You have just learned that your infant has congenital hypothyroidism. Naturally you are concerned and have many questions. This information sheet will help answer some of your questions. However, it should not take the place of an informed discussion with your baby’s doctor (primary care provider).

What is the thyroid gland?

The thyroid gland is located in the front, lower half of the neck. It produces thyroid hormone, also called thyroxine (T4). Thyroxine is absolutely essential for normal growth and development of the brain and body in infants. The thyroid gland is controlled by the pituitary gland which is located in the brain. The pituitary gland senses the amount of thyroxine in the blood, and if there is not enough or too much thyroxine, the pituitary gland signals the thyroid gland using a hormone called thyroid stimulating hormone (TSH) to make more or less thyroxine (T4). When there is not enough T4, the level of TSH increases. If there is enough T4, the level of TSH is normal.

What is congenital hypothyroidism?

Congenital hypothyroidism is a disorder that affects infants from birth (congenital). It is caused when the thyroid gland does not make enough thyroid hormone (thyroxine) after birth. Abnormal growth and development of the thyroid gland is the most common reason that enough thyroxine is not produced. Thyroxine is necessary for normal development of the brain in infants and normal body growth. If affected babies are not treated they will not grow and develop normally and will develop permanent mental retardation. When detected and treated early, babies with congenital hypothyroidism grow and develop normally.

What are signs and symptoms of congenital hypothyroidism?

Babies with congenital hypothyroidism often look completely normal up to three months of age, even though they may have this disorder. Most of the time you cannot tell an infant has a problem. This is the reason newborn screening is so important. However, infants with congenital hypothyroidism may have one or more of the following signs:

- large infant
- large fontanelle or soft spot
- large swollen tongue
- puffy face
- hoarse cry
- cold arms and legs, hard time staying warm
- decreased energy, very sleepy, difficulty staying awake
- poor muscle tone
- problems with jaundice or yellow color of skin and whites of eyes (sclera)
- poor feeding, hard time latching-on and sucking
- constipation, frequent vomiting, bloated abdomen
- little or no growth

Usually, infants born with signs and symptoms are at greater risk of developmental and growth delays than infants who are born with no signs or symptoms.
How often does congenital hypothyroidism occur?

Congenital hypothyroidism occurs in 1 out of every 3,000 to 4,000 live births. Mothers who have hypothyroidism may pass TSH receptor blocking antibodies through the placenta, causing temporary hypothyroidism in the infant. All cases of primary and secondary congenital hypothyroidism are permanent. Congenital hypothyroidism occurs most often in families that do not have a history of the disorder. About 15 to 20 percent are due to heredity.

What testing was done?

Screening: In Utah, an infant receives two newborn screening tests that check for congenital hypothyroidism, in addition to other disorders. A few drops of blood are collected from the infant’s heel. This screening test makes it possible to detect hypothyroidism before signs and symptoms appear. The first specimen is usually collected before hospital discharge and the second specimen between 7 and 28 days of age. The newborn screening test measures the level of thyroid stimulating hormone (TSH). If the TSH is abnormal, a thyroxine (T4) is done. The results are reported out immediately to the baby’s doctor as abnormal when the TSH level and T4 level are outside of their normal ranges, indicating the baby may have congenital hypothyroidism.

Confirmatory testing: Abnormal newborn screening results are referred to the baby’s doctor with a recommendation for a physical examination and evaluation and more testing. A blood specimen is drawn to test the levels of the serum free T4 and serum TSH. These tests are more specific and help your doctor to know if your baby has congenital hypothyroidism. Once this disorder is confirmed, treatment is begun.

How is congenital hypothyroidism treated?

Congenital hypothyroidism is treated by replacing the thyroxine the infant’s thyroid gland does not produce by using a synthetic or man-made thyroxine called levothyroxine. Synthroid, levoxyl and levothroid are examples of levothyroxine. It is a tablet that can be crushed or dissolved in a small amount of water and placed in the infant’s mouth. It works just like the thyroid hormone normally made by our bodies and must be taken by your baby every day for his or her entire lifetime.

Frequent monitoring of serum levels of T4 and TSH are necessary to adjust the hormone dosage needed because babies are growing rapidly. Once normal levels are reached and stable, your doctor will check your baby’s blood levels of T4 and TSH every 2 to 3 months for the first 3 years of life to make sure your baby is getting enough hormone replacement. Thyroid medication can be given with soy formula, but soy may decrease medication absorption and result in a need for a higher dose of levothyroxine. It is imperative that your baby’s blood levels are checked if a change is made to soy formula from a non-soy formula. Iron supplements may also interfere with medication absorption.

Successful treatment of congenital hypothyroidism depends on early and life long daily hormone replacement with close follow-up by your baby’s doctor of hormone levels. This will ensure your baby grows and develops normally.