NAIL PATELLA SYNDROME

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A 14-year-old male having nail-patella syndrome, manifested as deficient nails on the ulnar aspect of thumbs, V-shaped half-moons, rudimentry patella on right side and absence on left side. X-ray of pelvis showed iliac horns. Family history was suggestive of autosomal dominant type of inheritence.

Key words : Nail-patella syndrome.

This syndrome characterized by a defect in the mesodermal and ectodermal structures, is manifested by absence or hypoplasia of the nails and patella, iliac horns, abnormalities of the elbows and in a minority of the cases with renal changes.[1] The nails are grossly defective, usually reduced in size, and never reach the finger-tips. Ulnar half is more frequently involved. Finger nails are more commonly involved, the severity of changes diminishing from the thumb to the little finger. Some nails may show a V-shaped half-moon.[2-4] Abnormalities of the patella can be either a reduction in the size or complete absence, or occasionally dislocation. Iliac horns are present in the pelvis, arising from the centre of the external aspect of ilium. Elbow joints may show limited supination and incomplete extension. Other features reported include, thickened scapulae, absent skin creases on distal fingers, webbing of the elbows, heterochromia of the iris and cataract. Rare features reported include club foot, spina bifida and plummer-vinson syndrome.[3,5,6] The disease is transmitted as an autosomal dominant trait with linkage between the loci controlling the gene and that of ABO blood group.[7]

Case Report

A 14-year-old male had deficient nails over both his thumbs on their ulnar aspect (Fig. 1).

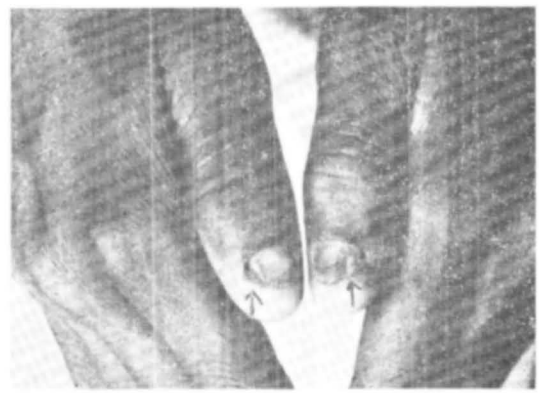

Fig. 1. Defective nails over both thumbs on their ulnar aspect.

The half-moon was triangular in almost all the finger nails. The rest of the finger nails and toe nails were normal. Iliac horns were palpable on both sides, on the posterior aspect of the iliac crest in its middle one third. Left patella was absent while the right patella was rudimentry. Patient was having genu valgum. There was no other associated abnormality. Family history was suggestive of autosomal dominant type of inheritence.

X-rays of the pelvis showed iliac horns (Fig. 2), and that of knee joints; rudimentry patella on right side and absence on left side (Fig. 3).

X-rays of elbow joints and hands showed no abnormality. Blood group was B (Rh+). Haematological examination was normal. Urine examination showed no abnormality. Fundus examination was normal.

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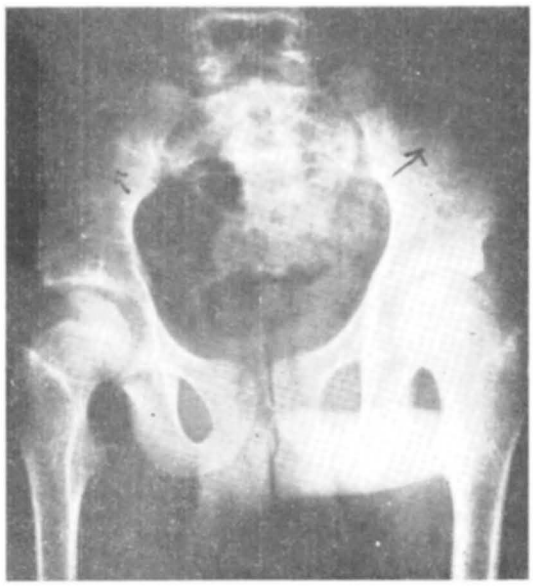

Fig. 2. X-ray of pelvis showing iliac horns.

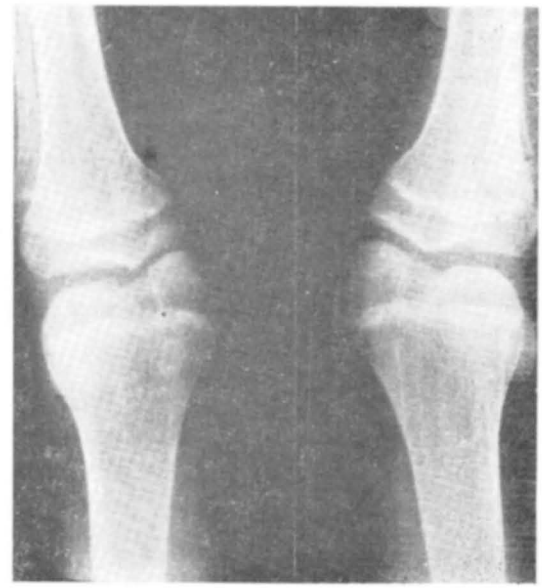

Fig. 3. X-ray of both knee joints showing rudimentary patella on right side and absence of patella on left side.

Comments

Chelatin[8] in 1820 observed a patient with a triad of abnormal nails, elbows and knees. In 1897, Little[9] quoted a description by Sedgwick of a family of which 18 members of 4 generations had no thumb nails and no patella, thus suggesting the hereditary nature of this disorder. Involvement of elbows in this hereditary defect was reported by Wrede.[10] Turner[11] observed flaring of the iliac crests and prominence of the anterior superior iliac spines in some of the affected patients. Fong[12] termed these prominences as iliac horns; however he did not associate them with any syndrome. After a few years, iliac horns were observed in association with knee, elbow and nail anomalies by other authors.[7,13] Thus, iliac horns were established as an important constituent of this syndrome. The popular name of nail patella syndrome has been applied to this triad of anomalies, but Love and Beiler[14] coined the more correct term of hereditary osteo-onychodysplasia.

It is inherited as an autosomal dominant trait with linkage between the loci controlling the gene and that of ABO blood group.[7] A family of five generations with 72 members, of whom 22 showed features of this syndrome, was reported in 1966 by Maini and Mittal.[15] Our case had typical nail changes and absent or rudimentary patella but no other abnormality.

References

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