

Congenital hypothyroidism screening



What is congenital hypothyroidism?

Congenital hypothyroidism is when a baby's body can't make enough thyroid hormone. For every 3,000 to 4,000 newborns, 1 is born with this problem. Thyroid hormone is important for your baby's brain to develop and grow. Without treatment, babies can't grow and develop normally.

What causes congenital hypothyroidism?

When the thyroid gland does not develop properly, it does not make thyroid hormone well. Sometimes the pituitary gland does not tell the thyroid to make the hormones. In a few cases, congenital hypothyroidism is caused by medicines taken by mom during pregnancy. This usually lasts for a few weeks to months after the baby is born. Congenital hypothyroidism is not usually passed on through families.

What are the signs and symptoms of congenital hypothyroidism?

The symptoms of congenital hypothyroidism are not obvious. Some babies don't have any symptoms. If it is serious, babies might have:

- Poor feeding
- A weak cry
- A hard time staying awake
- Constipation
- Yellow skin
- Puffy face
- Weak muscles
- Big tongue
- Big belly
- Large fontanelles (soft spots on head)

How do we diagnose congenital hypothyroidism?

It is hard to tell if babies have congenital hypothyroidism. So, all hospitals in the United States check for it with a blood test collected from your baby's heel. This process is called newborn screening. This test might miss congenital hypothyroidism in some babies (especially those who are sick, born premature, low birth weight, got blood transfusions, or are twins or triplets). To make sure we find all cases, we check for hypothyroidism again at 2 weeks of age for babies who are in the hospital. This is done using a blood test. If there is a positive result, your baby will need more blood tests before starting treatment. In some cases, the doctor may order a thyroid scan to see if the thyroid gland is missing or too small.

How is congenital hypothyroidism treated?

Thyroid hormone medicine treats congenital hypothyroidism. This medicine is a pill called levothyroxine. Many babies will need treatment for life. You should crush this medicine and give it to your child once a day mixed with water, human milk, or formula.

Endocrinology