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Aanvraag kopie / Boek te leen

ISSN p/e: 1326-5377 / MBSN:
Titel: medical journal of Australia
Jaar: 1983
Volume: 1
Aflevering: 6

Auteur: Lipson
Artikel: Fetal alcohol syndrome. A great paediatric imitator.
Pagina's: 266

Opmerking: 2015-06-24

Datum: 29-04-2015 17:52

(1) [ ] origineel gestuurd
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(5) [ ] niet aanwezig
(6) [ ] niet beschikbaar
(7) [ ] uitgeleend
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Groep: Elektronische levering

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Fetal alcohol syndrome

A great paediatric imitator

Anthony H. Lipson, David A. Walsh, and William S. Webster

ABSTRACT: Twenty children affected by prenatal exposure to alcohol are described. Their mothers either suffered from chronic alcoholism, or were binge or heavy drinkers while pregnant. Only in nine mothers (45%) was alcohol abuse recognized and associated with their babies' anomalies in the first months after birth; in some, the diagnosis of fetal alcohol syndrome was delayed for many years. Alcohol abuse in pregnancy is probably a significant cause of birth and developmental defects in Australian children.

(Med J Aust 1983; 1: 266-269)

ONLY RELATIVELY RECENTLY (during the past decade) have the teratogenic effects of alcohol been recognized as a significant cause of morbidity and mortality in human infants. Alcohol abuse during pregnancy has been associated with the birth of a baby with a pattern of malformations termed the fetal alcohol syndrome.[1] A range of anomalies is evident in these babies and children, such as central nervous system dysfunction, small-for-dates or low birthweight and subsequent failure to thrive, growth deficiency, a range of birth defects and a distinctive facial appearance.[1,2] The facial appearance is characterized by facial bone hypoplasia, small palpebral fissures, epicanthic folds, underdeveloped philtrum and a thin upper lip. Abuse of alcohol can often be associated with abuse of other drugs, excessive smoking, and malnutrition. However, recent clinical and experimental studies confirm that excessive alcohol consumption is the primary factor which leads to the fetal alcohol syndrome.[2-5]

Patients

Twenty patients with the fetal alcohol syndrome seen at the Royal Alexandra Hospital for Children, Sydney, by one of the writers (A.H.L.) from 1978 to 1981 are described. Of these, 10 were referred by other physicians with the diagnosis suspected or proven, three were detected during clinical surveys of birth defects, and in the remaining seven the diagnosis was ascertained during neonatal and general paediatric practice.

A prerequisite for diagnosis was a history of chronic alcoholism, binge drinking or heavy drinking during the pregnancy. Data on maternal alcohol intake was obtained at an interview with the mother, and from community agencies or local medical practitioners involved in the care of the mother. As children of mothers with untreated phenylketonuria can mimic features of the fetal alcohol syndrome,[6] blood phenylalanine estimations were performed when two or more children in one family were affected.

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Selected case reports

The details of 20 cases are listed in the Table (see pages 268-269). Diagnosis was often delayed when a maternal alcohol problem was not suspected and not connected with the child's anomalies. Consequently, several subspecialities were concerned with the care of these children. The following two case reports are typical of the varied presentation of the children and the unusual and delayed presentation of the mothers' alcohol problem.

CASE 1: A male child, small for gestational age at birth, had hypoplastic nipples, left microphthalmia, a wide metopic suture, and a large ventricular septal defect. Transfer to the Children's Hospital for treatment of the heart lesion was necessary at the age of three days. The mother accompanied the child, and was a resident within the mothers' quarters of the Hospital. Several days later, the ward sister mentioned that the mother was asking other resident mothers to go drinking at a hotel with her at 10 a.m. In a subsequent interview, the mother stated that she had consumed at least six cans of beer a day throughout pregnancy.

CASE 3: A girl with learning problems and temporal lobe epilepsy had been followed up for several years in a general paediatric clinic. Her facial features were noticed only when she was examined by another paediatrician at the age of seven years. She had prominent, underdeveloped ears and facial-bone hypoplasia, particularly of the maxilla. She had been small-for-gestational-age at birth, and current growth parameters placed her at the 10th percentile for height and weight and below the 3rd percentile for head circumference. The mother was questioned about the events of pregnancy, and admitted a beer intake of at least six glasses each afternoon and frequent drunkenness during the first trimester, associated with the break-up of her marriage.

Discussion

A recent government survey identified approximately 250 000 persons in Australia as problem drinkers.[7] A random survey among general practitioners in Sydney showed that, in 1974, less than 5% reported seeing a patient with an alcohol problem.[8] Such low recognition highlights the difficulties of diagnosis of a condition which is imitated by a variety of organic and psychological diseases and in which the patient is often unwilling to assist in diagnosis.[9] Only nine of the 20 mothers (45%) had their alcohol abuse recognized and associated with their babies, anomalies in the first months after birth; in some, the diagnosis of fetal alcohol syndrome in their children was delayed for several years.

A variety of defects is characteristic of teratogens, and alcohol is no exception.[1,2] This variety is probably related to the timing of alcohol abuse, the genetic make-up of the parents and fetus, and maternal-fetal metabolism. For a malformation to occur, alcohol abuse would need to take place during the organogenic period, which is between the 15th and 60th day of gestation. Features of the fetal alcohol syndrome which required medical care and led to recognition of the syndrome were congenital heart disease, which occurred in eight of the children (40%), cleft palate in three (15%), and failure to thrive in four (20%). Similar incidences of congenital heart disease and cleft palate in children with the fetal alcohol syndrome have been reported from the United States.[1,10] The fetal alcohol syndrome can also present as epilepsy (Patient 3) and developmental retardation (Patients 3, 10, 16, and 18). The identification of a child can bring to light affected sibs, as seen in Patients 12, 13, and 14. The fostering of three of the children (Patients 5, 14, and 17) reflects the social disruption in families with alcoholism. In a previous Australian study of six children with the fetal alcohol syndrome, all were fostered or adopted.[11]

Five children lacked the facial features associated with the syndrome, although four of the five were observed in the neonatal period in which the typical facies may not be obvious. An apparently normal facial configuration does not exclude a severe prenatal effect of alcohol, as major brain anomalies in the presence of normal facial features have been reported in the offspring of mothers suffering from chronic alcoholism.[12]

Three of the 20 children (15%) were Aboriginal, and, in another Australian report, two out of seven children with fetal alcohol syndrome had Aboriginal parents.[13] A recent study has indicated that the incidence of fetal alcohol syndrome is high in the American Indian population,[14] and there are similarities between the social and economic circumstances of the American Indians and the Australian Aborigines. Both are fragmented peoples living on the fringe of society, with a high incidence of alcohol abuse and alcohol-related diseases.[15] Studies are needed to define accurately the incidence of the syndrome in Australian Aborigines.

Epidemiological investigations have not indicated clearly either the amount of alcohol needed or over what time-scale its abuse needs to occur to put the fetus at risk. Although 22% to 43% of the newborn of women with chronic alcoholism in various studies are recognized at birth to be abnormal,[2] studies by Hanson,[4] and Majewski,[5] did not indicate a direct relationship between the daily consumption of alcohol and the occurrence or severity of the syndrome. What causes an abnormal outcome of prenatal alcohol abuse has yet to be determined.

The role of binge drinking has not been included in most epidemiological studies, though Hanson, who looked at a group of alcohol drinkers who took more than five drinks at a time, showed that abnormalities could occur.[4] The mothers of Patients 2, 3, and 4 in the study of Clarren *et al.* had brain abnormalities and some features of the fetal alcohol syndrome after intermittent and binge intake.[12] The mothers of Patients 6 and 7 in the study of Herman *et al.* had a binge pattern of drinking in the first trimester of pregnancy and abstention or light drinking in the second and third trimester.[16] In our study, the mothers of five patients (Patients 3, 5, 15, 16, and 18) had binge or heavy drinking confined to the first trimester.

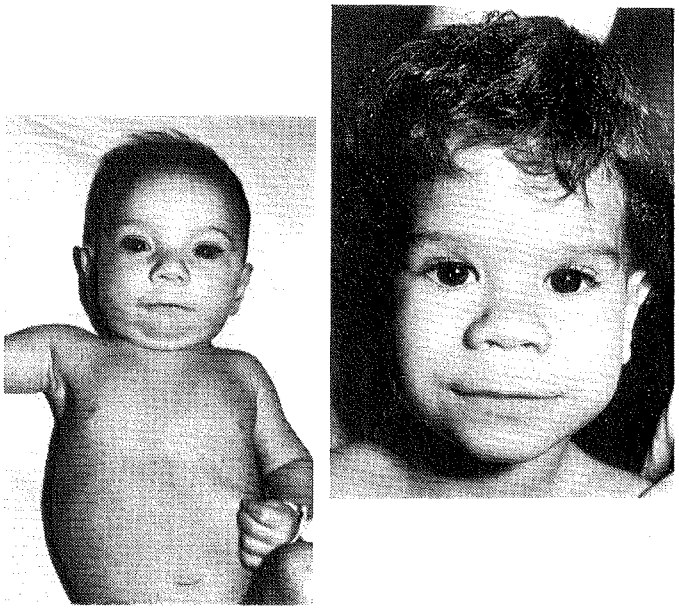

FIGURE 1: Patient 2 (left) and Patient 5 (right), both have the characteristic facial appearance of the fetal alcohol syndrome with an underdeveloped philtrum, maxillary and mandibular hypoplasia, and a small nose with anteverted nostrils and depressed nasal root.

Appreciation of the effect of alcohol abuse in pregnancy can often be delayed when attention is directed solely to a malformation, growth disorder, or central nervous system dysfunction. Recognition is important because of the need of a family with an alcohol problem for special help to identify the child at risk in order to facilitate identification of associated medical and educational problems, and, if abuse continues, to try to prevent further pregnancies.

References

1. Clarren SK, Smith DW. The fetal alcohol syndrome. *N Engl J Med* 1978; 298: 1063-1067.
2. Streissguth AP, Landesman-Dwyer S, Martin JC, Smith DW. Teratogenic effects of alcohol in human and laboratory animals. *Science* 1980; 209: 353-361.
3. Sokol RS, Miller SI, Reed G. Alcohol abuse during pregnancy: an epidemiological study. *Alcoholism (NY)* 1980: 4: 135-145.
4. Hanson JW, Streissguth AP, Smith DW. The effects of moderate alcohol consumption during pregnancy on fetal growth and morphogenesis. *J Pediatr* 1981; 92: 457-461.
5. Majewski F. Alcohol embryopathy. *Neurobehav Toxicol Teratol* 1981; 3: 129-144.
6. Lipson AH, Yu JS, O'Halloran M, Williams RD. Alcohol and phenylketonuria. *Lancet* 1981; 1: 717.
7. Senate Standing Committee on Social Welfare. Drug problems in Australia: an intoxicated society? Canberra: Australian Government Printing Service, 1977.
8. Reynolds I. Alcoholism and drug dependence: a survey of general practitioners' opinions. *Med J Aust* 1975; 1: 167-169.
9. Hore BD. Clinical features and treatment of alcoholism. *Br J Hosp Med* 1977; 18: 106-116.
10. Sandor GS, Smith DF, Macleod PM. Cardiac malformations in fetal alcohol syndrome. *J Paediatr* 1981; 98: 771-773.
11. Collins E, Turner G. Six children affected by maternal alcoholism. *Med J Aust* 1978; 2: 606-608.
12. Clarren SK, Alvord EV, Sunai SM, Streissguth AP. Brain malfunctions related to prenatal exposure to ethanol. *J Pediatr* 1978; 92: 64-67.
13. Walpole IR, Hockey A. Fetal alcohol syndrome: implications to family and society in Australia. *Aust Paediatr J* 1980; 16: 101-105.
14. Smith DF, Sandor GG, Macleod PH, *et al.* Intrinsic defects in the fetal alcohol syndrome. *Neurobehav Toxicol Teratol* 1981; 3: 145-152.
15. Hunt H. Alcoholism among Aboriginal people. *Med J Aust* 1981; 1 (suppl): 1-3.
16. Herman J, Pallister PO, Opitz JM. Tetracetrodactyly and other skeletal manifestations of the fetal alcohol syndrome. *Eur J Pediatr* 1980; 133: 221-226.

TABLE : Clinical features of 20 patients with fetal alcohol syndrome

Patient number and social details	Sex	Unit or clinic of presentation	Birthweight (grams)	Growth	Age at diagnosis	Typical facies	Birth defects	Retarded psychomotor development	Other
1. Mother admitted to drinking at least 170 g/day alcohol as beer during pregnancy	M	Neonatal	2500	NK	2 weeks	–	Ventricular septal defect; microphthalmia (unilateral); hypoplastic nipples	NK	
2. Both parents have alcohol problem	M	General paediatric	2300	HC, HT, WT <3% at 18 months	5 days	+	Cleft palate	+	Second cousin has cleft palate
3. Mother heavy drinker of beer in the first trimester; parents divorced; uncle died from alcoholic cirrhosis	F	General paediatric	2800	HC <3% HT and WT 10%	7 years	+		+	Temporal lobe epilepsy; learning problems
4. Mother suffers from agoraphobia and alcoholism	M	Cleft palate	1500 at 36 wks	HC, HT and WT <3% at 12 months	1 year	+	Cleft palate	+	Investigated because of failure to thrive and developmental delay in the first 12 months of life
5. Aboriginal parents, foster care; mother has alcohol problem	M	Endocrine	1870 at 42 wks	HC, HT and WT <3% at 2 years	2 years	+		+	Referred for growth failure
6. Mother has alcohol problem	M	Cardiac	2500	HC, HT and WT <3% at 8 months	5 days	–	Ventricular septal defect; persistent ductus arteriosus	NK	
7. Mother has alcohol problem	F	General paediatric	2700	HC, HT and WT = 3% at 2 years	2 years	+		+	
8. At least one binge in the first trimester and heavy drinking throughout pregnancy	M	Cardiac	2700 at 42 wks	NK	1 day	+	Tetralogy of Fallot; right coloboma; bulbar palsy; fusion of metopic suture	+	First cousin has hypoplastic aortic arch and ventricular septal defect
9. Mother has alcohol problem	M	Cardiac	2820	HC, HT and WT 3% at 8 months	3 mths	–	Transposition of great arteries	+	
10. Mother has alcohol problem	F	Neonatal	1600 at 32 wks	HC 10%; HT <3%; WT 25% at 22 months	1 day	–	Cleft palate	+	Myopia, cerebral palsy, finger contractures
11. Foster care; Aboriginal mother white father; both parents have alcohol problems	M	Cardiac	2600	WT and HT <3%; HC 3% at 9 months	2 wks	+	Tetralogy of Fallot	+	Pneumococcal meningitis at 3 months
12. One family;	M	Cardiac	2200	HC, HT and WT <3% at 18 months	8 mths	+	Tetralogy of Fallot	+	
13. both parents have alcohol problem;	M	Cardiac	2170	HC, HT and WT <3% at 3 years	2 years	–	Ventricular septal defect	NK	Infant second cousin has "hole in the heart"
14. foster care	M	General paediatric	2040	HC, HT and WT <3% at 4 years	4 years	+	Pulmonary stenosis	+	

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Patient number and social details	Sex	Unit or clinic of presentation	Birthweight (grams)	Growth	Age at diagnosis	Typical facies	Birth defects	Retarded psychomotor development	Other
15. Single mother; alcohol abuse in the first trimester	F	Cardiac	3140	HC, HT and WT <3% at 4 months	1 day	–	Transposition of great arteries; pulmonary stenosis; ASD, VSD, dextrocardia	+	Tube feedings until 3 months of age
16. One binge at approximately seven weeks' gestation — unconscious after wine party	F	Neonatal	2400	HC, HT and WT <3% at death	4 wks	+	Submucous cleft palate; unilateral cataract	+	Died at 4 months of aspiration pneumonia; spastic quadriplegia
17. Part-Aboriginal parents; foster care; mother has alcohol problem	M	Neonatal	2740	HC 10%, HT and WT <3% at 1 year	3 days	+		+	Hypotonia
18. Binge at five weeks' gestation	F	General paediatric	2265 at 36 wks	Corrected HC, WT and HT = 3% at 6 weeks	1 mth	+	Peripheral pulmonary artery stenosis	+	Cerebral palsy; gastro-oesophageal reflux; cerebral atrophy on ultrasound
19. Mother has alcohol problem	F	General paediatric	2300	HC, HT and WT = 3% at 6 months	6 mths	+		+	Hypoplastic toenails; investigated because of failure to thrive before diagnosis
20. Aboriginal parents; mother has alcohol problem	M	Neonatal	2400	HC 10% at birth	1 day	–	Hypoplastic right first metacarpal	NK	Hypoplastic toenails

% = percentile (Percentiles from NHMRC scales, 1972). WT = weight. ASD = atrial septal defect. VSD = ventricular septal defect. HC = head circumference. NK = not known. HT = height.

Looking back . . .

The Medical Journal of Australia
March 21, 1931

PROFESSIONAL SECRECY.

THE basis of the practice of medicine is the confidence of the patient in his physician. The patient cannot expect that the physician will understand the nature of an illness or know what remedy to apply unless he is made acquainted with the whole history of the malady. The wise patient does what he can to marshal the facts for the scrutiny of the physician and he does not demur when the latter finds it necessary to inquire minutely into the patient's mode of life, to ascertain what illnesses he has encountered, to discover, for example, whether he has worshipped at the shrine of Venus or Bacchus and whether the worship has been rewarded by the deities in question with pathological mementoes of the several pilgrimages. The patient would not be content to do these things did he not know that the physician will always lock up the details of the inquisitorial interviews in the "safe and sacred repository" of his memory, in other words, that he will be true to the Hippocratic oath. The clause in the Hippocratic oath relating to this question is as follows:

Whatever in connexion with my professional practice, or not in connexion with it, I may see or hear, I will not divulge, holding that all such things should be kept secret.