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Syndrome of Ankylosis, Facial Anomalies and Pulmonary Hypoplasia

Prof. Ashfaq Alam Khan, MBBS., FRCP., FRSH., DCH., DTM. & H., C.P.H.,
Head, Department of Paedriatics & Child Health.
University Teaching Hospital, Lusaka, Zambia.

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SUMMARY

A newly recognized syndrome, with a pattern of malformations (fixation of joints, facial anomalies, and pulmonary hypoplasia) is reported from Zambia Central Africa in an African child. Additional features of deafness and cataract are included in the congenital abnormalities and it is suggested that a form of genetic abnormality is responsible for this condition.

INTRODUCTION

This rare syndrome was first described by Pena and Shokeir in 1964 but has never been reported from Africa. Until recently only 4 cases have been recorded in America. Punnet and colleagues (1974) reviewing the literature on this syndrome were unable to define its aetiology, but found that it was closely related to Potter's syndrome which is a rare congenital malformation with isolated hypoplasia of the lungs together with renal agenesis. In view of the rarity of this condition, and the interesting spectrum of additional malformations it was considered worthwhile to record this case – the first from Africa.

The Syndrome

It occurs in the first born, and the affected infant shows compodactyly, ankylosis, facial anomalies and pulmonary hypoplasia.

CASE REPORT

E.M. an African male infant was born to a 25 year old gravida 1 mother and a 30 year old father, both of whom enjoyed normal health. Delivery was uncomplicated, at full term. Details of cord length and weight of placenta were not available. The birth weight was 2000G and the child was nursed in the Premature Nursery for 18 days and discharged with comments about multiple physical deformities. The child was admitted to the Paediatric Unit at six months of age because of a weeks cough, abdominal distension and, constipation. On clinical examination the child was found to have a host of congenital abnormalities and a detailed examination showed the following features.

The child weighed 3.8 Kgs, length 62.2cms, head circumference 41cms. He was rather sluggish and fed poorly. The cranial sutures and fontanelles were closed. There was microphthalmia, and hypertelorism. In addition he had a small mouth with depression of the bridge of the nose. The ears were low set and hypoplastic. There were clubbed feet, and fixed flexion of terminal and interphalangeal

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FIG. I

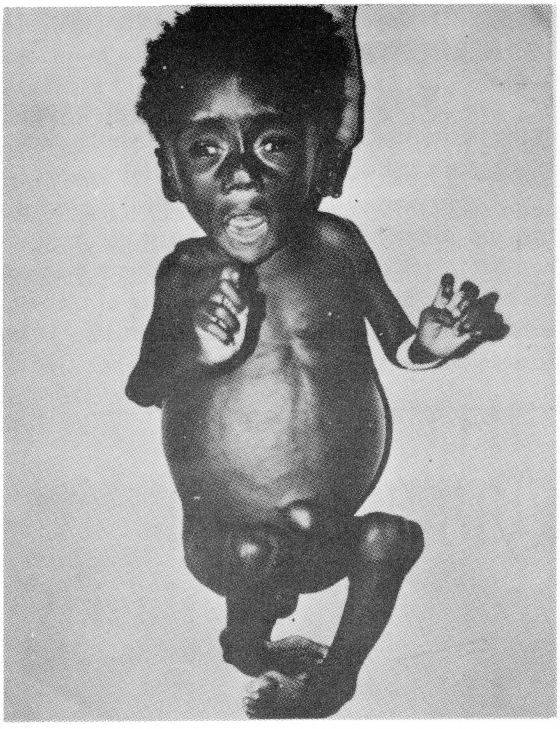


FIG. II

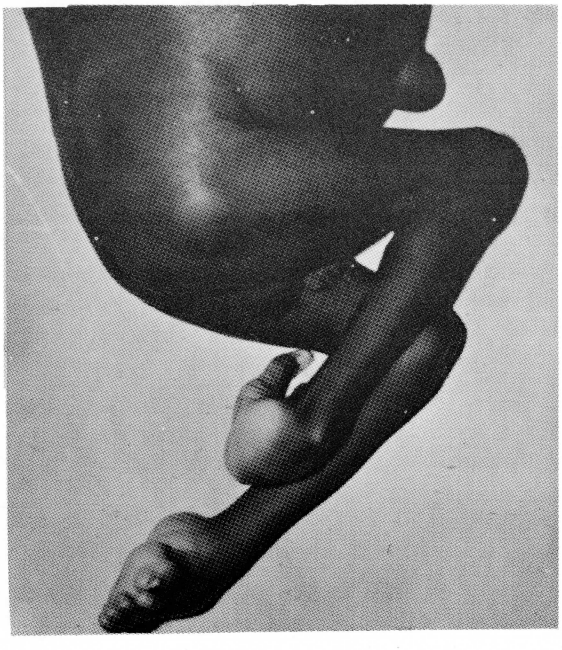
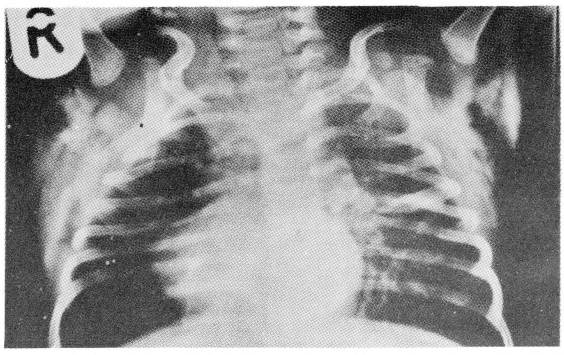


FIG. III



joints. Further there was a fixed flexion of the elbow, the wrist, the hip, the knee and the ankle joints. Clinically there was no abnormality of the cardiovascular system.

The child was dyspnoeic, and the thoracic cage was small in size, showing poor movement and lack of air entry on the right side. The liver was 1cms. The abdomen was thin and distended with visible peristalsis. The bowel sounds were present. The testes were in the scrotum and the kidneys were not palpable. The child was deaf. The eyes had bilateral cataract.

Investigations

X-ray Chest:	Hypoplastic R. lung.
	– Hypoplastic R. Pulmonary vessels
I.V.P.	– N.A.D.
E.C.G.	– Right axis deviation with R.V. preponderance.

A comparison of clinical defects in the 4 reported cases and the present one is shown in tabular form in Table I.

DISCUSSION

The major clinical abnormalities found in this child are remarkably similar to those reported by Pena & Shokeir (1974) and Hope, and associates (1974). The latter agreed that this pattern of malformations (fixation of joints, facial abnormalities, and pulmonary hypoplasia) represented a new syndrome.

Hypoplasia of the lungs had been seen more often but in a review of 38 cases, Oyamada et al (1953) found only four cases with any abnormality of the extremities, and Winberg (1974) found no instance of this combination of anomalies among the 31 infants with pulmonary hypoplasia born in Sweden during the period 1965-1971. Punnet and associates (1974)

TABLE I
Clinical Features

	Pena & Shokeir		Punnet et al		Alam Khan
	I	II	I	II	
SEX	F	F	M	M	M
Intrauterine growth retardation	+	+	+	+	+
FACIES					
Low set malformed	+	+	+	+	+
Hyperteleorism	+	+	+	+	+
Epicanthal folds			+	+	+
Small mandible	+	+	+	+	+
Depressed tip of toe	+	+	+	+	+
EXTREMITIES					
Arthrogryposis	+	+	+	+	+
Club feet	+	+	+	+	+
Camptodactyly	+	+	+	+	–
OTHERS					
Hypoplastic lung	+	+	+	+	+
Undescended testis			+	+	–
Hypoplastic pulmonary vessels	–	–	–	–	+
Deafness	–	–	–	–	+
Cataract	–	–	–	–	+

commented that unusual facies and flexion deformities are also a feature of Trisomy 18, and that perhaps some infants reported as clinical trisomy 18 with normal chromosomes had this syndrome.

The full spectrum of this syndrome, and the aetiology are still not clear, but most of the patients are the first born. The reporting of this syndrome outside the United States for the first time, may suggest involvement of some genetic factor.

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Tuberculous Arachnoiditis

—A CASE REPORT FROM CENTRAL AFRICA AND A BRIEF REVIEW OF THE DISEASE

Ben Umerah, M.B., B.Ch, BAO, D.M.R.D., F.R.C.R.
J. Singarayar M.B., B.S., M.R.C.P., From University Teaching Hospital, LUSAKA, ZAMBIA.

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INTRODUCTION

In most parts of Africa, where tuberculosis is endemic, tuberculous arachnoiditis remains a rare disease. This case is presented to illustrate the polymorphous course of the disease which probably due to its rarity is very scantily described in standard textbooks.

CASE REPORT

A young Zambian African female aged 21 years presented in a peripheral hospital with a history of acute and sudden onset of headache, three days prior to hospitalisation. The headache was accompanied by twitching of the right side of the face and vomiting. On examination she was found to have neck stiffness and flaccid weakness of the lower limbs. The cerebro spinal fluid was xanthochromic and protein content was 200mg%. A provisional diagnosis of subarachnoid haemorrhage was made and the patient was transferred to the University Teaching Hospital Lusaka, two days later. On arrival she was drowsy, disorientated and had marked neck rigidity and flaccid paralysis of both lower limbs. There were no signs of cerebellar or cranial nerve involvement and the sensory system as well as funduscopy were normal. The cerebrospinal fluid was still xanthochromic; proteins were now 4g%, sugar 48mg% and white blood cells 8/cu. mm.