Case report

NAIL PATELLA SYNDROME

V. Feier and C. Solovan

ABSTRACT

A case of nail-patella syndrome (NPS) with bone anomalies but without evident renal manifestations is presented. Additionally to nail dystrophy anomalies which have not been reported before are described: absence of the styloid apophysis of the cubitus and absence of the complete link of the iliac wings cartilage. It is emphasized that dermatologists and radiologists should be aware of diagnosing NPS early to anticipate possible renal involvement.

KEY WORDS
nail-patella syndrome, 23 year-old female, case report

INTRODUCTION

In the nail patella syndrome (NPS) or hereditary onycho-osteoarthroplasia, tissues of ectodermal as well as of mesodermal origin are involved. An autosomal dominant inheritance of variable expressivity but high penetrance is generally assumed (1). Females of short stature are more frequently affected. The NPS locus is at 9q34 linked to the ABO group (2). In 1965, 225 patients with this syndrome were living in Great Britain. The estimated prevalence is 22/million inhabitants, while the mutation rate is estimated 1.9 millions alele/generation (3).

The syndrome is characterized by four groups of symptoms:

1. Complete or partial absence of fingernails, the tumb-nail is absent or most severely affected. The severity of nail dystrophy is decreasing from index to the small finger (from the radial to the ulnar side)

2. Bony dysplasia of the knee aerea: hypoplasia or aplasia of patella, hypoplasia of the lateral femoral condylus, dislocation of the knee with subluxation of the patella.

3. Bony displasia of the elbow consisting of the hypoplasia of the capitellum and of the radial head.

4. Presence of iliac horns

In some families a nephropathy has been described as a complication of the syndrome: by light and electron microscopy a thickened glomerular basement membrane with irregular zones of rarefication (4, 5, 6).

CASE PRESENTATION

The family history was unremarkable

The 23 year-old patient L.A. presented herself for a dermatological consultation because several nail dystrophies on her hands. The alterations were present since birth.

A dermatological inspection revealed a complete absence of nails of both thumbs, pterigium unguium and onychoatrophy of fingers 2,3 and 4 on both hands as well as of the fifth finger on the right hand. The nail of the fifth finger of the left hand was normal. The patient complained that she could not extend completely her elbows (Fig. 1) and that she had difficulties carrying out the pronation and supination movements. At the age of 10 years she underwent a surgical treatment for recurrent dislocations of the left patella. The nails on the toes were normal. The 2nd, 3rd and 4th fingers on both hands displayed an axial deviation (Fig. 2).

The routine laboratory tests e.g. WBC, RBC, ESR, serum electrophoresis, glucose, thymol turbidity, serum calcium,

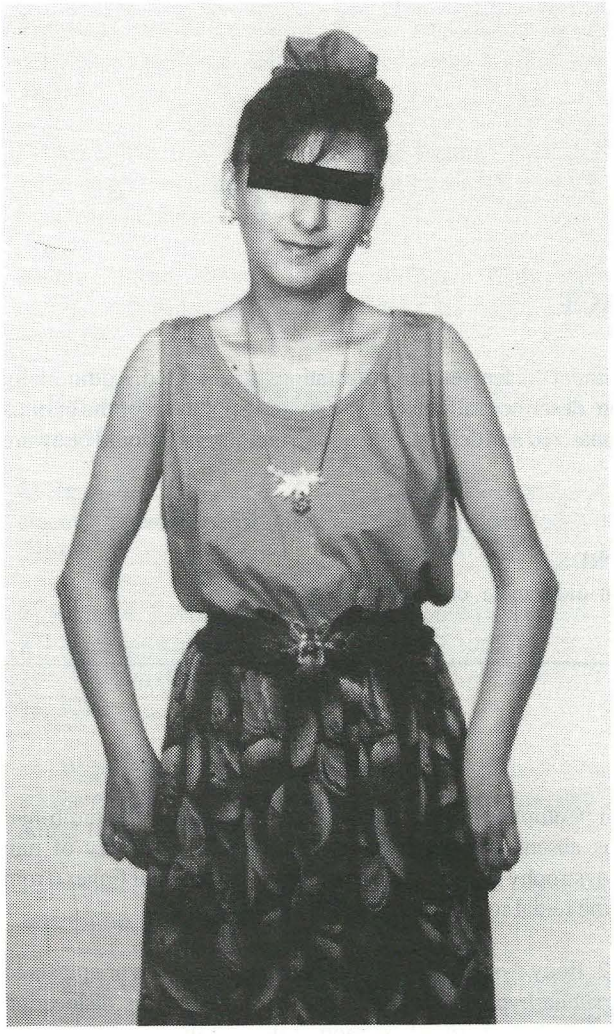

Figure 1: The patient is incapable of performing a complete extension in the elbow joint, and has difficulties in the pronation and supination movements, too.

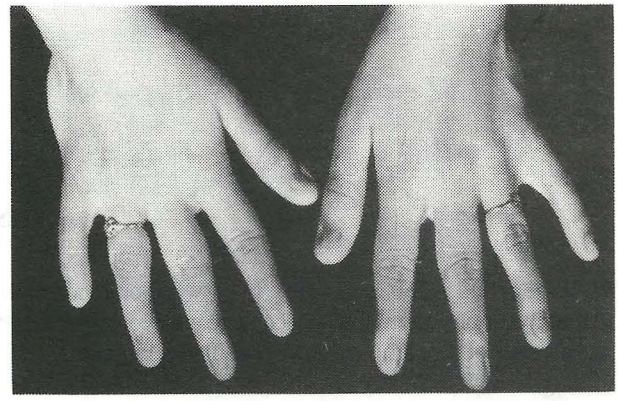

Figure 2: Absence of nails on both thumbs; dystrophy and pterigium unguium of fingers 2, 3 and 4 as well as of finger 5 on the right hand.

ALT and AST were within normal limits. Additionally the blood group A II, an eosinophylia of 6 %, a serum iron level of 28.2 umol (normal values up to 26 umol) and a proteinuria of less than 200 mg/24 h were found.

The cytogenetic investigation was also within the normal limits.

X-ray examination revealed the following changes:

The fingers 2 and 3 of the right hand and 3 and 4 of the left hand presented an axial deviation at the level of the proximal interphalangeal joint; there was a cubital deviation of the of the metacarpo-phalangeal joints of fingers 3 and 4 of the left hand and an osteosclerosis of the proximal apophysis of the phalanges of the right hand; the second phalanges of both fifth fingers were shorter; there was also a structural modification of carpal bones, especially the semilunar and the left big bone (periarticular osteosclerosis), demine-

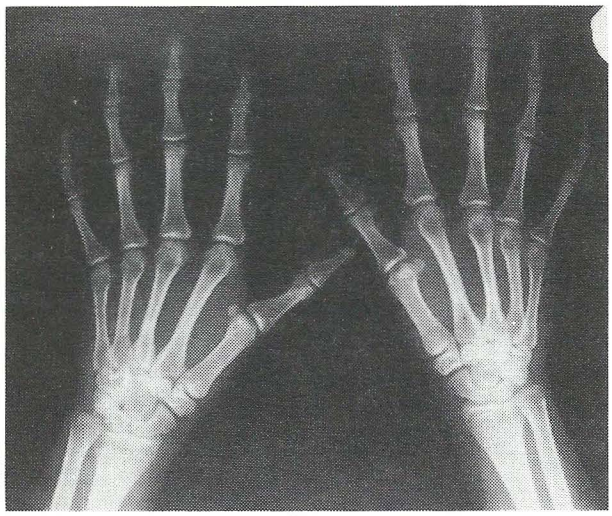

Figure 3: X - ray examination of the hands showed the absence of the styloid ulnar apophysis bilaterally.

ralisation of the distal epiphysis of the right radius and absence of the styloid cubital apophysis bilaterally (Fig. 3).

The gracil distal epiphyses of humerus were demineralised; a spotted demineralisation of the radius and ulna bilaterally, hypoplasia of the radial head with dislocation as well as a medial position of the olecranon were observed (Fig. 4).

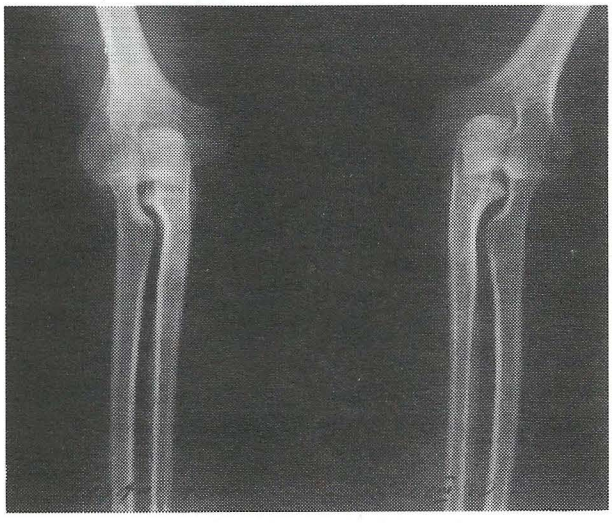

Figure 4: X - ray of the elbow articulation (joint) showed hypoplasia of the radial head and luxation in the joint, and an unusual position of the cubital (ulnar) head.

The acromial processes displayed a strong obliquity, a medial deviation as well as an asymmetry. The scapulae were more convex with a thickened external edge. On the right side there was also a slight acromio-clavicular disjunction.

The bones of the pelvis presented a periarticular sclerosis, incompletely linked growing cartilages of the iliac crests, sacralization of the L5 as well as a spina bifida. The heads of femur were slender bilaterally, giving the impression of coxa valga, the spinae iliacae posteriores inferiores were hypertrophic .

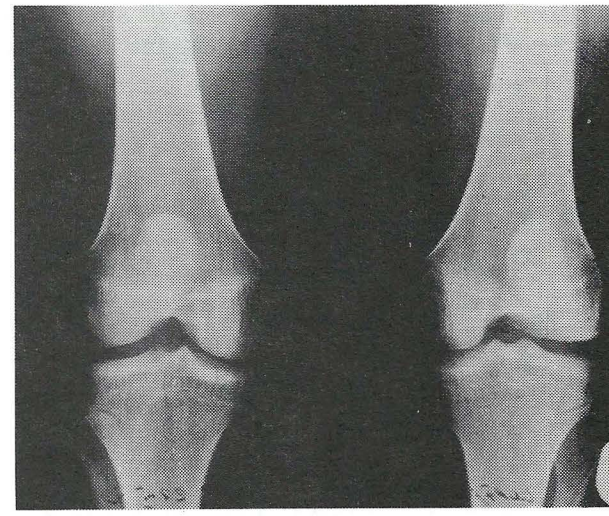

Figure 5: X - ray of the knee showed hypoplasia of the patella and of the lateral femular condyl, bilaterally.

The X-rays of the knees showed a demineralisation of the periarticular parts as well as hypoplasia of both patellae and lateral femoral condyles; both medial femoral condyles were hypertrophic, the left lateral tibial spine was flattened (Fig. 5). The ankles displayed a periarticular demineralisation with a few small areas of lysis at the distal epiphysis of the right fibula. There was a demineralisation of the metatarsal and phalangeal periarticular surfaces of the 2nd, 3rd, 4th and 5th toes bilaterally.

DISCUSSION

The case can be considered a complete NPS with subclinical renal alteration. The onychodystrophy was typical, the severity of lesions decreased from the radial to the ulnar side. The displasia of the nails is usually present from birth or since the first years of life.

Capillaroscopy revealed dilated capillary loops in the dorsal pterygium with formation of a microvascular system between them.

Certain symptoms which have been described in NPS like onychoschysis, hyponychium or triangular lunula (1) were missing in our patient. The presence of iliac horns on the posterior iliac crests which is one of the further characteristics

of NPS (7) was also missing in our case. Other bone dysplasias associated with NPS reported as: congenital absence of fibula (9) or shoulder dysplasia, were not manifest in our patient. There were however expressed a thickening of the lateral side of scapula, spina bifida as well as dysplasia of the proximal epiphysis of radius. Additionally our case presented bone dysplasias which were not mentioned previously in the literature: bilateral absence of styloid apophysis, asymmetry of acromial processes with a reduced glenoid cavity.

A glomerulopathy has been described in association with NPS. It is characterised by deposits of collagen-like material at the site of glomerular basement membrane as shown by light microscopy (3) and electron microscopy (5, 6). Proteinuria is asymptomatic in about 60 % of cases while in 5 to 8 % of patients the disease progresses towards a renal failure and hemodialysis becomes necessary. As our patient presented only a mild proteinuria a future evolution can not be foreseen. She declined renal biopsy.

In addition to the changes already mentioned a thickening of the epidermal basement membrane was detected by electron microscopy (11).

CONCLUSION

The review of the literature indicates that NPS is not so rare as it may appear. The majority of these patients are not seriously handicapped. Physicians however should bear in mind that due to changes observed in the basement membrane of glomeruli the condition may progress towards severe renal involvement. For this reason regular medical checkups are indicated.

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