Case report

NAIL PATELLA SYNDROME

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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.