

Spina bifida

What is spina bifida?^[1] ^[2]

Spina bifida is one of the possible neural tube defects that can occur during early embryological development. See the separate overview article on [Neural Tube Defects](#).

In spina bifida, the vertebral arch of the spinal column is either incompletely formed or absent. The defect can occur anywhere from the base of the skull to the sacrum. It is most commonly found in the lumbar region. Neurological symptoms and signs generally correspond to the level of the defect. Spina bifida can be classified based on the type of spinal defect:

- **Spina bifida occulta:** the overlying skin is intact, there is a bony vertebral arch defect but no visible external overlying sac. There is no protrusion of the spinal cord or its membranes. This may affect up to 10% of the population and is most common at the lumbosacral junction. This is a closed form of spina bifida.

- **Spina bifida cystica:** there is both a vertebral defect and a visible cystic mass on the back. This is an 'open' form of spina bifida. It can be subdivided into:
 - Meningocele - there is a cystic swelling of the dura and arachnoid mater which protrudes through the vertebral arch defect. No spinal neural tissue is present within the sac. There may be no neurological symptoms/signs.
 - Myelomeningocele - spinal neural tissue forms part of the sac. Excluding spina bifida occulta, this is the most common form of spina bifida.
 - Rachischisis - this is the most severe form of spina bifida cystica. The spine lies widely open and the neural plate has spread out on to the surface. It is often associated with anencephaly.

Arnold-Chiari type II malformation is often associated with myelomeningocele. Here there is cerebellar hypoplasia and displacement of the hindbrain through a widened foramen magnum. Cerebrospinal fluid (CSF) flow can be disrupted and hydrocephalus can result.

How common is spina bifida? (Epidemiology)

- The prevalence varies across time, by region and by both race and ethnicity. It also tends to be more common in girls. ^[3]
- Myelomeningocele affects about 1 in every 2,000 births worldwide. ^[4]
- The rate has declined markedly since folic acid supplementation was introduced and fetal ultrasound screening has increased. ^[1]
- Siblings of patients with spina bifida have an increased incidence of neural tube defects. ^[5]

Causes of spina bifida (aetiology) ^[2]

The cause of spina bifida is thought to be multifactorial:

- There appears to be a combination of **genetic susceptibility** with **environmental precipitants**, particularly shortage of folic acid in the mother's diet at a crucial stage in embryogenesis (days 17–30 when many mothers are unaware that they are pregnant). This is when the neural tube is forming and closing.
 - Supplementation of newly pregnant mothers' diets and periconceptual advice to increase **folic acid intake** have been shown to reduce the incidence of neural tube defects significantly.^[6]
 - **Chromosomal abnormalities** including trisomy 13 ([Patau's syndrome](#)), 18 ([Edwards' syndrome](#)) and 21 ([Down's syndrome](#)) have been associated with neural tube defects.
 - Risks are increased with maternal [diabetes](#) and obesity.^{[7] [8] [9]}
 - **Maternal use of anti-epileptic medications** – sodium valproate and carbamazepine. Risk is greater with valproate (11–14 times greater than controls) and increased further if more than one type of medication is used.^[10]
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Presentation of spina bifida^[1]

Spina bifida cystica

- The abnormal herniation of the dural sac/neural tissue is usually evident, either during antenatal ultrasound scanning or at birth.
- Classically, the disruption of spinal cord function causes sensory dysfunction, flaccid paralysis and areflexia below the affected level. An alternative pattern includes the preservation of some distal reflex activity which is usually exaggerated.
- In cases of meningocele alone, the herniation of the meninges is often covered by skin so the lesion may be more difficult to detect.
- Imbalanced muscle forces can lead to spinal deformity, limb contractures and joint dislocations.
- Arnold-Chiari II malformation may present with stridor or apnoea. Impaired cerebellar function can affect balance, co-ordination and walking. Hydrocephalus, seizures and impaired cognitive function may be present.^[3]

Spina bifida occulta

- This is common and more difficult to detect.
- There are usually no neurological sequelae or long-term consequence.
- There may be no cutaneous marker, or there may be an obvious abnormality along the spine including:
 - A fluid-filled cystic mass.
 - An area of hyperpigmentation or hypopigmentation.
 - Cutis aplasia.
 - Congenital dermal sinus (this may lead to meningitis or spinal abscess).
 - Capillary telangiectasias/haemangioma.
 - Hypertrichosis (a hairy patch of skin).
 - Skin appendages.
 - An asymmetrical gluteal cleft.
- The risk of significant spinal malformations in asymptomatic, healthy infants with an isolated simple sacral dimple is very low.^[11]
- Asymmetry of the legs/feet may be present.
- Scoliosis or other spinal deformities may develop.
- Progressive neurological motor and/or sensory deficits can develop with associated bladder or bowel disturbance (because of associated tethering of the spinal cord).
- There may be low back pain as the individual gets older.
- A sudden onset of pain, motor and sensory loss and bladder dysfunction can occur after acute trauma if there is spinal cord tethering.

Differential diagnosis

- The classical appearance of spina bifida cystica is not likely to be confused with other pathologies.

Examination

- Examine the spine and note the site and size of any lesion. Look for any spinal deformity.
- Perform a complete neurological examination of the newborn. Document any neurological abnormalities. This will act as a baseline:
 - Measure head circumference.
 - Assess cry and sucking reflex.
 - Assess anal sphincter.
 - Examine urinary stream.
 - Perform a full motor examination, including assessment of muscle bulk, spontaneous active movements, muscle tone and movements in response to stimulation.
 - Perform a full sensory examination.
- Look for foot and hip deformities.

Investigations

Prenatal diagnosis

- Raised levels of maternal serum alpha-fetoprotein (AFP) at 16–18 weeks of gestation are found in neural tube defects.
- The 18- to 20-week fetal anomaly screening ultrasound scan allows detection and diagnosis of neural tube defects and is much more specific.^[12]
- When amniocentesis is done, amniotic fluid AFP and acetylcholinesterase concentrations can be used to differentiate between open ventral wall defects (gastroschisis and omphalocele) and open neural tube defects.

Investigation of confirmed spina bifida

- Screening bloods can be carried out to detect any evidence of impairment of other organ systems, particularly renal impairment.
- Urine culture and urodynamics may be needed to detect any abnormality of the urinary tract caused by impaired bladder innervation.
- Latex allergy is relatively common among sufferers of spina bifida, probably due to inherent susceptibility and repeated exposure to surgical procedures.^[13]
- Enzyme-linked immunosorbent assay (ELISA) or skin-prick sensitivity testing may be needed to avoid illness caused by latex exposure. 40% of children with myelomeningocele may be latex-sensitive.
- Plain X-rays of the spine can help to detect any associated scoliosis and hip dysplasia or dislocation.
- CT and/or MRI scanning of the head and spinal cord may be conducted to look for evidence of the major complications of spina bifida, such as:
 - Hydrocephalus due to Arnold-Chiari II malformation.
 - Tethering of the spinal cord by fibrous bands.
- Gait analysis may be needed to evaluate a patient's functional mobility and allow intervention to improve independent mobility through the use of orthoses or surgery.

Management of spina bifida

General measures

- Nurse any newborn with an open neural tube defect in the prone position and cover the defect with a sterile wet saline dressing.
- A multidisciplinary team approach is needed in the management of an infant with spina bifida.

- Treatment aims are to maximise mobility, prevent or ameliorate complications of spina bifida (particularly hydrocephalus), encourage as normal as possible development, and to help the individual maintain as independent a life as possible.

Repair of the defect

- Fetal surgery for myelomeningocele before 26 weeks of gestation may preserve neurological function, reverse the hindbrain herniation of the Chiari II malformation, and prevent the need for postnatal placement of a ventriculoperitoneal shunt.^[14]
- Postnatal surgical treatment to correct the spinal cord malformation must be achieved during the first days of life.^[15]

Other interventions

- Ongoing management of mobility, utilising orthopaedic assessment, bracing and orthopaedic surgery, is often necessary. Spinal fusion, hip, pelvic or foot/ankle procedures are often needed.^[16]
- Prolonged physiotherapy, access to gym resources and/or adaptive training in children can be very helpful in maintaining independence and mobility.
- Developmental assessment by a paediatrician and help with maintaining a normal weight (weight gain is common due to impaired ambulation and can increase morbidity) are useful.
- Occupational therapy assessment and intervention can help to maximise function.
- Psychological input for the individual and their family to deal with the ramifications of their condition as they grow older is often needed.
- Neurosurgical follow-up is necessary to detect and treat complications such as hydrocephalus or a possible tethered cord.
- Bladder and bowel functions can be maintained or aided by the use of a regular bowel voiding regimen and intermittent self-catheterisation.^[17]

Complications of spina bifida

- [Meningitis](#) (especially in open neural tube defects).
- Fractures (particularly of lower-limb long bones) and [hip dislocations](#). These may be asymptomatic. There may be disuse osteoporosis and osteopenia.
- [Pressure sores](#) because of problems with mobility; 26% of people with spina bifida have them, 19% had one within the last 12 months.^[18]
- Skin ulceration around orthoses/braces.
- [Hydrocephalus](#) due to Arnold-Chiari II malformation causing developmental impairment.
- [Neurogenic bladder](#) causing incontinence and urinary tract infection.
- Constipation due to impaired bowel innervation and anal sphincter function.
- Latex allergy leading to [anaphylaxis](#).^[13]

Prognosis

With the advent of prenatal surgery, early repair of postnatal myelomeningocele, shunting to prevent hydrocephalus and expectant management of complications, most patients born with spina bifida survive into adulthood and develop relatively normally intellectually.

- Long-term survival depends on adherence to appropriate bowel and bladder regimens and careful management of urinary complications to prevent chronic kidney disease.
- Long-term outlook is very variable and depends on the degree of neurological deficit.^[19]
- Cognitive function (irrespective of intellectual disability) is important in everyday functioning.^[20]
- Patients with hydrocephalus and a lesion at the level of L2 or above seem to be more dependent with regards to sphincter control, locomotion, self-care, social cognition, and communication.^[21]

- Half of deaths (after the age of 5 years) are sudden and unexpected. Most occur in the community and the most frequent causes are epilepsy, pulmonary embolus, acute hydrocephalus and acute renal sepsis. [22]
- Women born with spina bifida benefit from lumbar ultrasonography prior to obstetric neuraxial anaesthesia. [23]

Prevention of spina bifida

- Periconceptual supplementation of folic acid and improved folate content in the diet of the general population (possibly through food fortification). [6]
- Folic acid may also help to reduce the severity of neural tube defects as well as preventing their occurrence. [24]
- Improved prenatal diagnosis and prenatal surgery or termination of some pregnancies may reduce the future burden of disability caused by this condition.

Further reading

- [Shepard CL, Yan PL, Hollingsworth JM, et al](#); Pregnancy among mothers with spina bifida. *J Pediatr Urol.* 2018 Feb;14(1):11.e1-11.e6. doi: 10.1016/j.jpuro.2017.08.001. Epub 2017 Sep 6.
- [Mazzone L, Moehrlen U, Casanova B, et al](#); Open Spina Bifida: Why Not Fetal Surgery? *Fetal Diagn Ther.* 2019;45(6):430-434. doi: 10.1159/000491751. Epub 2018 Sep 11.
- [Louden AR, Suhl J, Kancherla V, et al](#); Association between maternal periconceptual alcohol consumption and neural tube defects: Findings from the National Birth Defects Prevention Study, 1997-2011. *Birth Defects Res.* 2020 Mar;112(5):427-439. doi: 10.1002/bdr2.1656. Epub 2020 Feb 27.

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Authored by:	Peer Reviewed by: Dr Colin Tidy, MRCGP	
Originally Published: 20/11/2023	Next review date: 05/06/2023	Document ID: doc_2795

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