

## Clinical Report

# Czech Dysplasia Metatarsal Type

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**We report three further patients of the recently described new bone dysplasia—dominantly inherited pseudorheumatoid arthritis. The patients of this report have a similar clinical history, the same distinctive phenotype and almost identical radiographic findings. The only major difference is absence of weather dependent articular pain which characterized the family of the previous study. This report expands the clinical data of this bone dysplasia. All patients are Caucasians and originate from different parts of the Czech Republic. It seems that this disorder is quite a common constitutional bone disorder in this country. We propose the name of Czech Dysplasia Metatarsal Type for this unique disease.**

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**KEY WORDS:** bone dysplasia; hypoplasia; dominantly inherited pseudorheumatoid arthritis; platyspondyly

### INTRODUCTION

Recently we have reported a family with characteristic clinical history, unique phenotype, and progressive incapacitating bone dysplasia [Marik et al., 2004]. The history of this family was characterized by weather dependent articular pain and progressively disabling bone disease. The unique phenotypic hallmark was hypoplasia/dysplasia of the toes. Distinctive radiographic features included narrowing of the articular and intervertebral disc spaces, platyspondyly with rectangular lumbar spinal canal (lack of widening of the interpedicular distance as one descends from L1 to L5), pelvic and proximal femoral dysplasia. The upper extremities were minimally affected.

In this report we describe three patients with similar foot abnormalities but without history of rheumatoid-like pain and with more discreet radiographic changes. Similar to the previous family all the patients are from the Czech Republic. These patients expand our knowledge about this noteworthy disorder.

### CLINICAL REPORT

#### Patient I

This 19-year-old boy was born after an uncomplicated pregnancy and delivery. Birth weight was 3,250 g, length 49 cm. The patient was under medical supervision from the age of 1 year because of waddling gait secondary to shortening of the left leg. Radiographic examinations during his childhood disclosed hip dysplasia, irregularity of the vertebral plates of the thoracic spine with narrowing of the intervertebral discs and hypoplastic/dysplastic changes of the 4th toes. At 19 years of age he complained of hip and back pain after exercise. His height was 168 cm, weight 77 kg. There was accentuated thoracic kyphosis and limitation of hip movements. There was hypoplasia of the 4th toes (Fig. 1A). Radiographic examination documented minimal flattening of the thoracic vertebral bodies with irregularity of the vertebral plates and narrowing of the disc spaces. In the pelvis there was uneven outline of the acetabular cavities and erosion in the right femoral head (Fig. 1B,C). The upper extremities were grossly normal. His father has a history of "hip problems." His feet are normal. Radiographs of the hips showed abnormal, slightly bowed proximal femora and minor coxa vara. The mother and two siblings are normal.

#### Patient II

This 15-year-old girl was born by caesarean section at the 38th week of gestation to a gravida 1 para 1 mother. Birth weight was 2,600 g, length 45 cm. Hip dysplasia was recognized after birth. Her psychomotor development was normal in the first decade of life. Since the age of 10 she has complained of hip pain after prolonged walking. At the age of 15 her height was 169 cm, weight 54 kg. There was accentuated thoracic kyphosis and scoliosis convex to the right in the thoraco-lumbar spine with restriction of the spine and hip movements. There was hypoplasia of the 3rd and 4th toes (Fig. 2A). Radiographic examination documented flattened vertebral bodies with irregular vertebral plates and narrowing of the intervertebral disc spaces. The lumbar spinal canal was rectangular in A-P projection. There were dysplastic changes in the pelvis, hips, and proximal femora. There was minor flattening of the knee, ankle, distal radial epiphyses and hypoplastic/dysplastic changes in the 3rd and 4th toes (Fig. 2B–D). The upper extremities were grossly normal. Family history revealed that the mother has similar feet deformity (Fig. 2E). She had progressive spine pain and had undergone bilateral hip replacement. She refused to be examined. A younger brother is normal.

#### Patient III

This 14-year-old girl was born after an uneventful pregnancy and delivery to a gravida 3 para 1 mother. Birth weight was 3,200 g, length 43 cm. She has been under medical supervision

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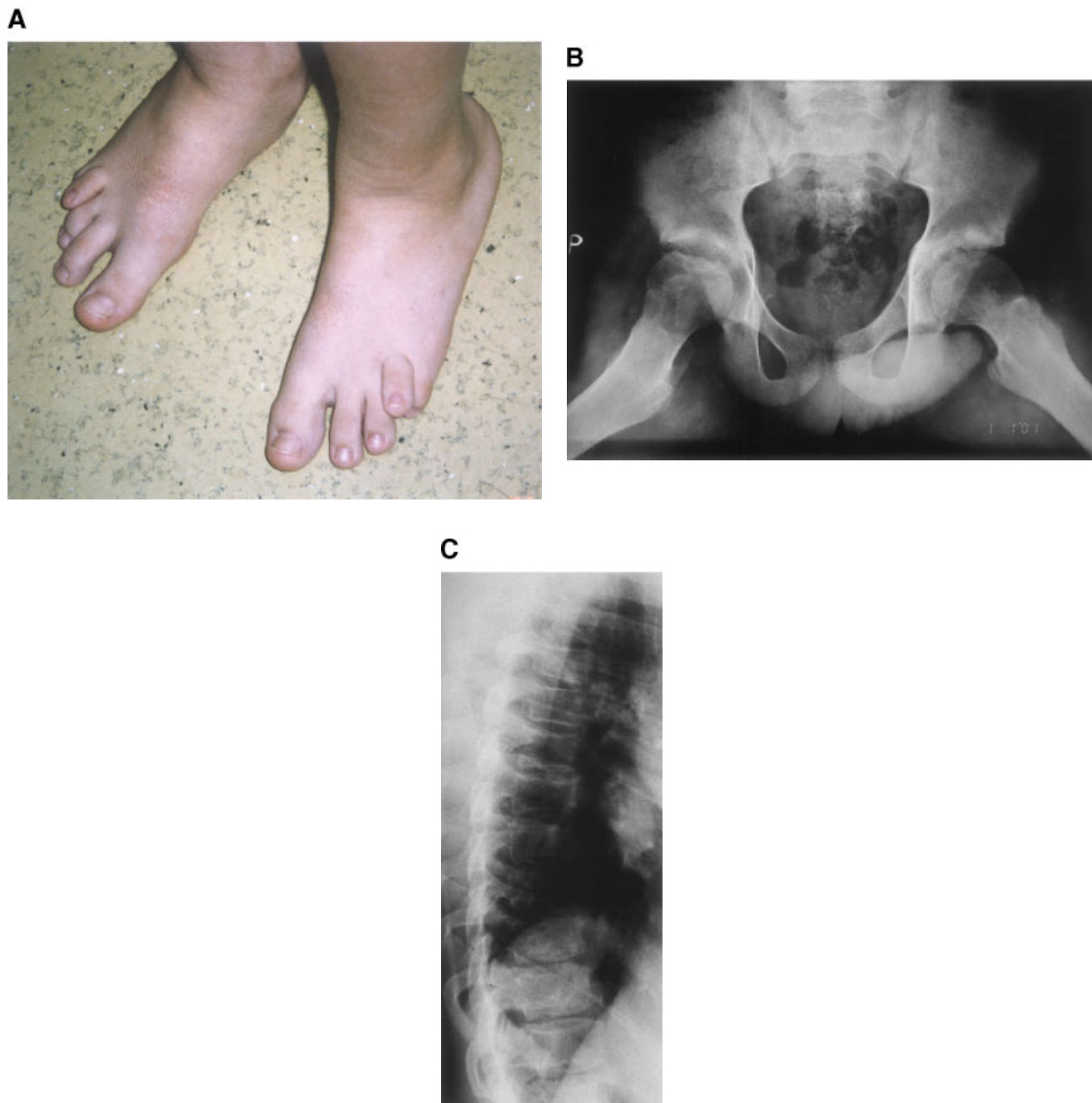


Fig. 1. **A–C:** A 13-year-old. Hypoplastic/dysplastic 4th toes. **B & C** 15-year-old. **B:** Platyspondyly. Irregular vertebral plates. Narrowed disc spaces. **C:** Irregular, sclerotic upper part of the acetabulae. Erosion at the upper part of the right femoral head. [Color figure can be viewed in the online issue, which is available at [www.interscience.wiley.com](http://www.interscience.wiley.com).]

for disproportionate short stature in the Paediatric Research Institute in Brno and Motol Teaching Hospital in Prague. Extensive biochemical investigations including mucopolysaccharides and oligosaccharides were normal. At the age of 14 her height was 134 cm, weight 41 kg. There was shortening of the trunk in relation to the extremities (upper height 70.5 cm = -4.2 SD, lower height 63.5 cm = -3.4 SD). There

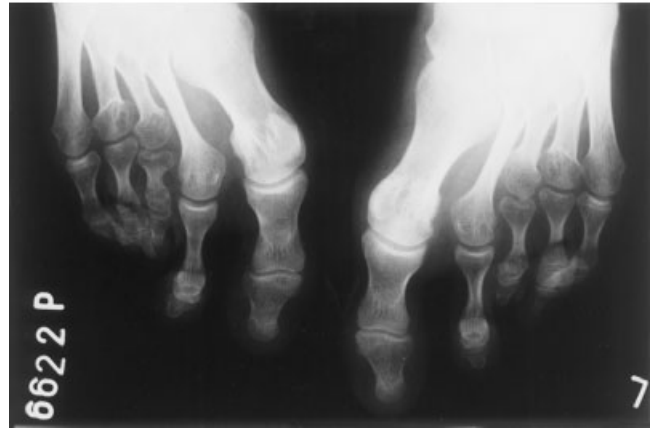
was scoliosis convex to the right in the thoraco-lumbar spine. There was restriction of the spine and ankle mobility. The hip range mobility was increased. The 3rd and 4th toes and the right and the 4th toe on the left were hypoplastic (Fig. 3A). Radiographic examination documented platyspondyly with irregularity of the vertebral plates and minor narrowing of the disc spaces. The lumbar spinal canal was rectangular in

Fig. 2. **A–E:** A–D: 16-year-old. **A** Hypoplasia of the 3rd and 4th toes. **B:** Hypoplastic/dysplastic 3rd and 4th metatarsals and toes. **C:** Severe platyspondyly. Irregular vertebral plates. Narrowed intervertebral disc spaces. **D:** Rectangular lumbar spinal canal. **E:** Narrow iliac wings. Narrowed hip joint spaces. Coxa valga. Narrow, short femoral necks. Prominent trochanters. The metallic artefact is a beauty ring. **F:** Feet of the mother of the patient, showing identical toe deformity to the daughter. [Color figure can be viewed in the online issue, which is available at [www.interscience.wiley.com](http://www.interscience.wiley.com).]

A



B



C



D



E



F



Fig. 2.

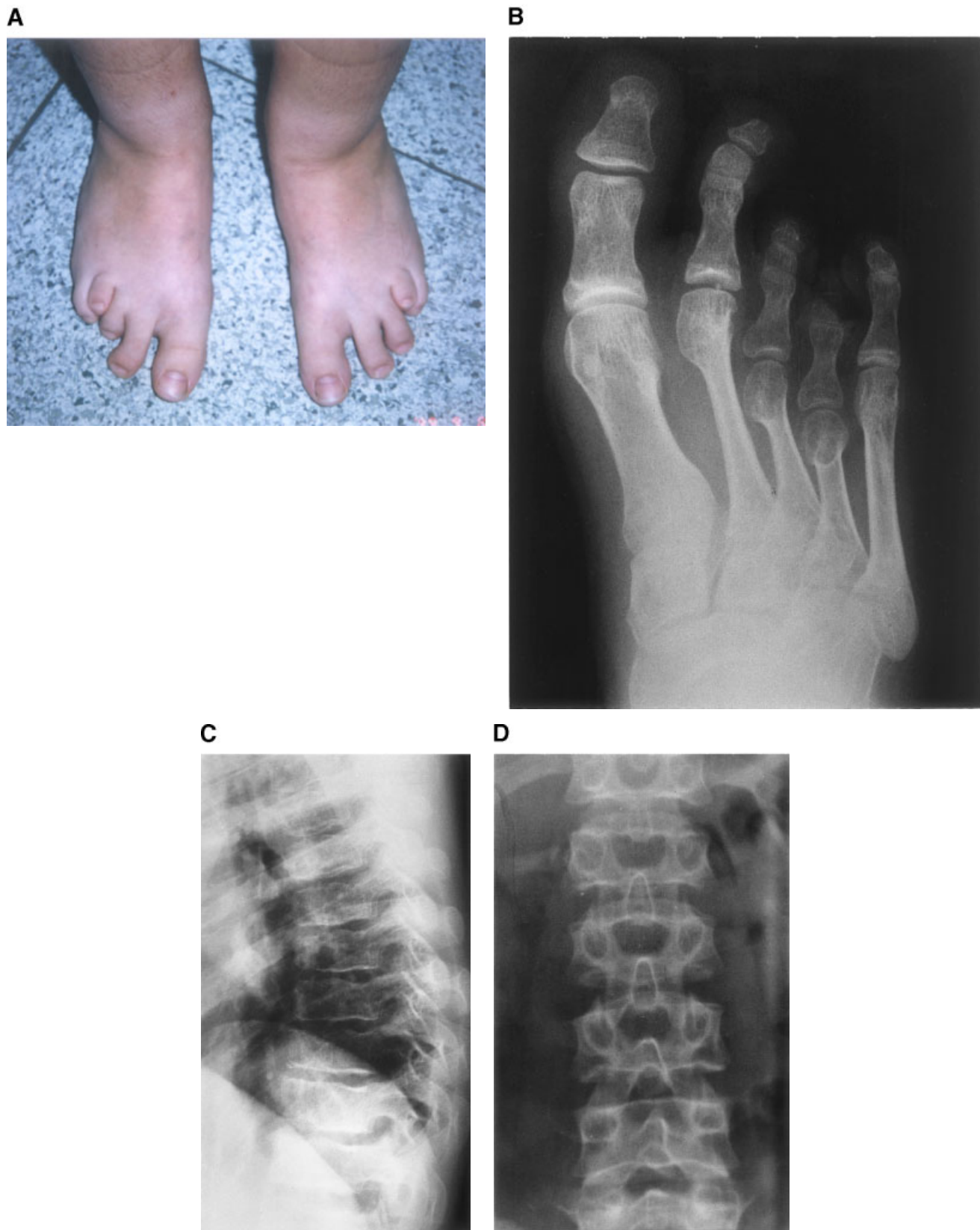


Fig. 3. A–E: 12-year-old. A: Hypoplasia of the right 3rd and 4th and left 4th toes. B: Hypoplastic/dysplastic 3rd and 4th metatarsals and toes. C: Platyspondyly. Irregular vertebral plates. Narrowed disc spaces. D: Rectangular lumbar spinal canal. E: Short iliac bodies. Flattened capital femoral epiphyses. Short femoral necks. Prominent trochanters. [Color figure can be viewed in the online issue, which is available at [www.interscience.wiley.com](http://www.interscience.wiley.com).]





Fig. 3. (Continued)

A-P projection. There were dysplastic changes in the pelvis, hips, and proximal femora. There was flattening of the medial part of the proximal tibial epiphyses and hypoplasia/dysplasia of the 3rd and 4th metatarsals on the right and 4th metatarsal on the left (Fig. 3B–E). The upper extremities were grossly normal.

Biochemical investigations disclosed elevated markers of osteosynthesis and normal values of markers of osteoresorption.

## DISCUSSION

The difference in clinical history between the previously reported family of 4 which we named “Dominantly Inherited Pseudorheumatoid Arthritis” [Marik et al., 2004] is the absence of skeletal pain related to weather conditions. The shared features are hip and back pain. The distinctive, common phenotypic feature is the hypoplasia/dysplasia of the toes. The skeletal abnormalities are similar. They are localized predominantly in the spine, pelvis, hips, and feet. They include mild platyspondyly with irregularity of the vertebral plates, narrowing of the joint and intervertebral disc spaces, rectangular shape of the lumbar spinal canal in the A-P projection, pelvic and proximal femoral dysplasia.

The two older members of the affected family reported as “Dominantly Inherited Pseudorheumatoid Arthritis” had a very severe, incapacitating clinical course. We are unsure about the clinical course in these patients as the mother of Patient II refused to be examined. We presume that she was severely affected, as she has required an early hip replacement.

In the patients reported here, skeletal problems were recognized early in life although no exact diagnosis was attempted. The clue to early diagnosis of the condition is

hypoplasia/dysplasia of the 3rd and/or 4th toes. Family history may reveal that similar toe malformations and “bone disease” has been present in other members of the family. Distinctive radiographic features in our patients are that the spine, pelvis, and lower extremities are affected whereas the upper extremities are grossly normal. The differential diagnosis of this disorder is with spondylo-epiphyseal dysplasias and progressive pseudorheumatoid dysplasia. Although in spondylo-epiphyseal dysplasias the lower extremities and the pelvis are often more severely affected than the upper extremities, the latter usually show epiphyseal involvement and retarded bone age. Some spondylo-epiphyseal dysplasias effect short tubular bones but none pertains exclusively to the fibular ray toes [Kozlowski and Beighton, 2001; Maroteaux and Le Merrer, 2002; Spranger et al., 2002]. Progressive pseudorheumatoid dysplasia is characterized by short stature, generalized progressive joint contractures, osseous expansion of the ends of the tubular bones, notably the phalanges. It is inherited as a recessive trait [El-Shanti et al., 1997]. Patients with Czech dysplasia metatarsal type are of normal height and their hands are grossly normal. Inheritance is dominant with variable expressivity. It is interesting that all the patients with hypoplasia/dysplasia of the toes and skeletal dysplasia are from different areas of the Czech Republic. Reviewing family histories we perceived that this bone dysplasia could be traced through several generations in some patients.

We conclude that patients in the previous study and those described in this report have the same disorder which is probably a relatively common constitutional bone disease in the Czech Republic. We think that the name “Czech dysplasia metatarsal type” would be a more suitable name than “Dominantly Inherited Pseudorheumatoid Arthritis,” as not all the members experience the pain associated with weather conditions. The different severity of the clinical course and radiographic changes in our patients is not surprising. Variable expressivity is a common feature of dominantly inherited diseases.

It would be interesting to know if similar patients were observed in other parts of the world, specifically countries neighboring the Czech Republic—Austria, Germany, Hungary, Poland, and Slovakia.

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