

Factsheet - CG05

The conditions associated with CHARGE

CHARGE is a rare condition that can affect different parts of the body. The most common problems are with the ears, eyes, heart, nasal passages, genitals, and with growth - although the condition, and its severity, does vary from person to person.

It is important to emphasise that not all these features are seen in every person with the condition, nor are these problems always severe.

It is believed that CHARGE is a genetic condition and very recently a mutation in the gene CHD7 has been found in approximately 50% of some children with CHARGE. Research into this is ongoing, and criteria for diagnosis are constantly under review.

How is a child diagnosed?

Sometimes children are diagnosed as having CHARGE quite quickly. In other cases it may take some time for medical professionals to put the different pieces of information together and make a firm diagnosis.

A child can be diagnosed as having CHARGE if he or she has three of the major characteristics listed below. Alternatively, if a child has two of the major characteristics, and three of the minor characteristics then they can also be diagnosed as having CHARGE. All of these medical terms are explained in the next section.

Major characteristics:

1. Coloboma
2. Choanal atresia
3. Characteristic ear anomalies
4. Cranial nerve dysfunction (facial palsy, vestibular dysfunction and swallowing difficulties);

Minor characteristics

- heart defect
- orofacial cleft
- genital hypoplasia
- growth deficiency
- a characteristic facial appearance
- Upper body hypotonia.

What do these terms mean?

Coloboma

This is an eye deformity where part of the eye has failed to develop properly and is missing. There is a notch-like defect (commonly described as a 'keyhole') in any part of the eye. It is present at birth and is non-progressive. Coloboma of the *iris* (at the front of the eye) may limit the person's ability to adjust to bright light, as the pupil is often deformed; coloboma of the *retina* (at the back of the eye) will create a blank area in the person's visual field. Sometimes the eye may be small (microphthalmia) and in rare cases missing altogether (anophthalmia).

Choanal atresia

This is a blockage of the passages at the back of the nose. The blockage may be on one or both sides, and may be formed of a membrane of skin or of bone. Again, surgery is often necessary immediately after birth to open these passages.

Ear anomalies

A child's hearing can also be affected. The external ear may be large, small or of an unusual cup shape. The middle ear may have bone malformations or chronic glue-ear infection. Also, the inner ear can be affected, resulting in a permanent hearing loss.

Hearing loss in people with CHARGE can be mixed and may be a combination of:

- a conductive loss in the middle-ear (sound isn't conducted effectively through the middle ear) combined with,
- a sensori-neural loss because of damage to the cochlea
- problems processing auditory information because of a missing or underdeveloped auditory nerve.

It is also possible that the semi-circular canals in the ear will be malformed or absent (in the inner ear) which means that a child's balance will be affected.

Cranial nerve anomalies

Cranial nerve problems are thought to be very common in children with CHARGE. In particular, problems with:

- the first cranial nerve which leads to a loss of the sense of smell. Although not an acute medical problem this may affect feeding, and may close off one more sense channel to a child who is already missing out on a lot of other sensory information.
- the seventh cranial nerve which causes facial palsy – a type of paralysis that can leave the face looking flat and expressionless, or drooping on one side.
- the eighth cranial nerve which causes sensori-neural hearing loss, and balance problems
- the ninth and tenth cranial nerves which cause swallowing problems. Children with facial palsy are more likely to have structural differences in the region of the larynx and pharynx in the throat. This may lead to swallowing problems and aspiration.

Heart defects

Heart defects may be of various kinds - from life threatening to minor. Sometimes these problems can resolve themselves over time, but often emergency surgery is needed very soon after a child is born.

Cleft lip and palate

It is common for children with CHARGE to have a cleft lip and palate (Orofacial cleft). This may make it harder to diagnose atresia of the choanae (a blockage of the passages at the back of the nose) if it is present. Surgery for a cleft lip may be required to repair severe clefts. Children with a cleft palate may also have problems with ear infections, feeding, and speech even after surgical corrections.

Genital differences

The incomplete or under-development of the external genitals, is very common in boys. Common problems are undescended testicles and/or small penis.

Some girls with CHARGE have small labia minora (inner vaginal lips).

Both males and females with CHARGE often experience hormonal problems. Evidence suggests that both sexes will experience difficulties with puberty, boys more so than girls.

Failure to adequately replace sex hormones in puberty may lead to brittle bones (osteoporosis) in adult life.

Growth deficiency and developmental delay

This means that the child grows and develops more slowly than expected.

There are many factors that can lead to a child growing more slowly - including severe feeding difficulties, breathing problems, chest infections and multiple surgical procedures with repeated and prolonged hospitalisation. Most of the people identified as having CHARGE are short (below the third percentile)

It is now recognised that many young people with CHARGE will not go through puberty without the support of hormone treatment.

Developmental delay means that a child needs more time to reach milestones such as sitting and standing and developing speech.

Facial Features

Facial asymmetry (the two sides of the face being different) may be present, even in the absence of facial palsy. Other characteristic facial features can include a square face with a broad prominent forehead, ptosis (droopy eyelids), a flat mid-face and small chin.

Upper body hypotonia.

Low muscle tone or very floppy muscles in the upper body.

Other conditions associated with CHARGE

As well as these main features of CHARGE there are a large number of other features.

These can get better or worse over time, and the impact they may have can also vary from child to child. The complexity of the condition means that each feature can affect another so that no two people with CHARGE are likely to be affected in the same way.

Again, it is important to emphasise that children with CHARGE may have some of these conditions – and these problems are not always severe.

Abdominal pain

Some teenagers with CHARGE get recurrent abdominal pain, although the reasons for this are unclear.

Abnormal Gait

Before they can walk, children with CHARGE often move by lying flat on their backs and scooting headfirst or by five-point crawling (i.e. two arms, two legs and forehead) backwards or forwards. Once walking, people with CHARGE often show the characteristic pattern of feet rather wide apart, knees slightly bent, feet being planted firmly on the ground, and the upper body and head slightly rolling from side to side. This characteristic gait is thought to result from a combination of balance problems, visual impairment, and poor muscle tone.

(See also: **Hypotonia**)

Chromosome Abnormalities

The CHD7 gene belongs to a well-known family of 'regulation genes' that function as a kind of relay panel in switching on and off other genes. A defect in this process causes problems in the early development of the embryo, which explains why so many organ systems are involved in CHARGE.

Corpus Callosum Agenesis

This is a rare neurological condition in which the corpus callosum - the bridge of white matter that joins the two halves of the brain - fails to form properly.

(See also: **Temperature Dysregulation**)

Delayed Puberty

This is very common in both boys and girls. (See also: **Pituitary Anomalies**)

Dental Anomalies

Dental development may be delayed with late cutting of teeth, which may be malformed and/or grow out of position and need surgery. There is also evidence that some children with CHARGE experience severe dental decay. Possible contributory factors to consider are high sugar content in medicines commonly taken by people with CHARGE, and acids caused by gastro-oesophageal reflux. (See also: **Gastro-Oesophageal Reflux**)

DiGeorge Sequence

This is a congenital absence of the thymus and parathyroid glands, along with heart defects. This anomaly leads to increased risk of infections, low blood calcium, and delayed development. There are other features which overlap with CHARGE such as

low set ears, **cleft palate and heart problems**. DiGeorge sequence is usually secondary to a problem affecting chromosome 22.

Epilepsy

Whilst rare there is growing evidence of epilepsy with some people with CHARGE.

Facial Palsy

This is a type of paralysis that can leave the face looking flat and expressionless, as the facial nerve that supplies the facial muscles does not work properly. This usually affects just one side of the face. This distinctive feature may become more or less apparent in people with CHARGE as they grow and develop. It has implications for communication because of the way that facial expressions and speech may be affected. (See also: **Cranial Nerve Anomalies**)

Gastro-Oesophageal Reflux

This is when the gastric contents of the stomach come back up into the gullet (oesophagus). This can be painful and also cause damage to the oesophagus because of acidity. It is quite common and may cause failure to thrive due to poor nutritional intake. Medical or surgical management may be required. (See also: **Oesophageal Atresia** and **Dental Anomalies**)

Hypocalcaemia

The inability of the body to absorb calcium from food and drink is believed to be a problem occasionally for some people with CHARGE, particularly if they show overlap with **DiGeorge Sequence** (which does have a chromosomal abnormality). There may be other problems with digestion and absorption of nutrition.

Hypoglycaemia

Episodes of low blood sugar are reported in children with CHARGE, and may be more common in those with feeding tubes (“dumping”). Periods of irritability or of drowsiness with sweating and pallor may suggest the need for food or drink to increase blood sugar levels.

Hypotonia

Low muscle tone or very floppy muscles are very characteristic of children with CHARGE and sometimes the problems associated with this persist into adulthood. It is thought that multi-sensory impairments, balance problems, health problems, persistent and repeated hospitalisation and surgery, breathing difficulties, and sensory defensiveness all play a part in this. (See also: **Abnormal Gait**)

Immune System Deficiency

This is seen in children who show an overlap with **DiGeorge Sequence**. They have an increased risk of infection.

Imperforate Anus

The anus is closed over at birth and needs to be opened surgically. Sometimes only a thin membrane needs to be opened. More often the blind end of the large bowel needs to be connected to the skin on the belly as a colostomy. Later the end of the bowel is put down through an artificial opening created where the anus ought to be.

Inguinal Hernia

This is also known as a “rupture” and appears as a lump in the groin. It is actually a small loop of intestine sticking out of a small hole connecting the inside of the

abdomen to the groin. This can result in obstruction or infection so preventive surgery is necessary.

(See also: **Omphalocele** and **Umbilical Hernia**)

Micrognathia

This means a small lower jaw that may contribute to feeding difficulties, and may be another factor to consider in the development of speech.

Mondini Defect

This is an abnormal opening from the semi-circular canal into the middle ear. It is a contributory cause of balance problems and can be surgically treated.

Nipple Anomalies

In rare cases extra nipples, or under-developed nipples, have been reported in people with CHARGE.

Oesophageal Atresia

The oesophagus, or gullet, may not be connected to the stomach and may just have a blind ending. Or it can be abnormally narrow or floppy leading to a range of feeding problems including reflux.

(See also: **Gastro-Oesophageal Reflux** and **Tracheo-Oesophageal Fistula**)

Omphalocele

This is a protrusion of variable amounts of the intestines, stomach, and the liver through the abdominal wall and requires immediate surgery.

(See also: **Inguinal Hernia** and **Umbilical Hernia**)

Otitis Media

Middle ear infections. These can occur when fluid accumulates in the middle ear behind the ear drum. If the fluid is infected the child will feel pain and there will be hearing loss that is usually temporary, but can be fluctuating or persistent. In CHARGE otitis media often lasts into the teenage years and requires close supervision. Repeated ear infections can also lead to permanent damage to hearing (so called "glue ear"), and insertion of grommets may be helpful.

Pituitary Anomalies

The pituitary is a gland at the base of the brain which produces several important hormones that help control growth, stress, metabolism as well as genital and pubertal development. The pituitary gland does not function properly in some children with CHARGE, and this can result in deficiencies in growth hormone and in the sex hormones which induce puberty. If left untreated these children will be short in stature and will not develop secondary sex characteristics. Hormone therapy has been used for these problems. (See also: **Delayed puberty**)

Renal Anomalies

There may be problems with kidney development or function, or even a missing kidney. Kidney reflux (i.e. leakage of urine from the bladder back into the ureter) is another feature which will not show up on ultrasound investigation and will need to be checked by having a Micturating Cystourethrogram (MCUG) and then carefully monitored.

If this kind of reflux occurs but is not detected for a considerable period of time (and there may be no obvious symptoms during this time) irreparable damage to one or both kidneys can occur.

Scoliosis

Progressive curvature of the spine mainly due to low muscle tone. Other contributory factors are balance problems, and visual impairment. These lead to poor use of the body and poor posture. This is reported to be very common in older children and adults with CHARGE and needs to be addressed as early as possible.

Semi-Circular Canal Anomalies

The semi-circular canals in the ears are the receptors of the balance sense. Balance problems due to absence or underdevelopment of the semi circular canals are thought to be very common in people with CHARGE. Severe hypotonia, unusual movement patterns in infants, and characteristic gait in older people with CHARGE may all indicate problems with the balance sense. (See also: **Cranial Nerve Anomalies**)

Sensory integration

Because people with CHARGE often have combined sight and hearing difficulties, many find receiving, processing and 'putting together' information from their environment very challenging. They may prefer to avoid objects or materials that provide unstable sensory information - such as soft toys that change and don't keep their shape.

Skeletal Anomalies

A small number of people with CHARGE are born with skeletal problems such as hands and feet which are clefted and may need corrective surgery. They may also have a very low set thumb or an extra finger or toe.

Sleep Apnoea

Disturbed sleep patterns due to children stopping breathing when they are asleep, causing them to wake up and then start breathing again.

Swallowing abnormalities

Some problems with co-ordinating breathing and swallowing can be attributed to damage to the ninth and tenth cranial nerves. Other contributory factors may be breathing difficulties, hypotonia, weak or malformed oesophagus and trachea, and sensory defensiveness.

(See also: **Cranial Nerve Anomalies**)

Tear ducts missing or narrow

The tear ducts may be unable to drain fluids from the eyes because of this problem and very sticky eyes will result.

Temperature Dysregulation

There are several possible reasons why people with CHARGE may be unable to regulate their body temperature consistently and effectively. One possibility is that the hypothalamus (the part of the brain that controls body temperature) may not be functioning correctly.

Another possibility may be abnormalities of the **Corpus Callosum** which are associated with episodes of severe hypothermia.

Finally, there may be problems with the part of the nervous system (autonomic) which regulates the functions of some of the internal organs, leading to wide swings in heart rates, temperature, blood pressure, and also high pain threshold.

(See also: **Corpus Callosum Agenesis**)

Tracheoeseophageal fistula

The oesophagus, or gullet, may not be connected to the stomach and may just have a blind ending. Or it can be abnormally narrow or floppy leading to a range of feeding problems including reflux. This is when the gastric contents of the stomach come back up into the food pipe (oesophagus).

Because of the complexity of this condition, children born with CHARGE may need emergency surgery and may experience long periods in hospital in their early years. The need for surgery and other medical intervention usually decreases over the years and the condition of some children can improve far beyond initial expectations.

(See also: **Oesophageal Atresia**) **Umbilical Hernia**

(See also: **Inguinal Hernia** and **Omphalocele**)

Webbed Neck

Webbed neck (shortened neck) and sloping shoulders are sometimes seen in people with CHARGE.

Where to go for help

Sense specialists have many years experience of providing assessments of babies and children affected by conditions that cause dual sensory impairment - including CHARGE. They also support families and offer consultancy services to schools. If you would like to find out more about CHARGE, or the support that is available in your area, please contact Sense.

See also Sense's website – www.sense.org.uk – which contains further information about CHARGE.

Sense also helps families by supporting a number of groups for families – including the CHARGE Family Support Group. This enables parents, children and others affected by CHARGE to come together, share information and offer mutual support.

The CHARGE Family Support Group: <http://www.chargesyndrome.org.uk/>

Or telephone Simon Howard on 020 8265 3604

Where can I go for help?

If you:

- would like to find out more about deafblindness or the services for deafblind people in your area
- require information in alternative formats including braille, large print, audio or disk - or would like this factsheet to be translated into your first language - please contact Sense's Information Team.

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