Fetal alcohol spectrum disorder (FASD): Perspective redefined

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INTRODUCTION:

Prenatal exposure to alcohol is the leading preventable cause of congenital anomalies and developmental disabilities. Fetal alcohol spectrum disorder (FASD) is a term that is used to describe the range of physical, behavioural, and neurodevelopmental effects that can occur in an individual who was prenatally exposed to alcohol and may have lifelong implications and high societal costs.

Pregnant persons who drink alcohol come from all socioeconomic strata, ages, and races . These individuals may also be using other addictive or illicit substances. Both the gravida and family benefit from factual, non-judgmental information about the maternal and Fetal risks of alcohol use and, if necessary, from counselling regarding strategies for cessation. Pregnant people are typically highly motivated to modify their behavior to help their unborn child. In one national survey from the United States, 87 percent of women who self-reported consumption of alcohol before pregnancy quit drinking during pregnancy, 6.6 percent reduced their alcohol intake, and approximately 6.4 percent reported no reduction.

Etiology

Fetal alcohol spectrum disorder is an umbrella term for a set of disorders caused by the consumption of alcohol by a mother whilst pregnant. These conditions range in diversity from the full presentation of fetal alcohol syndrome, involving a characteristic set of facial features combined with growth and neurocognitive deficits, to a range of conditions affecting the neurobehavioral presentations of the condition without all these features. Box 1 summarizes the different clinical criteria used, based on the USA Institute of Medicine diagnoses, as well as the common diagnostic methods used. O'Leary[3] recently summarized the epidemiological research in to Fetal alcohol spectrum disorders concluding that the estimated worldwide prevalence is around 1/100 for fetal alcohol spectrum disorders, making it the most common cause of learning difficulties.

Knowledge

levels of fetal alcohol spectrum disorders by the general public and health professionals in the UK are not accurately known. Most relevant studies have taken place in the USA and Canada where there is greater general awareness of the disorder. Nanson *et al.* surveyed a group of paediatricians and general practitioners (GPs). She showed that whilst most people had heard of fetal alcohol spectrum disorders, less than 50% knew much about how to recognize it. Ten per cent of those who did recognize the condition did nothing about it. Stohler studied 40 high risk pregnancies to see if fetal alcohol syndrome was detected in the offspring. A specially trained research assistant identified 16 cases resulting from these pregnancies. None had been identified by routine paediatric screening. Further, 73% of the case notes made no record of maternal alcohol consumption despite the mothers being known to be in a high-risk group.

Kesmodel *et al*. studied a group of pregnant Danish women. The majority (74%) felt that drinking in pregnancy was acceptable; 65% reported they had received little or no information from their midwife about possible dangers. This is consistent with data collected by the UK government in their alcohol reduction strategy: it was found that 61% of women drank during pregnancy to some level. MacKinnion studied a group of teenagers in America. Although 97% had heard of alcohol causing problems during pregnancy, 48% thought that the condition related to the baby being addicted to alcohol and just over 50% felt the condition could be cured. Similar information needs to be collected in the UK urgently in order to inform health promotion strategies.

Pathology

Since the naming of fetal alcohol syndrome in 1973 there has been some controversy as to its actual existence. Further uncertainty persists regarding the level of maternal alcohol consumption that can cause damage. Evidence for pathogenic mechanisms comes from mainly animal studies. These have been corroborated by some human investigations. The difficulty with human research lies in the ethics of the methodology and the subsequent biases inherent in available approaches. It is the combination of all the evidence that has given most insight into the condition's pathogenesis.

Maier and West suggest that it is the rise in alcohol levels, as well as the subsequent withdrawal, which cause damage. Both raised acetaldehyde levels as well as subsequent apoptotic damage from excess glutamate activity following GABA (gamma amino butyric acid) withdrawal are implicated. Ikonomidou *et al*. report that exposure of rat brain to ethanol for a period of hours during a specific developmental stage induces an apoptotic neurodegenerative reaction that deletes large neurons from several developing sites. This process is further complicated by individual genetic differences, diet, and hormonal interactions as some of the multifaceted risk factors. Thus, the prediction of individual risk is particularly difficult if not impossible. The UK binge-drinking culture and lack of awareness of true drink size by the general public are additional risks.

Reports have shown increasingly that there are vulnerable periods of neonatal development that can be affected by teratogenic ingestion. In terms of neural development, which occurs throughout pregnancy, it is often the inter-neurone connections that are damaged. This is especially the case at lower levels of consumption. Charness *et al* report that even at low concentrations of ethanol exposure, cell adhesion molecules are inhibited. These have subsequent effects on neuronal migration, fetal alcohol syndromeciculation and synaptogenesis, which are all vital to the developing brain. These risk factors, as well as protective factors, need further clarification.

More recently, work by Hepper *et al.* using ultrasound monitoring of fetal behaviour where mothers consumed alcohol at levels within current UK government guidance, showed effects on fetal startle which did not habituate to a level achieved by those that consumed no alcohol. They suggest that this is a sign that even at the low levels of alcohol consumption (an average of 4.3 units/week±1.9) permanent damage to the developing fetal brains is occurring.

Secondary disabilities

Jacobson summarized the cognitive deficits associated classically with fetal alcohol spectrum disorders. These deficits tend to be life long, and are evident in the absence of facial pathology. Box 2 summarizes the core deficits witnessed. Rasmussen[15] recently published a systematic review of the executive and working memory deficits associated with fetal alcohol spectrum disorders. Further, it was reported that the severity of these long-term core deficits is independent of the presence of facial features, meaning a diagnosis of alcohol related neurodevelopmental disorder can be as debilitating as full fetal alcohol syndrome. In many cases it can be more, because the person may appear superficially not to have a disability and will be expected to perform to a level of sophistication and ability they simply cannot manage. Clinical evidence suggests that this makes individuals more vulnerable to avoidable serious secondary disabilities.

Streissguth *et al*. have highlighted ongoing secondary difficulties. Intellectual tests show that average IQ is 85.9 for fetal alcohol spectrum disorders. This group have an uneven profile of abilities and disabilities that means their average level of intellectual functioning is not truly reflective or predictive of their pattern of cognitive strengths and needs. They further show this group are vulnerable to life events. Ninety per cent have some form of diagnosable mental disorder. These can be as diverse as ADHD (attention deficit hyperacitivity disorder), social and communicatory impairments, personality disorder, schizophrenia, addiction and depression. Fifty per cent have some form of confinement in mental health or criminal justice situations; 50% some form of sexually inappropriate behaviour. Much of this can be related to their inability to control and maintain their behaviour attributable to damage caused to their executive function abilities combined with difficulties in receptive language and inability to consolidate memories because of temporal/hippocampal damage.

Management

The management of fetal alcohol spectrum disorders classically is divided into two main areas. First, recognition of the dangers of alcohol consumption in pregnancy and the prevention of damage to the fetus. The second area is less well researched but relates to the management of people who have the condition. The emphasis on prevention has been the most highly publicized of the two with numerous authors stressing the level of risk that is harmful, early detection of at risk mothers, the need for information sharing between professionals and public as paramount priorities. Emerging methods such as the use of routine screening tools such as TWEAK, hair sampling, or meconium testing have been suggested. However, the ethical debate around their use is in its infancy thus clarification is required before they can be recommended routinely. Research into protective factors during pregnancy has been inconclusive and contradictory. The use of vitamin E as a potential antioxidant has been shown beneficial in some studies and ineffective in others. Clearly, much has still to be done before conclusive information can be given to mothers contemplating pregnancy. For this reason we continue to emphasize the general abstinence message.

With regard to children and adults who have fetal alcohol spectrum disorders, much work has been undertaken to categorize difficulties and establish diagnoses. Less research has been undertaken relating to clinical management. This work has mainly involved children in the USA and Canada. Chudley *et al*. recently reviewed the Canadian guidance on diagnosing and managing fetal alcohol spectrum disorders. They emphasize early recognition and psychometric testing combined with multidisciplinary intervention approaches.

CONCLUSION:

There are very few fetal alcohol spectrum disorder experts in the UK. Thus, obtaining specialist advice is restricted to the lucky few. As is the case for many, clinical service funding streams mean that for people with fetal alcohol spectrum disorders it is not always possible to obtain what they need. Nonetheless, by recognizing the condition, obtaining sufficient early evidence and using resources locally available in collaboration with multi-professional colleagues can reap important rewards.

The estimated extra cost of fetal alcohol spectrum disorders in USA in has been estimated at $500 000/individual over a 20-year period.[23] For a condition that can be prevented increasing awareness, education and UK-based research will help to allow access to local provisions and could be expected to reduce the prevalence of the condition as well as the human cost in the future.