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General Paediatric Issues

This information has been compiled from various resources including the Max Appeal Consensus Document on the Diagnosis and Management of 22q11.2 deletion. The main author of the chapter is Dr Alex Habel formerly Paediatric Consultant at Great Ormond Street Hospital. References and sources of evidence are freely available using the contact details above.

The purpose of this Information Leaflet is to inform parents, carers and the general public about the many problems associated with 22q11DS that need specialist care and other symptoms in childhood that may be associated with the syndrome that can be dealt with by a general paediatrician, who should refer problems to other specialists where necessary.

Breathing:

- Wheezing may be noticed from birth and this could be caused by parts of the throat being abnormal in structure or 'floppy'.
 - A soft larynx (laryngo-malacia) or soft trachea (trachea-malacia) may collapse as the baby tries to suck.
 - A similar weakness may occur in the large airways in the lungs (broncho-malacia).
- A web of tissue may partially block the vocal cords in the larynx (laryngeal web) but surgery is not always needed unless breathing is difficult.
- Sometimes a large blood vessel makes a ring around the wind pipe (trachea) and (gullet) oesophagus and this can constrict the passage of air and food. This is called a 'vascular ring'. In some cases the symptoms are not severe but in others urgent surgery may be needed.
- Palate and swallowing problems are frequently noted and some babies and young children can't control foods to pass into the stomach, so sometimes the food inadvertently enters the lungs ie 'goes down the wrong way', this is known as 'aspiration'. Babies may choke or cough during feeding, though many become accustomed to the sensation so the 'gag' reflex is lost. Such 'silent' aspiration might only be noticed because of a wheeze or

frequent chest infections. There are tests for this but feeding via a tube may be needed for a period of time.

Feeding:

Many children with 22q11DS find it difficult to feed until they are about 3 years old and weight gain can be problematical and can be caused by many problems:

- Weak muscles, a sub-mucous cleft palate and other defects with the heart and breathing may contribute to difficulties such as gastro-oesophageal reflux (GORD) when acid from the stomach enters the oesophagus can make swallowing painful (severe heartburn).
- Occasionally forceful vomiting might indicate a blockage due to the intestine being twisted. This is known as a malrotation of the bowel and can develop during pregnancy. While that may not cause any problems if the gut twists and causes a complete blockage of the digestive system (a volvulus) surgery will be needed.
- Feeds may need to be enriched with extra nutrients and tube feeding either by a nasogastric tube or by a tube directly into the stomach (gastromy or G tube, PEG or 'button') may be necessary. Some parents find these options difficult to come to terms with but sometimes the need for weight gain is paramount.
- Feeding difficulties may be linked to problems with breathing, a hidden cleft palate, dysfunction or heart problems.

Constipation:

This can result from poor muscle tone so things aren't moved through the intestine in the normal time, this can be made worse if high fibre foods are refused or to drink adequate amounts of liquids. The team who give advice on any feeding difficulties should have plenty of suggestions to help overcome constipation although it may be necessary to use laxatives on a regular basis.

Other problems need to be excluded such as an under active thyroid gland (hypothyroidism) or a structural blockage of the gut such as Hirschsprung's disease, or an anus in the wrong position or a narrowing of the anus (anal stenosis).

Growth:

This can be delayed in the pre-school years either because of problems with feeding and digestion or problems with thyroid or growth hormones. Most children catch up and when adult are normal in height but they may become overweight during their teens. Other causes of slow growth such as coeliac disease should be ruled out.

Problems with bones and muscles:

- Occasionally children with 22q11DS may be born with extra fingers or toes, which are removed early on.
- Sometimes babies are born with club feet (talipes) which can often be corrected with specialist splints, physiotherapy or boots.
- Sometimes one shoulder higher is than the other (Sprengel's shoulder), which is resolved early on by an operation.
- Scoliosis (a sideways curve of the spine) is the most common problem and children should be checked for this as they grow. In infancy scoliosis may be caused by when a bone or

bones in the spine are misshapen, in adolescence it may develop because of low muscle tone.

- Ligaments may be weak allowing more mobility in joints than is normal. Teenagers might suffer with dislocated knee caps. Flat feet and tight heel cords (the Achilles' tendon) are also fairly common but it's not known if these are responsible for the leg pains that are frequently reported by parents.

Sleep disturbance:

- Parents frequently tell us that their child wakes during the night and this can become a serious cause of tension in the household!
- Sleep may be disturbed by 'growing pains', leg pains or by 'restless legs'. Parents tell us that leg massages, warm baths before bed and applying ibuprofen type creams can help sometimes.
- Obstructive sleep apnoea (interrupted breathing) may occur occasionally after VPI and should be investigated.

Neurological aspects:

- 22q11DS children often show clumsiness and have trouble controlling both large muscles used in walking and running and smaller muscles like fingers, which make writing and using a keyboard more difficult to use, this dyspraxia will not get progressively worse. Exercises led by a therapist can help.
- Seizures may be due to low calcium levels (hypocalcaemia) which is a common endocrine (gland) problem in 22q deletion, rather than epilepsy. Any seizure should be investigated straight away.
- Occasionally badly formed vertebrae can cause spinal cord compression or trapped nerves, and investigations for this should be carried out urgently.

Kidney problems:

Although there may be some structural abnormalities these rarely cause any problems. Some children will have kidney anomalies, including a missing kidney, which can be detected by an ultrasound scan. Some doctors feel that advice should be sought where a child has problems with infections of the urinary tract, bedwetting or urinary frequency.

Genital problems:

In some boys, the opening of their penis may be on the underside rather than the end, this can also be associated with undescended testes and should be referred to a specialist.

Hearing problems:

Deafness resulting from 'glue ear' is common but does not usually cause long term problems. Some children may have a mild deafness due to poor development of the nerves and structure of the ear.

Problems with vision:

Long-sightedness might mean that glasses are needed to help with reading and to help judgment of distance, direction and location of objects. Conjunctivitis (pink eye) infections are also more common than with other children.

Dental care:

Tooth enamel is often poor in quality leading to cavities. Sometimes teeth are missing or are badly formed. Great attention should be paid to dental hygiene as soon as baby teeth come through. A referral to a dentist or hygienist skilled in handling children with special needs is helpful.

Autoimmune Diseases

An autoimmune disease is one where a person's immune system fights its own body and it is now becoming more frequently diagnosed in people with 22q deletion.:

- Juvenile idiopathic arthritis: pains in joints of arms legs and fingers should be investigated so that correct treatment can be given. A blood test is usually the first port of call.
- Thyroid disease: either an over active (hyperthyroidism) or an under (hypothyroidism) active thyroid gland.
- Raynaud's phenomenon: fingers go white in cold weather (due to poor circulation)
- Vitiligo: areas of skin lose the normal colouring/pigment (white patches or blotches)
- Coeliac disease: digestive condition where a person has an adverse reaction to gluten
- Autoimmune blood disorders: including low platelets, low levels of one type of white blood cell (neutrophils) and an anaemia resulting from the body destroying red cells.

Heart Problems:

Cardiology - if the child does not have a known cardiac defect then an evaluation by a cardiologist is important. The chances of finding a previously undiagnosed defect are very low; however a chest x-ray, an electrocardiogram and an echocardiogram may be carried out.

Developmental Delay:

Child Development and Psychology - many children with a 22q11.2 deletion have developmental delays and benefit from early years intervention. It is important therefore that regular monitoring is carried out in order to identify any problems and provide appropriate support. There are other psychological problems that may occur in a small number of patients and further information about these is available and research in this area is on-going.

Palate cleft or weakness:

Cleft Palate Team - all children should be seen by a plastic surgeon or ENT specialist who specialises cleft palates and VPI (velopharyngeal insufficiency), preferably at one of the recognised centres of excellence where the surgeon is familiar with the 22q11.2 deletion. Input from a speech therapist is also essential in order to assess speech and language and (in a very young child) feeding. Monitoring of the face, jaw and teeth by a growth specialist and a dentist may also be useful.

Endocrinology:

some new-born babies will have problems with low calcium levels and require supplements. This is often dealt with by a paediatrician or a cardiologist. If this problem continues then a hormone specialist may be required. Also some children with have short stature and growth problems which it may be possible to overcome with the assistance of hormone treatment.

Hearing loss, tonsils and throat problems:

ENT and Audiology - this is often dealt with by the cleft palate team. Some children will have repeated ear infections (glue ear is not uncommon) or suffer from problems with noisy breathing and advice from an ear, nose and throat doctor would be beneficial. Also some children suffer with hearing loss and it is helpful if a hearing test or audiogram is carried out as other anomalies can cause hearing loss.

Feeding problems:

These are often identified in the new-born period and the assistance of a paediatrician who specialises in this area is very useful as these problems can continue for some time and impact upon other areas of speech and development.

Genetics:

The geneticist is likely to have an overview of the diagnosis and will be able to advise on issues such as the chances of recurrence in future pregnancies, diagnosis during pregnancy and whether the condition is hereditary or a "new mutation". Genetic counselling is also important for those who have the deletion who wish to plan a family.

Immunology:

Every patient should have their immune system assessed at least once. Many new-born babies will have problems with infection and care should be taken with vaccines. Many children will outgrow this problem by their first birthday but others will continue to have trouble until later childhood and occasionally adulthood.

Kidneys:

Some children will have kidney anomalies, including a missing kidney, which can be detected by an ultrasound scan. A renal doctor or urologist should also be referred to where a child has problems with infections of the urinary tract, bedwetting or urinary frequency.

Neurology:

Occasionally children will have symptoms such as a seizure disorder or problems with balance. Advice from a neurology specialist may be helpful in some cases if any brain anomalies are suspected.

Other:

Other findings commonly noted include: constipation, leg pains and anomalies of the carotid arteries (blood vessels in the neck which lead to the brain). Less frequently there are problems noted with: bony abnormalities, such as extra fingers, extra ribs, and a missing bone in the forearm, other problems of the vertebrae which can lead to scoliosis, spina bifida, juvenile rheumatoid arthritis, eye problems, premature fusion of the soft spots or fontanel (craniosynotosis), and emotional or psychological problems. Specialist advice should be sought in any of these areas if a problem is suspected.