Oculo-auriculo-vertebral spectrum (Goldenhar syndrome)

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

What is Oculo-auriculo-vertebral spectrum (OAVS)?

A baby with oculo-auriculo-vertebral spectrum (OAVS) usually has multiple abnormalities of the face (mouth, eyes, palate, jaw, cheekbones), ears, and spine. OAVS includes diagnoses known as hemifacial microsomia (HFM) and Goldenhar syndrome (GS). The severity of the anomalies varies widely from case to case and may also include additional heart, renal, pulmonary, and nervous system problems. Babies with a very small chin may have breathing difficulties at birth that require delivery at a specialised center.

What is the likelihood of OAVS being detected in a routine anatomy scan?

Unfortunately, only the most severe cases of OAVS can be detected on routine prenatal ultrasound examinations. The typical findings are the presence of one side of the face smaller than the other, a very small chin, a wider mouth than normal, a partially formed or completely absent ear, and spinal deformities such as scoliosis (lateral deviation of the spine). In most cases, the anomalies affect only one side of the body, and sometimes they can be very subtle.

How does OAV happen?

The cause of most cases of OAVS is unknown. Generally, it occurs in families with no history of the condition. In rare cases, OAVS can be inherited from parents and follow an autosomal dominant or recessive pattern, that is, if one parent has OAVS, each of his or her children has a 50 or 25 percent chance of inheriting the disease respectively. However, in most families where one child has OAVS, the likelihood of another child being born with the same condition is very low.

How are the chromosomes relevant to OAV?

Most individuals with OAVS have a normal chromosome number. However, there are reports of abnormal chromosomes in patients with OAVS, so parents should consider the option of prenatal karyotype (or even better, microarray) testing. Microarrays are genetic tests that can detect smaller variations in DNA than karyotype and, therefore, can provide useful diagnostic information in fetuses with multiple structural defects.

Should I have more tests done?

A 3D ultrasound may be helpful in diagnosing facial abnormalities, and an MRI could help in further assessment of the mouth and upper airway in cases of severe OAVS. If the baby has a very small chin, a focused MRI examination of the mouth and upper airway could help the perinatal team to assess the need to perform an EXIT-to-airway procedure. This is a special type of delivery in which the baby is ventilated before the umbilical cord is cut from the placenta. As with most babies with multiple congenital defects, the ultrasound and MRI images will be discussed with a team of specialists, which may include a maternal-fetal medicine specialist, a geneticist, an ENT specialist, a neonatologist and a plastic surgeon.

What are the things to watch for during pregnancy?

If the baby has difficulty swallowing the amniotic fluid, you may feel that your belly is growing rapidly towards the end of pregnancy. This accumulation of amniotic fluid (polyhydramnios) may trigger preterm labour. Before this happens, your doctor may try to drain some of the amniotic fluid with a needle to prolong the pregnancy and give you corticosteroids to support the baby's lungs if you deliver too early.

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

Most babies with OAV have a normal life expectancy and a good prognosis. Because some patients with OAVS may have difficulty breathing and feeding in the early neonatal period, it is advisable that they are born in a center with neonatal and surgical capabilities, if available. There are several craniofacial syndromes whose findings may overlap with those of OAVS, so in most cases, the final diagnosis is not made until several days after birth. It is likely that most babies with OAVS will require multiple corrective surgeries and long-term follow-ups with ENT, orthopedics, and ophthalmologists.

Will it happen again?

Other than the unusual, well-known cases reported in the medical literature, the risk of recurrence is very low.

What other questions should I ask?

- Does this look like a severe case of OAV spectrum?
- Is it possible to terminate the pregnancy?
- Can I schedule a consultation with a geneticist to better understand what OAV means (what abnormalities I should expect, what my baby will look like, and the likelihood that my future children will have this syndrome)?
- Where should I deliver?
- Where is the best place to care for the baby after birth?
- Can I meet the medical team that will care for my baby at birth in advance?

Last updated June 2023