

Diastematomyelia

Patient Information Series – What you should know, what you should ask.

What is diastematomyelia?

Diastematomyelia is a very rare malformation, with complete or partial clefting (a longitudinal split) of the spinal cord, which is divided into two hemi-cords. It usually occurs to the incomplete fusion of the spinal elements and has a soft/bony tissue dividing the spinal cord into two.

How does diastematomyelia happen?

The spinal cord and the intradorsal nerve roots are split into two columns. This mass may be a bony, fibrous or cartilaginous septum that subdivides partially or completely the vertebral canal. The cleft may be found at any level, but in most cases is found in the lower thoracic or upper lumbar regions. Thoracic region is the one that runs from the base of the neck down to the abdomen and the lumbar region is the lower part of the back.

Diastematomyelia acts as a restraint that slows the normal growth of the spinal cord by impeding the upward migration of the neural elements, with progressive neurologic deficits (abnormal function) in the limbs. Rarely, it can involve the bowel and bladder function as well depending on the extent of the severity.

It also can be associated with development of abnormal curvature of the growing vertebral column leading to “congenital scoliosis”.

Should I have more tests done?

The tests available depend on where you are. A consultation with a clinical geneticist can be considered after getting an ultrasound done with a fetal medicine specialist. Tests to ask about include an amniocentesis to look for problems of the number of chromosomes and some genetic underlying conditions. You might be offered other genetic diagnostic tests such as chromosomal microarray testing or whole exome sequencing. Such testing can provide essential information regarding your individual case. When an amniocentesis is done, a needle is inserted in your abdomen to collect some fluid from around the baby and test it. Because diastematomyelia has been described in association with other various anomalies (such as open spina bifida, scoliosis, kyphosis, hemivertebrae, cutaneous manifestations and orthopaedic deformities of the feet, especially clubfoot), you should also ask for a detailed ultrasound examination.

What are the things to watch for during the pregnancy?

After a detailed ultrasonographic assessment, associated findings in the spine and in the organ systems are ruled out. Advanced imaging with use of Magnetic Resonance Imaging (MRI) can be considered on a case-by-case basis to study the presence of “tethered/stuck cord”, as the bony elements in diastematomyelia can prevent expected physiological migration of neural elements. MRI can also help detect the presence of “closed” spina bifida, which may be difficult to diagnose on ultrasound. This can also aid in prognostication with the paediatric orthopaedic surgeon.

The outcome is generally favourable, but neurosurgical and orthopaedic surgery may be necessary, and there is a chance of neurologic compromise based upon the presence of other spinal conditions with or without “tethering of the spinal cord”.

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What does it mean for my baby after it is born?

When diastematomyelia presents as a closed neural tube defect, the prognosis for neurological function may be enhanced by early surgical removal of the septum, so it is important to refer to an orthopaedic center to plan the correct timing of the surgery.

Will it happen again?

The risk of recurrence is unknown. Autosomal dominant transmission has been described in rare cases, so genetic counselling is recommended.

What other questions should I ask?

- Is diastematomyelia isolated or associated with other anomalies?
- Is open spina bifida present?
- Is it possible to terminate the pregnancy?
- What factors should be considered in making a decision for termination of pregnancy?
- How often will I have ultrasound examinations done?
- Is there a need for advanced imaging (MRI)?
- Is surgery after pregnancy available?
- Where and how should I deliver?
- Where will the baby receive the best care after delivery?
- Can I meet in advance the team of doctors that will be looking after my baby following delivery?

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