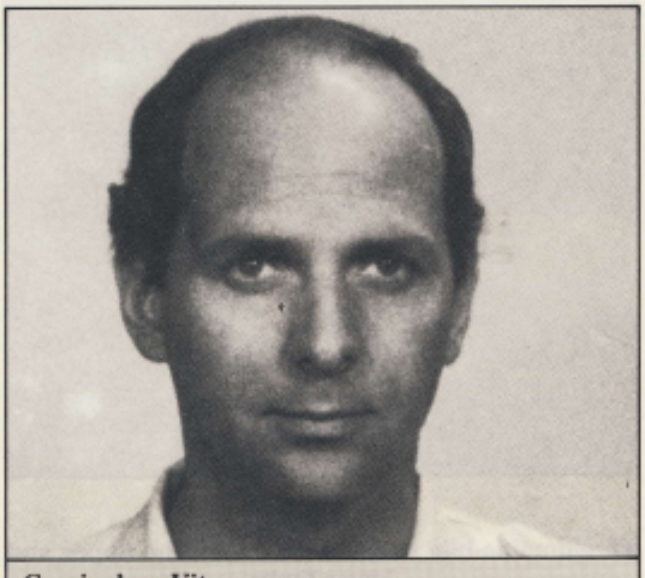
The Fetal Alcohol Syndrome

Part one of a two-part article

Leon J van Rensburg



Curriculum Vitae

Leon Janse van Rensburg kwalifiseer in 1975 aan die Universiteit van die Witwatersrand. Hy voltooi sy internskap aan die Bloemfonteinse Kompleks van Opleidingshospitale. Na voltydse poste in die Departement van Pediatrie en Anestesiologie werk hy by Themba-hospitaal, Witrivier (Tvl). Sedert die einde van 1978 praktiseer hy as huisarts in die Noord-Oos Kaap, Maclear. Gedurende 1978 word hy toegelaat as Gediplomeerde in Anestesiologie (DA) deur die Kollege van Geneeskunde van SA. In 1982 ontvang hy ook medelidmaatskap van die Fakulteit van Algemene Praktisyns (M F A P). Gedurende dieselfde jaar verwerf hy die graad magister in Huisartskunde (M Fam Med) met lof aan die Universiteit van die Oranje Vrystaat.

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Summary

This article is based on a thesis "The Fetal Alcohol Syndrome" which was submitted for the degree of M Fam Med at the University of the Orange Free State, Bloemfontein. Four case studies are described and the literature is critically reviewed with special emphasis on the problems facing primary care physicians.

KEYWORDS: Fetal Alcohol Syndrome; Fetal Growth Retardation: Physicians, Family.

INTRODUCTION

There is agreement that the Fetal Alcohol Syndrome (FAS) is a pattern of malformations observed in the offspring of women who drink alcohol during pregnancy[1,2,3]. Clinical experience and epidemiologic findings suggest a wide range of alcohol-induced effects on the developing embryo and fetus, with the FAS at the severe end of the spectrum[4].

Both clinicians and researchers need to define and agree on minimal criteria for the diagnosis of FAS. The Fetal Alcohol Study Group of the Research Society on

Alcoholism recommended that the diagnosis of FAS be made only when the patient has signs in each of three categories[5].

1. Prenatal and/or postnatal growth retardation (weight, length, and/or head circumference below the 10th percentile when corrected for gestational age).

2. Central nervous system involvement (signs of neurologic abnormality, developmental delay, or intellectual impairment).

3. Characteristic facial dysmorphology with at least two of these three signs:

(a) microcephaly (head circumference below the 3rd percentile);
(b) microophthalmia and/or short palpebral fissures;
(c) poorly developed philtrum, thin upper lip, and flattening of the maxillary area.

Many of these effects have been associated with other risk factors[6,7] that impair fetal growth and in the absence of the full FAS, the most accurate term might be "fetal alcohol effects". Current findings suggest that pre-natal exposure to alcohol is also associated with physical malformations such as joint, limb[8,9,10,11], cardiac[12,13] and renal[14] anomalies. The term alcohol embryopathy is perhaps more descriptive to include the full spectrum of alcohol effects on the fetus.

Mental retardation is the most sensitive and most serious manifestation of maternal alcohol abuse.

Although deformity and reduced size are the most visible aspects of the FAS, mental retardation is now recognised as "the most serious defect and probably the most sensitive manifestation of maternal alcohol abuse"[15,16]. Clarren and Smith[3] stated that maternal abuse of alcohol during gestation appears to be the most frequent known teratogenic cause of mental deficiency in the Western world, and the third most common known cause after Down's Syndrome and neural tube defects.

To maintain an understanding of the clinical perspective of the FAS is a problem facing practicing clinicians, clinical psychologists and research workers in all disciplines. The detrimental effect drinking has on one's offspring have been known for centuries[17], although the FAS was "rediscovered" during the last decade[2].

CASE PRESENTATIONS

Since October 1980, I have encountered and positively diagnosed eight (8) cases of FAS or fetal alcohol effects, that presented in my practice. Only four cases will be described to illustrate the spectrum of adverse effects due to maternal drinking during pregnancy. These cases are from different ethnic groups, with different socio-cultural backgrounds. In all cases the common denominators were maternal alcohol abuse during pregnancy and especially before the mothers knew they were pregnant, also total ignorance of the fact that alcohol is harmful or potentially lethal to unborn babies.

Case 1:

This case report has previously been published[8]. A full-term female infant was born before arrival at the hospital of a 33 year old unbooked Coloured woman, gravida 8, who was known to be an alcoholic. The infant weighed 1 600 g and the head circumference was 28 cm, well below the 10th percentile.

The infant had the following physical abnormalities:

Craniofacial: Short palpebral fissures, low-set retroverted ears, flat philtrum, smooth upper lip and micrognathia. There was a capillary haemangioma over the central forehead and the bridge of the nose.

Skeletal: Bilateral short phocomelia of the upper limbs and bilateral amelia of the lower limbs.

Genital: Hypertrophy of the clitoris and abnormally small labia majors.

FIGURE 1:
Thalidomide-like skeletal defects of the upper and lower limbs

The baby was extremely jittery and had a high-pitched cry. She followed a progressively downhill course and died 20 hours after birth. The mother admitted to heavy drinking throughout the pregnancy, but more so during the first trimester before she knew she was pregnant. Her other children are apparently normal. (I have subsequently examined six of her children and found no anatomical features of FAS. Their school progress seems normal at this stage, but IQ studies have not been done.)

An autopsy was requested and the mother gave informed consent. The examination revealed a patent foramen ovale as the only abnormal anatomical finding. The brain was histologically normal, the lungs showed partial atelectasis and the liver extramedullary haemopoiesis. The abnormalities in this case bear a close resemblance to the wide spectrum of thalidomide-induced deformities in humans.

Maternal abuse of alcohol during gestation appears to be thé known teratogenic cause of mental deficiency in the Western world.

Case 2:
The parents of a 13 year old Black girl from Transkei sought medical attention for the child because of a deformed left arm. The child had the following physical features:

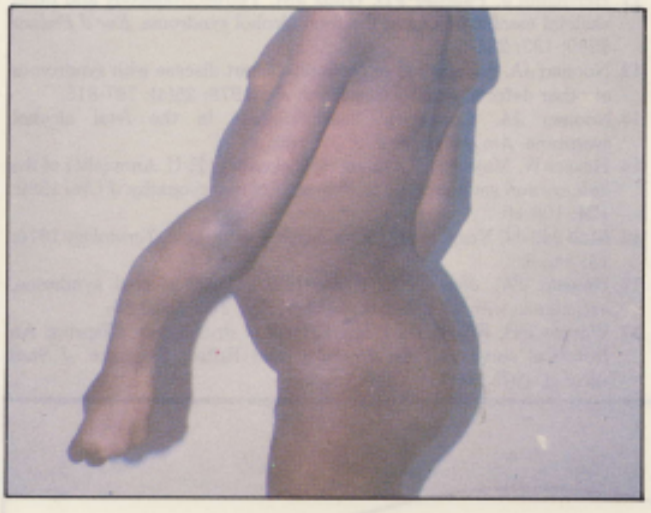

FIGURE 2:
Photograph showing smooth upper lip and underdeveloped philtrum, low-set ears and bow-shaped, deformed left forearm

1. Smooth upper lip and underdeveloped philtrum;
2. Low-set retroverted ears;
3. A bow-shaped, deformed left forearm.

Radiographic investigation revealed:

(i) underdevelopment of the left distal ulna;
(ii) dislocation of the left radial head;
(iii) fusion of the right capitate and hamate bones (described by Jaffer et al[2]).

In the differential diagnosis the following conditions were considered:

(i) Holt-Oram Syndrome
(ii) Ehlers-Danlos Syndrome
(iii) Fong Syndrome.

The mother was not an alcoholic per se, but admitted to the drinking of traditional alcoholic beverages throughout pregnancy, especially before she knew she was pregnant. This child was referred to a specialist centre and lost in follow up.

Case 3:
A female infant was born in hospital with the following physical features:

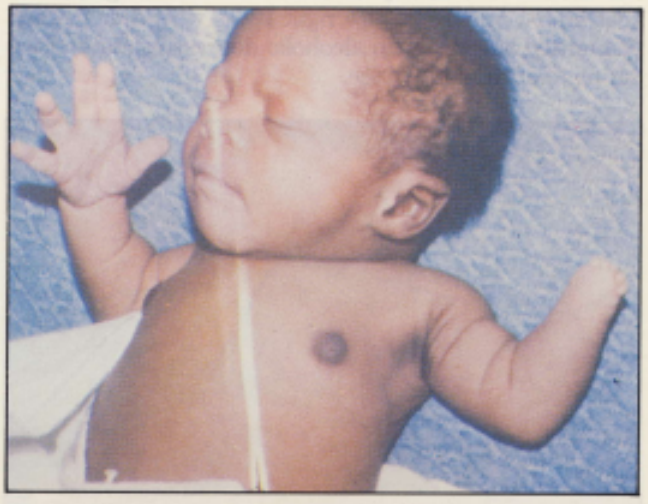

FIGURE 3
Photograph showing smooth upper lip, underdeveloped philtrum and hemimelia of the left arm with rudimentary finger buds.

(i) The infant weighed 2,8kg;
(ii) head circumference was 39,5 cm;
(iii) Smooth upper lip and underdeveloped philtrum;
(iv) Low-set, retroverted ears;
(v) Hemimelia of the left arm, with rudimentary finger buds. The radiograph showed complete agenesis of the carpal, metacarpal and phalangeal bones. (Unfortunately the radiograph got lost.)

The mother admitted to the drinking of traditional Xhosa beer throughout pregnancy, but more on occasions such as weddings, funerals or feasts.

The child was taken home to Transkei and reported dead at the age of 13 months.

Case 4:
This 11 year old girl and her family are well known to me. The patient was initially brought to me for recurrent tonsillitis. During one such visit, the mother, a known and previously rehabilitated alcoholic, ventured the question as to the aetiology of the patient's deformed left hand.

On examination the patient had:

(i) a wrist-drop deformity of her left hand (Figure 4);
(ii) shortening of the left index and middle fingers
(iii) radiographic examination revealed a decrease in length of the metacarpal bones, and absent epiphyses at the proximal interphalangeal joints of the left index and middle fingers.

The mother admitted to constant drinking during pregnancy, with episodes of intermittant binge drinking.

I did not tell the patient the diagnosis and mentioned the possibility of alcohol effects to the mother. A notable feature was the intense guilt feelings that the

mother suffered, at the thought that she could have been responsible for her child's disability.

I considered it not to be in the interest of the patient, her mother and family to know the true diagnosis. A year later I found out how right I was when her mother committed suicide in an alcoholic depressive state, after yet another drinking binge.

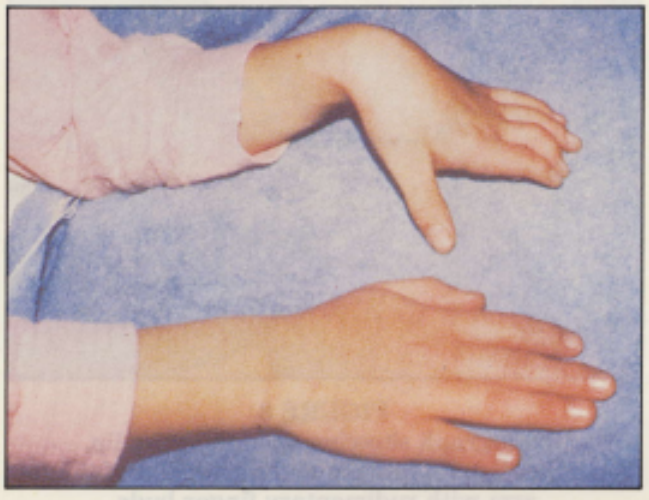

FIGURE 4
Wrist-drop deformity of the left hand

Part two of this article will appear in the next issue.

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