Structural Differences of the Spinal Cord

Structural problems of the spinal cord are rare among individuals with 22q differences but can have severe implications. This sheet provides general information on the more common spinal cord malformations in children with 22q11.2 deletion syndrome (22q11.2DS).

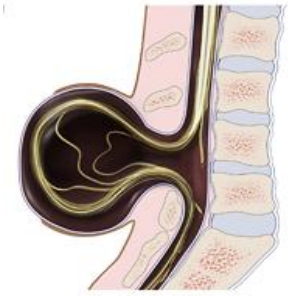
Spina Bifida (including Meningomyelocele)

- When a fetus is developing in the womb, the neural tube is the part that will form the brain and spinal cord [see video]. The two sides of the neural plate fuse together to form the neural tube within the first 28 days of pregnancy.
- **Spina bifida** is a birth defect that occurs when the **neural tube fails to close completely** in the fetus, and the spinal cord and its covering (meninges) form a sac. As a result:
  - The backbone is misshapen (needs to form around bulge)
  - The **sac protrude out of the backbone** (in some cases)
- Spina bifida is a **neural tube defect** because it results from the malformation of the neural tube.
- The exact cause of spina bifida is not known, but may involve genetic and nutritional factors.
- To check if a fetus has spina bifida, a blood test or ultrasound imaging can be done during pregnancy
- There are different types of spina bifida, depending on type and severity of problems. Some types (e.g. **spina bifida occulta**) can be very mild and may not be easily noticed.
- For some types of spina bifida, fetal surgery (in the womb) can repair the spinal cord before birth.
- Spina bifida occulta have been found in individuals with 22q11.2 deletion and duplication syndromes.
- To minimize the chances of spina bifida in a child, it is recommended that women of childbearing age start taking **folate** daily before becoming pregnant and continue during pregnancy.

Sacral Dimple

- A small indentation just above the crease of the buttocks, visible at birth
- Occurs in 2-4% of newborns
- Usually harmless
- In rare cases, **a sacral dimple may signal serious conditions like spina bifida and tethered cord**, especially if these are found near the dimple: a tuft of hair, extra skin, or the discoloration of the skin

Meningomyelocele or myelomeningocele (MM)



- MM is the most severe type of spina bifida but the causes are unknown.
- Part of the spinal cord is in the fluid sac that protrudes from the back. The sac is usually **not covered by skin,** and the **nerves and tissues are exposed to the outside world.**
- Babies with MM likely have:
  - **Hydrocephalus** – Fluid buildup in the brain that puts pressure on the brain. A tube (inserted via surgery) can drain the fluid
  - A higher risk of getting dangerous infections
  - Life-long incontinence problems with bladder and bowels – the person cannot control when urine and feces come out
  - Weakness or loss of movement in the legs
  - An increased risk of tethered cord syndrome (see next page)
- **Surgery is needed within 72 hours of birth** to close the opening and lower the risk of infections

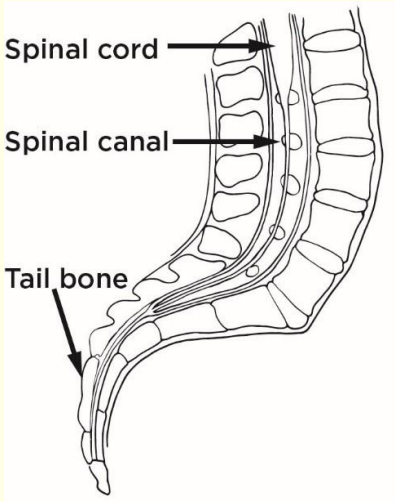
MM and 22q11.2DS

- Patients with MM are ~23 times more likely to have a 22q11.2 deletion than the general population
- Patients with 22q11.2DS have a 12 to 15 times higher risk of having MM compared to the general population
- The increase in the risk of MM may be due to the loss of 1 copy of the *CRKL* gene in the C-D region of chromosome 22q11.2.
- Individuals with 22q11.2 deletions involving A-D, B-D, and C-D regions have lost a copy of the *CRKL* gene
- *CRKL* encodes a protein involved in communication inside the cell.

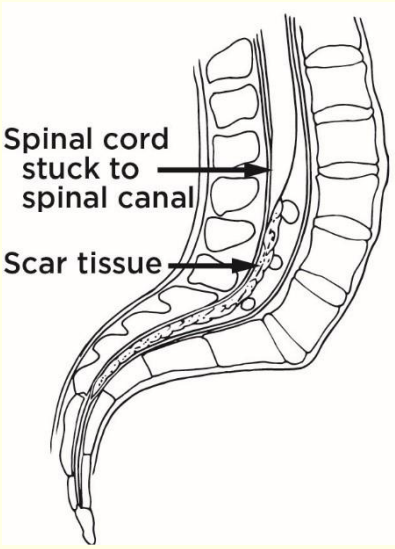
Structural Differences of the Spinal Cord (continued)

Tethered Cord Syndrome
(Tethered spinal cord syndrome)

- The **spinal cord** is a compact collection of nerves and nerve cells that run from the base of the brain downwards inside the backbone to the lower back. Nerve signals travel from the brain to the rest of the body and back.
- Normally, the spinal cord floats freely within the spinal column, allowing it to move as the body grows and bends (see top figure on the right).
- In tethered cord syndrome, **the spinal cord is abnormally attached** ("tethered") to tissues around the spine, usually at the lower back (see bottom figure on the right).
- A tethered spinal cord is **no longer free to move around**. Instead it stretches as a person grows and moves.
- Symptoms include back pain, leg weakness, numbness, difficulties with bladder or bowel control, and unusual marks on the back / bum.
- Tethered cord can happen:
  - At birth – Tethered cord alone or together with spina bifida (including Meningomyelocele)
  - Later in life – Cord becomes attached to tumors, infections, trauma or scars from surgeries (e.g. surgeries for spina bifida)
- Tethered cord can be diagnosed using spine imaging (e.g. MRI)
- Treatment typically involves surgery to release the cord from the surrounding tissues. This relieves symptoms and prevents further damage to the spinal nerves.
- If the tethered cord is not treated, the pull on the cord can limit the flow of fluids in the spinal cord and create a **syrinx** (a small sac of spinal fluid). This condition is called **syringomyelia**. The syrinx can in turn damage and affect the function of the spinal cord.
- Researchers do not know how common tethered cord syndrome is among individuals with 22q11.2DS. However, it is more common than meningomyelocele in this population.



Above: Normally, the spinal cord floats freely in the spinal canal.
Below: In tethered cord syndrome, the spinal cord is stuck to tissues.



Resources

- Sacral Dimple – Cleveland Clinic
- Sacral Dimple – Mayo Clinic
- Spina bifida – Mayo Clinic
- What is spina bifida? – Children's Hospital of Philadelphia
- Spina Bifida – National Institute of Neurological Disorders and Stroke
- Myelomeningocele – Cleveland Clinic
- Tethered Spinal Cord – Cleveland Clinic
- Tethered Spinal Cord Syndrome – Children's Hospital of Philadelphia
- Syringomyelia – Mayo Clinic
- 2-Minute Neuroscience: Early Neural development (video) – Neuroscientifically Challenged
- MRI (Magnetic Resonance Imaging) – Cleveland Clinic
- Risk of meningomyelocele mediated by the common 22q11.2 deletion – 2024
- Neurologic challenges in 22q11.2 deletion syndrome – 2018
- Neural Tube Defects and Atypical Deletion on 22q11.2 – 2014
- Microduplication 22q11.2: A Benign Polymorphism or a Syndrome With a Very Large Clinical Variability and Reduced Penetrance – 2008
- Updated clinical practice recommendations for managing [children][adults] with 22q11.2 deletion syndrome – 2023
- ❖ Graphic: Types of spina bifida – Wikipedia / CDC
- ❖ Graphic: Normal spinal cord and tethered cord – Christopher Reeve Foundation

International 22q11.2 Foundation

The mission of the International 22q11.2 Foundation is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.
This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.

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