



Factsheet 9

Clinical assessment of vision for children with CHARGE

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The 'C' of CHARGE is for Coloboma

Coloboma is from a Greek word meaning 'not fully formed'. An eye forms as a bubble from the brain. The blood vessels of the eye enter the centre by forming a dip in the lower part of the eye. The two sides of this groove seal, starting in the middle. Eventually, at the front end, the future iris closes, turning an upside down horseshoe into a round pupil. If the groove does not close at the other end, the retina has a defect that may involve the macula and the optic nerve.

At the most severe, the eye is not formed at all – anophthalmos. At the most mild, the eye is of normal size but the iris has a keyhole shaped pupil, has good vision and slightly increased sensitivity to bright light. In CHARGE the right and left eye are frequently affected to a different degree.

Acuity

Assessment starts with acuity – a measure of the ability to see detail in the distance and near. This is tested in older children by reading letters on an eye chart. Younger children can match letters or identify pictures and infants can be tested with pictures or stripy cards using the preferential looking technique. In CHARGE, acuity ranges from normal to no light perception. It is impaired due to a reduced number of light sensitive cells in the retina. Generally, the smaller the eye the fewer the light sensitive cells there are. Acuity is severely affected if the macula doesn't form.

Recording near vision:

- Record the near vision in good lighting, with the light coming over the child's shoulder.



- Use high contrast print of standard sizes on non-reflective paper.
- Measure the distance the child is most comfortable holding the book.
- Glasses are of limited benefit and are challenging to prescribe and to fit, as the eye is distorted in shape and ears may be abnormal. An analogy is that a photograph taken with a basic camera will not have as much detail as one taken with high megapixels, whatever lens the camera has, or how much it is magnified.

Delayed Vision Maturation (DVM)

Children with CHARGE often have a form of DVM, particularly if they suffer from multiple disabilities. That is, their use of vision may dramatically improve during the first year of life after initially being feared



to be blind. It is difficult to predict an infant's vision potential from observation of their behaviour, or assessment of the retinal and optic nerve defect.

It can be helpful for a child to be referred to a specialist ophthalmologist for pattern visual evoked potential testing (VEP). This assesses the signal quality input into the brain from the eye stimulated with various check patterns on a screen.

Dazzle

Acuity varies with lighting due to 'ocular straylight' (loss of retinal image contrast as a result of intraocular light scatter). If there is an iris coloboma, as more light enters the eye, less of it is absorbed by the retinal pigment epithelium. In this case, the white retinal coloboma acts like a mirror inside the eye.

To assess this, the acuity test can be repeated whilst torchlight is shone at the child's eye from the side. Alternatively, simply observe behaviour in various lighting conditions.

Transmitted light can be reduced by the use of sunglasses and a hat with a long brim with a matt dark fabric interior.

Visual field

The portion of the retina and underlying choroid that did not form is inferonasal to the optic nerve. The defect in the field of vision is usually a generalised constriction. Often this results in difficulty on steps and stairs. Additional mobility problems are caused by photophobia and lack of contrast between surfaces, such as steps and pavement edges.

Visual field can be assessed by a two observer technique, with an assistant silently bringing a bright toy into a child's field of vision from each side at a low and then a high position. Whilst this is done, the observer is making eye contact with the child and encouraging them to look straight ahead. As the toy approaches, the child will glance in its direction or even turn their head.

Corneal health

Children with CHARGE often have weakness of a facial nerve that results in reduced blink, reduced eyelid closure and tears spilling down the cheek. The cornea may become dry and develop scarring with loss of vision and blinding ulceration. The signs and symptoms are redness of the eye, pain and loss of the bright light reflex on the cornea. If the white of the eye is red, check that the eye looks the same colour as the other eye and that the cornea is not duller.

Retinal detachment

There is a thin membrane, where the retina and choroid should have developed, that may spontaneously, or after a blow to the eye, develop a tear that results in detachment of the retina.

To try to prevent this blinding complication, this area can be sealed by laser treatment to the edges of the retina. However, this treatment has a risk that needs to be weighed against potential benefit.

Monitoring

Acuity should be regularly assessed to provide information for family and educators. Checking each eye separately once every week can detect early retinal detachment in a child who cannot say they have a new problem. By simply covering one eye, and then asking the child to do a visual task that they can normally perform with the uncovered eye, carers can be alerted to any marked change.

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GLOSSARY

Choroid: a thin vascular layer that supplies blood to the retina and conducts arteries and nerves to other structures in the eye.

Inferonasal: below the eyes midline and towards the nose.

Intraocular light scatter: light scatters as it enters the eye adversely affecting the focal point on the retina.

Retinal pigment epithelium: the pigmented cell layer just outside the neurosensory retina that nourishes retinal visual cells, and is firmly attached to the underlying choroid and overlying retinal visual cells.