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April 2002

Reg Charity No 1058461
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EYE ASPECTS OF CRANIOFACIAL CONDITIONS

Introduction

The various Craniofacial syndromes are a group of disorders characterised by premature fusion of one or more of the suture lines, where the bones of the skull vault and the facial skeleton join. They embrace other features (such as the hand malformations seen in Apert’s syndrome) but it is the effect on the skull and those structures housed within the skull that are of paramount importance.

Probably the greatest fear for the parents of affected children is that the eyes or visual pathways will become damaged, thus compromising their child’s eyesight. This article considers the threat to vision in the craniofacial syndromes and reviews some of the treatment options available.

Ophalmic features

- **Proptosis/ Exophthalmos** (forward protrusion of eyes)
  Prominent eyes are a part of the obvious and distinctive facial features. It results from premature closure of the bony sutures causing a shallow orbit and reduced orbital space. The orbit cannot, therefore, accommodate all the structures within it, and, as a consequence, the eyes are displaced forwards. Very rarely the situation can be so extreme that a complete dislocation of the globe beyond the eyelids is caused.

- **Lagophthalmos** is the inability to shut the eyes completely. The forward protrusion of the globe makes it difficult for the upper and lower lids to close completely and this can lead to exposure keratopathy, when the cornea becomes dry because of the poor lid closure.

- **Squint surgery** to correct the abnormal alignment of eyes, as part of the treatment for ‘lazy eye’. The surgery may be undertaken with the intention of relieving double vision or enabling the developing of stereoscopic vision. In some cases it is really just for the improved cosmetics, though this is an important goal for many of the children.

Summary

Children with craniofacial anomalies will often require little input from Ophthalmologists other than monitoring vision and ensuring that parents and teachers are aware of the situation. These conditions do, however, carry the potential for severe and permanent visual loss, as well as the cosmetic impact of the prominent and poorly aligned eyes. All parents should have easy access to a Unit, which will be able to assess the children accurately, if problems arise.

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Treatment of exposure keratopathy – regular use of artificial tear supplement to avoid dry spots on the corneal epithelium (the surface layer of the cornea). This will usually be sufficient to maintain the integrity of the cornea as a clear and smooth focusing surface. Intensive antibiotics therapy is urgently needed in cases with superimposed infection to avoid any cornea scarring. Tarsorrhaphy (the surgical closure of the eye lids) may be indicated for severe corneal exposure if all other measure fails. Tarsorrhaphy is usually temporary until craniofacial surgery is undertaken to increase the orbital volume.

Treatment of amblyopia –

- The correction of any refractive error with glasses to maximise the quality of image sent to the brain.
- To start patching of the stronger eye on detection of unequal vision, to stimulate the ‘switched off’ portion of brain. This is only effective during the period of active visual development, which is normally complete by between 6 and 8 years of age.

Detection of papilloedema can be due to exposure keratopathy and strabismus induced Amblyopia (‘lazy eye’).

- Exposure keratopathy is the excess exposure of the surface of the eyes to the dry atmosphere. It is caused by imperfect wetting of the corneal surface by the tear film because of the inability of the lids to resurface the cornea with each blink. It’s severity can range from inferior punctate epitheliopathy (spots of surface irregularity in the lower portion of the cornea) to severe corneal ulceration, neovascularization (abnormal new vessel formation onto the clear cornea, causing clouding). Very rarely severe infection and even globe perforation can occur depending on concurrent management.

- Ocular motility abnormalities and Strabismus or squint, means the two eyes are not aligned in an appropriate manner, or do not move together as a symmetrical unit. A variety of horizontal and vertical deviations of the eyes can be seen. Most commonly the children show a ‘V’ pattern of eye movements, in which the eyes tend to be divergent in the up-gaze, less divergent in the straight ahead position and less divergent still (or even convergent) in down-gaze. Very often excyclorotation (outward rotation of the globes) is found because of the changes to orbital structure alter the mechanics of the ocular muscles and hence their direction of pull.

- Hypotelorism is the increased separation of the orbits, due to the premature fusion of the bony sutures. It causes the characteristic facial appearance of the eyes being more widely spaced. It can be associated with lacrimal (tear) system anomalies, with symptomatic watering eyes and recurrent infection.

- Reduced vision can be due to exposure keratopathy and strabismus induced Amblyopia (‘lazy eye’).
Exposure keratopathy causes irregularity on the cornea surface (irregular astigmatism) affecting the sharpness of images focused onto the retina. The imperfect images cause reduced vision in both eyes. In cases where one eye is more affected than the other, the difference in the quality of blurred image may produce amblyopia. This would result in one eye having poorer vision than the fellow eye, even with spectacle correction- the so-called 'lazy eye'.

A similar process happens in children with strabismus (Squint), with the developing visual system tending to ignore the image from the squinting eye. Amblyopia is usually reversible with active management in early life (before the age of 6 and as early as possible).

**Rare Complications**

- **Optic nerve disease – Papilloedema** (optic nerve swelling secondary to raised pressure inside the brain) and **Optic atrophy** (wasting away of the optic nerve). Closure of the coronal cranial suture in particular, can result in raised intracranial pressure. That raised pressure is transmitted to the sheath surrounding the optic nerve causing papilloedema and, if not relieved, consecutive optic atrophy. Papilloedema is nearly always bilateral, although it may be asymmetrical. If this is not reversed, optic atrophy inevitably would occur which results in permanent decrease in vision.

- **Other ocular features reported are** (these are the findings not related to the extent of craniofacial disorder.)
  - **Down slanting palpebral fissure**: typically inferior (downward) and lateral (outward) canthal (outer corner of eye) malposition.
  - **Anirida**: congenital absence of iris
  - **Blue sclera**: describes the unusually thin sclera (the white part of the eye), therefore the underlying choroidal pigment can be seen more readily.
  - **Cataract**: opacity in the lens inside the eye.
  - **Ectopia lentis**: abnormal position of the lens of the eye.
  - **Glaucoma**: an eye disease characterised by raised intraocular pressure, which may cause optic nerve damage.
  - **Megalocornea**: congenital abnormal enlargement of the cornea.
  - **Keratoconus**: conical protrusion of the central part of the cornea.
  - **Coloboma**: absence of some ocular tissue due to failure of closure of foetal fissure in the developing eye. It may affect various ocular tissues e.g. eye lid, iris, Ciliary body, choroids, lens, retina and optic disc.
  - **Nystagmus**: involuntary rapid movements of the eyeball, which are usually the consequence of reduced vision in early life.
  - **Optic nerve hypoplasia**: incomplete development of the optic nerve

**Management**

The management of the eye problems requires the involvement of the paediatrician, cranio-facial surgeon and a neurosurgeon. This multidisciplinary approach is especially important in cases with raised intracranial pressure. The primary aim of the eye care is to prevent permanent visual loss by targeting the process of amblyopia and avoiding damage to the cornea and optic nerve.