

## What is Congenital Hypothyroidism?

Congenital hypothyroidism is a disorder that occurs when a baby's thyroid gland, which is located in the neck, does not produce enough thyroxine (T4). Thyroxine is a hormone (body substance) which is necessary for normal physical growth and mental development.

## What causes Congenital Hypothyroidism?

The thyroid gland is unable to produce enough thyroxine because it has not developed properly. In some cases, a baby's thyroid gland is absent entirely, so there is no thyroxine produced. About one of every 4,000 babies is born with congenital hypothyroidism.

## How is Congenital Hypothyroidism found?

Shortly after birth, several drops of blood are taken from a baby's heel. The dried blood is sent to the State Department of Health and Senior Services Inborn Errors of Metabolism Laboratory, where it is tested for congenital hypothyroidism and several other conditions. These tests are all part of the State's Newborn Screening Program. If the results are not within an acceptable range, the baby's doctor and parents are notified.

## What does the lab look for?

The lab tests the baby's blood for:

1. Thyroxine (T4)-the hormone made in the thyroid gland. When a newborn has hypothyroidism, the T4 level is low (not enough thyroxine is being made).
2. Thyroid stimulating hormone (TSH)-this hormone is produced by the pituitary gland in the brain and stimulates the thyroid gland to produce T4. When the thyroid gland is not producing enough T4, the pituitary gland produces more TSH, signaling the thyroid gland to work harder. If the T4 level is low, the TSH level is usually high.

## Does an abnormal screening test mean that my baby has CH?

Screening tests always need to be confirmed by additional testing and medical evaluation.

**IF YOU ARE ASKED TO HAVE YOUR BABY RE-TESTED, ACT QUICKLY AND FOLLOW YOUR DOCTOR'S ADVICE**

## What will my baby's doctor do?

Depending on the results of the lab test, your doctor may repeat the testing or refer your baby to a specialist (pediatric endocrinologist). Your doctor and the specialist will confirm the diagnosis of CH by doing one or more of the following:

- Getting information about your baby
- Examining your baby
- Doing a special test known as a thyroid scan or ultrasound to see whether the baby has thyroid gland, where it is located, and how well it is working.

## How is Congenital Hypothyroidism treated?

If congenital hypothyroidism is found and treated early, then problems such as mental retardation and poor growth can be prevented. Congenital hypothyroidism is treated by giving the baby the thyroxine that the baby cannot make on its own. Your baby's doctor or pediatric endocrinologist will prescribe a thyroid medication by mouth that only needs to be given once a day. The earlier treatment is started, the better chance a baby has for reaching his/her full potential. Frequent medical checkups are essential to be sure that your child is growing and developing normally.

## Is Congenital Hypothyroidism permanent?

In most cases congenital hypothyroidism is permanent and treatment must be continued throughout life. Occasionally the condition is temporary and the child's supervising doctor is able to discontinue medication after 2-3 years.

For more information, please contact:

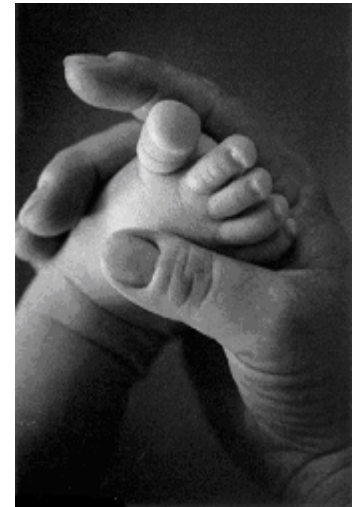
The New Jersey Department of Health and  
Senior Services

Newborn Screening and Genetic Services at  
(609) 292-1582

or

The Inborn Errors of Metabolism Laboratory  
at (609) 292-3090

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## Congenital Hypothyroidism

Important Information for  
Parents

