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THE NAIL-PATELLA-ELBOW SYNDROME

A Case Report

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The nail-patella-elbow syndrome, also called hereditary osteo-onycho-chondro-dysplasia, arthro-onycho-dysplasia or onycho-mesodysplasia, is of interest to clinicians and geneticists because of the variety of congenital abnormalities it presents (Brixey & Burke 1950). Many hundreds of cases, the majority assembled within a few families, have been reported in the international orthopaedic literature, but none before in Scandinavia. In the patient presented here many of the characteristic deformities were found.

CASE REPORT

The patient is a boy who at 15 years of age was sent to our clinic from a neighbouring orthopaedic hospital. At birth he was found to have foot deformities, bilateral pes varo-adductus, which were treated using corrective plasters followed by splints. At 6 months of age restricted extension was noticed in both elbows; X-ray showed nothing pathological. A tentative diagnosis of arthrogryphosis multiplex congenita was made on that occasion. One year later it was noticed that he had small thumb nails. At 5 years of age the absence of both patellae was discovered. At 8 years of age an accentuated lumbar lordosis was noticed together with general weakness of the muscles and an eye deformity similar to arcus senilis. At 10 years of age a lengthening of the right achilles tendon was performed because of the equinus deformity. Proteinuria was accidentally discovered at this age but otherwise the urine was normal. Biochemical findings showed normal serum cholesterol, plasma proteins, urea clearance and creatinine in serum, and normal urine concentration and dilution on thirst provocation. An intravenous pyelograph was normal. He was admitted to our clinic because of his elbow deformities. The parents denied hereditary deformities in the family. The boy had no subjective complaints regarding his deformities and his mental development was normal.

On examination we found him to be a slender boy with an accentuated lumbar lordosis (Figure 1). A web formation across the cubital fossa was noticed in both elbows as well as an extension defect in the right elbow of 45° and in the left of 35°, and restricted supination in the right elbow of 30° and in the left of 20°.

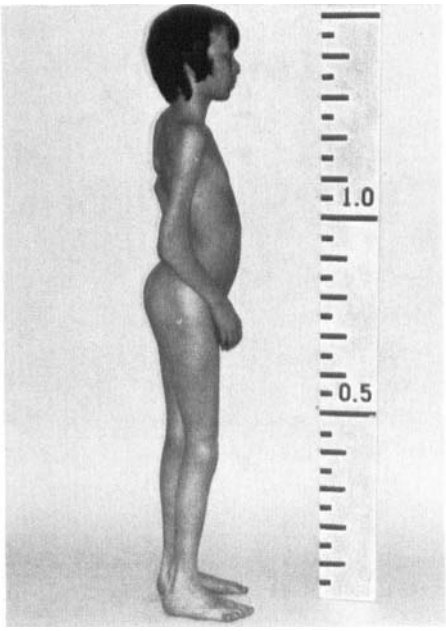

Figure 1. Cubital web and accentuated lumbar lordosis.

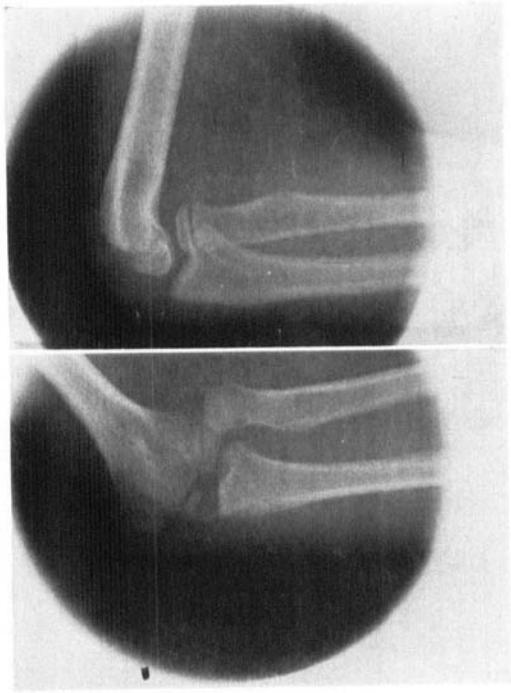

Figure 2. Right elbow with dislocation of the radius.

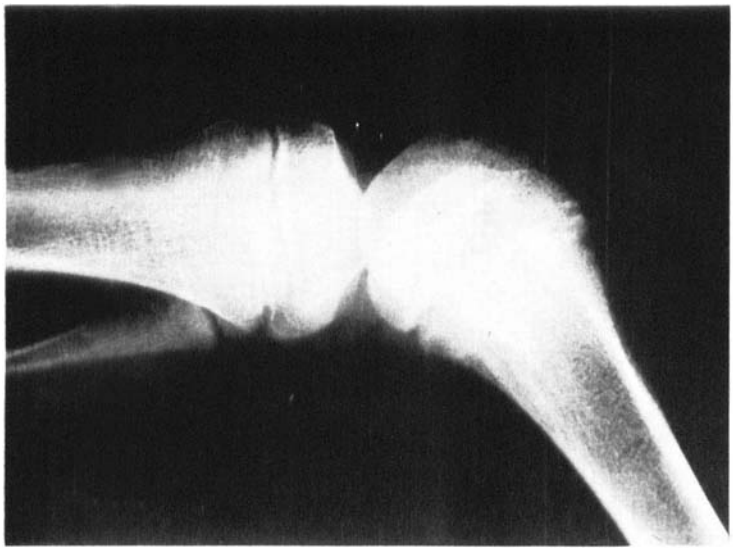

Figure 3. Left knee; absence of the patella.

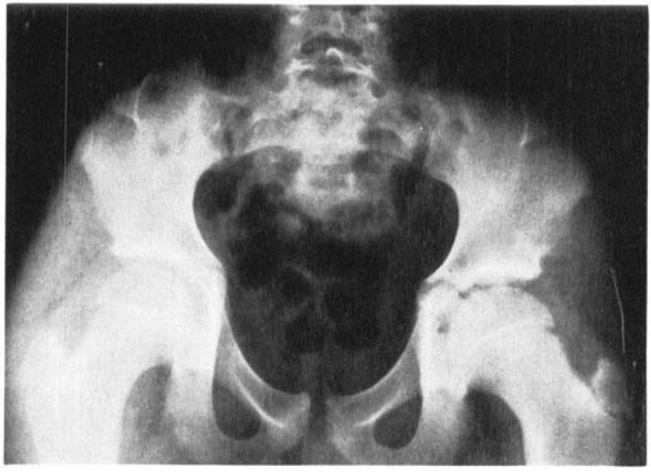

Figure 4. Iliac horns on both sides.

Marked atrophy of the triceps and biceps as well as the deltoid muscles of both arms was also noticed. The circumference of the upper arm was 14.5 cm on both sides compared with the circumference of the webbed elbow which was maximally 22 cm on the right and 24 cm on the left side. Radiographs showed bilateral dislocation of the radius together with asymmetrical humeral condyles (Figure 2). Knee deformity was also found. There was a moderate genu valgum, the left patella was absent and the right was hypoplastic, measuring 2 × 1 cm. The tibial tubercles were prominent

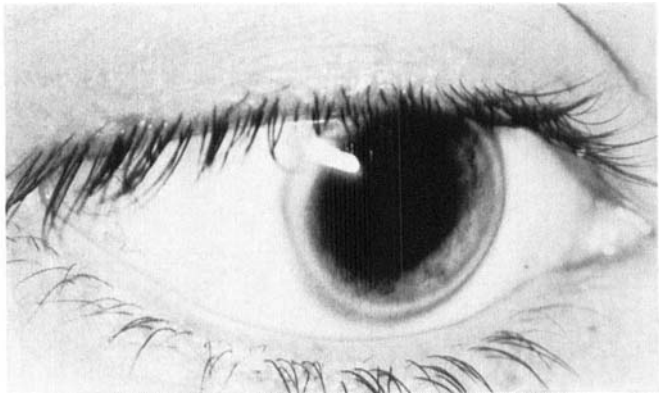

Figure 5. The Lester iris.

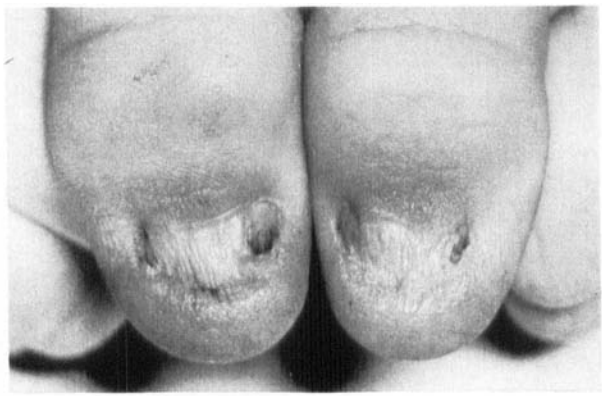

Figure 6. Both thumbs; absence of the ulnar part of the nails.

as were also the medial femoral condyles. A very noticeable quadriceps atrophy was found. The radiographs showed a hypoplastic laterally displaced patella on the right side and absence of the patella on the left side as well as small lateral femoral condyles (Figure 3). On the pelvis one could easily palpate the pathognomonic iliac horns on both sides. The radiographs showed these horns and flaring of the iliac crests (Figure 4). Other deformities found were a shortening of the right leg by 1.5 cm, a slight pelvic slope and compensatory scoliosis, and also winging of the scapulae. The right foot had a slight valgus deformity. In both eyes the Lester iris was noticed (Figure 5). The nails of the thumbs showed a characteristic absence of the ulnar half (Figure 6). The other nails exhibited triangular lunulae and were thin with a central groove (Figure 7). The dorsal creases were absent on the distal interphalangeal joints of the second and third fingers on both hands.

As the boy had no complaints and lived a perfectly normal life we only registered the deformities and have not planned any corrective operations.

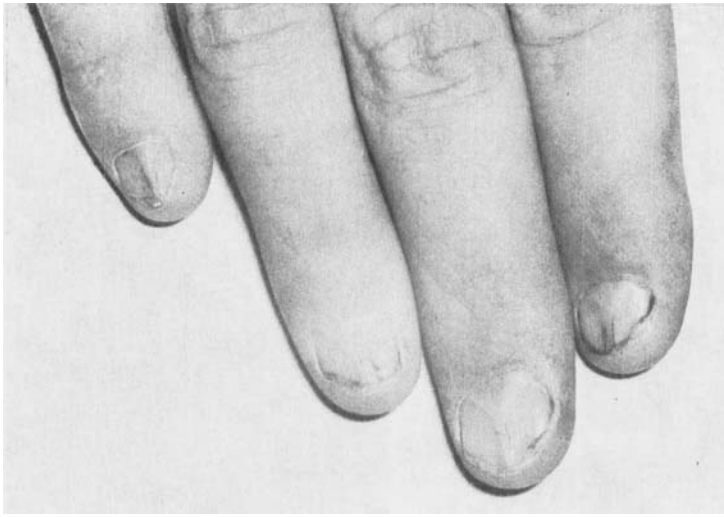

Figure 7. Right hand with thin nails with triangular lunulae and a central groove.

DISCUSSION

The first description of this syndrome was given by Chatelain in 1820 and Sedgwick quoted by Little (1897). Turner (1933) gave the first comprehensive report of the syndrome.

The syndrome is transmitted as an autosomal dominant trait with a characteristic 100 per cent penetrance. Definitive linkage has been established between the nail-patella-elbow-geme locus and that of the AB O blood groups (Jameson et al. 1956).

Skeletal dysplasias

Knee. Absence or hypoplasia of the patella, asymmetrical development of the femoral condyles, sloping tibial plateaux and prominent tibial tubercles, osteochondritis dissecans and loose bodies have been reported. These abnormalities may cause deformity, instability of the patella, genu valgum or walking difficulties (Love & Beiler 1957, Valdueza 1973).

Elbow. Hypoplasia of the capitulum of the humerus, often with some degree of posterior or anterior dislocation and elongation of the radius, is a special feature. The elbow deformities often cause little or no disability and often the patients are unaware of them (Eisenberg 1972, Valdueza 1973).

Pelvis. Iliac horns are pathognomonic of the syndrome. They are usually symmetrical and well shaped, quite unlike the irregular projections of exostoses and have only been described together with the nail-patella-elbow syndrome. They may represent a pelvic spinose process, analogous to the spinose and acromial processes of the scapula. The horns cause no disability (Darlington & Hawkins 1967). Another pelvic deformity is straightening of the anterior part of the iliac crest or outward flaring of the iliac crest (Turner 1933, Jameson et al. 1956).

Soft tissue

Soft tissue changes with web formation around the elbows and axilla have been described as have also flexion contractures of the hip, the knee, the elbow and the fingers. Foot deformities: equino-varus, calcaneo-valgus and flat feet have been described in association with this syndrome. Hypoplasia of the deltoid, triceps and brachio-radialis muscles have been reported as well as hypoplasia of the quadriceps or absence of a proper rectus femoris muscle (Love & Beiler 1957, Maini & Mittal 1966, Beals & Eckhardt 1969, Aggarwal & Mittal 1970).

Hand

Laxity of the metacarpal-phalangeal joints or congenital contractures of the interphalangeal joints, mostly in the little fingers, may occur. Nail deformities, complete absence or absence of the ulnar half of the nail, or merely a thin grooved nail may occur, this being most marked in the thumb and decreasing in severity in the ulnar direction. Triangular lunulae are often seen and absence of the dorsal creases over the distal interphalangeal joints may occur. Madelung's deformity and superfluous metacarpal bones have also been described (Montant & Eggerman 1937, Silverman et al. 1967).

Renal dysplasia

The nephropathy in this syndrome is classed in the group of chronic nephritis (Whalen & McIntosh 1962). Proteinuria is the cardinal manifestation and casts and blood cells may be present. This may be asymptomatic and associated with normal health. Impairment of glomerular and tubular function has ben reported (Muth 1965, Norton & Mescon 1968). Renal biopsies have confirmed the non-specific glomerular nephritic nature of the lesion in patients with and without proteinuria.

The severity of the nephropathia is quite variable, it may be serious or it may be harmless (Eisenberg et al. 1972).

Eye deformity

Heterochromia of the iris was first described by Lester (1936); it is quite common and its relationship to this syndrome is not clear.

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