Tuberosus Sclerosis with Cortical Hamartomas (TSC)

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

What is tuberous sclerosis (TSC) and how can it affect my baby?

Tuberous Sclerosis Complex (TSC) is a genetic disorder that affects various organs, including the brain, heart, and kidneys. It occurs in approximately 1 in 5,000 to 10,000 births. The neurological symptoms are often the most severe. Many children with TSC experience seizures that are difficult to control, intellectual disabilities, and/or autism spectrum disorder. In some cases with severe seizures, they can become life-threatening.

How do cortical hamartomas in TSC develop?

Cortical hamartomas are brain lesions that result from mutations in the **TSC1** or **TSC2** genes, which normally regulate cell growth. Mutations in these genes lead to uncontrolled cell growth, abnormal brain development, and the formation of benign tumors, particularly in the cerebral cortex (the brain's outer layer). These tumors can disrupt normal brain function, contributing to seizures and developmental delays.

How are chromosomes involved in TSC?

Chromosomes are where most of our genetic information is kept. Our genetic information is organized into 46 chromosomes, inherited from both parents. Mutations in either the TSC1 or TSC2 genes, located on these chromosomes, cause tuberous sclerosis. The "two-hit hypothesis" explains how this condition happens. The first "hit" is a mutation present in the sperm or egg cells, which can cause mild symptoms or none at all. A second "hit" during early development, particularly in brain or heart cells, can lead to more severe manifestations like brain tumors or heart abnormalities.

Should I have additional tests done?

Your caregiver may refer you to additional tests that can help assess the baby's condition more thoroughly. Fetal echocardiography, a specialized ultrasound to detect tumors in the baby's heart, called rhabdomyomas, that often occur with TSC. Fetal neurosonography, a detailed brain ultrasound to detect cortical hamartomas or other structural changes. Magnetic Resonance Imaging (MRI), if available, an MRI scan provides 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, can help identify mutations in the TSC1 or TSC2 genes. You might be referred for advanced genetic tests like chromosomal microarray or exome sequencing, for in-depth investigation of the genetic cause of the TSC.

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What should I watch for during pregnancy?

While there is currently no treatment for TSC during pregnancy, close monitoring is essential. Regular ultrasounds can track the progression of any brain or heart tumors. Your caregivers will also monitor for signs of abnormal heart rhythms (arrhythmias), fluid buildup in the brain (hydrocephalus), or heart failure (fetal hydrops).

If there are no complications, delivery is typically recommended at a hospital with a neonatal intensive care unit (NICU). Vaginal delivery is possible unless complications like fetal hydrops are present, in which case a cesarean section may be recommended. Your caregiver can advise you what is best for you and your baby.

What does it mean for my baby after birth?

The severity of TSC symptoms can vary greatly between patients, from mild to severe, depending on the location and size of the lesions. Some children may have behavioral, cognitive, or psychiatric challenges, while others may experience seizures that are difficult to manage.

Will this happen again in future pregnancies?

If either parent carries a mutation in the TSC gene (inherited in an autosomal dominant manner), there is a 50% chance of passing the condition on to future children. However, in about 65% of cases, the mutation occurs spontaneously (de novo), meaning the parents do not have the mutation, and it arises for the first time in the child, and there is no increased risk of recurrence. Your genetic counselor can advise you about the risk for your future children, based on the results of your genetic testing.

What other questions should I ask?

- What prenatal testing is recommended for my baby?
- Is there any prenatal treatment available?
- How often will I need ultrasounds?
- Where should I deliver my baby?
- Where will my baby receive the best postnatal care?
- Can I meet the medical team that will care for my baby after birth, before my delivery?

Last updated September 2024