Review

Eye Abnormalities in Fetal Alcohol Syndrome

Abdelmageed Abdelrahman, Richard Conn

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BACKGROUND

Fetal alcohol syndrome (FAS) – a condition caused by chronic maternal alcohol consumption during pregnancy – is important both in terms of its prevalence and its effects: it is estimated to be the commonest non-inherited cause of learning disability. Diagnosis is based on the presence of characteristic facial dysmorphism, postnatal growth retardation and functional or structural central nervous system deficits. In this review, however, we propose that the eye is a sensitive and reliable marker of teratogenesis and provides a useful adjunct to the diagnosis of FAS, with eye abnormalities having been shown to occur in over 90% of children with the condition. It is our objective, then, to describe the effects of prenatal alcohol exposure on the eye, quantify their incidence and comment on their importance in diagnosis of children with FAS.

METHODS

We searched the electronic library Medline from its inception to March 2009 for original research and review articles relating to prenatal alcohol exposure, using combinations of the terms ‘fetal alcohol’, ‘eye’, ‘ophthalmic’ and ‘alcohol teratogenesis’. We applied no language restrictions. We also searched reference lists of identified articles. Whilst this is not an exhaustive review, we have attempted to make it representative of the literature in this field.

RESULTS

Short palpebral fissures: A shortened distance between the inner and outer corners of each eye, defined as a length two or more standard deviations below the mean. Normal measurements for palpebral fissure length are provided by the Hall Caucasian Charts. Measurement can be easily performed with a small ruler, although the examiner must ensure that the patient’s eye is fully open. The presence of short palpebral fissures is of particular discriminant value in FAS.

Epicanthus: This is a lateral extension of the skin of the bridge of the nose over the endocanthion. It is important to remember that epicanthal folds may occur naturally in some races and are more commonly noted in childhood. A study found 80% of children prenatally exposed to ethanol had epicanthus; degree of epicanthus may be quantified with a Likert scale.

Ocular hypertelorism: Defined as an increased interorbital distance, and may be measured as the distance from right endocanthion to left endocanthion. It is a commonly reported finding in FAS, although not pathognomonic of the condition.

Coloboma: Normally the choroid fissure closes during the seventh week of development - failure of closure results in the formation of a distinctive cleft in the iris known as coloboma iridis. This is one of the key extensive malformations that may be found in children with FAS.

Strabismus: An abnormal alignment of the two eyes. While it is a non-specific finding, it is common in FAS and may be diagnostically useful in conjunction with other features; Strömland found that of thirty children with FAS, 13 had strabismus, 12 of which had a horizontal convergent form (esotropia).

Blepharoptosis: (or ptosis), is drooping of the upper eyelid. Although it is a non-specific sign, Strömland found that 20% of children with FAS had blepharoptosis.

Microphthalmia: An abnormally small eye – is a frequent finding in FAS and was included in the Fetal Alcohol Study Group diagnostic criteria. However, the diagnostic usefulness of this condition is limited by difficulty in detection, particularly in the presence of confounding factors such
as microcephaly and short palpebral fissures. Objective methods such as ocular axial length measurement under ultrasonography may be necessary.\textsuperscript{2,3}

**Abnormalities of the fundus:** The fundus may be affected by various abnormalities - the most common findings are hypoplasia of the optic nerve and increased tortuosity of the retinal vessels.\textsuperscript{8,9} In a cohort of Swedish children with FAS, Strömland found optic nerve hypoplasia in 48% and increased tortuosity in 49%.\textsuperscript{2} More recently, Hug et al suggested that prenatal alcohol exposure leads to disturbed retinal function on the basis of abnormal electroretinograms in ten children with FAS.\textsuperscript{10} These conditions are important primarily because of their association with visual loss, but also because they may be useful diagnostically via ophthalmoscopic examination of the fundus.

**DISCUSSION**

The detrimental effects of alcohol on the fetal eye are perhaps less well recognised than its other manifestations, but are common and important from the perspective of both diagnosis and management.

While the relationship between alcohol and teratogenesis is not disputed, there is conflicting evidence on the effect of very small amounts of alcohol on mother and baby. Consequently, the threshold of safe alcohol consumption during pregnancy is unclear. The Royal College of Obstetricians and Gynaecologists (RCOG) states that women should avoid drinking excessive amounts of alcohol when pregnant but there is no definite evidence that drinking 1 to 2 units once or twice a week is actually harmful.\textsuperscript{11}

To this end, we recommend a full ophthalmic examination in all children suspected of having a fetal alcohol spectrum disorder, whether this is on the basis of developmental concerns or the possibility of maternal alcohol consumption during pregnancy. This should comprise:

- Inspection for periocular features, possibly supplemented by morphometric analysis
- Measurement of visual acuity (using visual evoked potentials), visual fields and eye movements
- Slit lamp examination of anterior segment and media
- Ophthalmoscopic examination of anterior segment and media
- Ophthalmoscopic examination with particular attention paid to the optic disc

Not only is this examination useful diagnostically, but it also allows timely and appropriate management of problems, which may cause profound visual loss and contribute significantly to the handicap of those affected.

We recognize that, aside from those described here, a myriad of other eye abnormalities have been described in FAS. Nevertheless, it is our hope that this review raises awareness of the effects of alcohol on the developing eye and their clinical importance.

The authors have no conflict of interest. Abdelmageed Abdelrahman and Richard Conn are final year medical students at Queen’s University Belfast, Northern Ireland

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