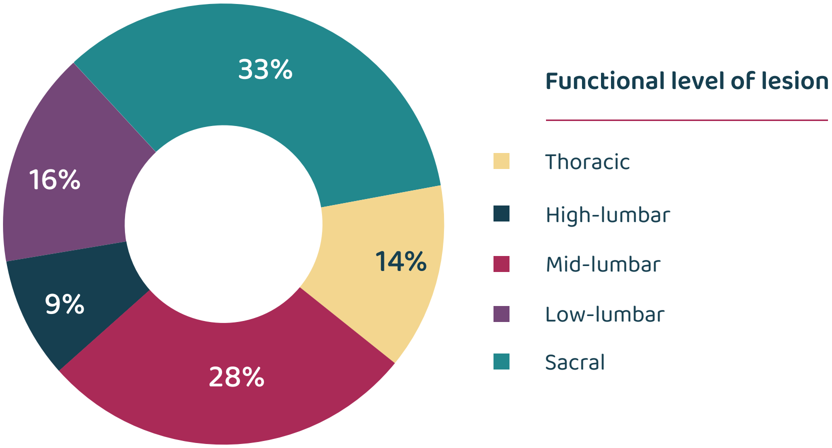

Figure 2.5.1 Functional level of lesion in individuals with spina bifida in US registry, 2009–2022. Source: National Center on Birth Defects and Developmental Disabilities. Materials developed by CDC, available in the public domain at https://www.cdc.gov/spina-bifida/data/findings-from-the-nsbpr-registry.html

Functional mobility

A subset of functional ability is functional mobility, which is the ability to move from place to place to perform daily tasks and activities. It includes navigating various environments (e.g., home, school, work, the community).

Walking is part of functional mobility. More than half of all children with spina bifida with lesions at or below the thoracic level of the spine are able to walk.[54] Many use mobility aids* or orthoses.† In general, the lower the level of the lesion in the spine, the more likely the individual will successfully walk. Walking ability, however, tends to decrease once the individual enters adolescence, but it stabilizes through early adulthood. Table 2.5.1 shows the functional level of the lesion and associated walking ability at different points in time.[55–57]

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* Devices that enhance mobility, independence, and safety. Also termed assistive mobility devices, assistive devices, walking aids, and gait aids.

† Devices designed to hold specific body parts in position to modify their structure or function. Also termed orthotics, splints, or braces.

Table 2.5.1 Functional level of lesion and associated walking ability

FUNCTIONAL LEVEL OF LESION	WALKING ABILITY
Thoracic/High-lumbar	• Not common • May walk very short distances in childhood with mobility aid and hip-spanning orthoses* • Wheelchair mobility
High-lumbar	• May walk short distances in childhood with mobility aid and orthoses • Wheelchair mobility in most settings
Mid-lumbar	• May walk in childhood with mobility aid and orthoses. • Wheelchair mobility for longer distances
Low-lumbar	• Likely will walk with ankle-foot orthoses† • May require mobility aid for walking and wheelchair for longer distances • Majority maintain walking in adulthood
Lumbo-sacral	• Walk with or without orthoses • Majority maintain walking in adulthood
Sacral	• Walk without orthoses or support • Majority maintain walking in adulthood

* A type of orthosis that spans from the hip to the foot, providing support at the hip, knee, and ankle.

† A type of orthosis that controls the ankle and foot.

Each individual with spina bifida is unique, and level of the lesion cannot fully predict their ability. It is important that the individual's ability not be underestimated as the functional ability may be greater than predicted. It is also important to focus on the many things the child *can* do rather than on what they may not be able to do.

2.6

Associated conditions

"Associated conditions" refers to conditions that occur in individuals in addition to their primary condition. Associated conditions commonly occur in individuals with spina bifida and range from mild to severe depending on the size, level, and severity of the lesion. The higher the level of the lesion, the more likely multiple associated conditions occur.

Associated conditions, either present at birth or that develop as the individual grows, may include the following:

- **Neurological conditions:** These are conditions that impact the nervous system—the brain, spinal cord, and nerves. Neurological conditions are present in the majority of individuals with spina bifida and are caused by the altered flow or absorption of CSF, scarring around the spinal cord after the repair of the lesion, or damage to nerves and other structures. Neurological conditions may also include seizures. Neurological conditions are addressed in Chapter 6.
- **Neurogenic bowel and bladder:** The term "neurogenic" refers to the alteration of typical nerve function that causes impaired motor and sensory function.

  - An individual with a neurogenic bowel may have difficulties either storing feces or effectively emptying the bowel.
  - An individual with a neurogenic bladder may have difficulties either storing urine or effectively emptying the bladder.

  Neurogenic bowel and bladder are present in the majority of individuals with spina bifida. Other conditions that impact the bowel, bladder, and the urinary system may also develop. Neurogenic bowel and bladder are addressed in Chapters 7, 8, and 9.
- **Reproductive system conditions:** These are conditions that impact the reproductive system, puberty, and sexual health and include pelvic organ prolapse, precocious puberty, fertility, and sexual dysfunction. They occur due to the impact of the lesion on the nerves that regulate reproductive organs and contribute to sexual function. Reproductive system conditions are addressed in Chapter 10.
- **Musculoskeletal conditions:** These are conditions that impact the muscles, bones, joints, and their related structures. They develop due to disrupted messages between the brain and the muscles, resulting in muscle paralysis, weakness, and imbalance; uncoordinated movements; and shorter stature than typically developing peers. Musculoskeletal conditions impact functional mobility. They are addressed in Chapters 11 and 12.
- **Skin conditions:** These often occur due to insensate skin, which is skin with diminished or absent sensation. Skin conditions are addressed in Chapter 13.
- **Other physical conditions:** Obesity, metabolic syndrome, lymphedema, sleep-related breathing disorders, pain, latex allergy, and eye conditions also may occur. They are addressed in Chapter 14.
- **Neuropsychological and mental health conditions:** Neuropsychological conditions affect how the brain functions, impacting thinking, memory, behavior, emotions, or learning abilities. They may include cognitive impairment, speech and language challenges, executive dysfunction, attention problems, and motor delays. Challenges with social functioning may also occur. Mental health conditions impact an individual's thoughts, feelings, and behavior. They include depression and anxiety. Neuropsychological and mental health conditions are addressed in Chapter 15.

In hindsight, even though Maddy's diagnosis came as a shock, I am grateful that my physician had accurate information about spina bifida and that I learned about it when I did. I was immediately referred to a specialty clinic that could best support me moving forward. But I left the doctor's office numb and robbed of the joy I had dreamed of. Tears streamed down my face. I was angry, sad, confused, and unsure of what this all meant for my life, for my child's life, or if they would have a life. I screamed to God, "Why? Why my baby? What did I do wrong?!" Not knowing what to do, I called my mother, the person that I knew was always there for me. She knew I had the appointment that day, so when I called, she excitedly asked how it went. I tried to hold it all in, all of the pain, sadness, and fear. I couldn't do it. I felt my lip fall and start to tremble, tears falling from my face. In an instant I went from a soon-to-be-mama to a child needing my own mother.

I cried, "Mom, something is wrong with my baby." My mom cried with me but said something to me that is forever ingrained in my being. She said, "Rachelle, God knows you and would never put anything in your life that you could not handle." I left the conversation feeling like I had a Band-Aid on my wound: the pain was still there, but I felt a little better.

After driving aimlessly around the city awhile, I went to the library to try and educate myself about omphaloceles, clubfoot, and spina bifida. I wasn't as concerned about the omphalocele and clubfoot after reading about them as they seemed repairable. It was the spina bifida that frightened me the most, and since I had no idea what type it was, I was left in the dark. She could have a mild form with very few symptoms, or a severe form of myelomeningocele causing many issues. But as I drove away from the library, I had a feeling of strength come over me. There was nothing I could do now other than educate myself about my daughter's condition, go to the doctor, follow their recommendations, pray for the best, and prepare for the worst. The specialist scheduled me for a C-section to be done around mid-January, near my due date.

Key points Chapter 2

- The initial management of spina bifida focuses on restoring protection for the spinal cord and nerves by surgically closing the lesion either before birth or shortly after.
- Individuals with spina bifida have impaired motor, sensory, and autonomic function in areas of the body that are at and below the level of the lesion. The higher the lesion level, the greater the level of impairment.
- Spina bifida can occur in any region of the spinal column, but it most commonly occurs in the lumbar and sacral regions.
- In an individual with spina bifida, the CSF flow and absorption within and around the brain are disrupted, which results in excess volume of CSF in the ventricles.
- Hydrocephalus occurs when enlarged ventricles in the brain (a condition known as ventriculomegaly) result in increased intracranial pressure (ICP). Prolonged increased ICP can cause brain damage.
- Chiari malformation type II is a condition in which contents of the brain (the cerebellar tonsils) are displaced downward into the foramen magnum (a bony opening at the base of the skull).
- The three types of spina bifida are differentiated by which contents are displaced from their typical position in the spinal column. In general, the more contents that are displaced, the more severe the signs and symptoms the individual will experience.
- Spina bifida occulta is a closed neural tube defect. The spinal cord and nerves remain in their normal position. A layer of skin is present and may include a sacral dimple, bulge, patch of hair, or red discoloration in the skin.
- Meningocele is generally considered an open neural tube defect even if a layer of skin is present. Meninges are displaced from their normal position, filling with CSF. The spinal cord and nerves usually remain in their normal position. Skin may or may not cover the meninges.
- Myelomeningocele is an open neural tube defect. The meninges, along with the spinal cord and nerves, are displaced from their normal position, filling with CSF. With myelomeningocele, skin is typically absent.

- Approximately 90 percent of infants born with spina bifida survive the first year of life, and 83 percent survive to age 18.
- The functional level of the lesion is the lowest level of spinal nerves with preserved function and is more often used to describe individuals with spina bifida than is the anatomical level of the lesion as it better predicts functional ability.
- For children with spina bifida with lesions at or below the thoracic level of the spine, more than half will be able to walk. Many require mobility aids or orthoses.
- Associated conditions, which often accompany spina bifida, vary in severity based on the size, level, and severity of the lesion, with higher lesions increasing the likelihood of multiple associated conditions.

Key points Chapter 8

- In general, the philosophy of managing neurogenic bowel in individuals with spina bifida is to start with the least invasive treatment before progressing to more invasive treatment.
- Bowel management for an individual with spina bifida is often done within a bowel program—a structured regimen used to maintain regular, controlled bowel movements. Adaptive toileting equipment may be included in a bowel program.
- Bowel management is generally divided into nonsurgical and surgical management. The needs of individuals with spina bifida change through life and some treatment options are more appropriate at certain ages. A bowel program that can be performed independently (at age-appropriate times) should be prioritized when possible.
- A bowel cleanout is a procedure used to clear all the stool from the bowel. It may be done to prepare for a medical test or surgery or to treat severe constipation and impacted stool.
- Bowel-related complications may occur and include rectal bleeding, rectal prolapse, defecation syncope, and autonomic dysreflexia.

Key points Chapter 11

- Musculoskeletal conditions occur in individuals with spina bifida due to disrupted messages between the brain and the muscles as a result of the lesion. These disrupted messages can lead to the muscles inadequately contracting, resulting in muscle paralysis, weakness, muscle imbalance, and uncoordinated movements.
- Individuals with spina bifida tend to have lower bone density than those without spina bifida, and early-onset osteoporosis (osteoporosis beginning in the 20s or 30s) develops in almost half of all adults with spina bifida.
- A fracture is a break or crack in a bone. Individuals with spina bifida experience fractures at a reported rate of 11 to 30 percent, and fractures commonly occur below the level of the lesion.
- Individuals with spina bifida may have atypical bone or joint positions that can lead to stress on the joints and may make walking difficult or painful.
- Lower extremity conditions that may occur in individuals with spina bifida include femur and tibia conditions such as femoral and tibial torsion, knee conditions such as knee flexion or extension contractures, and ankle and foot conditions such as clubfoot.
- Hip and spine conditions that may occur in individuals with spina bifida include hip contractures, pelvic obliquity, hip displacement, scoliosis, and kyphosis.
- Upper extremity conditions that may occur in individuals with spina bifida include rotator cuff tear, carpal tunnel syndrome, joint degeneration, and damage to the nerves in the elbow.
- Individuals with spina bifida tend to be shorter in stature compared to their peers.

Chapter 12

Management of musculoskeletal conditions

12.1

Introduction

> Determine that the thing can and shall be done,
> and then we shall find the way.
> **Abraham Lincoln**

The approach to treating musculoskeletal conditions is often unique to individuals with spina bifida. The goal is to optimize mobility, functional ability, and independence.[199] The approach to treating the same conditions in an individual without spina bifida may be very different because of differences in sensation in the individual and the likely success of the treatment.

Considerations relevant to the management of musculoskeletal conditions for those with spina bifida include the following:

- **Lower extremity conditions:** The goal in managing foot and ankle conditions is to achieve a foot that is in plantigrade position (parallel to the ground) whether or not the individual walks, but it is particularly important for those who do walk. Treatment may begin with bracing (wearing orthoses) or casting and include surgery if the bones and joints become fixed in an atypical position.[181]

- **Hip and spine conditions:**
    - Preserving function and range of motion (ROM) is the most important goal in managing hip conditions in spina bifida.
    - Hip displacement (subluxation or dislocation) requires different management in an individual with spina bifida. Due to decreased sensation, individuals with spina bifida may not typically experience pain with this condition. If their ability to walk is not impacted (and often it is not), surgery may not be recommended.[181] The hip muscles of an individual with spina bifida may not adequately support the hip even after surgical repair, which can lead to recurrence of hip displacement.
    - Maintaining pelvic alignment is important as individuals with spina bifida are at increased risk for pressure sores and skin breakdown.
    - The degree to which an individual with spina bifida can sit balanced in their wheelchair may affect the need for surgical treatment of scoliosis.[195] Hip contractures may need to be addressed before surgery to treat scoliosis.[195]
- **Upper extremity conditions:** Physical therapy and exercises focused on optimizing movement and preserving the ROM in the shoulder, elbow, and wrist joints should be a priority for those who use mobility aids such as manual wheelchairs or forearm crutches. Using padded gloves or hand grips and properly positioning the wrist and hand can help prevent carpal tunnel syndrome.[197] As well, properly positioning the elbow, stretching frequently, and avoiding excessive pressure on the elbow can help prevent and manage nerve damage.[198] For some individuals, transitioning from a manual wheelchair to a power-assist wheelchair may help protect joints.

In general, management of musculoskeletal conditions may include:

- **Physical therapy**
- **Assistive technology**
- **Medical management, casting, and orthopedic surgery**

Each of these topics is addressed separately in the following sections, but in practice, they overlap. For example, physical therapy often includes the use of orthoses and mobility aids, and orthopedic surgery often includes casting.

USEFUL WEB RESOURCES

12.2

Physical therapy

Physical therapists provide services that develop, maintain, and restore a person's maximum movement and functional ability.[200] Physical therapists have different titles in different countries: in many countries they are called physiotherapists.[*] In this book, the terms "physical therapist" and "physical therapy" are used.

Physical therapists work as part of the multidisciplinary team taking care of the individual with spina bifida. Therapy services often begin during infancy with treatment focused on maximizing joint ROM and strength, while also supporting the child's progress in reaching developmental milestones. As the child progresses in age and skill, therapy activities progress to support the child's unique needs and functional abilities.[†]

Though physical therapists may select treatments designed to improve functional activities,[‡] the treatments themselves are often not strictly

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[*] In Ireland, the term "physical therapist" and "physiotherapist" are not interchangeable. There, a physical therapist is someone trained specifically in the manual treatment of soft tissues, mostly massage.

[†] Occupational therapy is also relevant for individuals with spina bifida but less so for management of musculoskeletal conditions. Occupational therapy is addressed in Chapter 15.

[‡] Tasks such as mobility, feeding, and personal care, or other activities of daily living.

functional activities. For example, a child's goal may be to walk for short distances in their home. During the evaluation, the physical therapist may identify moderate strength in the hip flexor and knee extensor muscles, significant weakness in the ankle and foot, and impaired balance as the underlying problems preventing the child from meeting their goal. As a result, in the plan of care for that child, the physical therapist might include specific strength training for the hip flexors and knee extensors (whose nerve control is not as affected by the spinal lesion and have the ability to gain strength); assessment for orthoses to address the weakness in the ankle and foot muscles (whose nerve control is affected by the spinal lesion and do not have the ability to gain strength); balance training; and trying to use a mobility aid. These treatments address the underlying problems, but they, themselves, are not functional activities.

As the underlying problems are reduced, the treatment can shift to task-specific functional activities. In this example, the child would begin to practice standing and walking with their new orthoses and mobility aid. Physical therapists (in collaboration with other members of the multidisciplinary team) are a great source of guidance on orthoses and mobility aids in daily life.

Specific gait training used during therapy may include treadmill training, partial body weight support treadmill training, and various forms of assisted and even robotic training. The emphasis during these treatments is not only on strengthening and weight-bearing to build bone strength but also on practicing a high number of repetitions, or steps, providing the opportunity to retrain the muscles with decreasing support in a variety of environments.

The following are all elements of physical therapy and are covered below in more detail:

- **Strengthening**
- **Stretching**
- **Postural control and balance training**
- **Functional mobility training and transfers**

Strengthening

Individuals with spina bifida often experience decreased muscle strength, particularly in the lower extremities. Muscle strength is related to the level of the lesion in individuals with spina bifida: those with higher-level lesions (e.g., thoracic level) are more impacted than those with lower-level lesions (e.g., lumbo-sacral level). Muscle strength is further affected by abnormal muscle growth and bone development.

The physical therapist can determine appropriate exercises based on the individual's muscle function and developmental stage. Muscles can be strengthened in different positions with functional activities. For example, for an individual who has intact nerve control of their knee extensors, strengthening may include moving from a sitting to a standing position with or without assistance. Strengthening for an older child or adolescent often includes a combination of specific floor or mat-based exercises as well as functional activities. It may also include weight-bearing activities, especially in young children, to help with muscle growth.

In addition to strengthening exercises being done as part of a focused routine, strengthening must be built into everyday life. For the small child, a variety of positions can provide an opportunity to strengthen the muscles during play. Often, those same positions can be used to achieve both stretch and strength.

Stretching

Stretching is not a task-specific functional activity, but it is still important for individuals with spina bifida. Stretching is required throughout growth for the young child, the older child, and the adolescent with spina bifida. How this stretching is achieved may vary over the years, but the need for it remains constant. There are also critical periods when stretching is especially important, which coincide with the periods of most rapid growth: the first three years and during puberty.

The physical therapist can provide guidance for stretching, but it must be built into the activities of everyday life. Two to four hours of stretching per day is required for normal muscle growth, which a typically developing child gets during the day when they get up and start to move about, run, and play. (Note that while reference is usually to stretching muscles, what is actually being stretched is the muscle-tendon unit and the associated joint.)

The methods used for stretching depend on several factors, including muscle tightness, age, and developmental stage. Positioning and active movement are both types of stretching that may be used in individuals with spina bifida.

a) Positioning

A variety of positions can be used throughout the day to achieve sustained muscle stretching to promote muscle growth as well as to ensure pressure relief for areas of insensate skin (skin with diminished or absent sensation). The child or adolescent should be in a variety of positions throughout the day and not spend too much time in one position. They may have favorite positions, but varying positions is important.

Positioning may include using a stander to stand upright or sit in various positions, such as legs extended or elevated. It may also include prone positioning, which means the individual is lying on their stomach. Nighttime positioning can be done using orthoses or splints. However, if using orthoses, splinting, or special positioning at night interferes with sleep, a balance between nighttime stretching and good sleep needs to be found. The benefits and drawbacks of each need to be weighed.

b) Active movement

Active movement is exactly what it sounds like. Optimally, the child or adolescent should get plenty of active movement through the entire ROM of the joints. Individuals can remove orthoses if they get in the way of doing a task, and then put them back on during downtime for a prolonged muscle stretch. Often, a balance between active movement without orthoses and static stretch with orthoses is needed.

Postural control and balance training

Individuals with spina bifida may struggle with posture and sitting balance, specifically those with a lesion in the thoracic spine affecting nerves that control trunk strength. Physical therapy often involves equipment for positioning support and balance. In addition, the physical therapist may use supported sitting exercises to help improve trunk control, strength, and balance.

Functional mobility training and transfers

A significant amount of physical therapy focuses on practicing functional mobility. The physical therapist selects specific mobility activities or tasks based on the individual's age, function, and their or the family's goals. For very young children, physical therapists emphasize practicing developmental activities such as rolling or crawling if the motor abilities allow. For older children, adolescents, and adults, functional mobility may include how a person moves from one position to another, known as a transfer; for example, moving from bed to wheelchair.

For individuals with spina bifida, functional mobility training includes using mobility aids such as forearm crutches, reverse walkers, standers, gait trainers, adaptive strollers, and wheelchairs (see section 12.3). Other equipment such as parallel bars may be used as standing support during functional therapeutic activities (see Figure 12.2.1). While standing and gait training support bone health and mobility, they may not lead to independent walking. However, they are important for enabling stand-pivot transfers, a type of transfer that involves a person standing up, pivoting on their feet, and sitting down on a new surface, such as moving from a wheelchair to a bed.

Figure 12.2.1 Parallel bars for mobility training.

Individuals who are able to walk independently can likely transfer independently; in fact, a review of a large sample of adults with spina bifida found that 78 percent can do so.[201] Those who use wheelchairs may need transfer assistance. Both the level of lesion and presence of hydrocephalus are associated with the ability to transfer independently:[202]

- Individuals with a lesion level below L2 are likely to be independent in transfers.
- Approximately 38 percent of individuals with a lesion above L2 and hydrocephalus require help with transfers.

Physical therapists assess an individual's ability to transfer and provide recommendations and training for the most appropriate method. For those who can transfer independently, the training may include instructions for how to move between surfaces safely and independently. For individuals who require transfer assistance, the recommendations may require training caregivers to assist with a stand-pivot transfer or in the use of a sliding board.* Depending on the individual's function and environmental demands, physical therapists may teach the individual to

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* A piece of equipment that facilitates movement between surfaces, such as from a bed to a chair or from a wheelchair to a toilet.

use compensatory movements* or recommend using orthoses or mobility aids to maximize their ability to assist in their movement.

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* Movements that the body uses when the typical movements are not possible. For example, when a person is transferring from sitting on the floor to standing using a surface such as a chair, typically they lift one foot forward first to push up into standing. A person with leg muscle weakness may push up from the floor with both feet together as a compensatory movement to allow them to complete this transfer.

12.3

Assistive technology

"Assistive technology"* refers to products and services designed to enhance the functional capabilities and independence of individuals with disabilities to allow them to participate.[203] Assistive technology commonly used by individuals with spina bifida include:

- **Orthoses**
- **Mobility aids**
- **Adaptive recreational equipment**

Assistive technology should be selected based on assessments performed by a multidisciplinary team, including medical professionals (e.g., physician, physical and occupational therapists, orthotist) in conjunction with the individual and their family. An individual's unique health needs and the family's care and function goals help guide what assistive technology is best suited for the individual. Regular reevaluation is important, the frequency of which depends on the individual and the product.

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* In the US, some assistive technology is referred to as "durable medical equipment" (DME) for medical insurance purposes.

Orthoses

Orthoses are designed to hold specific body parts in position in order to modify their structure or function. (The singular term "orthosis" comes from the Greek word "ortho," which means "to straighten or align.") Orthotics is the branch of medicine concerned with the design, manufacture, and management of orthoses. An orthotist is a professional in this specialty. The word "orthotic" is sometimes used to mean the device; "orthosis" is the more correct term, but given how alike the two terms are, their interchangeability is understandable. The terms "brace" or "splint" are also sometimes used (the former particularly for spinal braces). Orthoses are commonly used to address musculoskeletal conditions in individuals with spina bifida.

Different orthoses have different functions. The goals of treatment with orthoses may include some of the following:[204]

- Maintain or improve ROM at a joint through a prolonged stretch
- Provide stability or support to a joint
- Improve function of a limb
- Improve balance
- Improve gait
- Minimize the effects of hypertonia (including spasticity)
- Provide protection for a limb with decreased sensation
- Accommodate or minimize a joint alignment problem
- Prepare for surgery
- Facilitate positioning after surgery

Choosing an orthosis involves collaboration of many members of the team—often including the orthotist, therapist, physician, and family. Collaborative goal-setting is important[205] as there is frequently a trade-off with orthoses. For example, using a spinal brace to help stabilize an individual during sitting activities may weaken their independent trunk strength, causing issues when they are not wearing the device. There may be no perfect solution for these competing goals in the treatment plan: one goal may be to offer support for the trunk during some activities, but another may be to see the individual strengthen their trunk muscles. A balance must be identified with the understanding that a perfect orthosis for all goals rarely exists.

Orthoses can be custom-made (molded to a specific individual's body) or prefabricated (fit based on size and already made). Different orthoses may be prescribed over the years as the child grows and as their goals, body structure, and function change. A custom device such as an ankle-foot orthosis (AFO) requires that a mold of the leg and foot first be taken, then the individual returns in a couple of weeks to be fitted with the new orthosis. Adjustments are made to confirm that it fits comfortably and functions well. Adherence to the prescribed wear time is critical to ensure that the individual receives the full benefit of the device. After the initial fitting, further adjustments may be needed if the individual is experiencing any of the following:

- Discomfort
- Redness of the skin
- Skin breakdown
- A growth spurt
- A change in functionality
- A change in ROM

Additionally, children are likely to outgrow their devices before they wear them out and may require new devices every year or so until they stop growing.

Most commonly, individuals with spina bifida require lower extremity orthoses and spinal braces, addressed below. Occasionally, upper extremities orthoses may be required.

a) Lower extremity orthoses

Lower extremity orthoses are used for the feet, ankles, knees, and hips. They are intended to maintain ROM at the joint and provide support and positioning during standing, walking, and transfers while weight-bearing. The following types of lower extremity orthoses may be used by individuals with spina bifida and are described in Table 12.3.1:

- Foot orthosis (FO)
- Supramalleolar orthosis (SMO)
- Ankle-foot orthosis (AFO)
- Knee-ankle-foot orthosis (KAFO)
- Derotation straps or cables

- Hip-knee-ankle-foot orthosis (HKAFO)
- Reciprocating gait orthosis (RGO)
- Knee immobilizer (KI)

Shoes that fit well must be used with foot orthoses, especially those intended for standing or walking. If the shoes do not fit well, the orthoses may not function correctly and may not be comfortable. In most cases, a slightly larger shoe is needed. Certain stores, including some available online, allow purchase of two different-sized shoes. Athletic shoes that can be zipped, laced, or fastened snugly can be a good option. Some manufacturers of athletic shoes now offer models that are specifically designed to be easy to get on and off with orthoses, such as BILLY shoes or the Nike FlyEase.

To get a child to consistently wear an orthosis, it must be comfortable; it must not cause any rubbing or blisters, for example. Parents should not hesitate to speak up if they notice any problems, even minor ones. Orthoses must also be acceptable in appearance to the wearer. It helps if the child can choose from a variety of colors and designs. An orthosis would not (and should not) be prescribed unless it's necessary, so although the child may not love wearing it, they should wear it as prescribed.

Table 12.3.1 Lower extremity orthoses

ORTHOSIS TYPE	SUBTYPE/ IMAGE	DESCRIPTION
Foot orthosis (FO)	Prefabricated foot orthosis 	A prefabricated foot orthosis is inserted into the shoe to provide support, distribute pressure, and help maintain proper positioning of the midfoot, especially in the arch.
	UCBL 	A custom UCBL foot orthosis (named for the University of California Berkeley where it was developed) is inserted into the shoe to provide support, distribute pressure, and help maintain proper positioning of the foot, especially the heel and the arch.
Supramalleolar orthosis (SMO)		"Supra" means above and "malleolar" refers to the malleoli (ankle bones). The SMO is a custom-molded flexible or rigid plastic shell that extends just above the ankle. An SMO provides more support than the UCBL and helps maintain proper positioning of the ankle and foot.

Cont'd.

ORTHOSIS TYPE	SUBTYPE/ IMAGE	DESCRIPTION
Ankle-foot orthosis (AFO) Extends above the ankle joint and stops before the knee. It protects the foot, manages foot malalignments, prevents the toes from dragging, provides varying support and stability to the ankle and knee, and can help prevent the progression of ankle contractures.	Articulated AFO 	An articulated AFO has a hinge at the ankle joint to allow ankle motion. It often has a plastic posterior "stop" that blocks plantarflexion, preventing the wearer from moving the foot down or dragging their toes. Dorsiflexion at the ankle joint has many benefits but may also allow crouch gait* in individuals with hamstring tightness or weakness of the ankle plantarflexor. Articulated AFOs are also worn by adolescents and adults who would benefit from support and the added ROM while still preventing their toes from dragging.
	Solid AFO (SAFO) 	A SAFO is the most supportive type of AFO and is commonly used for individuals with spina bifida. Typically made of a rigid, durable plastic, it provides greater stability for the ankle and knee. This AFO does not allow ankle movement, which can make some functional movements difficult to perform while wearing it (e.g., going up and down stairs).
	Ground reaction AFO (GRAFO or floor reaction AFO) 	A GRAFO is used for an individual with crouch gait due to quadriceps or ankle plantarflexor weakness who needs more support than is offered by a SAFO. The anterior shell gives the orthosis more leverage, distributes pressure more evenly, and encourages knee extension more fully during gait. The goal is to improve knee stability and reduce crouch gait. It works best for individuals with good hip and knee strength who do not have significant knee flexion contractures. SAFO and GRAFO can also be used for the same reasons.
	Nighttime AFO or stretching splint 	A prefabricated nighttime AFO typically has a plastic shell, soft inner liner, and adjustable straps. It is worn just at night and designed to help stretch the calf muscles, maintain ankle ROM, and prevent the progression of ankle joint contractures.

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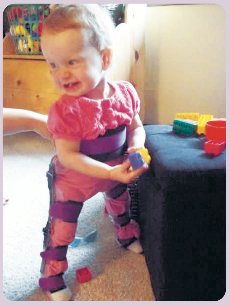
* A walking pattern characterized by excessive bending at the hips, knees, and ankles; the individual appears to be crouched while walking.

Cont'd.

ORTHOSIS TYPE	SUBTYPE/ IMAGE	DESCRIPTION
Knee-ankle-foot orthosis (KAFO)		A KAFO typically consists of a custom-molded plastic shell extending above the knee but stopping before the hip. It typically has contoured metal uprights with a choice of knee joints. It provides support, control, and correction of the foot, ankle, and knee joints. The metal uprights of a KAFO may also be removable so the AFO can be worn separately.
Derotation straps or cables (also known as twister straps or cable)		Most derotation straps are made of a waist belt and leg straps that wrap around the legs and attach to an individual's shoes or AFOs. The fabric is wrapped in the direction needed to correct excessive inward or outward rotation of the feet or legs.
Hip-knee-ankle-foot orthosis (HKAFO)		The HKAFO adds a custom-molded, contoured pelvic section and metal hip joints to a KAFO to support and control the foot, ankle, knee, and hip joints. Due to the weight of the device, it is often used only in therapeutic walking and commonly with a mobility aid. 
Reciprocating gait orthosis (RGO)		Similar in design to an HKAFO but with a different pelvic section and hip joints interconnected by cables. Hip extension on one side propels hip flexion on the opposite side, resulting in alternating flexion and extension at the hips and an assisted walking motion partially driven by the orthosis. Due to the weight of the device, it is often used only in therapeutic walking and commonly with a mobility aid.

Cont'd.

ORTHOSIS TYPE	SUBTYPE/ IMAGE	DESCRIPTION
Knee immobilizer (KI)		A KI consists of a soft knee wrap, rigid aluminum struts, and straps to adjust the fit. As the name suggests, the KI prevents the knee from moving. It is used to stretch the knee flexors (hamstrings) or to maintain ROM after surgery. Wearing one KI each night and alternating between right and left may help if wearing two at once is too much. Alternatively, these can be worn for stretching during the day.

Adapted from Ward 2021.

b) Spinal braces

A spinal brace is designed to provide support and improve alignment of the spine by applying specific pressures to the torso to slow scoliosis curve progression and prevent or delay surgery. It is often used in the management of mild scoliosis or kyphosis curve progression. The overall goal of scoliosis and kyphosis management is to maintain a balanced spine and posture.

There are several types of spinal braces, each named for the region of the spine they impact. The most common type is the thoraco-lumbo-sacral orthosis (TLSO), which is typically made from molded, rigid plastic that extends from the armpits to the pelvis, not limiting hip motion. TLSOs are custom-made for the individual to provide the best comfort and scoliosis curve control. A TLSO may be fabricated at the medical care center or at a company that specializes in making them. Once the TLSO is made, it is fit to the child by the orthotist.

TLSOs can vary in design, type of padding, and material choice (type of plastic); however, regardless of the details, the TLSO must be fitted well and designed appropriately for the individual's specific curve characteristics.

The two types of TLSO are full-time and nighttime hypercorrective TLSOs. A full-time TLSO is prescribed to be worn during the day and night, typically 18 to 23 hours per day. It should be removed for physical activity such as sports, physical therapy, workouts, and for hygiene.

For individuals who are unable to stand to put the brace on, a TLSO may be designed to be in two pieces, typically a front and a back piece, to make that easier.

A nighttime hypercorrective TLSO is prescribed to be worn only while the individual is lying down, typically when sleeping. The nighttime hypercorrective TLSO takes advantage of the elimination of gravity. When lying down, the spine is more correctible, and greater corrective forces can comfortably be applied to the scoliosis curve—hence the term “hypercorrective.”

The two types, both of which may be used by individuals with spina bifida, are described in Table 12.3.2. Both are used to manage scoliosis, but only the full-time TLSO is used to manage kyphosis curve progression.

Table 12.3.2 Spinal braces

TYPE	CURVE LOCATION	WEAR TIME	IMAGE
Full-time thoraco-lumbo-sacral orthosis (TLSO)	Primary curve in the thoracic and lumbar spine	18 to 23 hours a day (worn day and night)	
Nighttime hypercorrective thoraco-lumbo-sacral orthosis (TLSO)	Primary curve in the thoracic and lumbar spine	8 to 12 hours a day (worn at night only, while sleeping)	

Mobility aids

Mobility aids (also termed "assistive mobility devices," "assistive devices," "walking aids," and "gait aids") are types of equipment that enhance mobility, independence, and safety, and they vary in the level of support they provide. The following mobility aids are addressed below:

- **Forearm crutches**
- **Reverse walkers**
- **Gait trainers**
- **Standers**
- **Adaptive strollers**
- **Wheelchairs**

Information on transfer aids and vehicle transportation is included in Appendix 5 (online).

a) Forearm crutches

Forearm crutches (also known as Lofstrand or elbow crutches), have a cuff that wraps around the forearm and a handgrip for the individual to grab onto. They can be adjusted in height as the individual grows. They help distribute weight from the lower body to the upper body so require good upper body strength. (See Figure 12.3.1.)

Figure 12.3.1 Forearm crutches.

b) Reverse walkers

Reverse walkers provide support behind the individual, placing their body in the front, open part of the frame. These are also termed posterior-wheeled walkers. (See Figure 12.3.2.)

Figure 12.3.2 Reverse walker.

c) Gait trainers

Gait trainers are similar to walkers but offer increased postural support, weight-bearing capabilities, and mobility assistance while walking. Gait trainers typically consist of a wheeled frame with weight-bearing support straps and upper extremity supports such as a tray, handle, or handrail. They are appropriate for individuals who are incapable of walking unsupported.[204] Gait trainers can be customized to suit an individual's functional ability and needs. (See Figure 12.3.3.)

A gait trainer can be used to develop or improve an individual's walking ability but is often used primarily for exercise. Walking with a gait trainer provides the benefits of improved strength, ROM, episodes of lower extremity weight-bearing, self-esteem, participation, and bowel and bladder function.[204]

- Snow skiing (e.g., outriggers)
- Sled hockey (e.g., sitting sled for ice hockey)
- Snow boarding
- Swimming (e.g., back float belts, neck floats and specialized aquatic wheelchairs)
- Frame running (also known as race running), where individuals compete with running frames on an athletics track
- Waterskiing (e.g., kneel or sit skis)
- Golf (e.g., adaptive golf carts)
- Kayaking (e.g., outriggers, trolling motors)
- Surfing (e.g., specialized systems)
- Horseback riding (e.g., high back saddles for balance)

Figure 12.3.12 Adaptive equipment for recreational activities.

"This book does a phenomenal job of describing all the different ways the condition can affect a person. It also caters to different audiences—people with spina bifida, parents, and those who take care of people with the condition."

—GRACIE HADLICH, AN ADULT LIVING WITH SPINA BIFIDA

"This handsome resource belongs in the hands of every individual with spina bifida and on the shelf of every professional serving this complex population."

—JONATHAN CASTILLO, MD, MPH, FAAP

"*Spina Bifida* is a highly accessible resource that weaves together medical expertise with practical information for people living with the condition."

—NIENKE DOSA, MD, MPH

Spina bifida is a neural tube defect that results in incomplete closure of the vertebrae, muscles, and skin that normally surround and protect the spinal cord and nerves. Initial management focuses on restoring protection for the spinal cord and nerves and monitoring the brain closely.

This practical guide addresses the most common type of spina bifida, myelomeningocele, its associated conditions across the lifespan, and evidence-based best practice. It also includes the lived experience of families. The writing of *Spina Bifida* was led by Kelly Cho, MD, Pediatric Rehabilitation Medicine Physician at Gillette Children's, a world-renowned center of excellence for the treatment of brain, bone, and movement conditions. This book is part of the Gillette Children's Healthcare Series, a series of books for families who are looking for clear, comprehensive information. Health care professionals, researchers, educators, students, and extended family members will also benefit from reading it. Other titles in the series include:

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