

NAIL-PATELLA SYNDROME: A CASE REPORT

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ABSTRACT

An otherwise healthy 33 days old girl, displaying features of the Nail-Patella syndrome is reported. This rare autosomal dominant disorder is characterised by nail and bone abnormalities, with nail hypoplasia or dysplasia and absent or hypoplastic patella as essential features for the diagnosis. The patient had bilateral absent palpable patella with hypoplasia of the fingernails. Our patient did not have the typical iliac horns and there was no evidence of renal involvement.

Key words: Nail-Patella syndrome

INTRODUCTION

The nail-patella syndrome, also known as hereditary osteo-onychodysplasia, is a rare autosomal dominant disorder involving tissue of both ectodermal and mesodermal origin^{1,2}. This disorder was first described by Little in 1837². It is characterised by nail and bone abnormalities and, frequently renal disease. Nail hypoplasia or dysplasia and absent or hypoplastic patella are essential features for the diagnosis³. Other features are deformation or subluxation of the head of radius, iliac horns that are said to be pathognomonic but reported to be present in only 70% of cases³. Other skeletal anomalies are pes equinovarus and dislocated hip joints. The most serious components of Nail-Patella Syndrome is the nephropathy which, however, does not occur in all patients with the syndrome³. The nephritis associated with Nail-Patella Syndrome resemble chronic glomerulonephritis with a wide spectrum of severity.⁴

Some patients present in early childhood whereas others are asymptomatic as adults. This is an uncommon condition, which has not been previously reported from this centre. We report one such case.

CASE REPORT

A 33 days girl who was brought to the paediatric out patient department by her mother with complaint of abnormal shape and movement of both lower limbs noticed since birth. She was the 2nd child of a monogamous family with no history of consanguinity. She was delivered after a term-unsupervised pregnancy at home. The pregnancy was said to be unremarkable and delivery was uneventful. At birth, patient was noticed to have abnormally shaped feet and abnormal extension of lower limb at the knee joint. There was no history of similar anomaly in both families.

On examination, the patient appeared normal for age, with weight of

3.1kg, length 51cm, occipito-frontal circumference 34.5cm and a chest circumference of 33cm. The anterior fontanale was patent and normotensive measuring 2x3cm. Patient was alert but slightly hypotonic with normal deep tendon reflexes. There were scarification marks on the anterior abdominal wall, umbilical hernia and soft hepatomegaly. Musculoskeletal system examination revealed hypoplasia of the nails, absent palpable patella with hyperextension at the knee joint and talipes equinovarus deformity of both feet (Figure 1). Other systems were normal.

Figure 1: Clinical appearance of patient



Full blood count revealed a Pack cell Volume of 37%, WBC 11.4×10^9 /L with Neutrophils of 23% Lymphocyte of 72%, Monocyte of 03% and Eosinophils of 02%. Blood urea nitrogen (BUN) was 1.5 mmol /L, Na^+ 145mmol /L, K^+ 5.0 mmol/L, Cl^- 108 mmol/L, and HCO_3^- 26mmol/L. Serum creatinine was 104 umol /L, Ca^{2+} 2.02 mmol /L, Phosphorus 2.10mmol /L, Alkaline phosphatase 142 IU /L. Urine analysis for protein and sugar were negative. The X-ray showed dislocation of both hip joint and clubfeet deformity. Iliac horns, which are bilateral bony projections from the central area of the

external iliac fossae directed posterolaterally, were not observed on the X-ray. The baby remained healthy until she was lost to follow up.

DISCUSSION

Although the diagnosis of osteo-onychodysplasia [Nail-patella syndrome] can be established in early infancy through the radiographic demonstration of iliac horns⁵, this disorder is rarely diagnosed in infancy. This is because the abnormalities of the nail and patella, which are essential for diagnosis, may not become apparent until later in life. The diagnosis of this case in the early infancy was necessitated by the need to carefully scrutinize a child who have presented with talipes equinovarus when the absent palpable patella was noticed. Absent patella is not a feature of talipes equinovarus whether congenital or positional. Talipes equinovarus however, is a rare association of nail – patella syndrome^{3,6}. The presence of hypoplastic nails and absent palpable patella in this patient is consistent with diagnosis of nail – patella syndrome. Other stigmata in this patient which has been described in patients with the Nail-Patella syndrome includes bilateral hip joint dislocation and joint hyperextensibility, although they do not contribute to the diagnosis.^{3,7}

Since the skeletal deformities of nail-patella syndrome characteristically cause no significant disability,⁸ the importance of recognising this syndrome lies with the associated nephropathy. Proteinuria, which is present from infancy, is the commonest presenting feature⁶ and has been reported in more than 60% of cases³. In this patient, the urine was negative for protein. Despite the unusually favorable prognosis associated with this nephropathy, death due to hereditary nephropathy of osteo-

onychodysplasia have been reported. Leahy⁸ reported the death of a 9-year-old girl from nephropathy of osteo-onychodysplasia, whose renal disease started as early as 2 years of age. Consequently, there is the need for early recognition of this clinical entity and as such, should be included in the differential diagnosis of infants with significant deformities of the knees or feet, since this may be the first clinical manifestation of this disease.

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