ONYCHO-OSTEODYSPLASIA—THE NAIL-PATELLA SYNDROME

A REPORT OF TWO CASES

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SYNOPSIS

Hereditary onycho-osteodysplasia is characterised by absent or defective nails, dislocated radial heads, absent or hypoplastic patellae and iliac horns.

Two new cases showing typical features are presented. In one, a history was given though not corroborated, of the disease having affected several members of the family. In the other case, the immediate relatives were found to be free of the disease and the patient was presumably in receipt of a spontaneous mutant from one of his parents. This condition was discovered in both instances as a result of recognition of the iliac horns on routine radiography.

Onycho-osteodysplasia is a clinical entity characterised by absent or defective nails, dislocation of radial heads, absent or hypoplastic patellae and the presence of iliac horns. This condition, popularly referred to as the nail-patella syndrome has been known for a long time.

Little (1897) cited a report by Sedgwick of a family in which 18 members in four generations lacked thumb nails and patellae, thus suggesting that such ectodermal and mesodermal defects could be hereditary. Wrede, in 1909, first recorded the coexistence of elbow deformity with malformation of the nails and knees. Detailed study of this triad of defects was subsequently made by Osterreicher (1931) and Turner (1933). Fong (1946) noticed the occurrence of conical bony projections arising from the posterior surface of the iliac bones and called these structures iliac horns. A few years later, Mino *et al* (1948) and Thompson *et al* (1949) independently showed that iliac horns were, in fact, an integral feature of the nail-patella syndrome. Since then, many other subsidiary defects have been described as occurring in association with the main tetrad of lesions. A comprehensive review of the whole range of anomalies associated with onycho-osteodysplasia was given by Duncan and Souter (1963).

Genetic studies suggested that the disease was transmitted as a non-sex-linked dominant inheritance and an extensive survey by Renwick and Lawler (1955) and Lawler *et al* (1957) of nine large families showed linkage between the gene responsible for the disease and that of the ABO blood groups.

The first local report of onycho-osteodysplasia in a Chinese family was made by Pillay (1965). He noted two new features of the disease, namely, delayed ossification and shortening of quadriceps. Of special interest was the fact that this condition affected two sibs, the parents of whom showed no signs of the disease and he ascribed this phenomenon to spontaneous mutation in the gonads of one parent.

CASE REPORTS

Two new cases of onycho-osteodysplasia seen locally are presented. The diagnosis was made in both cases from routine abdomen X-rays in which iliac horns were recognised.

Case 1

M.C., an 84 year old female Eurasian, was extremely conscious of her nail and elbow deformity and gave a detailed account of the disease having afflicted several members of her family for six generations. Her history, however, could not be corroborated as her affected mother and grandmother were deceased and the few living relatives who exhibited features of the disease declined to come up for medical examination. This patient was found to have abnormal nails and dysplasia of the elbows, knees and pelvis.

Hands and Feet: The thumbs (Fig. 1) showed almost complete absence of nails with the terminal pulp extending on to the dorsal surface, distal to the dystrophic nail. The nail of the left index finger was less severely affected while the rest of the finger nails were spared. The dorsal skin creases over the distal inter-phalangeal joints

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were absent or poorly formed. No laxity of the ligaments of the metacarpo-phalangeal and inter-phalangeal joints was noted. Radiographs of the hands showed that on both sides the second metacarpal was longer than the third. There was no abnormality of the toe nails and digits of the feet.

Elbows: Both elbows showed an increased carrying angle and anterior webbing was evident on the right side (Fig. 2). Pronation and supination were severely restricted but only minor limitation of flexion and extension was detected. Radiographs revealed that on both sides, the capitellum and lateral epicondyle of the humerus were hypoplastic and the medial epicondyles markedly prominent. Posterior dislocation of the heads of each radius was also noted associated with hypoplasia of head and elongation of neck (Fig. 3).

Knees: The patellae were normal in size but showed recurrent dislocation with knees fully flexed (Fig. 4). The medial femoral condyles were large while the lateral condyles were relatively hypoplastic. The changes were not sufficiently pronounced to cause genu valgum.

Pelvis: Large iliac horns were present and were readily palpable through the gluteal muscles. The iliac crests were also flared out with prominence of the anterior superior iliac spines (Fig. 5).

Case 2

C.W.O., a 31 year old male Chinese, exhibited minor nail and knee dysplasia but had characteristic features of the disease in the elbows and pelvis. Clinical examination showed that his parents, brothers and sisters were normal. Also, no signs of any nail or bone defect were apparent in the immediate relatives. It is probable that this condition arose as a result of spontaneous mutation.

Hands and Feet: The nails of the hands were normal in size but the thumb nails showed a longitudinal groove on the radial side. The eponychium of both thumbs and right middle finger was incompletely formed (Figs. 6 and 7). X-ray studies revealed long second metacarpals on both sides. Minor nail dystrophy was encountered in the right 3rd, 4th and 5th digits and left 2nd and 4th digits of the feet.

Elbows: Dysplasic changes were found in both elbows resembling those in Case 1 (Fig. 8). No webbing was present and the radial heads were laterally dislocated. Severe crepitus was also elicited on movement of the joints.

Knees: The patellae were not hypoplastic but were slightly high in position. Mild hypoplasia of the lateral femoral condyles was evident.

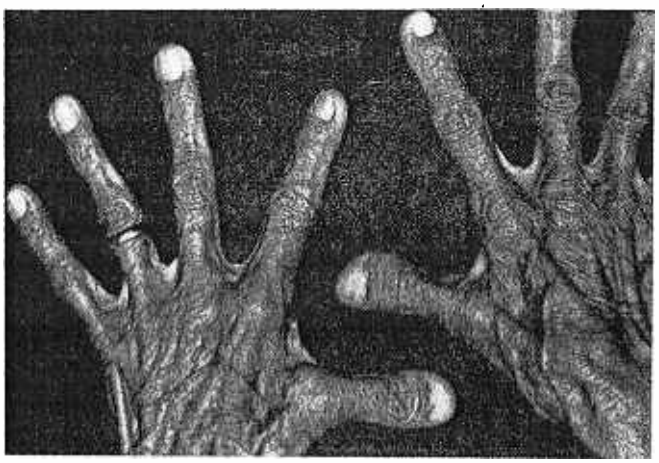


Fig. 1. Case 1: The thumbs show almost complete absence of the nails with dorsal overgrowth of the digital pulp. The nail of the left index finger is less severely affected. Skin creases over the distal inter-phalangeal joints are poorly formed.

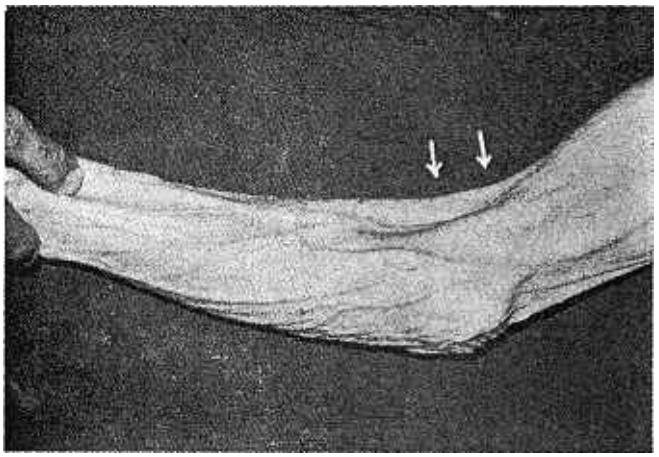


Fig. 2. Case 1: Anterior webbing of the right elbow is shown (arrowed).

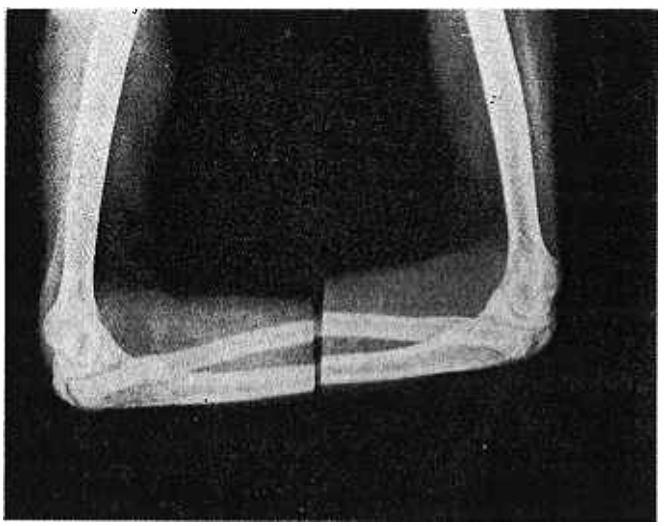
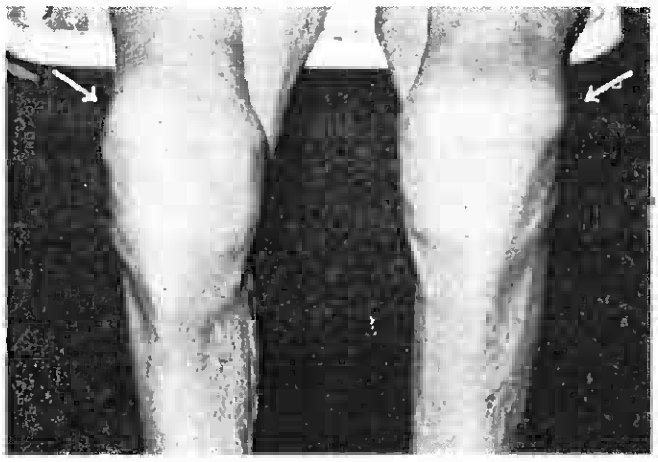


Fig. 3. Case 1: Lateral X-ray view of the elbows showing that the radial heads are hypoplastic and posteriorly dislocated and the necks somewhat elongated.



F 4. Case 1: Flexion of the knees causes lateral and upward dislocation of the patellae (as arrowed).

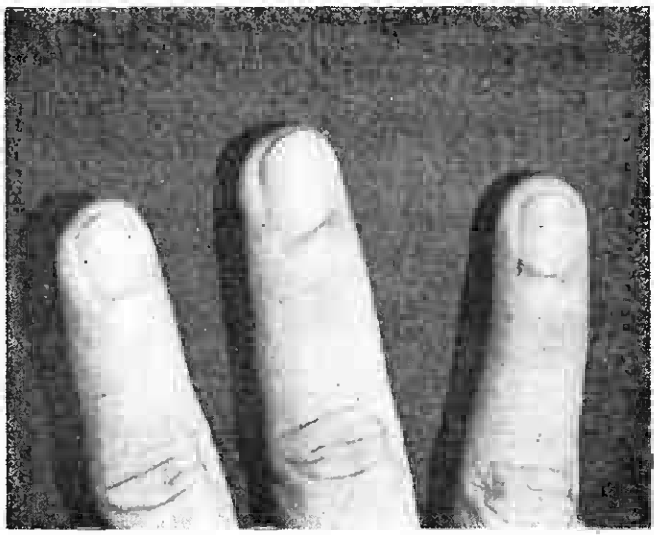


Fig. 7. Case 2: Right index, middle and ring fingers showing normal nail formation but deficiency of the eponychium of the middle finger. Note the absence of the normal skin creases over the distal inter-phalangeal joints of the index and middle fingers.

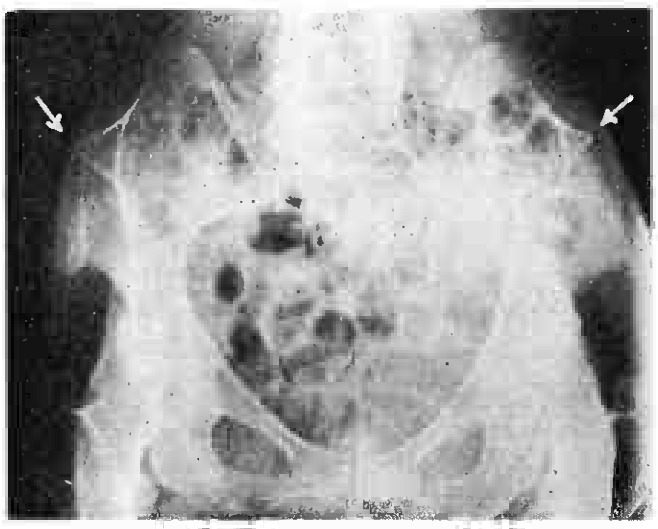


Fig. 5. Case 1: Radiograph of the pelvis showing prominent iliac horns (arrowed) with the iliac crests markedly flared out.

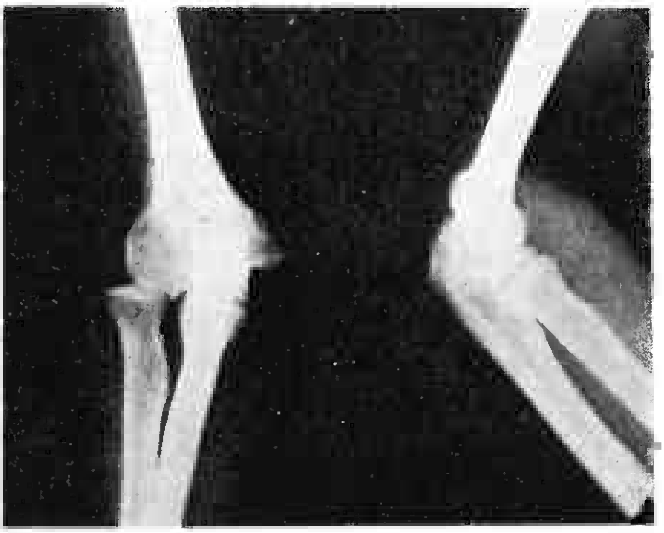


Fig. 8. Case 2: The lateral dislocation of the radial head is well shown in the right elbow.

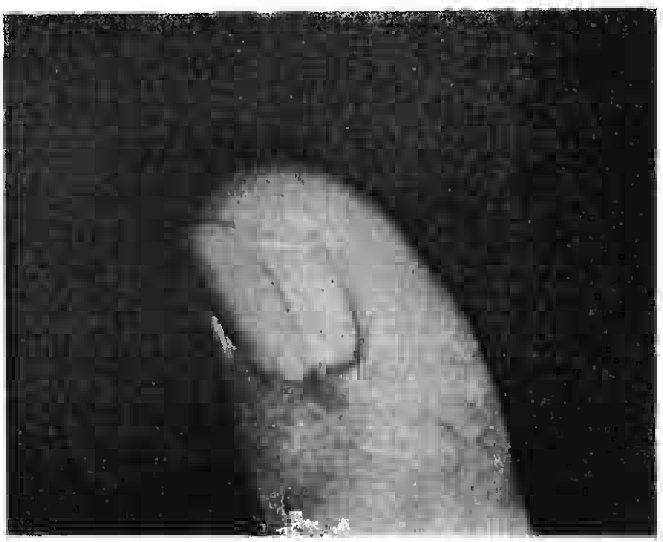


Fig. 6. Case 2: Atypical dystrophy of the right thumb nail taking the form of a single deep longitudinal crack and defective eponychium.

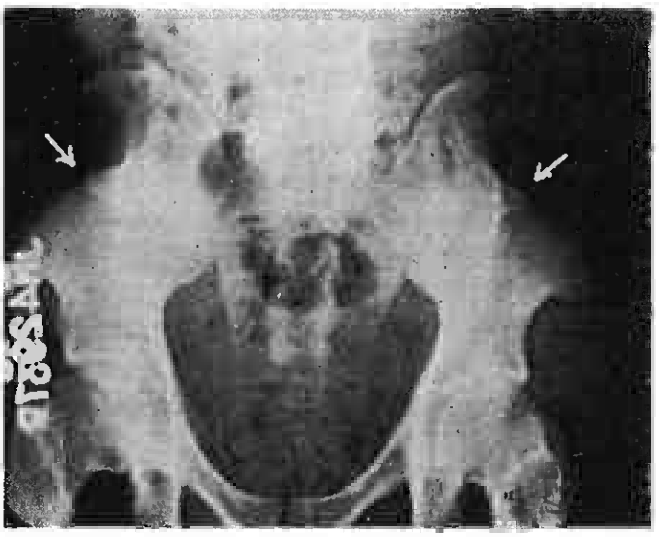


Fig. 9. Case 2: The pelvis showing bilateral Fong's horns.

Pelvis: The two forms of pelvic anomaly were also present and were quite marked (Fig. 9).

DISCUSSION

Onycho-osteodysplasia or the nail-patella syndrome has excited much attention and interest among orthopaedists, radiologists and geneticists. Certain clinical and radiological features of the disease are considered pathognomonic and this enables the condition to be recognised on routine health and radiographic examinations.

It has been established that the disease is due to a single, autosomal, dominant gene, closely linked with the ABO blood groups, with the loci of both genes residing in the same chromosome (Duthie and Hecht, 1963). The simple, dominant transmission of the condition is shown by the fact that generally affected persons have an affected parent; the transmission of the trait through successive generations is complete; and since all genes are paired, the dominant trait is passed on to half the offsprings. The gene is autosomal, i.e. non-sex-linked, and both sexes are shown to be equally affected.

A survey of the family of Case 1 would have produced interesting results but the pedigree study was not possible due to lack of co-operation on the part of the affected relatives. The parents, brothers and sisters of Case 2 were examined and found to be free of the disease. It would, therefore, appear that the propositus (Case 2) was the first member of the family to exhibit this disease, and that, most probably, he was in receipt of a spontaneous mutant from either his father or mother.

Variability in the clinical picture of the disease is well documented by Duncan and Souter (1963). While the present cases showed the complete tetrad of anomalies, significant nail dystrophy was absent in Case 2 and the knee dysplasia was mild. The pelvic dysplasia, consisting of flaring of the iliac crests with prominence of the anterior superior iliac spines and iliac horns was pronounced in both cases. In fact, these anomalies were highly developed and the pelvis assumed a winged appearance, likened by Lacroux *et al* (1960) to an elephant's ear. The diagnosis of the two cases was made on the basis of these marked pelvic changes found on routine radiographic examination.

ACKNOWLEDGEMENTS

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