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Retardation of Osseous Maturation in Noonan's Syndrome

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Summary

Jaiyesimi, F. (1976). *Nigerian Journal of Paediatrics*, **3** (1), 33. **Retardation of Osseous Maturation in Noonan's Syndrome.** A case of Noonan's syndrome with retardation of osseous maturation is reported. The probable mechanism of osseous retardation in the patient is discussed, and mention is made of the nosological implication in terms of the close similarity with Turner's syndrome.

THE eponym, Noonan's syndrome, is used to describe patients with Turner's phenotype (Turner, 1938) but normal sex karyotype. The cardinal features of the syndrome are small stature, valvular pulmonary stenosis, hypertelorism, mental retardation, ptosis, undescended testes in males and skeletal anomalies (Noonan, 1968). The main difference between Noonan's and Turner's syndromes have been reviewed recently (BMJ, 1974). This communication concerns a case of Noonan's syndrome with retardation of osseous maturation, which, to our knowledge, has not been reported previously.

Case Report

A 3-year old Nigerian male child was referred to the University College Hospital (UCH), Ibadan, because of a chest deformity which was noticed at birth. Both the pregnancy and the delivery were normal, but he was said to be very small at birth. The developmental milestones were normal except for a delay in acquiring speech. His entire vocabulary at the age of three years comprised only two words. He was the fifth child of his parents. All the other members of the family were normal.

Physical examination revealed a fairly well-nourished child with short stature. The height (78cm) was below the 5th per centile for children of similar social class and age (Janes, 1970). There was moderate dolicocephaly and microcephaly, the head circumference being 45cm. His growth velocity, as measured by the increase in his height and weight over a period of $2\frac{1}{2}$ years was, however, normal. He had bilateral ptosis, hypertelorism, epicanthic folds and webbing of the neck (Fig. 1). The nipples were widely separated. There was pectus excavatum. Clinical assessment of his cardiovascular system revealed normal peripheral pulses but a moderate left parasternal heave. The first heart sound was normal, the pulmonary closure sound was soft and there was a harsh pulmonary ejection murmur. His blood pressure was normal. There was no neurological deficit except for the speech retardation and bilateral ptosis.

The haematocrit and white blood cell count were normal; the haemoglobin genotype was A. The serum proteins and cholesterol were 6.9gm.

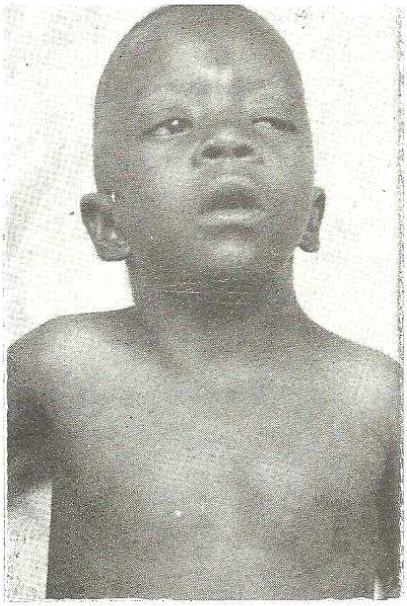

Fig. 1. A 3-year old Nigerian male child with Noonan's syndrome. Note the dolicocephaly, bilateral ptosis, hypertelorism, and webbing of the neck.

and 105mg per 100ml respectively. The serum calcium was 9.5mg/100ml, phosphate 4.0mg, and alkaline phosphatase was 13 K. A. units. The resin triiodothyronine uptake was normal (97 per cent). Chromosome studies revealed a normal 46-XY karyotype. There was electrocardiographic evidence of moderate right ventricular hypertrophy. Chest radiograph showed slight cardiomegaly and normal pulmonary vasculature. Radiographs of the wrists showed complete absence of the carpal ossific centres and demineralization of the metacarpals (Fig. 2).

The diagnosis of Noonan's syndrome was based on the characteristic phenotypical features, clinical, radiologic and electrocardiographic evidence of pulmonary stenosis, and normal sex karyotype.

Comments

Assessment of osseous maturation in Noonan's syndrome has been mentioned in only one previous report (Heller, 1965), in which the bone age was stated to be normal in three out of five patients; the 4th patient had advanced bone age, and bone age was not estimated in the 5th patient. In our patient, there was evidence of severe retardation of osseous maturation. Although the ossific centre in the head of the humerus was present, the centres in the carpal bones (hamate and capitate), which normally appear during the 4th and 6th month of life respectively (Wilkins, 1965), were absent. The bone age in the present case was about three months, contrasting markedly with his chronological age of three years.

The pathogenesis of the osseous retardation in the present case is not clear. Retardation of osseous maturation is common in hypothyroidism and hypopituitarism. There was, however, no clinical or biochemical evidence of hypothyroidism in our patient and the normal growth velocity excluded a deficiency of growth hormone. There was no evidence of any disturbance of calcium metabolism either and the serum proteins were normal. The possibility also exists that the delay in osseous maturation is due to a

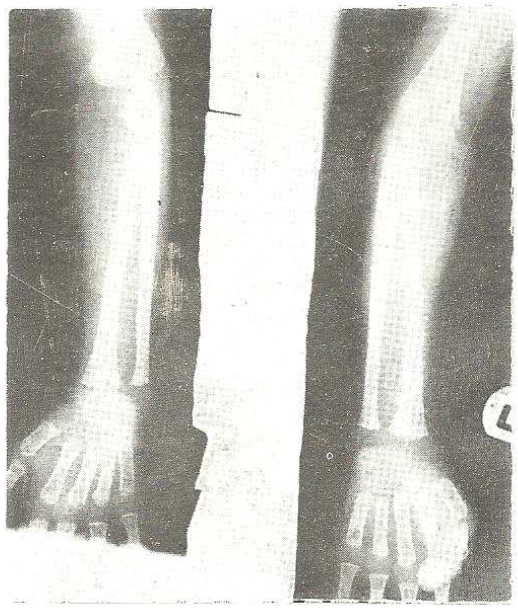

Fig. 2. X-ray of the wrists of patient. Note complete absence of carpal ossific centres and demineralization of the metacarpals.

genetically controlled disturbance of bone maturation. Similar, though less severe, osseous lesions have been reported in patients with Turner's syndrome (Lemli and Smith, 1963). The present case therefore further illustrates the close similarity between Turner's and Noonan's syndromes.

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