BASIC MEDICAL SURVEILLANCE ESSENTIALS

adapted draft recommendations
of the Down's Syndrome Medical Interest Group (DSMIG(UK))

CERVICAL SPINE INSTABILITY (Atlanto-axial instability)

This condition presents as a very small risk throughout life.

There is an increased mobility of the 2 bones directly under the skull (Atlas and Axis), which in extreme cases can become compressed. Compression can occur gradually or rapidly causing dislocation and even paralysis.

Symptoms of cord compression

Neck pain, restricted neck movement, unsteadiness in walking, deterioration in bowel and bladder control. These symptoms warrant urgent medical attention.

Treatment

In extreme cases, the 2 bones may be fused together to prevent further movement.

Screening

There is no screening procedure that can predict those at risk. X-rays are not useful. The annual physical examination should include a check for signs of cord compression.

Precautions

People with Down’s syndrome should not be barred from sporting activities except intensive trampolining, intensive horse riding and high diving.

When undergoing any treatment requiring anaesthesia, a collar should be used to keep the neck stable. Collars should also be used if a person with Down’s syndrome is involved in a road traffic accident, until examination has taken place.

HYPOTHYROIDISM

Hypothyroidism is very common through all ages affecting between 10% and 20% of people with Down's syndrome, although it occurs more with age.

The thyroid gland in the neck fails to secrete enough thyroid hormone to meet the body's need. Thyroid is the hormone that regulates metabolism and affects heart rate and growth.

Symptoms

Weight gain, lethargy, dry skin, puffiness under the eyes, confusion, memory impairment and intolerance of cold.

Untreated hypothyroidism can lead to hallucinations and coma.

Screening and Diagnosis

Newborn babies have a neonatal screen for hypothyroidism using the heel prick (Guthrie) test.

From age 1, and then every 2 years throughout life, a blood test should be taken. A fingerprick blood test can be taken which is less distressing for the patient.

Treatment

Thyroid supplement in tablet form (Thyroxin).

Patient will be monitored regularly to ensure that the dose is correct.

THE HEART

Incidence of heart defects

About 1 in 3 children born with Down's syndrome will have a heart defect that is present from birth (congenital). If no problem is detected in early life, it will not develop later on.

Newborn babies with Down's syndrome are checked for heart defects by the Paediatrician. As it is difficult to pick up any problems so early on, a further examination and electrocardiogram (ECG) plus chest X-ray will be carried out at 6 weeks.

Not every child with a heart defect will need surgery. Many children and adults with Down's syndrome lead full lives despite a heart disorder, and merely return to the Consultant periodically for check ups.

ATRIO-VENTRICULAR SEPTAL DEFECT (AVSD)
This is the most common heart defect in children with Down's syndrome, affecting 1 in 6. It is a hole between the two atria and in some cases between the two ventricles as well. Both partial and complete canal defects can be corrected by surgery in infancy.

VENTRICULAR SEPTAL DEFECT (VSD)
This occurs in about 1 in 10 infants with Down's syndrome. It is a hole between the two ventricles. In some cases the hole is so small it causes no problem and some small holes close themselves. Holes causing problems can be repaired by surgery.

PERSISTANT DUCTUS ARTERIOSUS (PDA)
This occurs in about 1 in 50 children with Down's syndrome. A duct lying above the heart fails to close after birth, allowing excess blood to flow into the lungs. The duct is closed by surgery.

TETRALOGY OF FALLOT
Only about 1 in 100 children with Down's syndrome have this combination of four heart defects, including a large hole between the ventricles and a narrowing in the blood vessel from the heart to the lungs.

Repair involves a complex open-heart operation. Total correction is difficult in an infant, so temporary repair is often carried out until the child is older.

Symptoms of heart defects

Most babies will not have symptoms at birth, they usually occur at a few months of age. In infants the earliest symptoms are difficulty in feeding and poor weight gain. Breathing may be rapid or laboured during feeding, baby may perspire and become tired while eating. Breathing may be shallow at rest. Skin colour may be pale with a blue tinge (cyanosis).

Screening for heart defects

Either – Thorough examination by experienced Paediatricians and echocardiogram (ultrasound study) in the first few days of life.

Or – Thorough examination plus electrocardiogram (ECG – graphs the electrical activity of the heart) and chest x-ray in the first few days of life and again at 6 weeks. Followed by an echocardiogram for those with any abnormalities.

Heart problems in later life (Acquired Cardiac Disease)

As in the general population, people with Down's syndrome can develop heart problems in later life.

The most common is mitral valve prolapse (MVP). The mitral valve has two flaps which swing open to let blood flow from the upper-left chamber of the heart to the lower-left chamber. The flaps then close neatly together. In mitral valve prolapse, the flaps cannot close properly, either because on of the flaps is larger than the other, or because the 'hinges' of the flaps are damaged. As a result, one or both of the flaps closes in the wrong direction and blood can leak back into the upper chamber.

Mitral valve prolapse is also very common in the general population. It is rarely serious and many people (60%) with the condition show no effects or symptoms.

Symptoms of mitral valve prolapse

Irregular heartbeat. May feel palpitations when lying on the left side. Increased heartbeat. Pounding chest. Non-specific chest pain occurring at rest rather than on exertion. Fatigue and weakness even after slight exertion. Migraine headaches

Antibiotic precautions

Precautions should be taken when a child or adult with a heart defect has any operation, particularly where bacteria may be entering the bloodstream. For example, drilling or extraction of teeth may allow bacteria in the mouth to enter the bloodstream, causing an infection in the heart (endocarditis).

Dentists should always be informed of any heart defect, so that antibiotic cover can be arranged if necessary.

HEARING LOSS

Over 50% of children and adults with Down's syndrome will have a significant hearing impairment, which may be mild, moderate or severe.

There are two types of hearing loss.

Conductive hearing loss

This is when sound does not reach the inner ear.

It is caused because children and adults with Down's syndrome have small, narrow ear canals. They are also prone to wax build up, glue ear, ear infections and problems with the formation of bones in the middle ear.

Treatment

Removal of any impacted earwax. Antibiotic and/or decongestant medication to treat ear infections. Hearing aids.

Sensorineural hearing loss

This is caused by problems with the cochlea or the auditory nerve that carries impulses to the brain. Information about incoming sounds is not effectively transmitted to the brain.

This type of hearing loss is usually permanent.

Treatment

More difficult to help. Some people may benefit from a cochlear implant or hearing aids. Input from speech and language therapy to increase understanding and teach of signing and symbols.

Symptoms of hearing loss

Failure to react to sounds, difficulty in understanding, problems with learning, withdrawal, being quiet, needing TV and radio volume louder, not responding when called, increase in emotional and behavioural problems.

Hearing Screening

Newborn babies will be screened as part of the neonatal assessment.
Full audiological assessment between 6 and 10 months of age. This ensures that any degree of hearing loss is identified quickly.
Yearly reviews until child reaches the age of 5.
2 yearly reviews throughout life. More frequently if problems exist.

VISION

Over 90% of children and adults with Down's syndrome have some type of vision problem.

Refractive error and/or squint (strabismus) may be present from an early age.

Refractive error causes short sightedness (myopia), long sightedness (hypermetropia) or astigmatism.

1. Squint (strabismus)

There is abnormal alignment of the eye which may turn into the corner towards the nose, or outwards. This causes double vision but the brain will

ignore the image produced by the lazy eye. The good eye eventually takes over from the lazy eye, which in turn becomes blind.

Treatment
Glasses, which will reduce the squint, sometimes by patching the good eye. Corrective surgery is possible.

2. Short sightedness (myopia)

Objects that are near are clearly seen but those in the distance are out of focus and blurred.

Symptoms
Eyestrain, headaches, deterioration in day-to-day activities.

Treatment
Glasses or contact lenses. Check up every 1 – 2 years.

3. Long sightedness (hypermetropia)

Difficulty in seeing objects that are close, but far vision can also be affected.

Symptoms
Occasionally eyestrain and/or headache.

Treatment
Glasses or contact lenses. Check up every 1 – 2 years.

4. Astigmatism

A condition where the surface of the cornea is not smoothly spherical.

Symptoms
Blurred vision

Treatment
A particular type of glasses.

5. Cataracts

Cloudiness (opacity) of the lens in one or both eyes.

Symptoms
Gradual loss of vision, cloudy lens.

Treatment
Good lighting, glasses if the case is not too severe. In severe cases the lens in the eye can be removed. Vision can be corrected with thick glasses, contact lenses or artificial lens implants.

6. Keratoconus

The central part of the eye protrudes and has a conical shape. This is not particularly noticeable when facing a person but is obvious in profile. It can affect one or both eyes.

Symptoms
Difficulty in seeing, eyestrain, headaches, deterioration in day-to-day activities.

Treatment

Eye lubricants, padding and pain relief. In chronic cases glasses may be helpful. Contact lenses can be useful if the person can tolerate them. Surgery may also be an option. If scarring is present corneal transplant may be needed.

Screening for vision problems

Newborn babies are examined for **congenital cataract** and the Paediatrician monitors visual behaviour.

During the second year of life a thorough opthalmological review is carried out. Many children with Down's syndrome will have some visual defect and will be kept under close review.

Those with no obvious defect will have a further opthalmological review at around 4 years.

After the age of 4 vision should be checked at least every 2 years throughout life.

Regular eye examination will identify any changes in vision. In addition, it will monitor the health of the eye and aid early identification of cataracts or other problems.