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Foetal Alcohol Syndrome

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INTRODUCTION

Foetal alcohol syndrome (FAS) is a condition of mental and physical defects that occurs when alcohol crosses the placental barrier and enters the foetal circulation due to the intake of alcohol by the mother during pregnancy. Tragically, the consequences of this prenatal alcohol exposure are irreversible and last a lifetime. Up to 90% of children with FAS present with ocular manifestations[1].

According to the World Health Organization (WHO), there is NO safe level of alcohol that may be consumed during pregnancy and recommend total abstention of alcohol during pregnancy. In addition to pregnancy, alcohol intake during breast feeding may have a negative impact on the quality of milk produced by the mother[2].

PREVALENCE

South Africa has documented the highest rate of FAS in the world, with a figure greater than 45 per 1000 births in the Western Cape[3] and 20 per 1000 births in Gauteng[4]. Comparably, prevalence studies in other parts of the world show a rate of 0.2-2 births per 1000[5].

In South Africa, nearly one million adults and children are affected by the advanced mental or physical manifestations of FAS. The spectrum of disorders arising from FAS in South Africa, and particularly in the Western Cape, is even more common than Down's syndrome, making it the most common birth defect and cause of mental retardation[6]. As more evidence is coming to light of the staggering rate of FAS in South Africa, it is undoubtedly an issue that requires immediate and decisive intervention[6].

It has been postulated that the high rate of FAS in rural areas of South Africa may be attributed to the *dop system*- a 400 year old practice where labourers, especially in the agricultural sector, were historically given their wages in the form of cheap wine. Although this system was abolished in 1961, this pattern of binge drinking remains a common practice on farms, particularly in the Cape[7]. According to Croxford et al[8], heavy alcohol intake very commonly in a binge drinking pattern (more than 5 drinks per sitting), is seen in over 20% of women in the Western Cape.

Also, the lack of available information from primary care givers and the lack of surveillance and available data across the country's population suggest that these figures may actually be much higher.

DIAGNOSIS

Diagnosis of FAS is based largely on three diagnostic criteria that are characteristic of the syndrome, viz: dysmorphism of facial and body features, diminished structural growth for age and neurological (intelligence and social) defects. A history of maternal alcohol consumption is helpful although not necessary and sometimes unattainable[4]. Milder forms of the condition, in which the full triad of signs do not manifest as a full syndrome, are known as Foetal Alcohol Effects (FAE) and also present with various developmental and behavioural problems.

1. **Dysmorphia**: Dysmorphia refers to congenital malformations that occur due to a disturbance in the development of the foetus. The National Centre for Birth Defects and Developmental disabilities of America has identified small palpebral apertures, a smooth philtrum (which is an underdeveloped groove between the nose and the upper lip) and thin vermillion border as the three cardinal features of facial dysmorphia that is consistent with FAS[9]. These give the 'monkey face' appearance associated with FAS. Furthermore, flattened cheekbones, cleft palates, low nasal bridges and auditory disorders are common, although less diagnostic features. These facial anomalies are most noticeable between the ages of 2- 10 years and are difficult to identify at birth and later on in life[10].

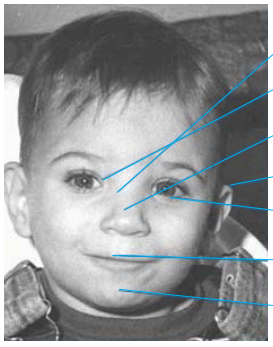

Figure 1: the characteristic facial dysmorphia seen in FAS.

2. **Diminished Structural Growth for Age:** Low birth weight or height (or both) at or below the 10th percentile at any point in time assists in supporting the diagnosis of FAS. Small head circumference (microcephaly) and a failure to thrive are also seen very commonly in infants with a prenatal history of maternal alcohol or substance abuse. Children with FAS tend to have a short stature and are often underweight for their age throughout childhood.

3. **Neurological:** Neurological defects that are not associated with fever or injury are seen in FAS. In infants, this is seen as poor suckling when feeding. Delayed milestones such as crawling, walking or talking are also noted. Later, poor IQ, behavioural and learning disabilities which manifest in different ways give rise to secondary disability. An inability to grasp concepts such as time and money, inept social skills such as difficulty making and keeping friendships, hyperactivity and impulsivity, poor fine motor and balance skills and speech impediments are also commonly seen in FAS. It is different from Attention Deficit Hyperactivity Disorders (ADHD) in that children with FAS tend to have difficulty encoding information and shifting attention whilst ADHD presents with difficulty focusing and sustaining attention. Since they are often unable to distinguish right from wrong, they often become involved in criminal behaviour.

Foetal Alcohol Syndrome

Many children born to parents who abuse alcohol are fostered or adopted and the new parents are unaware of the lifestyle of the biological mother making it sometimes difficult to diagnose the condition. Evaluation often requires a multi disciplinary approach between health and educational professionals. Of concern, foetal alcohol exposure may increase the risk for later alcohol, tobacco, and drug dependence in these adults[11].

Even a small amount of alcohol consumption of 2-3 glasses a day can put the child at risk for the development of foetal alcohol syndrome. Unfortunately, many expectant mothers discover that they are pregnant some weeks into gestation, by which time significant damage to the developing embryo has occurred in the case of habitual alcohol consumers.

THE EFFECT OF ALCOHOL DURING THE PREGNANCY TIMELINE:

The negative impact of alcohol as a teratogen occurs in all trimesters of pregnancy. There is not a single period where alcohol exposure remains without consequence to the development of the foetus. Not only is the dose of alcohol related to the severity of the damage on the foetus, the timing and the metabolism of alcohol exposure has an effect on the prenatal development.

1st trimester (0 - 12 weeks):

This is the most critical period for the structural development of the foetus. The teratogenic effects of alcohol during this period are most severe and alter the way in which cells grow and arrange themselves resulting in the characteristic facial features and small head circumference found in FAS. The foetus, in unplanned pregnancies, often goes through this critical period without changes in lifestyle by the expectant mother and this may cause the most severe and irreversible damage to embryonic development.

2nd trimester (12 - 24 weeks):

Foetal exposure to alcohol may result in miscarriage. Drinking during this trimester will affect the physical size of the child.

3rd trimester (24 weeks – to delivery):

This is the period when the foetus undergoes substantial growth. The cerebellum is one of the last structures of the brain to develop and alcohol exposure during this period is most likely to cause mental retardation and nystagmus.

OCULAR MANIFESTATIONS:

External Ocular findings:

Short palpebral fissures: This is a critical diagnostic feature that defines the unique facial phenotype of FAS. Palpebral fissures may be measured using a PD ruler.

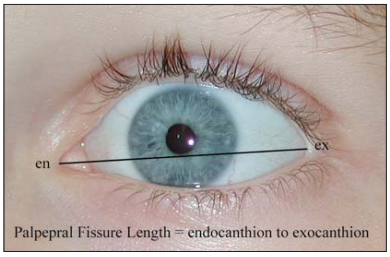

Figure 2: Measurement of the palpebral fissure.

Source: American Academy of Paediatrics

Strabismus, telecanthus, blepharoptsosis, epicanthus and iris coloboma are also ocular indicators used in the diagnosis and assessment of FAS. Due to higher presence of epicanthus and telecanthus in FAS, a pseudostrabismus may be present and needs to differentiated from a true strabismus by measurement of the angle kappa.

Internal ocular findings:

Optic nerve hypoplasia: This is one of the most common ocular findings in FAS, seen in approximately 48% of cases[1]. Optic nerve hypoplasia is a result of a failure of optic nerve axons to be produced and sustained, with premature apoptosis of cells. Optic nerve hypoplasia is one of the leading causes of low vision and blindness in children. It is diagnosed through the classic small, pale appearance of the optic nerve. In general, it remains stable and does not become progressively worse over time. Using retinal imaging such as fundus photography, previous studies have confirmed optic nerve hypoplasia by measuring the ratio of disc diameter (DD) to the disc macula distance (DM). DD: DM ratios less than 0.35 and reduced vision are considered to be diagnostic of optic nerve hypoplasia[12]. The peripapillary double ring, a band of often pigmented tissue is also commonly seen although not diagnostic of optic nerve hypoplasia.

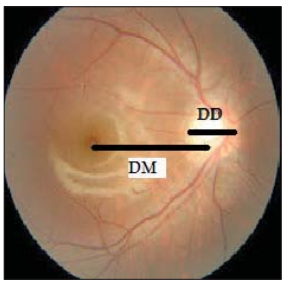

Figure 3: Optic nerve hypoplasia which is easily diagnosed using the ratio of DD: DM.

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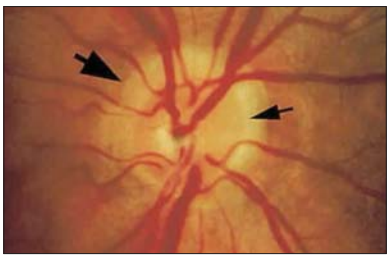

Figure 4: The Double Ring Sign which is commonly seen in optic nerve hypoplasia.

Optic nerve hypoplasia is also commonly associated with retinal tortuosity. These findings result in reduced VA's, increased light sensitivity and nystagmus in severe cases. Strabismus from poor vision, most commonly esotropias are associated with optic nerve hypoplasia.

MANAGEMENT OF THE PATIENT WITH FAS:

- All children suspected of having FAS or with a history of prenatal maternal alcohol abuse should have a full workup done which includes[13]:

1. A physical observation of facial features,
2. VA testing,
3. Slit lamp examination and
4. Posterior visual assessment with particular focus on the optic nerve head.

- FAS requires a multi disciplinary approach and optometrists may have to work with audiologists, speech therapists, occupational therapists and other health care professionals to ensure that the patient receives holistic treatment and management. Furthermore, eradication of FAS in communities where there is a high prevalence of alcoholism due to poverty conditions will take much effort. Alcohol has a numbing effect which is used as a means of escapism from reality and this will require a total change in the long term mindset and structure of society.
- Education- since FAS has been identified as alarmingly high in certain rural areas of South Africa, it is important to make parents and care givers aware of the cause and effect of this condition. Since this remains a lifelong handicap, it is imperative that care givers are educated so that the best possible approach to treatment may be reached. It is found that children who are diagnosed before the age of 6 years and who have a stable home environment cope much better with FAS and have a lower risk of secondary disabilities arising from the syndrome.
- Spectacles in cases of myopia or low vision aids in cases of poor vision that is associated with optic nerve hypoplasia.
- Prism may be required in cases of strabismus.

CONCLUSION:

Substantial evidence from various research studies has confirmed the teratogenic nature of alcohol on the development of the foetus. FAS is a totally preventable condition and also unfortunately a totally irreversible condition. The shockingly high prevalence of FAS in the South African setting makes this a condition that we ought to be familiar with. Despite some intervention and increased awareness, the consumption of alcohol in an irresponsible manner remains wide spread. We are still a long way from eradicating this mindset in our population. The presence of eye related manifestations are sometimes less recognized but are nonetheless important from the aspect of both diagnosis in the clinical setting and the management of this syndrome[13].

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