

Nail Patellar Syndrome (NPS)

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Abstract

The Nail Patellar Syndrome (NPS) is a genetic disorder primarily involving the bones, nails and kidneys. Inheritance is autosomal dominant with unknown pathogenesis. Diagnostic features are hypoplastic or absent nails, patella, iliac horns and elbow abnormalities. Multiple other bony deformities, eye structure abnormalities and nephropathy are seen. A case of 3-year old female child with nail patellar syndrome is presented.

Keywords: Onychoosteodysplasia; autosomal dominant; mucopolysaccharides; genetic counseling.

Introduction

The nail patellar syndrome (NPS), also known as hereditary onychoosteodysplasia (HOOD), Turner-Kaiser Syndrome or Fong's syndrome, is a genetic disorder primarily involving the bones, nails and kidneys. Many other organ systems may demonstrate abnormalities.¹

The incidence of NPS has been reported to be from 4.5 per million in the United States to as high as 22 per million in the United Kingdom.² This syndrome is a hereditary disorder with an autosomal dominant mode of inheritance. There is virtually complete penetrance of the nail dysplasia, but variable expression of the other manifestations of the syndrome. In NPS, the genetic locus is situated on chromosome 9, closely linked to the genes of ABO blood groups and the adenylate cyclase gene.³ The pathogenesis of NPS remains unknown, although several mechanisms have been proposed.^{4,5,6,7} Excessive secretion of mucopolysaccharides has been observed in some but not all patients.^{8,1} No pre-natal diagnosis test is available, although one can somewhat assess the risk by being aware of ABO blood types.

Case Report

A 3-year female child presented in the Siddhi Memorial Hospital, Bhaktapur with a limp and abnormal nails in all the fingers. She was the second child of her parents with a history of full-term normal home delivery but with Low Birthweight. Birth history was normal and she was completely immunized with EPI schedule. Her milestone of development was delayed in the form of gross motor. There was history of delayed sitting and walking. Mother was on regular ante-natal check up and there was no history of teratogenic drug intake, radiation or viral infection during pregnancy. She was born from non-consanguineous parents.

On general examination, the child had normal face. Family history showed her elder sister had difficulty in walking with some fingers having no nails especially in the big toes. (Fig. 1, 2, 3)

Fig. 1: Shows dystrophy of nail in big toes

Fig. 2: Shows dystrophy of nails in thumbs

Fig. 3: Shows hypoplastic and dystrophic finger nails

Anthropometric measurements showed wt. = 12 kg., length = 82 cm and Occipito Frontal Circumference (OFC) 48 cms. Musculoskeletal system examination revealed the knee joint hyper extended with depression in the centre. On palpation, patella was absent (Fig. 4). Examination of hands showed dystrophy of the nails in all the fingers of hands and legs. Other systemic examinations revealed no abnormalities.

Fig. 4: Shows hypoplastic patella

On investigation, CBC-Normal, Hb = 14.5 gms platelet-normal. The X-ray of the knee showed absent patella in both knees. Impression-Nail Patellar Syndrome was the possible diagnosis.

Discussion

Nail Patellar Syndrome was first described in 1820 by Chatelain.⁹ The syndrome consists of a tetrad of diagnostic features: dysplastic, hypoplastic, or absent nails; hypoplastic, or absent patellae; iliac horns, and elbow abnormalities.^{2,1} Nail dystrophy occurs in 90 percent of cases, usually present at birth, and is greatest in the thumb-nail, progressively decreasing

from the second to the fifth digit. These changes are usually bilateral and symmetrical. Similar changes can occur in the toe nails, but these are less frequent.^{10,11}

Triangular lunulae hypoplastic or absent patellae leading to gait disturbances have been reported to be pathognomonic and universal finding respectively.¹² Multiple other bony abnormalities that have been reported are scoliosis, straightened clavicles, hypoplasia of the first rib, scapular abnormalities, sternal deformities, spina bifida occulta and high arched palate.^{10,13,11,1}

Eye involvement like irregular iris, microcornea, cataracts, glaucoma, strabismus has been reported.¹⁴

NPS has special interest for nephrologists as nephropathy occurs in 30 to 55 percent of the patients.¹

References

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