

ISCHIOPUBIC HYPOPLASIA : A RARE CONSTITUENT OF CONGENITAL SYNDROMES

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Hypoplasia of the ischiopubic region is described in four patients. An adolescent was referred for spinal dysraphism and scoliosis, another one for bilateral aplasia of the patella and scoliosis, and finally two children were treated for congenital dislocation of the hip. The osseous malformation of the ischiopubic bones was not associated with any other intrapelvic disorders, and remained unchanged until skeletal maturity. The appearance of this extremely rare anomaly is usually reported as a syndromic constituent of a limited number of congenital malformation syndromes that can be widened to include congenital dislocation of the hip.

INTRODUCTION

In the chaos of malformation syndromes the radiographic detection of ischiopubic hypoplasia, which is an extremely rare anomaly, is particularly helpful in the diagnostic investigation (6, 8).

The appearance of this anomaly in association with either dysplastic scoliosis with a significant kyphotic element or with a more heterogeneous group of disorders including multiple segmental defects of the spine, rib anomalies and less pronounced facial abnormalities has been described as ischiovertebral dysplasia (IVD) or ischiospinal dysostosis (ISD), respectively (3, 4, 9).

An entity combining abnormalities of the pelvic girdle and aplasia/hypoplasia of the patella was differentiated from the onycho-osteodysplasia (nail-patella) syndrome and has been called ischio-patellar dysplasia (IPD) or the "small patella" syndrome (2, 5, 10, 11).

Furthermore, hypoplasia of the ischiopubic region has already been described in association with acetabular dysplasia in three related cases (1).

Four patients with ischiopubic hypoplasia that remained unchanged until skeletal maturity are presented. In two of them spinal dysraphism, IVD and IPD respectively, was diagnosed. The clinical and radiographic findings which indicated the diagnosis are discussed. Finally, failure of development of the ischiopubic bones was associated with congenital dislocation of the hip (CDH) in two other patients.

CASE REPORTS

Patient 1

An 11-year-old girl presented with a history of low back discomfort. Physical examination revealed a peculiar face with retrognathia and high-arched palate but no hand or foot deformities. There was a local ill-defined area of pigmentation on the left side of the thoracolumbar region of the spine. Lumbar pain was mild and was more pronounced with intense activity. There was neither muscular weakness and sensory deficit nor any manifestation of bladder dysfunction. A pelvic

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Fig. 1. a. — AP X-ray of the pelvis shows bilateral absence of ossification in the region of the ischiopubic synchondrosis ; **b.** — AP X-ray of the spine showing a mild scoliosis and spina bifida of the thoracolumbar vertebrae.

radiograph showed bilateral incomplete ossification of the ischiopubic region and a secondary ossification center in the inferior margins of the ischia (fig 1a). Anteroposterior standing radiograph of the spine revealed a thoracolumbar scoliotic curve of 8° with minimal vertebral rotation and asymmetrical orientation of the lower ribs as well as a defect of the posterior arches wider from the first to fourth lumbar vertebrae and less in the lower thoracic vertebrae (fig 1b). No other skeletal abnormalities were found. Her parents and sister did not show similar findings.

The patient was seen again five years later. She remained symptom free but with a progression of the scoliotic curve to 17° . A pelvic X-ray showed failure of ossification of the ischiopubic region. MRI of her spine revealed spinal dysraphism. Bony anomalies of the vertebral column included maldevelopment of the posterior elements consisting of partial absence of the lumbar spinous processes and slight widening of the laminae. Neural abnormalities included diastematomyelia of the lower cord. The appearance of a fibrous septum splitting the spinal cord was indicated on coronal T1-weighted images of the thoracolumbar spine. Sagittal T1-weighted images of the lumbosacral spine revealed a tethered cord (fig 2a). In axial T1-weighted images of the lumbar spine the division of the cord in two almost symmetrical parts as well as a posterior location and tethering of the conus medullaris was noted (fig 2b).

Patient 2

An 8-year-old boy was referred with a history of abnormal gait. On clinical examination there was flattening of the anterior aspect of the knees more pronounced in flexion. Weakness through the last 10° of extension was detected on the left side. Participation in school activities was unrestricted. There were no hand, foot or nail deformities. A complete radiographic skeletal survey showed bilateral absence of the patella, a bilateral defect at the level of the ischiopubic synchondrosis (fig 3) and a thoracolumbar scoliotic curve of 7° . The MRI examination of the knees indicated, apart from the bilateral absence of the patellae, hypoplasia of the

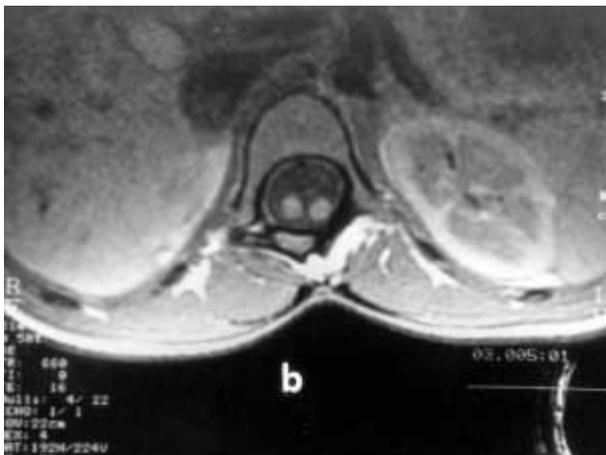


Fig. 2. a. — Sagittal T1-weighted image of the lumbar spine showing the conus medullaris to be as low as the disc between the second and third lumbar vertebra and located posteriorly to the dura mater. The filum terminale internum is also attached posteriorly to the dura (tethered cord); **b.** — Axial T1-weighted image of the caudal end of the first lumbar vertebra showing two hemicords and abnormal segmentation of the posterior vertebral arch.



Fig. 3. — AP X-ray of the pelvis shows bilateral partial absence of the ischio-pubic segment.

vastus medialis on the left side. The hamstring and adductor muscles appeared normal. Routine blood and urine examinations were normal. No similar findings or any other obvious hereditary disease could be traced in his family. The range of motion of both knees had not deteriorated four years later. By that time, there was a progression of the scoliotic curve to 12°, and a pelvic X-ray indicated incomplete fusion of the ischiopubic region.

Patients 3 and 4

An 8-month-old girl was referred for congenital dislocation of the left hip. The pelvic X-ray indicated bilateral absence of both superior and inferior pubic rami (fig 4a).

Finally, a 5-year-old girl presented with a bilateral CDH. Radiographic control and a CT-scan of the pelvis showed hypoplasia of the inferior pubic and ischial rami (fig 4b).

Physical examination in both patients revealed no abnormal findings, and the pulmonary, cardiac and abdominal examinations were normal. There was no evidence of kidney or bladder dysfunction. Routine blood and urine examinations were normal. Both patients were followed until skeletal maturity, and the radiographic appearance of ischiopubic hypoplasia has not changed.

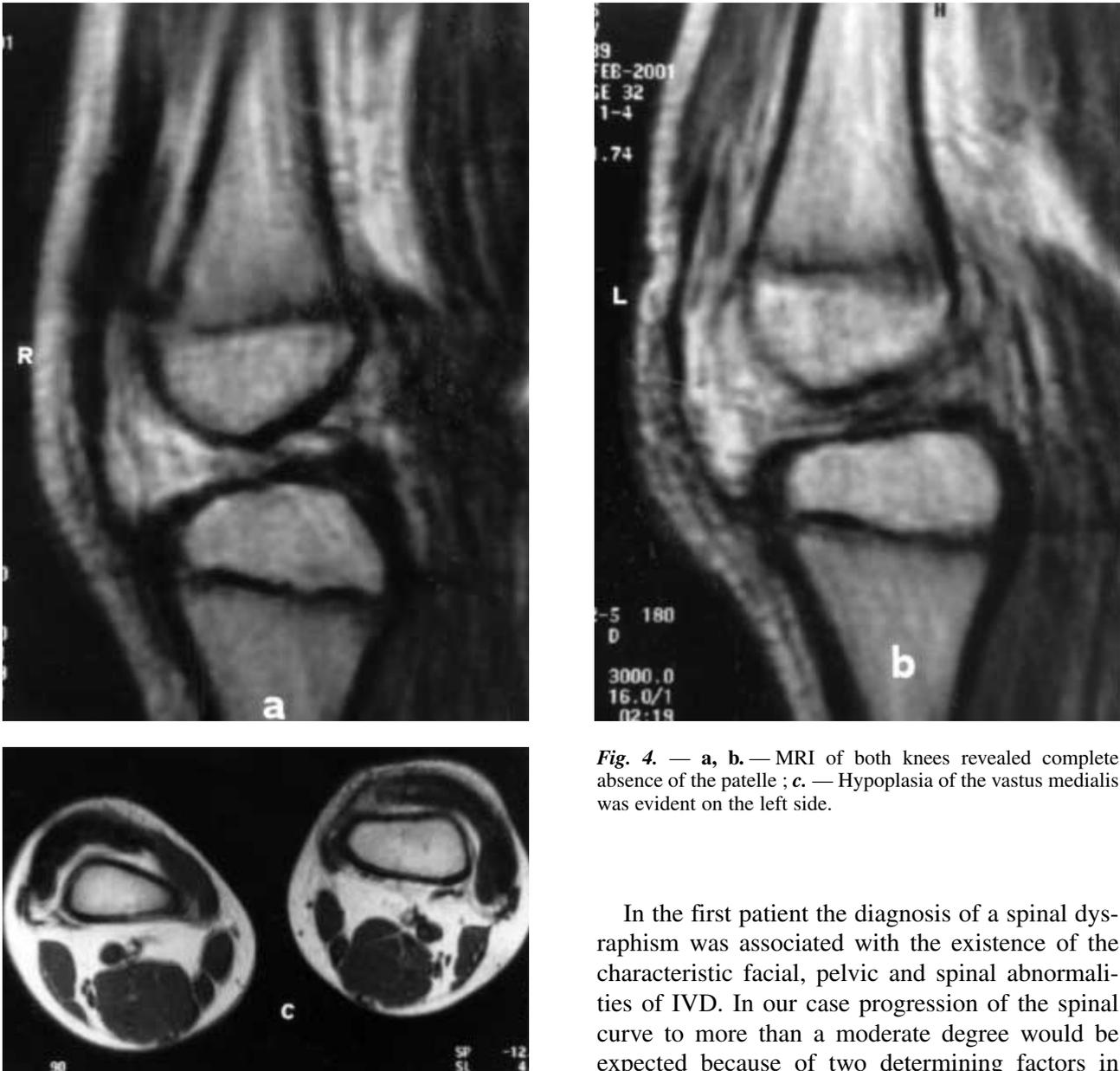


Fig. 4. — **a, b.** — MRI of both knees revealed complete absence of the patelle ; **c.** — Hypoplasia of the vastus medialis was evident on the left side.

DISCUSSION

True failure of ossification of the ischiopubic region is uncommon. It has been described in association with several syndromes including acetabular dysplasia, cleidocranial dysplasia, IPD or the “small patella” syndrome, IVD, ISD, hypophosphatasia, metatropic dwarfism, campomelic and spondyloepiphyseal dysplasia (1, 2, 3, 4, 5, 8, 9, 10).

In the first patient the diagnosis of a spinal dysraphism was associated with the existence of the characteristic facial, pelvic and spinal abnormalities of IVD. In our case progression of the spinal curve to more than a moderate degree would be expected because of two determining factors in prognosis : IVD and diastematomyelia. In patients with either IVD or diastematomyelia there is an increasing tendency for scoliosis to develop with age (3, 4, 7). However, in our patient there was neither a significant progression of the scoliotic curve, nor any neurologic deficit. On the other hand, diastematomyelia is a well known entity associated with tethered cord. The fixation or tethering of the spinal cord apart from the inability of the conus to ascend to its normal level, significantly reduces cord mobility. The latter may cause vascular insuf-



Fig. 5. a. — AP X-ray of the pelvis indicated bilateral absence of most of the pubis; **b.** — a CT-scan showed bilateral hypoplasia of the ischiopubic segment.

iciency with stretching of the spinal cord. Rapid growth is therefore very likely to produce neurological deterioration in previously asymptomatic children. However, even if a patient escapes deficits during ageing, like the one presented in this report, neurological and musculoskeletal evaluation should be performed at periodic intervals, especially during the fifth and sixth decade when the onset of kyphosis with ageing may lead to neurological deterioration (12).

In the second patient a sporadic case of IPD or the “small patella” syndrome was diagnosed. Differential diagnosis from the more common and potentially more serious nail–patella syndrome (hereditary osteo-onychodysplasia or Turner –

Fong syndrome) was based on the absence of nail dysplasia, elbow deformities or iliac horns (11). In most reported cases IPD is associated with other malformations such as anomalies of the hips, including CDH, and feet, such as talipes equinovarus (2, 12). The patient presented exhibited a combination of the syndrome with scoliosis that has not been previously reported.

The two patients with CDH associated with ischiopubic hypoplasia were considered to be sporadic cases. This rare coincidence has not been previously detected.

The ischial and pubic changes that have already been identified by various authors include absence of most of the ischium and inferior pubis to irregularity of the ischiopubic synchondrosis. However, a variable expression of the disorder may occasionally be expected (10). That was indicated in patient 3 who presented with complete bilateral absence of the entire pubic bone.

Although major defects of the ischiopubic bones may occasionally be identified, ischiopubic hypoplasia usually appears as a minor defect of ossification of the ischiopubic synchondrosis. The appearance of this rare anomaly in young children should always be regarded as a syndromic constituent, and the patient should be carefully evaluated and followed until skeletal maturity.

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