

## Congenital Aplasia of the Patella

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### ABSTRACT

Congenital absence or marked reduction of the patella may exist in association with other congenital anomalies, most likely the nail-patella syndrome and the ischiopatellar dysplasia or as an isolated entity described in only a few families to date. It may also appear in other orthopaedic malformations, such as the congenital dislocation of the knee. A careful physical examination is essential in the diagnosis of the syndrome, which is mainly clinical. The purpose of this review is to present a range of the clinical syndromes that may be associated with aplasia of the patella, in both children and adults, and to review the related literature.

**Key words:** Congenital patella aplasia; Nail-patella syndrome; Ischiopatellar dysplasia; Isolated familial patella aplasia; Congenital dislocation of the knee

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### EDITORIAL

Congenital aplasia of the patella is an extremely rare disorder, characterized by congenital absence or marked reduction of the patella. The prevalence rate of patellar aplasia in the newborn child is not readily diagnosed because the patella is completely cartilaginous at birth and starts its ossification process between the third and the sixth year. Aplasia of the patella has been documented in the papers of Little and Mayer since 1897. Congenital absence of the patella is usually observed bilaterally and is usually associated with other malformations, although it may occasionally present as an isolated lesion. Congenital anomalies characterized by patellar aplasia belong to a clinically diverse and genetically heterogeneous group of disorders. The most common syndromes associated with congenital patellar aplasia is the nail-patella syndrome (NPS), also known as hereditary onychoosteodysplasia (HOOD syndrome), as Fong disease or as Turner-Kieser syndrome, and the ischiopatellar dysplasia (IPD), also known as the 'small patella' syndrome (SPS)<sup>[1-9]</sup>.

Patients with NPS also present with ungula (nail) dysplasia from birth, elbow dysplasia, exostoses of the ilia (iliac horns), swan-neck deformity and absent skin creases on the dorsal aspect of the distal interphalangeal joints of the fingers, tingling, numbness, and in some cases renal insufficiency. The disease is transmitted as a simple autosomal dominant gene resulting from symmetrical mesodermal and ectodermal abnormalities. There is a definite linkage between the locus of the nail-patella gene and the ABO blood groups. LMX1B (LIM homeobox transcription factor 1 beta) is a unique mesenchymal determinant of dorsal patterning during vertebrate limb development, so mutations in the LMX1B gene on chromosome 9q34 result in skeletal dysplasia affecting the dorsal tissues, especially the nails and patellae. Most patients function without difficulty, while some individuals may have flexion contractures and recurrent knee dislocations. Diagnostic radiographic findings include absent or severely hypoplastic patellae (with a tendency to

recurrent dislocation), hypoplasia of the radial head or capitellum (leading to subluxation/dislocation), bilateral posterior iliac horns and protuberant anterior iliac spines (Figure 1). The bilateral posterior iliac horns, due to exostoses arising from the posterior aspect of the iliac bones, are present in as many as 80% of patients; this finding is considered pathognomonic for the syndrome. The horns may be capped by an epiphysis. Iliac horns were initially described by Edward Everett Fong, an American radiologist in 1912. Various other skeletal anomalies, including pes equinovarus, dislocated hips, and contractures of major joints have been described in NPS but do not contribute to the diagnosis. Other features include: scoliosis, genu valgum deformity, flared iliac crests, prominent tibial tuberosities, clinodactyly, short 5<sup>th</sup> metacarpals, open angle glaucoma and renal osteodystrophy<sup>[10-35]</sup>.

Ischiopubic hypoplasia is a rare constituent of congenital syndromes. Ischiopubic hypoplasia is associated, in most cases, with other malformations such as anomalies of the hips, including CDH, of the feet, including talipes equinovarus and scoliosis. Congenital anomalies of the pelvic bones and hips associated with aplasia of the patellae are diagnosed as IPD or as the SPS<sup>[36-39]</sup>. They are also known as coxopodopatellar syndrome or Scott-Taor syndrome (Figure 2). It is a rare autosomal dominant disorder, characterized by aplasia of the patellae and various anomalies of the pelvis and feet. The syndrome was first described by Scott and Taor in 1979 in a large family with

bilateral small or absent patellae accompanied by anomalies of the pelvic girdle and upper femora, in most of the affected subjects. Other major signs were a wide gap between the first and second toes, short fourth and fifth rays of the feet, and pes planus. Various other skeletal anomalies have been reported, such as elongated femoral necks, flattened and widened proximal femoral epiphyses, hypoplasia of the lesser trochanter and tarsal anomalies<sup>[40-50]</sup>.

Other congenital anomalies associated with patellar aplasia include the Meier-Gorlin syndrome<sup>[51,52]</sup>, the Rapadilino syndrome<sup>[53]</sup>, the genitopatellar syndrome<sup>[54]</sup>, aniridia<sup>[55]</sup>, and trisomy 8q syndrome determined on chromosome analysis<sup>[56]</sup>. Patellar aplasia may be also occasionally associated with various orthopaedic malformations, but the most common one is congenital dislocation of the knee (Figures 3 and 4)<sup>[57-61]</sup>.

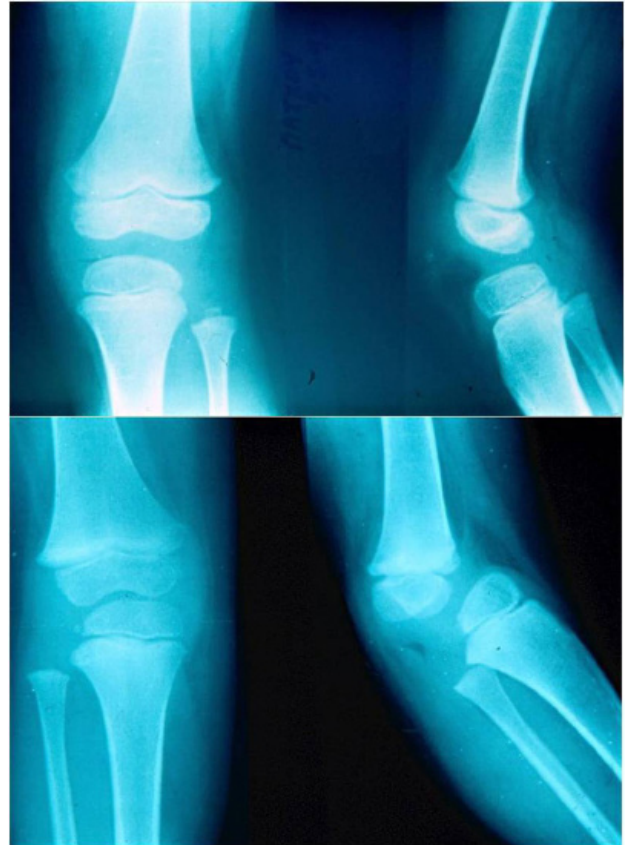
Isolated familial patella aplasia or congenital patellar syndrome is a rare autosomal dominant disorder in which congenital aplasia of the patellae is the only clinical and radiographic finding. Congenital absence or marked reduction of the patellar bone (Patellar Aplasia-Hypoplasia-PTLAH) has been described in only a few families to date. It may cause no major dysfunction, and surgical or major treatment is not always required. Whenever the absence of the patella is associated with atrophy of the quadriceps mechanism, patients suffer from weakness of knee extension and may develop progressive flexion deformity of the knee. A locus for PTLAH has been identified on



**Figure 1** A 57-year-old man with a diagnosed nail-patella syndrome. The nails are markedly ridged, abnormally structured and almost completely absent in the thumbs. The skin shows a loss of creases on the dorsal aspect of the distal interphalangeal joints of the fingers. Radiographs showed complete absence of the patella on the right and a hypoplastic patella on the left knee. Elbow radiographs indicated hypoplasia of the radial head and capitellum. Anteroposterior radiograph and computed tomography of the pelvis demonstrated protuberant anterior iliac spines and a bilateral bony exostosis or posterior iliac horn, which is pathognomonic of NPS.



**Figure 2** An 8-year-old boy with bilateral congenital absence of the patella. Ischiopubic hypoplasia and scoliosis were also evident in the patient leading to the diagnosis of a sporadic case of IPD or SPS.



**Figure 4** A 5-year-old girl with congenital dislocation of the right knee (bottom). The range of joint motion of the left knee (top) was normal. Posterior slope of the right tibial surface was evident on the lateral views. There is aplasia of the right patella, while the left one seems to be hypoplastic. However, further follow-up is required to secure the diagnosis of the congenital patellar anomalies.



**Figure 3** Typical clinical appearance and radiographic features of congenital dislocation of the knee in a 4-day-old girl.

chromosome 17q21-22<sup>[62-71]</sup>.

Differential diagnosis of patients with aplasia of the patellae requires careful clinical and radiographic evaluation, and primarily involves the NPS, IPD and the isolated familial PTLAH. In NPS, patellar aplasia is associated with nail anomalies since birth, deformation or luxation of the head of the radius resulting in impaired mobility of the elbow,

iliac horns and, frequently, nephropathy. Radiological examination of the pelvis and feet should be performed in all patients with aplasia of the patellae in order to differentiate SPS from PTLAH, and to further evaluate the diagnostic value of pelvic and feet anomalies in SPS. Pelvic anomalies, including anomalies of the ischiopubic joint or infra-acetabular notches have never been mentioned in PTLAH. Linkage

studies excluded allelism of IPD with NPS, while allelism between IPD and PTLAH cannot be excluded so far<sup>[72]</sup>.

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