DEFINITION OF CONGENITAL HYPOTHYROIDISM

Congenital hypothyroidism is a condition of thyroid hormone deficiency present at birth. It represents one of the most common preventable causes of mental retardation. Approximately 1 in 4000 newborn infants has a severe deficiency of thyroid function, while even more have mild or partial degrees. If untreated for several months after birth, severe congenital hypothyroidism can lead to growth failure and permanent intellectual disability.

The fetal hypothalamic-pituitary-thyroid axis begins to function by midgestation and is mature in the term infant at delivery. If fetal hypothyroidism develops, untoward effects may be demonstrated in certain organ systems, including the central nervous system and skeleton. However, most infants with Congenital Hypothyroidism appear normal at birth. Recent data suggest that the hypothyroid fetus is protected to a certain extent by placental transfer of maternal thyroid hormone; serum thyroxine ($T_4$) levels in the cord blood of thyroid fetuses approximate one third of maternal levels.

CAUSES OF CONGENITAL HYPOTHYROIDISM

Congenital hypothyroidism can be caused by the formation of an abnormal thyroid gland during pregnancy whether it is:
• Does not exist
• Outside the normal position
• The thyroid gland is not functioning

SIGNS OF CONGENITAL HYPOTHYROIDISM

Typically, most babies with Congenital Hypothyroidism did not show any unusual signs at birth. However, early signs of Congenital Hypothyroidism are:
• Umbilical Hernia
• Constipation
• Difficulty feeding
• Less active
• Prolonged Neonatal Jaundice
Symptoms of untreated Congenital Hypothyroidism can be detected when a baby is about 3 to 6 months old; such as:
- Rough face expression
- Guttural cries
- Dry and rough skin
- Dry hair
- Stunted development and growth
- Mental retardation

SCREENING PROGRAMME FOR CONGENITAL HYPOTHYROIDISM
With the introduction of newborn screening programmes for Congenital Hypothyroidism, newborns with hypothyroidism are detected early before clinical manifestations are evident; this enables thyroxine replacement treatment to be instituted, ideally within two weeks of birth, thus reducing the risk for cognitive problems. With the changing priorities in health care, the improvement in economy and lifestyle will lead to preventive issues that include national screening programs which have been well established in many industrialized countries.

Screening programs for Congenital Hypothyroidism were started in USA, Canada and Western Europe. It is now an established national programme in many industrialised
countries; in Europe from 1974 to 1979, United Kingdom in 1982, Australia and New Zealand in 1983, Hong Kong in 1984 and Singapore in 1989. Apart from Singapore and Hong Kong, congenital hypothyroid screening is not being done routinely on a nation-wide scale in the South East Asian region. Malaysia, too, has yet to develop a national screening programme for this condition. In October 1998, Ministry of Health (MOH) commenced newborn screening for Congenital Hypothyroidism in Malaysia. The screening methodology thus differs from country to country with respect to:

(i) site of sample collection - cord blood or capillary heel prick
(ii) timing of sample collection
(iii) test strategies, and
(iv) recall criteria

TEST DETECTION OF CONGENITAL HYPOTHYROIDISM

Congenital Hypothyroidism can be detected through a blood screening test from the umbilical cord immediately after birth (must be within half an hour from the delivery period to prevent blood clots and TSH surge that occurs after \( \frac{1}{2} \) to 72 hours). If blood sample was not taken at birth for some reason, an appointment should be given to parents to take their baby to the paediatric clinic after the third day of birth where blood samples are taken for screening. Screening test performed on a blood sample is TSH (Thyroid Stimulating Hormone). If it is found to be abnormal, a confirmatory tests (T4 and TSH) will be performed. Screening and verification is done at all state hospitals and most district hospitals. Hospitals that do not have laboratory facilities to carry out the screening and confirmation testing, blood samples will be sent to the reference hospital.

TREATMENT FOR CONGENITAL HYPOTHYROIDISM

Infants who suffer from Congenital Hypothyroidism should be treated as soon as possible. The delay in starting treatment can cause mental retardation and development. Congenital Hypothyroidism is treated by replacing thyroxin that cannot be produced by the thyroid gland, usually in the form of L-thyroxin tablets which will be set by the doctor treating the baby. Follow-up on the appointed date is very important to make sure the baby gets the proper treatment. L-thyroxin medication is lifelong.

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