

Sacral Agenesis

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

What is sacral agenesis?

Sacral agenesis (also known as sacral hypoplasia) is a rare condition, occurring in approximately 1 in 25,000 births, in which there is an abnormality in the development of the lower part of the spine. This can vary from partial absence or underdevelopment of the lower spine or complete absence of the lower spinal elements. Depending on severity, the lower extremities may be underdeveloped or even absent.

The condition can present with a wide variation from patient to patient involving the function of lower limbs, bowel, bladder and genitals.

How does sacral agenesis happen?

The cause of sacral agenesis is unclear, but it has a well-known association with diabetes in pregnancy. Mothers exposed to organic fat solvents, embryonal trauma, radiation, lithium salts, etc. may be at risk of having babies with sacral agenesis. Rare genetic associations have also been seen in babies with congenital sacral agenesis.

Should I have more tests done?

Sacral agenesis can be diagnosed by an ultrasound scan usually in the second trimester of the pregnancy, but an earlier scan may also rarely show features suggestive of sacral agenesis. Use of 3D sonography can aid in further delineation of the condition to better understand the extent of the disease.

A detailed scan of your baby's anatomy is recommended to look for other abnormalities which may be associated with sacral agenesis. An echocardiogram or an MRI may also be performed to complete the fetal evaluation.

You may be referred to genetic counselling and genetic testing. This may include tests like amniocentesis or chorionic villus sampling (CVS) to check for chromosomal abnormalities and other genetic diagnostic tests such as chromosomal microarray testing or whole exome sequencing. Such testing can provide essential information regarding your individual case.

What are the things to watch for during the pregnancy?

Sacral agenesis may be associated with maternal diabetes as well as an increase in the amount of amniotic fluid around the baby. Screening for diabetes should be done if not already available at the time of ultrasonographic diagnosis of sacral agenesis. These should be looked out during the pregnancy.

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

There is no known cure for sacral agenesis and treatment is guided as per the symptoms encountered, during the development of the baby. In continuing pregnancies, no modification of standard obstetric management is necessary, and delivery can occur vaginally at term. The baby will require multi-disciplinary care involving the paediatricians, orthopaedic surgeons, physiotherapists and radiologists to provide optimal care. Additional tests, including imaging modalities such as X-Ray of the spine and MRI can be required to assess the extent of the condition for planning further care.

Your child may suffer from lower limb motor deficit, urinary incontinence or sexual dysfunction. The treatment and long-term outcomes of the condition depends on the extent of the malformation, and may include orthopaedic and urologic surgery. The child may also require more than one surgery to manage symptoms as they grow older.

Will it happen again?

Sacral agenesis occurs sporadically but is much more common in infants born to mothers with pregestational diabetes. It is therefore most important to have adequate blood glucose control in the preconception period.

Targeted screening in subsequent pregnancies can aid in timely identification of a recurrence.

What other questions should I ask?

- Is sacral agenesis isolated or associated with other anomalies?
- Is it a complete or partial form?
- Should I have other tests?
- Can the pregnancy be terminated?
- How often should ultrasound examinations be performed?
- Is surgery after pregnancy available?
- Where should I deliver?
- Where will the baby receive the best care after it is born?
- Can I meet in advance the team of doctors that will be looking after my baby when it is born?

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