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Factsheet 7

Hearing and audiological assessment in CHARGE syndrome

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The CHARGE ear

CHARGE syndrome is associated with cranio-facial abnormalities and can affect all parts of the auditory pathway from the outer ear to the auditory nerves.

The pinna or outer ear may be an unusual shape with small lobes and a malformed helix (Lanson *et al.* 2007).

Middle ear malformations may be present – for example, the ossicles or small bones which transmit sound from the eardrum to the inner ear may be incomplete or fused (Arndt *et al.* 2009; Collins and Buchman, 2002).

Patients may suffer from recurrent ear infections or 'glue ear' resulting from poor ventilation of the middle ear caused by eustachian tube dysfunction (Lanson *et al.* 2007).

Children with CHARGE syndrome may present with congenital inner ear malformations which affect both the hearing and balance systems. The semi-circular canals, the balance organ, may be completely absent and the inner ear or cochlea, may be incomplete (cochlear dysplasia).

Some children with CHARGE may have a cochlea which consists of a single cavity, unlike the usual snail shape with several turns, which is called a Mondini deformity.

Additionally the auditory (cochlear) nerve which transmits auditory information from the cochlea to the brain may be thin ('hypoplastic') or absent entirely ('cochlear nerve aplasia'). (Collins and Buchman, 2002).

CT/MRI scans

Children who have been diagnosed with CHARGE syndrome, or who are undergoing diagnostic assessment where hearing loss is suspected, should undergo Computed Tomography (CT) scanning and, where possible, Magnetic Resonance Imaging (MRI) to assess the status of the inner and middle ears.

MRI assessment is particularly important in cases of suspected profound hearing loss where there may be a concern that the auditory nerve is hypoplastic or absent (Arndt *et al.* 2009; Holcomb *et al.* 2012).

Absence of semi-circular canals is considered a key criteria for the differential diagnosis of CHARGE from other cranio-facial abnormalities, along with cochlear dysplasia (Arndt *et al.* 2009).

Audiometric testing

Children may have a moderate conductive loss which arises from the malformation of the middle ear, a sensori-neural loss arising from the malformation of the cochlea and/or its associated nerve or a combination of the two – resulting in a mixed hearing loss. Thus, hearing loss may range from moderate to profound and careful assessment is necessary (Arndt *et al.* 2010; Holcomb *et al.* 2012).

As with all audiometric testing, it should be adapted to the age and special profile of each patient, taking into account any other sensory difficulties, developmental age and motor difficulties. It may require a series of sessions over a period of time to obtain accurate and reliable results which should be considered in conjunction with observations from carers.

Behavioural testing may not be the most appropriate. It can be a very challenging part of the overall audiological assessment of children with CHARGE and so reliance may be placed on objective measures where the child does not have to give a response.

This is often carried out under general anaesthetic. In the first instance, this will be Auditory Brainstem Response (ABR) testing. It is always preferable however, to confirm these results with behavioural testing where possible as there is some evidence that an abnormal or missing ABR can occur where neurological damage is present (Stein and Kraus, 1995).

Amplification options

Results from ABR tests can provide information for the preliminary fitting of hearing aids where appropriate. If cochlear implantation is being considered as an option, further assessment using eABR (electrically evoked ABR) may be used, particularly where there is concern regarding the condition of the auditory nerve (Warren *et al.* 2010).

Where the hearing loss is found to be primarily conductive in origin arising from malformation of the middle ear, or is a moderate to severe mixed loss, hearing aid fitting will depend on the nature of the cranio-facial abnormalities.

For example, if the pinna is severely deformed, or there are chronic middle ear problems, this may make the use of standard air conduction hearing aids difficult. In this case, bone conduction hearing aids may be tried where the sound is transmitted directly to the inner ear via the bones of the skull. Increasingly a Bone Anchored Hearing Aid (BAHA) is offered, usually with a trial with a softband version first followed by surgical implantation of a device where appropriate (McDermott *et al.* 2009).

For children with a severe to profound loss, a Cochlear Implant (CI) may be an appropriate option but only after very careful assessment of the condition of the cochlea and the auditory nerve (Arndt *et al.* 2009; Bamiou *et al.* 2001; MacArdle *et al.* 2002)..

It is a feature of CHARGE syndrome that more than one element of the hearing pathway is often affected, making audiological management complex.

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