

Fetal Alcohol Spectrum Disorder(FASD)

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ABSTRACT

Fetal alcohol syndrome (FAS) is the leading cause of mental retardation worldwide but is also the foremost preventable cause of neurobehavioral and developmental abnormalities. It is equally important to know spectrum of disorders due to maternal alcoholism during pregnancy such as Fetal alcohol spectrum disorder (FASD) in order to identify and treat affected child and family effectively. This article aims to create awareness among practising clinicians most of whom are only aware of phenotypical variant of FASD which is FAS. In this article we discuss those aspects of FASD relevant to the clinician such as: terminological ambiguity, assessment, diagnosis and prevention. [Indian J Pediatr 2009; 76 (11) : 1173-1175] E-mail: mkbakar76@rediffmail.com

Key words: Fetal alcohol syndrome (FAS); Fetal alcohol spectrum disorder (FASD); Mental retardation (MR); Neuro-behavioural abnormalities

References to the association between maternal alcohol consumption and fetal malformation date back to early Greek and Roman mythology. However, the syndrome "Fetal Alcohol Syndrome" (FAS) as triad of facial dysmorphism, growth retardation and central nervous system deficits was first described by Jones and Smith¹ in 1973. Though FAS is a well-recognised disorder, it is still in its infancy in medical literature. As the leading cause of preventable mental retardation, given a rate of nearly 2 per 1000 births², and as Indian women are increasingly prone to effects of globalisation such as alcoholism during pregnancy it is high time that FAS and Fetal Alcohol Spectrum Disorder (FASD) should receive more clinical attention. Although entirely preventable, the lifelong deficits and disabilities it confers are irreversible and adversely affect the child, family and society at large. Since its formal recognition over 30 years ago, considerable progress has been made in understanding its etiopathogenesis, clinical presentations and related developmental dysfunctions. But in terms of effective prevention and intervention strategies, there is still a long way to go because of lack of awareness about FAS and FASD among both public and medical community which is further complicated by the Terminological ambiguity and varied presentation. From a clinician's perspective, the take-home message is: at every opportunity, advise women

on the risks of alcohol consumption in pregnancy, maintain a high index of suspicion in its early recognition and diagnosis, and refer to a specialist to ensure that a comprehensive assessment and treatment package is made available for these children and their families.

In this paper, we focus on the clinical aspects of FAS and FASD such as Terminological ambiguity, assessment, diagnosis and prevention.

Terminological Ambiguity: There is now universal agreement that FAS should only be used to describe the triad of facial dysmorphism, growth retardation and neuro-cognitive deficits, with confirmed history of maternal alcohol consumption during pregnancy. But there is less agreement on the choice of terminology to describe children with some but not all of the characteristic features of classic FAS. National Organisation on Fetal Alcohol Syndrome (NOFAS) in 2004 agreed to use the term Fetal Alcohol Spectrum Disorder (FASD) as an umbrella term, not as a diagnostic category but to encompass various other diagnostic categories such as FAS, alcohol related birth defects (ARBD), alcohol related neurodevelopmental disorder (ARND) etc. FASD was defined as "the range of effects that can occur in a person, whose mother drank alcohol during pregnancy, including physical, mental, behaviour and learning difficulties, with possible lifelong implications". Table 1 summarises the terminological complexity and provides a short description of each concept by its proponents.

Assessment: It is clinically important to note that it is

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TABLE 1. Terminologies and Descriptors of Various FASD Categories

FAS – Fetal Alcohol Syndrome	Facial features, growth restriction, CNS abnormalities with or without confirmed maternal alcohol exposure.
FAE – Fetal Alcohol Effects	Absence of facial features, some of the other features present, confirmed maternal alcohol exposure.
ARND- Alcohol Related Neurodevelopmental Disorder	CNS abnormalities, evidence of complex patterns of behaviour or cognitive abnormalities inconsistent with developmental level and that cannot be explained by genetic predisposition, family background or environment alone. Confirmed history of prenatal alcohol exposure.
FAAS – Fetal Alcohol Abuse Syndrome	Abel proposed to change the nomenclature from FAS to FAAS, as he was of the opinion that FAS occurs only in children whose mothers are alcohol abusers. Hence Abel proposed that FAAS would avoid the notion that FAS can result from low levels of alcohol use.
AARBE – Alcohol Abuse Related Birth Effects	Abel’s opinion that only very high level of drinking would result in harm to the foetus and proposed to name these resulting effects as AARBE and hoped for much less uncertainty towards the aetiology.
PFAS – Partial Fetal Alcohol Syndrome	Facial features, evidence of growth retardation and CNS abnormalities may or may not be present, may present with complex pattern of behaviour or cognitive abnormalities inconsistent with developmental level and that cannot be explained by genetic predisposition, family background or environment alone. Evidence of maternal alcohol exposure may or may not be present.
ARBD – Alcohol Related Birth Defects	Confirmed evidence of maternal alcohol exposure. Facial features, Specific congenital structural defects in at least 1 organ system, if minor anomalies, at least 2 must be present. Normal growth and CNS features.
Alcoholic Embryopathy	Majewski and his colleagues proposed a weighted scoring system ranging from 1 to 8 of 25 items. Based on total score, a child is graded 1, 2, or 3 alcohol embryopathy.
FASD, Fetal Alcohol Spectrum Disorder	a concise, uniform definition for conditions caused by prenatal alcohol exposure and not intended for use as clinical diagnosis; an umbrella term describing the range of effects that occur in an individual whose mother drank alcohol during pregnancy.

TABLE 2. Age Related Additional Presentations of FASD

Newborn	Early Childhood/ Pre-School	Middle Childhood	Adolescence and Adulthood
Sleeping & Feeding Difficulties	Talkative & friendly	Small for age	Characteristic facies may disappear
Weak, sick, irritable & tremulous	Temper tantrums	Impulsive, impaired attention	Poor school performance
Excessive crying	Hyperactive	Poor social skills	Impaired judgement
Hypersensitive to light & sound	Small for age	Specific learning difficulties	Behavioural problems
Seizures	Speech delay	Language deficits	Poor peer relationships
Failure to thrive	Fine motor abnormalities	Lack of organisation	More prone to substance misuse, depression, teenage pregnancy, etc
	Mental retardation	Impaired abstract thinking	Difficulties with living skills
		Mental Retardation	Mental retardation

an interplay of a host of maternal risk factors and protective factors that go into determining the development, or not, of FASD. Hence the prediction of an individual child’s risk is difficult. The characteristics may only appear with age and the degree of severity varies from individual to individual and that some of these abnormalities change with age. Table 2 lists some of the key age dependant presentation features of FAS (in addition to the characteristic triad of symptoms) in the newborn, through early and middle childhood to adolescence and adulthood. As FASD is a complex and multi-faceted disorder, to achieve a comprehensive

assessment and appropriate/ definitive diagnosis, often a multi-disciplinary team approach involving physicians, psychologists, speech and language therapists, occupational therapists and physiotherapists is required.^{3,4,5}

Diagnostic Criteria: The three most commonly used diagnostic criteria for making a diagnosis of FAS are the Institute of Medicine Criteria (IOM),³ Centre for Disease Control (CDC) criteria ⁴ and the four-digit diagnostic criteria.⁵ The IOM criteria identifies 5 categories of alcohol related disorders:

1. FAS with confirmed maternal alcohol exposure –

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requires evidence of the following features: confirmed maternal alcohol exposure, characteristic pattern of facial abnormalities, growth retardation and neuro-developmental abnormalities (e.g., Head Circumference \leq 10th percentile, structural brain abnormalities, neurological hard or soft signs).

2. FAS without confirmed maternal alcohol exposure – all of the above features but without confirmed maternal alcohol exposure.

3. Partial FAS with confirmed maternal alcohol exposure – requires evidence of the following: confirmed maternal alcohol exposure and characteristic facial abnormalities and some of the following: growth retardation, neuro-developmental abnormalities and behavioural / cognitive abnormalities not explained by family background or environment alone (e.g., impaired impulse control, deficits in social perception, difficulties in higher level receptive and expressive language, and impairments in attention, judgement, abstraction, memory and metacognition).

4. Alcohol related birth defects (ARBD) - require presence of these: confirmed exposure to alcohol and congenital abnormalities as a result of this affecting various organ systems such as heart (atrial septal defects, ventricular septal defects, etc), kidneys (hypoplastic kidneys, hydronephrosis, etc), eyes (strabismus, retinal vascular abnormalities, etc), bones (scoliosis, hemivertebrae, clinodactyly, etc) and ears (conductive or sensory hearing loss).

5. Alcohol related neurodevelopmental disorders (ARND): - presents with maternal alcohol exposure, CNS neuro-developmental abnormalities (as listed above) and behavioural and cognitive abnormalities (as listed above). For detailed look at the complete CDC diagnostic criteria, refer to www.cdc.gov/ncbddd/fas/documents/fas_guidelines_accessible.pdf.⁴

Differential Diagnosis: FAS should as far as possible be a diagnosis of exclusion. Some of the disorders that overlap with FAS are⁶ : DeLonge syndrome, Noonan Syndrome, maternal Phenylketonuria, malnutrition, failure to thrive, other causes for pre-natal and post-natal growth retardation; Williams Syndrome and Down Syndrome.

Prevention: Prevention at three possible levels: Level 1 being primary prevention^{7,8} which is intervening to

prevent the onset or development of FAS (examples: raising public awareness, primary care physicians advising pregnant women that there is no suggested safe level of alcohol intake for pregnant women and they should abstain alcohol, etc); secondary prevention⁸ which includes strategies and interventions aimed at early diagnosis and treatment of children with FAS (examples of effective measures include improving clinicians' ability to recognise symptoms of FAS early, and specific targeted psychological treatments to help such children and families cope with their developmental deficits); and tertiary prevention strategies aimed at reducing the onset and impact of secondary disabilities such as confinement, mental health difficulties, dependant living, etc.

Contributions: MKB; Review of articles from year 1973-2007, contribution to terminological ambiguity and diagnosis of FASD, Preparing and submitting the manuscript. NSK; collecting data and input with regard to psychological aspects of FASD. SG; collecting data and input to preventive aspects of FASD.

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