

# Septo-Optic Dysplasia (SOD)

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

## **What is a septo-optic dysplasia (SOD)?**

Septo-optic dysplasia (SOD) is a rare developmental disorder of the fetal brain. It involves abnormal development of the optic nerves, which may lead to poor vision after birth. In some cases, SOD can also affect the endocrine system, leading to hormonal imbalances.

## **How does SOD happen?**

SOD occurs when part of the brain called the cavum septum pellucidum (CSP) fails to develop properly. This structure is located in the anterior (front) part of the brain, and when it is underdeveloped or absent, it is known as agenesis of the septum pellucidum (ASP). ASP can be associated with abnormal development of other brain structures, such as the optic nerves. ASP and SOD are often first suspected during routine mid-pregnancy ultrasounds.

## **Should I have more tests done?**

Your caregiver may refer you to additional tests that can help assess the baby's condition more thoroughly. You may be referred to fetal neurosonography, a detailed brain ultrasound, and/or Magnetic Resonance Imaging (MRI), if available, which can provide more detailed images of the fetus's brain and other organs. You might also have prenatal genetic counseling and testing. Genetic tests like amniocentesis, where a small amount of amniotic fluid is drawn and sent for analysis of the baby's chromosomes, where most of our genetic information is kept. You might also be referred for advanced genetic tests like chromosomal microarray or exome sequencing, for in-depth investigation, to determine if there is a genetic cause of the ASP or SOD.

## **What is the prognosis of SOD and ASP?**

When ASP is isolated (occurring without other abnormalities), the prognosis is generally favorable. During pregnancy, ultrasound and MRI can help evaluate the development of the optic nerves, which are crucial in determining the baby's potential vision. However, it is not always possible to completely rule out the diagnosis of SOD before birth.

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

If the optic nerves appear normal on prenatal imaging, the baby is likely to have good vision. However, a definitive assessment of vision can only be made after birth through a detailed eye examination (ophthalmologic evaluation). Additionally, blood tests may be needed to assess hormone levels and check for any endocrine dysfunction. The risk of this condition recurring in future pregnancies is extremely low. During genetic counselling, your counsellor will provide detailed information based on the results of genetic testing.

## **What other questions should I ask?**

- What prenatal testing is recommended for my baby?
- How often will I need ultrasounds?
- Where should I deliver my baby?
- Can I consult with the medical team that will care for my baby after birth, before my delivery?

*Last updated September 2024*