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Thyroid UK

Congenital Hypothyroidism

Your Notes

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Congenital Hypothyroidism

Congenital Hypothyroidism (CH) is the name for babies born with hypothyroidism due to the thyroid gland being absent or severely deficient. If it is not detected and treated early irreversible neurological problems and poor growth can occur.

Some children may develop primary hypothyroidism after birth rather than CH and these children do not experience the irreversible neurological problems due to untreated CH.

CH is found in around 1 in 4,000 live births and it is twice as common in females. There is a possibility that it is more prevalent in multiple pregnancies.

Causes

75% of cases of CH may be due to a total or incomplete development of the thyroid, which is not inherited or a failure of pituitary control (TSH is absent).

10% of cases are due to an enzymic failure in the manufacture of thyroid hormones, (defects in thyroglobulin), which is usually inherited so there may be a risk to further infants.

5% of cases are due to hypothalamic-pituitary dysfunction and this type of dysfunction usually causes other disorders such as lack of growth hormone etc. This type of cause may include tumours, arterial blockage and congenital defects.

5% of patients will also have other congenital defects and in some cases there are genetic defects that have caused CH.

Lack of early treatment may cause delayed mental development, learning difficulties and poor co-ordination.

Some cases are due to other causes such as the mother taking medications or having thyroid antibodies that have crossed the placenta. This kind of problem improves after delivery of the baby.

Symptoms

- Unresponsive
- Feeding difficulties
- Constipated
- Doesn't cry very much

Signs

- Puffy appearance
- Tongue may appear enlarged and may protrude
- Broadened and flattened nose
- Large fontanelles
- Nasal obstruction
- Low temperature (often under 35^{o}C)
- Cold and mottled skin
- Jaundice
- Umbilical hernia
- Weak and floppy muscles (hypotonia)
- Hoarse voice

- Enlarged heart
- Slow heart rate
- Fluid around the heart
- Failure of fusion of distal femoral epiphyses
- Older children will have short stature, hypertelorism (distance between the eyes) and narrow palpebral fissures and swollen eyelids
- Goitre

Diagnosis

All babies are screened at birth (the heel prick test) for hypothyroidism and if a high TSH is detected a diagnosis will be confirmed. Sometimes infants are tested for thyroid auto-antibodies and given ultrasound scans as well.

Some babies may be subclinical, in which case they need to be tested again a few months later.

Management

Early detection means early treatment to ensure that infants do not develop irreversible neurological disability.

Levothyroxine is given with dosage increases until thyroid function tests return to normal and then testing is done regularly until the infant is two years of age. FT4 should be kept in the upper half of the normal range.

Children's growth and mental development should be monitored throughout their childhood.