



Down's Syndrome Association

Medical Series

1. THYROID DISORDER AMONG PEOPLE WITH DOWN'S SYNDROME

Notes for parents & carers

WHAT IS THE THYROID GLAND?

The thyroid gland is situated in the neck in front of the trachea (windpipe), just below the larynx (voice box). It produces several chemical substances called hormones, which circulate round the body in the blood. These hormones, one of which is thyroxine, help to regulate the body's energy level. Thyroxine has a major influence on physical and mental development and on general wellbeing. This is because it helps to control the rate of chemical reactions in all the body cells.

WHAT IS THYROID DISORDER?

If the thyroid gland is overactive (hyperthyroidism) a person may be agitated and jittery, lose weight and suffer palpitations. If the gland is underactive (hypothyroidism) a person can become tired, overweight and generally sluggish with slow physical and mental reactions.

People with Down's syndrome do sometimes have an overactive thyroid, but it is far more common among them for the gland to be underactive. We know that in 15 – 20% of adolescents with Down's syndrome the thyroid gland is not working properly. Not enough thyroxine is produced to keep the body running at an optimum rate. These people benefit greatly from thyroid replacement therapy. This involves taking one or more tablets of a thyroid preparation every day.

HOW DO YOU KNOW IF THERE IS A PROBLEM WITH THE THYROID GLAND?

It is far more difficult to diagnose thyroid deficiency in children and adults with Down's syndrome than in the general population. Some classic features of poor thyroid function – tiredness, overweight and general sluggishness – have already been mentioned. In addition the person may feel the cold, have a tendency to constipation, have dry skin, sparse hair and a rather hoarse voice. Those of you who have a relative or friend with Down's syndrome will recognise that similar features can in any case be quite common in people with the syndrome, so we cannot rely on these signs to identify those who would benefit from thyroid replacement therapy. Therefore *the only way we can be sure of recognising those in need of treatment is to carry out regular blood tests.*

Testing For Thyroid Disorder

AT BIRTH

In the UK it is national policy for all newborn babies to have their thyroid function tested. A spot of blood from a heel prick is collected on a piece of filter paper and allowed to dry. This is sufficient to allow several tests to be carried out to check for some serious disorders, which, if left untreated, are known to cause learning disabilities. Thanks to the newborn thyroid test (often called the Guthrie test) we now rarely have children who suffer grave long-term effects of thyroid deficiency at birth. You will probably not be aware that your child had this test just after he/she was born. There is a very efficient system for notifying people if the tests are abnormal, so if you were not notified of anything untoward then all the screening tests were normal. Some people worry that because a baby has Down's syndrome perhaps these tests were not carried out. This is not so. They are carried out on all babies.

HOW OFTEN SHOULD TESTS BE CARRIED OUT LATER IN CHILDHOOD AND IN ADULT LIFE?

Different doctors follow different schedules and timings, and as long as they have some system in place all should be well. We assume that all babies will have thyroid function tested as new-borns. Thereafter screening blood tests are necessary from time to time throughout life. Traditionally this involves taking blood from a vein and testing for two or three substances (see below).

The UK Down's Syndrome Medical Interest Group (DSMIG) recommends that if venous blood testing is used this should take place at age 1 and thereafter at least every two years for life. If certain warning signs – which we mention later – show up in the blood tests this will alert your doctor to carry out more frequent checks. Some doctors prefer to do venous tests every year or so.

It is now possible to carry out limited thyroid testing using a few drops of fingerprick blood instead of a larger amount from a vein. DSMIG recommends that if this screening method is used it should be repeated every year. You can find information about how to organise fingerprick testing on www.dsmig.org.uk

WHAT TESTS ARE CARRIED OUT?

There are several tests of thyroid function known collectively as "thyroid function tests" (TFTs). The two most frequently used are:

- i. A test which measures the level of thyroid hormone – thyroxine (often written as T4) – in the blood**

- ii. A test which measures the level of a hormone called thyroid stimulating hormone (TSH)**

TSH is produced by a gland underneath the brain and it 'drives' the thyroid gland to produce thyroxine. If the thyroid gland is not working well the TSH levels may be unduly high because more TSH than usual is being required to 'drive' it. Raised TSH levels can therefore be a warning that the thyroid gland, though currently producing enough thyroxine, may become underactive in the future. TSH levels, however, can be difficult to interpret in children with Down's syndrome. Quite a number of these children seem to produce raised TSH levels in early childhood for no obvious reason and these levels subsequently return to normal. If your child, or an adult, has a raised level of TSH in the blood this does not necessarily mean that there is a shortage of thyroxine. It does mean that your doctor will probably want to check the thyroxine levels more often than otherwise. Sometimes additional tests may be carried out.

iii. A blood test to detect the presence of thyroid antibodies

This is another test which your doctor may use. It is based on one of the causes of thyroid problems. There are several different causes of thyroid underactivity but the most common among people with Down's syndrome is that they themselves start to produce substances called thyroid antibodies. These antibodies harm a person's own thyroid gland and prevent it producing enough thyroxine. This type of disorder, which also occurs in the general population, is called autoimmune disorder.

At least 30% of adults with Down's syndrome produce these antibodies. About half of them continue to produce enough thyroxine for the body's needs and never need thyroid replacement therapy. However, for the other half the presence of these antibodies in early blood tests can be a useful warning sign that thyroid deficiency may develop in the future. If thyroid antibodies are found in a blood test your doctor may want to check thyroxine levels more often than otherwise.

All three tests can be done on a single blood sample if the blood is taken from a vein. When fingerprick testing is used it is usually only possible to do test (ii).

How do we treat thyroid dysfunction in Down's syndrome?

Thyroid dysfunction is much the same in Down's syndrome as in the general population. The same treatment is used. If a person has an underactive thyroid gland this can be treated by giving one or two tablets a day of replacement thyroxine. Some people with Down's syndrome respond more slowly to treatment than other people and advice from a specialist – an endocrinologist – may be needed. The far less common situation of overactivity can usually be treated with medicines to stop the overproduction of thyroid hormones but occasionally an operation may be necessary to remove part of the gland.

Summary

The main problem about disturbed thyroid function in people with Down's syndrome is that it can be very difficult to diagnose on clinical grounds – that is, by a doctor asking questions and carrying out a physical examination - particularly in the early stages. Doctors have to rely on the results of blood test to make sure they are not missing any cases. If thyroid deficiency

is diagnosed the problem then, from a medical point of view, is the same as for anyone in the general population and treatment is both possible and very worthwhile.

ACTION FOR PARENTS AND CARERS

It is very important that parents, carers and other non-medical professionals should be aware of the possibility of thyroid underactivity in a person with Down's syndrome. You may be the first to notice early changes. If a person with Down's syndrome starts to slow down physically and intellectually and put on a lot of weight, then consult a doctor. You should tell the doctor that you are worried about the possibility that the thyroid gland may not be working properly and that you wonder if a blood test should be done. It is more likely, however, that you will have no immediate worries about thyroid problems. In that case just check at some convenient moment with your family doctor, paediatrician, Child Development Clinic or Learning Disability Team what the local policy is about regular thyroid tests. You can then make sure that the person with Down's syndrome with whom you are involved is tested at the appropriate time.

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