FANCONI SYNDROM
A CASE REPORT

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Introduction

The complex nature of this hereditary disease is best illustrated by its description under a variety of different names. These include lignac-Fanconi disease, the syndrome of de Toni, Fanconi and Debre, cystine rickets, cystinosis, cystine storage disease with aminoaciduria, amino acid diabetes (1). The disease, usually encountered in infants and young children, is characterized by failure to thrive, rickets, glycosuria, generalized aminoaciduria, polyuria hypophosphataemia, protein uria and often electrolyte disturbances with hypokalaemia (2). A generalized defect in the renal reabsorption mechanism is believed to be the underlying cause of these abnormalities, (3,4,5). The available evidence is in favour of a recessive mode of inheritance and according to the data of shih (6) the incidence of this syndrome is 1 : 245 000.

In this article is presented, what we believe to be, the first case report of Fanconi Syndrome in the Sudanese medical literature.

The Case :

A.H.K., is a nine months old boy from the west of the Sudan. He was the product of the sixth pregnancy, the first of which terminated in still birth. Two of his sisters died at the age of 2-3 years of acute gastroenteritis without showing any notable abnormality prior to the attack of gastroenteritis. When the child was six months old his mother noticed that he passed a sticky urine with a sweet taste. Few months later she took him to a local hospital where the presence of sugar in his urine was confirmed. At this point they were adviced to proceed to Khartoum and on arrival the child was admitted to Soba University Hospital and further investigated.

Clinical findings :

On admission the child weighed 7.5 Kg and generally he looked well and cheerful (Fig I). He was not anaemic (Hb 13.5). The only positive finding on examination was thickening of both wrists (Fig. II) His skull was normal and there was no evidence of rachitic rosary or Harrison's groove. Further laboratory investigations were then carried out.

esults and discussion :

The results of the laboratory investigations are presented in the two tables shown and in (Fig. III). It can be concluded from the results in table I that the patient is not diabetic since the changes in his blood glucose level are within the normal range. The remarkable finding, however, is the heavy glucosuria which was evident even before the load of glucose was given. It is worthy to note that in spite of this heavy glucosuria, ketone bodies could not be petected in any of the urine samples. Since in juvenile diabetics ketosis is a common complication, this finding would further exclude diabetes. In addition to the glucosuria there was a dramatic increase in the urinary excretion of amino acids (Fig. III) This aminoaciduria was of a generalized type. Protein was also detected in urine; a total of 825 mg. was excreted in one day. There was notable increase in the daily excretion of inorganic phosphate (8mg per Kg body weight per day).

With the exception of uric acid and inorganic phosphate which are decreasedand alkaline phosphatase which is increased, all the results presented in table 2 are within the normal range. The increased alkaline phosphatase activity is due to the rachitic bone changes which are known to occur in this condition (Fig IV). The fall in the level of uric acid and inorganic phosphate in serum is a manifestation of the renal tubular defect which leads to their wash out in urine.

We can conclude from these results that the diagnosis of Fanconi synddrome is fully established in this case.

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TABLE (1)

Changes in the level of glucose in blood and urine after the adminstration of 15 grammes glucose by a gastric tube after an over night fast. Blood and urine were analysed for glucose at the time intervals shown. The concentration is expressed in mgm ber 100 ml blood or urine. Glucose was determined by the glucose oxidase method.

Time (Minutes)	0	30	60	120
Blood glucose	80	—	137	103
Urinary glucose	227	343	571	—

TABLE (2)

Other relevant biochemical parameters investigated for plasma proteins, the results are expressed in grammes per 100 ml. serum. The units for alkaline phosphatase are King Armstrong Units. All the others are expressed in mgm . per 100 ml serum.

Total proteins	7.00
Albumen	3.8
Urea	24.00
Uric acid	1.8
Inorganic phosphate	2.3
Cholesterol	198.0
Alkaline phosphase	81.0

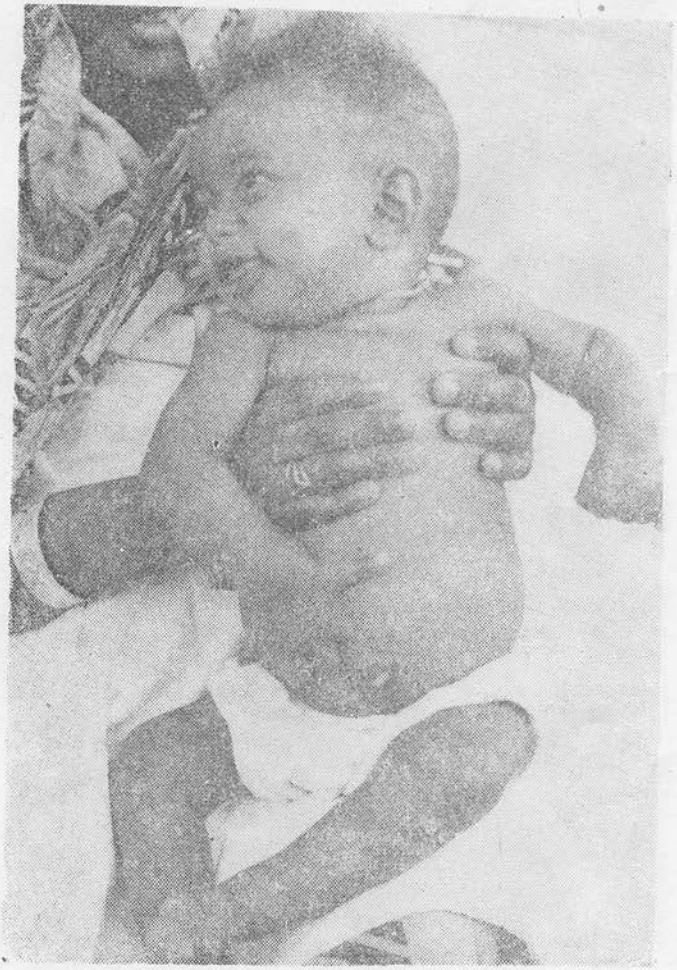

Fig. 1 Photograph showing the general appearance of the baby.

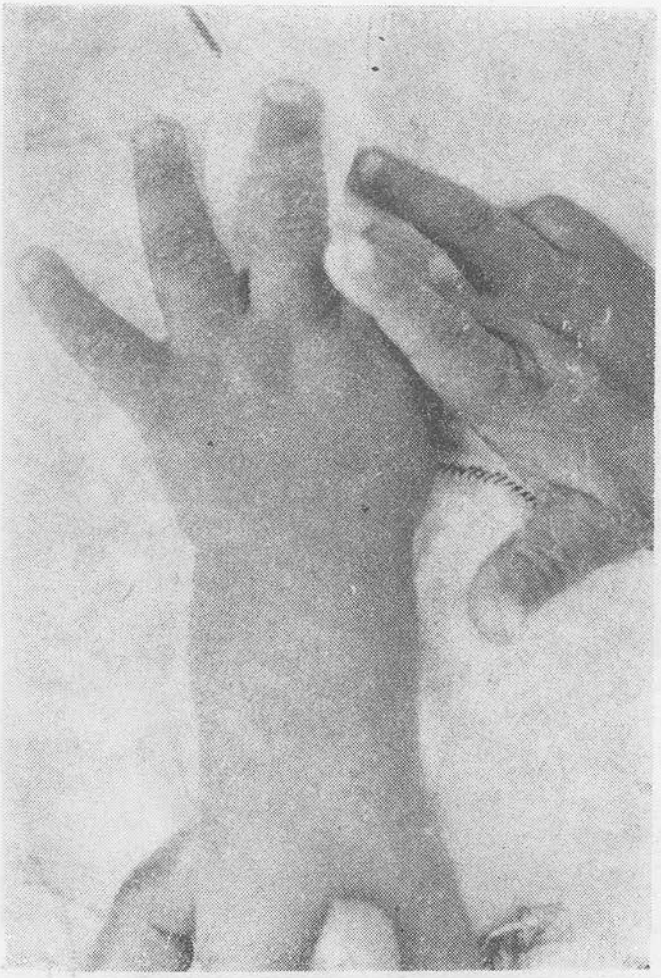

Fig. II Photograph showing widening of the child's left wrist.

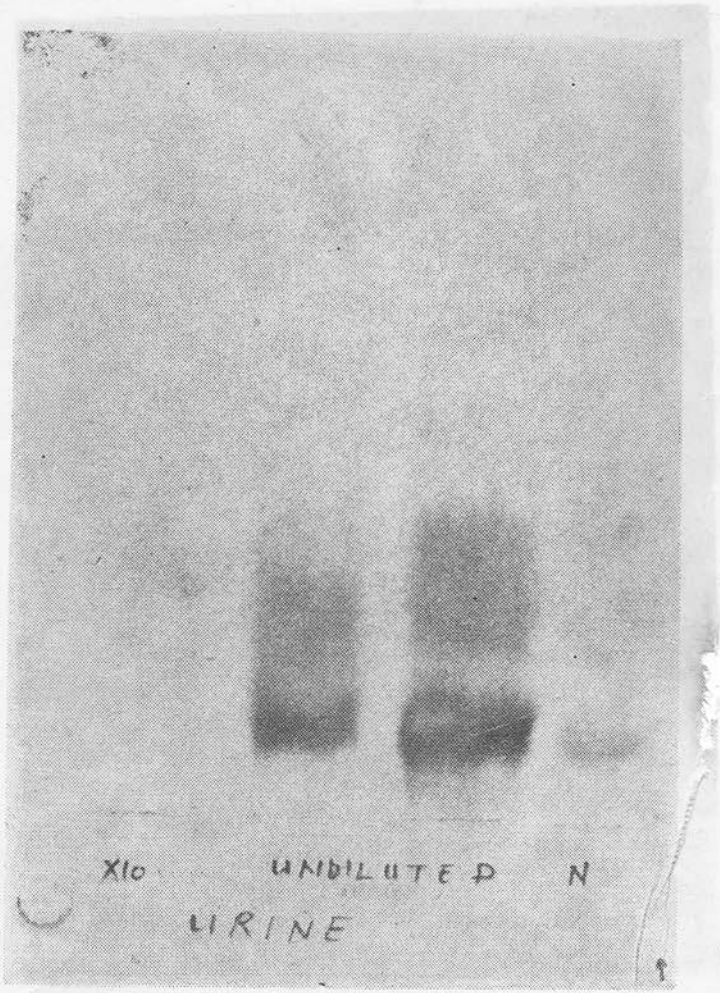

Fig. III Excreti on of amino acids in urine as revealed by paper chromatography. On the far right (N) is shown the pattern of amion acid excretion in a normal subject. The central two positions show the pattren of excretion in the undiluted urine of the child, On the far left the result obtained after diluting the infant's urine 10 times.

Fig. IV : X-ray appearance of the wrists showing some degree of rarefaction together with widening of the epiphy sial lines and cupping of diaphysical ends.

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