British Society for Paediatric Endocrinology and Diabetes

CONGENITAL HYPOTHYROIDISM (CHT)

As you know a small heel prick blood sample for chemical testing was taken from your baby around the fifth day of life. The tests on that sample have shown that your child may have congenital hypothyroidism (CHT). This is a condition in which there is underactivity of the thyroid gland (hypothyroidism) present at the time birth (congenital).

The thyroid gland lies in the front of the neck across the upper part of the trachea (windpipe). Its main function is to produce and transfer into the blood a chemical (hormone) called thyroxine. After birth this hormone has an important role in regulating the growth and development and the chemical activity of nearly all the cells in the body, including the brain. Lack of thyroxine causes slowing of these processes. Thyroxine is an essential chemical at all times of life but particularly in the growing child. Before birth the baby receives a supply of thyroid hormone from mother but after birth the baby's well-being will be dependent on the thyroid hormone that it can make for itself.

Cause

In the UK about 1 child in 3,500 is thought to be affected by CHT. In most babies with CHT (80%) the thyroid gland fails to develop properly before birth. This is often referred to as thyroid gland 'dysgenesis'. The cause of thyroid dysgenesis is usually unknown. In a minority of babies (20%) the thyroid gland fails to work adequately because of a problem with the thyroid hormone 'production line' – a 'dyshormonogenesis'. CHT is seldom apparent from the baby's physique or behaviour until some weeks after birth but it is extremely important to identify the problem early because if it remains undetected and untreated there is slowing of the development of all organ systems including the brain. If thyroxine is not given within the first few weeks after birth then brain damage can occur which is why all babies are tested for this condition. If the screening test shows any indication of underactivity then thyroid function must be checked more accurately on a blood sample taken from a vein. If this confirms underactivity then treatment with thyroxine is started immediately. As long as the thyroxine is started within the first weeks after birth then growth and development is almost always normal. Some paediatricians will also suggest that your baby has a special scan in the first weeks of life. This is to work out in more detail what the problem with the thyroid gland actually is.

Treatment

Thyroxine is made chemically and is available as small white tablets in 25, 50 and 100 microgramme (mcg) sizes. Liquid preparations (solutions) are also available and are used by some doctors. Most babies are started on a dose of 25-50 mcg thyroxine daily. A dose equivalent to 37.5mg a day can be given by administering 25 and 50 mcg tablets on alternate days or by giving one and a half of the 25mcg tablets daily. The tablets are easily crushed and given with some milk off a spoon. On treatment further blood samples are needed to allow adjustment of the dose to produce a normal level of thyroxine and a normal level of the thyroid controlling hormone (TSH) in the blood. Frequent blood tests are needed at first but are required less often as the child grows older. The dose of thyroxine usually needs to be increased as the child grows but most adults only need 150-200mcg per day. Do not be misled by the small size and unimpressive nature of the thyroxine tablets; this hormone is absolutely essential for the normal growth and development of your child. Establish a foolproof routine for giving the tablet or solution daily.

Occasionally CHT is transient and the function of the gland turns out to be normal in the longer term. If there is any doubt about whether lifelong thyroxine treatment is needed then a further check on the function of the gland can be made. This requires stopping treatment briefly and is best done when the child is a little older, usually after the age of two or three years. No harm at all will have resulted from giving thyroxine up till then because the natural secretion from the gland is adjusted to maintain a normal level.

Outlook

On treatment the child with CHT is normal. Thyroxine is not really a medication - it is simply an exact replacement of a missing chemical. The child can therefore have the usual set of immunizations and treatments needed for other conditions without any problems. There is a natural tendency to worry that quirks of physique or behaviour may relate to the CHT or its treatment but be assured that as long as the thyroxine dose is appropriate then this is not the case.