ENDOCRINE DISORDERS:

CONGENITAL HYPOTHYROIDISM (CH)

What is Congenital Hypothyroidism?

CH stands for "Congenital Hypothyroidism". Congenital means present at birth. Hypothyroidism is a condition in which the person does not make enough thyroid hormone. The thyroid gland is a butterfly-shaped organ at the base of the neck. Its job is to make specific hormones that help the cells of the body function correctly. The main hormone made by the thyroid gland is thyroid hormone, also called 'thyroxine', or T4. It is released by the thyroid gland into the bloodstream whenever it is needed by the body. In babies and young children, thyroid hormone is important for normal growth and development of the body and brain. It helps cells work more efficiently and also helps maintain body temperature. Babies who do not have enough thyroid hormone and are left untreated will have mental retardation, developmental delay, and stunted growth.

WHAT ARE THE CAUSES OF CONGENITAL HYPOTHYROIDISM?

There are a number of different causes for CH which are mentioned below.

1. Missing or misplaced thyroid gland

The normal thyroid is a butterfly shaped gland located at the center of the front of the neck, above the windpipe. Most babies with CH have a missing, abnormally located, or a smaller than usual thyroid gland (Figure 1).

2. Maternal iodine deficiency

If the mother has low iodine intake during pregnancy, the fetal thyroid gland may not be able to make enough thyroid hormone, and develop CH. This is a problem in some parts of the Philippines where people do not get enough iodine in their diet. This is rare now, because table salt is usually fortified with iodine ('iodized salt'), and other foods such as dairy products, contain iodine as well.

3. Maternal thyroid condition and medications

In a small number of cases, CH can occur when the mother has a thyroid condition or is given an anti-thyroid medication during pregnancy.

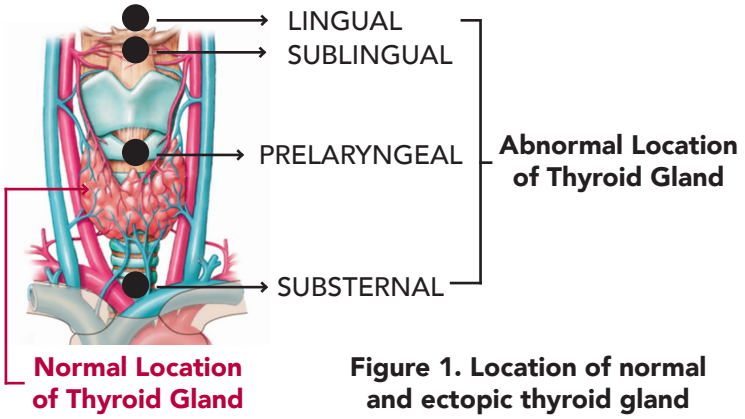

Figure 1. Location of normal and ectopic thyroid gland

IS CONGENITAL HYPOTHYROIDISM INHERITED?

Less often, CH is caused by inherited changes in a gene or pair of genes. Children with the inherited type of CH do not make enough thyroid hormone even though their thyroid gland appears normal in size and shape. About 15% of children with CH have an inherited form.

CONGENITAL HYPOTHYROIDISM (CH)

IF CH IS NOT TREATED, WHAT PROBLEMS OCCUR?

Most babies with CH do not have symptoms right after birth but some may present with the following:

- Jaundice or yellowish discoloration of the skin
- Low activity level - babies sleep more than usual and don't move as much
- Poor feeding and poor suck
- Fewer bowel movements or constipation
- Floppy muscle tone (hypotonia)
- Swelling around the eyes and a puffy face
- Large swollen tongue
- Cool, pale, dry skin
- Large soft spot on the skull (the fontanel) that closes late
- Large belly with protruding navel ('umbilical hernia')
- Hoarse cry

Children who remain untreated usually become mentally retarded and are much shorter than average. They may have spasticity and an unsteady gait. Most have speech delays, some have behavioral problems and poor school performance.

If left untreated, babies may develop some or all of the following effects over time:

- Delayed teething
- Breathing problems
- Goiter (enlarged thyroid gland causing a lump in the neck)
- Pale skin
- Hearing loss

WHAT IS THE TREATMENT OF CH?

Your baby's doctor may work with a pediatric endocrinologist, a doctor with training in treating children with thyroid and other hormone problems, to care for your child. The main treatment for CH is thyroid hormone replacement, which should be initiated within 2 weeks after birth. It is safe and easy to take. If your child is diagnosed and treated early, the developmental delay, growth stunting and mental retardation can be prevented.

1. Medication

L-thyroxine is a synthetic form of thyroid hormone which comes in tablet form. Your doctor will decide how much L-thyroxine your baby needs. L-thyroxine tablets are small and can be crushed into food or dissolved into a small amount of formula, juice or other liquid. Young children can easily chew and swallow the pills. It is important to give your child the correct amount of L-thyroxine. Tell your doctor if your baby is given a soy-based formula or iron supplements so that dose adjustments can be made.

2. Monitoring

Your child will need regular visits to the doctor to check his or her weight, height, development and overall health. Your child will also likely need regular blood tests to check the level of thyroid hormone. Blood tests are usually done every one to three months until age one, and then every two to four months until age three. They can usually be done less often after age three. It is encouraged to breastfeed your baby and should receive the usual recommended immunizations for age.

3. Developmental monitoring

Your doctor may suggest a formal evaluation of your child's development. If your child show delays in certain areas of learning or speech, a proper referral can be made.

CONGENITAL HYPOTHYROIDISM (CH)

What will be the outcome for patients with CH?

Children with CH who start treatment two weeks after birth usually have normal growth and intelligence and can live typical and healthy lives. If treatment is not started early or irregularly given, delays or learning problems may occur.

Reference:
http://newbornscreening.info/Parents/otherdisorders/CH.html Accessed on 7 November 2014