Spina bifida

Spina bifida is a condition in which the neural tube, a layer of cells that ultimately develops into the brain and spinal cord, fails to close completely during the first few weeks of embryonic development. As a result, when the spine forms, the bones of the spinal column do not close completely around the developing nerves of the spinal cord. Part of the spinal cord may stick out through an opening in the spine, leading to permanent nerve damage. Because spina bifida is caused by abnormalities of the neural tube, it is classified as a neural tube defect.

Children born with spina bifida often have a fluid-filled sac on their back that is covered by skin, called a meningocele. If the sac contains part of the spinal cord and its protective covering, it is known as a myelomeningocele. The signs and symptoms of these abnormalities range from mild to severe, depending on where the opening in the spinal column is located and how much of the spinal cord is contained in the sac. Related problems can include a loss of feeling below the level of the opening, weakness or paralysis of the feet or legs, and problems with bladder and bowel control. Some affected individuals have additional complications, including a buildup of excess fluid around the brain (hydrocephalus) and learning problems. With surgery and other forms of treatment, many people with spina bifida live into adulthood.

In a milder form of the condition, called spina bifida occulta, the bones of the spinal column are abnormally formed, but the nerves of the spinal cord usually develop normally. Unlike in the more severe form of spina bifida, the spinal cord does not stick out through an opening in the spine. Spina bifida occulta most often causes no health problems, although rarely it can cause back pain or changes in bladder function.

Frequency

Spina bifida is one of the most common types of neural tube defect, affecting an estimated 1 in 2,500 newborns worldwide. For unknown reasons, the prevalence of spina bifida varies among different geographic regions and ethnic groups. In the United States, this condition occurs more frequently in Hispanics and non-Hispanic whites than in African Americans.

Causes

Spina bifida is a complex condition that is likely caused by the interaction of multiple genetic and environmental factors. Some of these factors have been identified, but many remain unknown.

Changes in dozens of genes in individuals with spina bifida and in those of their mothers may influence the risk of developing this type of neural tube defect. The best-studied of these genes is MTHFR, which provides instructions for making a protein
that is involved in processing the vitamin folate (also called vitamin B9). A shortage (deficiency) of this vitamin is an established risk factor for neural tube defects like spina bifida. Changes in other genes related to folate processing and genes involved in the development of the neural tube have also been studied as potential risk factors for spina bifida. However, none of these genes appears to play a major role in causing the condition.

Researchers have also examined environmental factors that could contribute to the risk of spina bifida. As mentioned above, folate deficiency appears to play a significant role. Studies have shown that women who take supplements containing folic acid (the synthetic form of folate) before they get pregnant and very early in their pregnancy are significantly less likely to have a baby with spina bifida or a related neural tube defect. Other possible maternal risk factors for spina bifida include diabetes mellitus, obesity, exposure to high heat (such as a fever or use of a hot tub or sauna) in early pregnancy, and the use of certain anti-seizure medications during pregnancy. However, it is unclear how these factors may influence the risk of spina bifida.

Inheritance Pattern

Most cases of spina bifida are sporadic, which means they occur in people with no history of the disorder in their family. A small percentage of cases have been reported to run in families; however, the condition does not have a clear pattern of inheritance. First-degree relatives (such as siblings and children) of people with spina bifida have an increased risk of the condition compared with people in the general population.

Other Names for This Condition

- cleft spine
- open spine
- rachischisis
- spinal dysraphism

Diagnosis & Management

Formal Treatment/Management Guidelines


Genetic Testing Information

- What is genetic testing? /primer/testing/genetictesting
• Genetic Testing Registry: Neural tube defects, folate-sensitive

• Hickey SE, Curry CJ, Toriello HV. ACMG Practice Guideline: lack of evidence for
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23288205

Research Studies from ClinicalTrials.gov
• ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22spina+bifida%22

Other Diagnosis and Management Resources
• Benioff Children's Hospital, University of California, San Francisco Fetal Treatment
  Center
  https://fetus.ucsf.edu/spina-bifida

• Benioff Children's Hospital, University of California, San Francisco: Treatment of
  Spina Bifida
  https://www.ucsfbenioffchildrens.org/conditions/spina_bifida/treatment.html

• Centers for Disease Control and Prevention: Living with Spina Bifida
  https://www.cdc.gov/ncbddd/spinabifida/living.html

• Spina Bifida Association: Urologic Care and Management
  https://www.spinabifidaassociation.org/resource/urologic-care/

Additional Information & Resources
  Health Information from MedlinePlus
• Encyclopedia: Folic Acid in Diet
  https://medlineplus.gov/ency/article/002408.htm

• Encyclopedia: Spina Bifida (Image)
  https://medlineplus.gov/ency/imagepages/19086.htm

• Health Topic: Neural Tube Defects
  https://medlineplus.gov/neuraltubedefects.html

• Health Topic: Spina Bifida
  https://medlineplus.gov/spinabifida.html

Genetic and Rare Diseases Information Center
• Spina bifida
  https://rarediseases.info.nih.gov/diseases/7673/spina-bifida

• Spina bifida occulta
  https://rarediseases.info.nih.gov/diseases/10787/spina-bifida-occulta
Additional NIH Resources
- National Institute of Neurological Disorders and Stroke
  https://www.ninds.nih.gov/Disorders/All-Disorders/Spina-bifida-Information-Page
- Office of Dietary Supplements: Folate
  https://ods.od.nih.gov/factsheets/Folate-Consumer/

Educational Resources
- Boston Children’s Hospital
  http://www.childrenshospital.org/conditions-and-treatments/conditions/s/spina-bifida
- Centers for Disease Control and Prevention: Folic Acid
  https://www.cdc.gov/ncbddd/folicacid/index.html
- Centers for Disease Control and Prevention: Spina Bifida Facts
  https://www.cdc.gov/NCBDDD/spinabifida/facts.html
- Centre for Genetics Education, New South Wales, Australia
- JAMA Patient Page
  https://jamanetwork.com/journals/jama/fullarticle/193939
- KidsHealth from the Nemours Foundation
- MalaCards: spina bifida aperta
  https://www.malacards.org/card/spina_bifida_aperta
- MalaCards: spina bifida occulta
  https://www.malacards.org/card/spina_bifida_occulta
- March of Dimes
  https://www.marchofdimes.org/complications/spina-bifida.aspx
- Merck Manual Consumer Version
- Orphanet: Isolated spina bifida
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=823

Patient Support and Advocacy Resources
- Center for Parent Information and Resources
  https://www.parentcenterhub.org/spinabifida/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/spina-bifida/
• Shine: Spina bifida - Hydrocephalus - Information - Networking - Equality (UK)  
  https://www.shinecharity.org.uk/
• Spina Bifida & Hydrocephalus Canada  
  http://sbhac.ca/
• Spina Bifida Association  
  https://www.spinabifidaassociation.org/

**Scientific Articles on PubMed**

• PubMed  
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Spinal+Dysraphism%5BMAJR%5D%29+AND+%28spina+bifida%5BTI%5D%29+AND+review%5Bpt%5D+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

**Catalog of Genes and Diseases from OMIM**

• NEURAL TUBE DEFECTS, FOLATE-SENSITIVE  
  http://omim.org/entry/601634
• NEURAL TUBE DEFECTS, SUSCEPTIBILITY TO  
  http://omim.org/entry/182940

**Medical Genetics Database from MedGen**

• Spina bifida  

**Sources for This Summary**

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10559453
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19918803  
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4239538/
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Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2790326/

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Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5388593/

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