

Esetismertetés

Czech dysplasia metatarsal type

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We report a further female patient with the recently described new bone disease, Czech dysplasia metatarsal type. Czech dysplasia metatarsal type (CDMT) is an autosomal dominant debilitating disorder. Its constant phenotypic trait is hypoplasia/dysplasia of the 3rd and/or 4th toes. "Congenital hip dysplasia" or "hip disease" is commonly evoked in the family history. The clinical course may be severe, incapacitating the patients early in life, or progress slowly with increasing hip and spine pain. This girl's phenotype and radiographic findings are similar to the seven previously reported cases.

**Czech dysplasia metatarsal type,
spondyloepiphyseal dysplasia,
autosomal dominant spondyloarthropathy,
brachydactylyies**

Cseh dysplasia metatarsalis típusa

Egy nőbeteg esetét ismertetjük, aki egy újonnan leírt csontbetegség, a cseh dysplasia metatarsalis típusában szenvedett. A cseh dysplasia metatarsalis típusa (CDMT) autoszomálisan öröklődő, gyengességgel járó betegség. Állandó fenotípusú jellemzője a 3. és 4. ujj hypo-, illetve dysplasiája. Congenitalis csípődysplasia vagy csípőbetegség gyakran előfordul a családi anamnézisben.

A klinikai lefolyás súlyos lehet, a betegség mozgásképtelenné teheti a beteget az élete elején, vagy lassan progresszióval fokozódó csípő- és gerincfájdalommal. A leány fenotípusa és a betegség radiológiai képe hasonló a korábban közölt hét esetéhez.

**Cseh dysplasia metatarsalis típusa,
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Recently we reported a family with progressive disabling bone dysplasia resembling pseudorheumatic arthritis¹. A phenotypic hallmark of this family were the hypoplastic/dysplastic 3rd and 4th toes. This report was followed by description of three unrelated children with much the same phenotype but without rheumatic-like clinical history and less disabling X-ray findings². We concluded that all the patients have the same bone dysplasia which we named Czech dysplasia metatarsal type (CDMT). All patients were from Czech Republic.

In this paper we report a girl with Czech dysplasia metatarsal type, and discuss the differential diagnosis with bone dysplasias and brachydactylies.

CASE REPORT

This three years two months old girl presented to the Ambulant Center for Defects of Locomotor Apparatus in Prague because of genu varum, planovalgus position of the feet and slight lateral displacement of the patellae with the diagnosis Schmidt metaphyseal dysplasia.

She was born after an uneventful 38 week pregnancy and delivery to a 26 year-old mother and 33 year-old father. Birth weight 3500 g, length 45 cm. A long trunk and short lower extremities were noted after the birth. There is a history of "hip dysplasia" in the mother's family (Figure 1). The 157.6

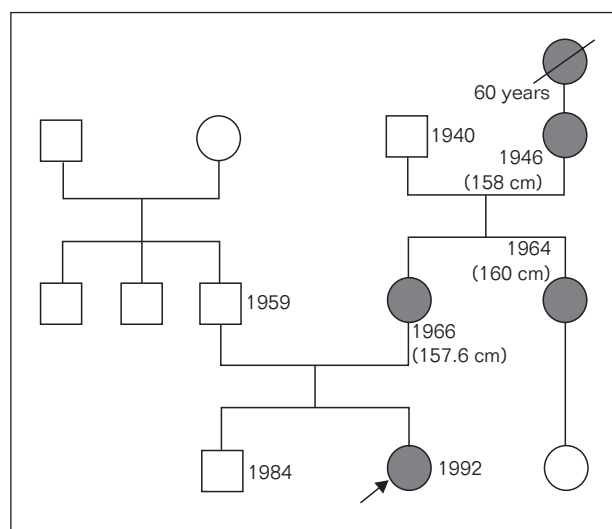


Figure 1. Family tree

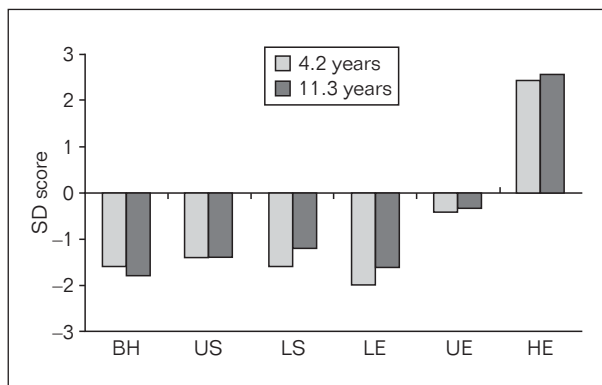


Figure 2. Morphogram

BH: height; US: upper segment; LS: lower segment; LE: lower extremity; UE: upper extremity; HE: epicondyle distance of humerus



Figure 3. 13 year-old. Hypoplasia of the left 3rd and right 4th toes

cm tall mother was diagnosed as "pseudo Perthes disease" at the age of seven years. The mother, her sister and grandmother were similarly affected. The living members of the family refused to be examined. The 33 year-old father and older son are healthy.

At the time of examination some further mild phenotypic abnormalities were noted – wide nasal bridge, narrow gothic palate, epicanthus, lumbosacral hyperlordosis, slight shortening of the extremities. Additionally hypoplasia/dysplasia of



Figure 4. 11 year-old. Hypoplasia/dysplasia of the left 4th metatarsal



Figure 5. Six year-old. Skeletal maturity is delayed

the right 4th and left 3rd toes was noted. Her height was 90.5 cm (3–10 centile), weight 15.5 (90–95 centile). Because of mild genu varum and 2nd degree planovalgus deformity of the feet she underwent corrective orthopedic treatment.

She was re-examined at the ages of four years and two months and 11 years and three months (*Figure 2*). At the age of 11 years three months her height was 136 cm (10–25 centile), weight 38 kg. There was shortening of the trunk and the lower extremities. Her face was slightly dysmorphic – wider nasal bridge and epicanthus. Her palate was high. The eyes, ears and teeth were normal. There was hyperlordosis, decreased muscle tone and hyperlaxity of the joints.

The distal phalanx of the thumb was slightly shortened. The most noticeable phenotypic feature was shortening of the 4th right and 3rd left toes (*Figure 3*).

The routine blood and urine examination and extensive biochemical examinations including markers of bone metabolism were all normal.



Figure 6. Three year-old. Oval shaped vertebral bodies. Unequal width of the intervertebral disc spaces

The X-ray examination demonstrated minimal platyspondyly with some narrowing of the intervertebral disc spaces and flattening of the capital



Figure 7. 13 year-old patient. *a)* Minor platyspondyly. *b)* Flattening capital femoral epiphyses

femoral epiphyses. The fibulae were thin, elongated distally and there was hypoplasia/dysplasia of the right 4th and left 3rd metatarsals. The carpal bone age was delayed and there was some shortening of the distal phalanx of the thumb (*Figures 4–9*).

DISCUSSION

Czech dysplasia metatarsal type is an autosomal dominant debilitating disorder. Its constant phenotypic trait is hypoplasia/dysplasia of the 3rd and/or 4th toes. “Congenital hip dysplasia” or “hip disease” is commonly evoked in the family history. The clinical course may be severe, incapacitating the patients early in life¹, or progress slowly with increasing hip and spine pain². Other phenotypic features such as shortening of the trunk and lower extremities are usually minor, inconsistent and uncharacteristic. Radiographic changes are localised to the lower extremities and the spine. The upper extremities and the skull are grossly normal.

The common feature of the present and the previously reported patients^{1, 2} is the hypoplasia/dysplasia of the 3rd and/or 4th toes and the history of dominantly inherited “hip disease”. The generalised bony changes in this patient are similar to those of the previous reports.

The differential diagnosis of Czech dysplasia metatarsal type is with spondylo-epiphyseal dysplasias (SED)^{3, 4} and autosomal dominant spondyloarthropathy (ADSA)³.



Figure 8. 11 year-old patient. Flattening of the medial part of the proximal tibial epiphyses

In spondylo-epiphyseal dysplasias the lower extremities are often more severely affected than the upper extremities but none of spondylo-epiphyseal dysplasias shows hypoplasia/dysplasia of the toes.

The clinical features of autosomal dominant

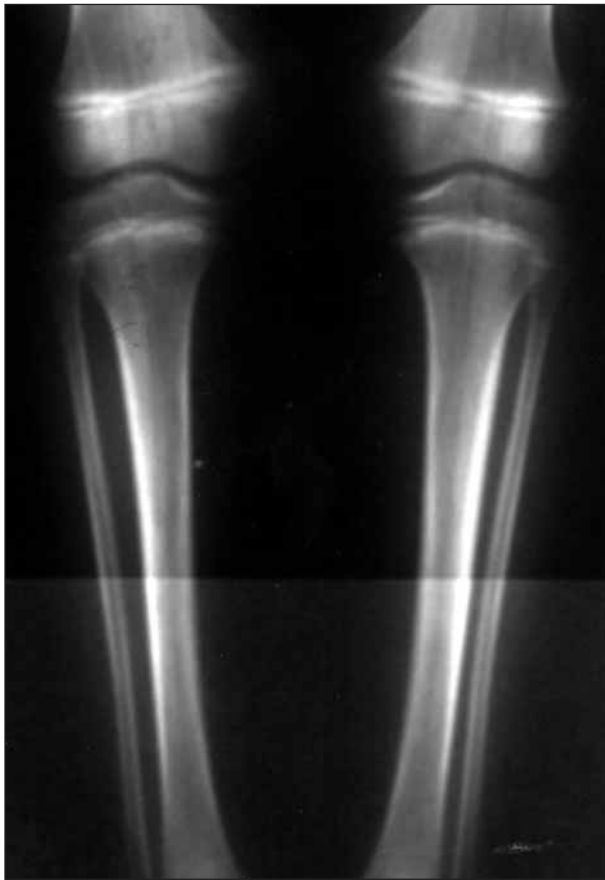


Figure 9. Six year-old. Thin fibulae

spondyloarthropathy are similar to Czech dysplasia metatarsal type. Short, broad metacarpals may be present in autosomal dominant osteoarthropathy



Figure 10. Albright hereditary osteodystrophy. 10 year-old patient. a) Hypoplasia of both 3rd and 4th toes. b) Hypoplasia/dysplasia of the right 3-5th and left 4-5th metacarpals, middle phalanges of the 2nd and 5th fingers and the distal phalanges. Note cone shaped epiphyses of the middle phalanx of the 2nd finger

whereas in Czech dysplasia metatarsal type the upper extremities are grossly normal. Increase epicondyle humeral distance in our case was an interesting finding. The major differential hallmark is however the hypoplasia/aplasia of the 3rd and/or 4th toes features absent in autosomal dominant spondyloarthropathy and present in all patients with Czech dysplasia metatarsal type.

Other bone dysplasia are unlikely diagnoses. Progressive pseudorheumatoid chondrodysplasia is inherited as an autosomal recessive trait. Rheumatoid arthritis has more debilitating clinical signs; positive rheumatoid tests and radiographic osteoporosis are other distinctive features. None of them has hypoplasia/dysplasia of the toes.

Hypoplasia/dysplasia of the toes is a feature of some of the brachydactylies, namely, Albright hereditary osteodystrophy (AHO) (includes various forms of hypoparathyroidism and pseudohypoparathyroidism) and brachydactyly E (BE). In Albright hereditary osteodystrophy, shortening of 3–5th metatarsals is accompanied by shortening of the metacarpals, distal phalanges and middle phalanx of the index finger (Figure 10). Distinctive phenotype (short stature, obesity, mental retardation, seizures) and abnormal biochemistry are usually present. Symmetrical hypoplasia/dysplasia of the metacarpals/metatarsals and shortening of the distal phalanges of the fingers is characteristic of brachydactyly E (Figure 11). However there are no other major clinical or radiographic findings and the height of the patients is normal.

Czech dysplasia metatarsal type is clinically and radiologically a *distinct entity*. The similarity in



Figure 11. Brachydactyly E. 13 year-old patient. a) Hypoplasia of both 4th toes. **b)** Shortening of the metacarpals. The left 5th metacarpal is most severely affected. Short distal phalanx of the thumb

abnormalities of the toes suggests that *there might be some molecular affinity* between the latter, Albright hereditary osteodystrophy and brachydactyly E. Because of logistical and financial reasons molecular studies could not be performed in our patients.

All our patients with Czech dysplasia metatarsal type were from different parts of Czech Republic. It is possible that this disorder might be present in the neighbouring countries such as Austria, Hungary, Slovakia and Poland.

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