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Cone-shaped knee epiphyses – important diagnostic sign

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Summary

Background:	The first case of Bellini bone dysplasia in the Polish literature is reported.
Case Report:	Skeletal survey of a 3½ year old girl with a history of mental retardation, short stature, dysmorphic face and short hands and feet was received for consultation.
Conclusions:	Cone shaped knee epiphyses is a diagnostic radiographic sign of Bellini bone dysplasias. This disorder is recognizable on the basis of skeletal survey alone. For diagnosis of the type of Bellini bone dysplasia, concurrent phenotypic evaluation of the patient is necessary.
Key words:	cone-shaped epiphyses • knee • osteomyelitis • trauma • vitamin A
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Background

Cone-shaped knee epiphyses is a diagnostic sign of Bellini bone dysplasia (BBD) [1–8]. Three types of this bone dysplasia are recognized.

Radiographs of a 3½ year old girl with the history of mental retardation, short stature, dysmorphic face, and short hands and feet were received for consultation. The changes were characteristic for BBD Type II (Figures 1,2).

BBD type I

In 1966 Bellini et al. [1] described a patient with short stature, alopecia and distinctive bony abnormalities consisting of *cone-shaped knee epiphyses*, cupping of all metaphyses of the tubular bones of the hands and feet, and advanced bone age. Jequier et al. [2] confirmed the identity of the disorder in 1981. Verloes et al. [3] suggested for Bellini type I the name *trichoscyphodysplasia*.

BBD type II

In 1984 Bellini et al reported two sibs with short stature, normal face, *normal hair* in the older sibling and *sparse hair* in the younger brother, short limbs, knee flexion due to hypomobility of knee joint and disproportion between trunk and lower extremities [4]. The *radiographic abnormalities were similar to BBD type I*. Additionally there was

marked *shortening of the tubular bones* of the hands and feet – distinctive constant feature of TYPE II. Other clinical data of TYPE II are variable (Table 1). Usually the face is dysmorphic, the mental development delayed and the bone age advanced [3–5]. Verloes et al. [3] suggested the name *acroscyphodysplasia* for BBD type II.

BBD type III

In the following years patients were reported with *similar knee changes to BBD Type I and II* – cone-shaped knee epiphyses – but with *normal hair* and *normal hands* [6–8].

Discussion

Our patient showed all the major radiographic features of BBD Type II – cone-shaped epiphyses of the knees, hypoplasia of the facial bones, short tubular bones of the hands and feet and advanced carpal/tarsal bone age [3–5]. Her mental development was retarded.

Skeletal survey performed at the age of 1½ years showed diagnostic radiographic findings – cone shaped knee epiphyses, short tubular bones of the hands and advanced carpal/tarsal bone age. There was also shortening of the tibia with relative proximal and distal elongation of the fibula. The vertebral bodies were almost square shaped and there was hypoplasia of the antero-superior aspect of L2. The lateral parts of the clavicles were hypoplastic (Figure 1A–G).

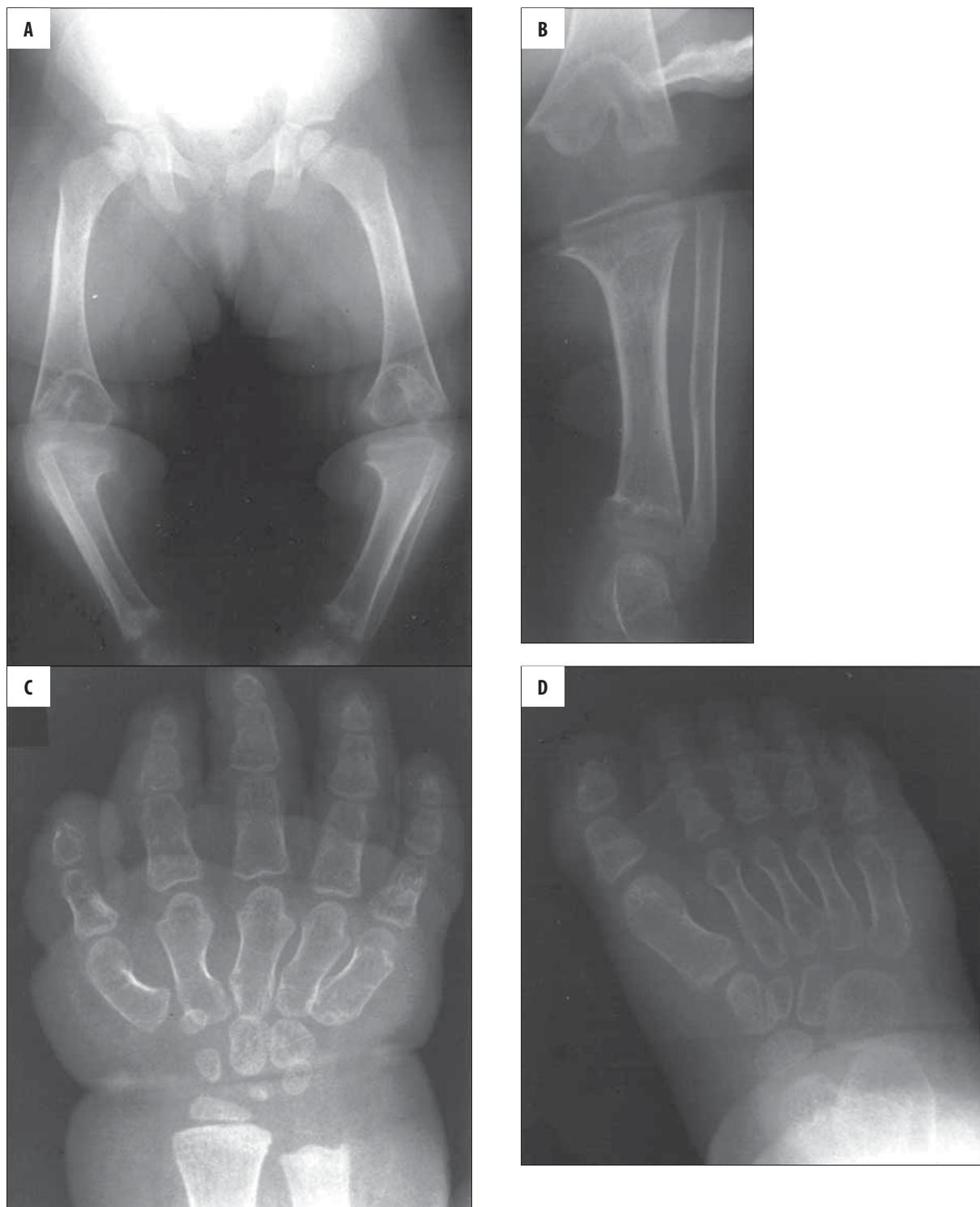


Figure 1. 1 year old. (A,B) Cone shaped distal femoral epiphyses. Short tibia. Both distal and proximal elongation of fibula. (C,D) Short tubular bones of the hand and foot. Advanced carpal & tarsal bone age.

At the age of 3½ years there was little further growth of the hands. There was some regress of the spinal abnormality (Figure 2A,B).

Although for reasons of clarity, separation of BBD into three types is warranted, there is some blurring between the three types. The second sibling of Bellini Type II

[4] had sparse hair and the first patient with Type II of Verloes et al. [3] was mentally normal and her bone age was not advanced. Two sisters of Kozłowski et al. [7] had cataracts and pes equino-varus. The older girl had additionally imperforate anus and absent vagina. In our girl the antero-superior aspect of L2 vertebral body was hypoplastic.

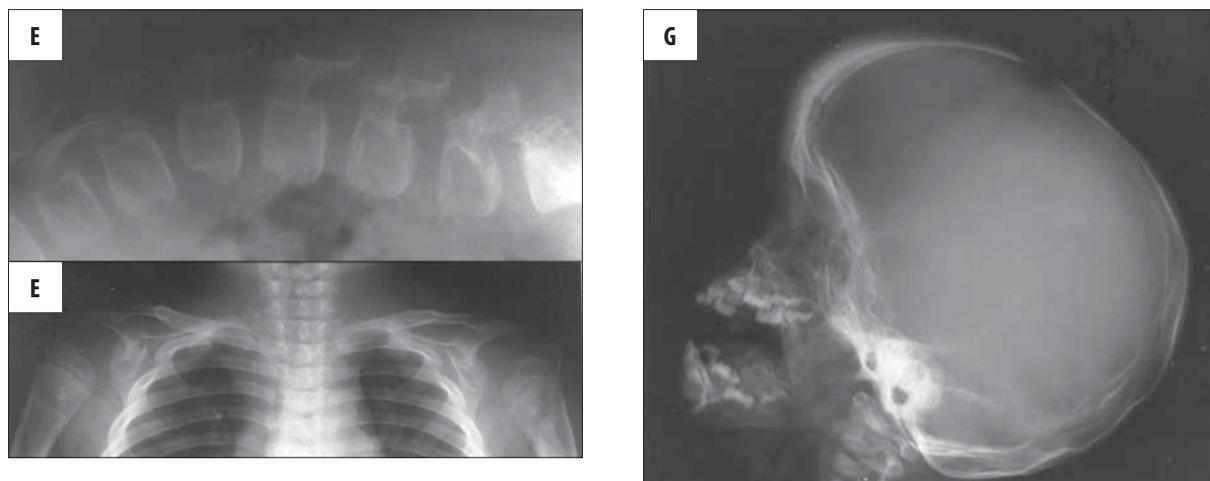


Figure 1. 1 year old. (E) Hypoplasia of the antero-superior aspect of L2 (F) Hypoplasia of the lateral part of the clavicle. (G) Hypoplasia of the facial bones. Small anterior cranial fossa. Prominent frontal and parietal bones.

Table 1. Review of clinical presentation in previous and present patient with Bellini bone dysplasia Types I–III.

Type I	Face	ED	MR	Hands	Bone age
Bellini & Bardare 1966	N	+	–	N	>
Jequier et al. 1981	N	+	–	N	>
Type II					
Bellini et al. 1984					
Pt 1	N	–	+	S	>
Pt 2	N	+/-	+	S	>
Frediani et al. 1986	D	–	+	S	>
Verloes et al. 1991					
Pt 1	D	–	–	S	N
Pt 2	D	–	+	S	>
Kozłowski 2009	D	–	+	S	>
Type III					
Kozłowski et al. 1995a					
Pt 1	N	–	–	N	N
Pt 2	N	–	–	N	N
Kozłowski et al. 1995b					
Pt 1	N	–	–	N	N
Pt 2	N	–	–	N	N
Verloes et al. 2004					
Pt 1	N	–	–	N	N
Pt 2	N	–	–	N	N
Pt 3	N	–	–	N	N

ED – ectodermal dysplasia; MR – mental retardation; N – normal; D – dysplastic; S – small; > – advanced.

The differential diagnosis of Bellini bone dysplasias is predominantly from disorders affecting the knees such as metaphyseal and spondylometaphyseal dysplasias. Bellini

Type II characterized by short hands and feet is to be differentiated from acrodysostosis, acro-mesomelic dysplasias, pseudohypoparathyroidism and pseudo-pseudohypopara-



Figure 2. 3 year old. (A) Short tubular bones. Cone shaped epiphyses are best seen in the proximal phalanges. Advanced carpal bone age. (B) The vertebral bodies are high and almost square shaped. Hypoplasia of the antero-superior aspect of L2.

thyroidism. Presence of symmetrical cone-shaped epiphyses of the knees is a diagnostic sign of BBD. For designation

of the type of BBD, phenotypic evaluation of the patient is necessary. Ectodermal dysplasia is a hallmark of Type I [1,2]. *Facial dysmorphism, short hands and feet and mental retardation* are features of Type II [3-5]. Type III is associated with *normal face, normal hands and feet and normal mental development* [6-8] (Table 1).

Some overlapping of the BBD types and additional variable abnormalities are suggestive for polygenic recessive inheritance of BBD with variable expressivity in affected individuals. Until the genetic defect of BBD is found, it remains uncertain if BBD is a single disorder or a group of related but different diseases.

Cone shaped knee epiphyses do occur as a result of infection, trauma, Vit A poisoning and multisystem inflammatory disease [9-11]. These are usually *asymmetrical* and have a distinctive *positive etiological clinical history*.

Conclusions

A 3½ year old girl with short stature, facial dysmorphism and mental retardation is reported. Radiographic examination documented symmetrical cone shaped knee epiphyses, short tubular bones and advanced bone age.. Presence of this unique malformation pattern *allows a spot diagnosis* of BBD Type II and makes further tests unnecessary. As cone shaped knee epiphyses are present already at birth, early diagnosis of BBD is possible if the disorder is known to the observer.

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