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Case Report

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SED-brachydactyly and distinctive speech: Report of a new familial case

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Summary

Background: Spondyloepiphyseal dysplasia-brachydactyly and distinctive speech (SED-BDS) is a syndrome characterized by short stature, disproportionately short limbs, peculiar face, thick and abundant hair, high-pitched and coarse voice, small epiphyses, brachymetacarpalia, brachymetatarsalia and brachyphalangia of fingers and toes, small pelvis and delayed carpal bone age, among other features.

Case Report: We report a Brazilian patient with father, brother and sister presenting with the same typical features of the syndrome. Clinically, he showed disproportionately short stature, rhizo-meso-acromelic shortness of the extremities, short hands and feet, a peculiar distinctive high-pitched voice, peculiar facies, and other features already reported as characteristic of this syndrome. Radiographic findings included shape anomalies of the vertebral bodies such as cuboid-shaped vertebral bodies, mild scoliosis, short and broad tubular bones, brachymetacarpalia, brachymetatarsalia, and brachydactyly, lumbar hyperlordosis, generalized osteopenia, and hypoplastic iliac wings.

Conclusions: Few cases have been described, as this is a rare skeletal dysplasia. This paper describes a new familial case of SED-BDS.

key words: **SED-BDS • Tattoo dysplasia • Fantasy Island syndrome**

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Background

Spondyloepiphyseal dysplasia-brachydactyly and distinctive speech (SED-BDS), also known as Fantasy Island syndrome or Tattoo Dysplasia (OMIM 611717) [1], was initially described as a "new" osteochondrodysplasia in 2 unrelated patients by Cantu et al. (1991) [2] during the 8th International Congress of Human Genetics. However, the syndrome was first defined with a case reported by J. R. Gorling – Hervé Villechaize, the actor known as Tattoo in the U.S. television series "Fantasy Island". Four years later, the 2 patients in the report by Cantu et al. in 1991 were described as having the Fantasy Island syndrome [3].

It was proposed that SED-BDS is inherited in an autosomal dominant manner, as both sexes were affected and parental consanguinity was never reported [4]. The last description was a case presented by Nunez-Reveles et al. (2009) [5] during the 59th Annual American Society of Human Genetics Meeting. We describe here a new familial case of SED-BDS that presents typical clinical and radiological features of the syndrome. This case report was approved by the Research Ethics Committee of the institution (Process no. 0447/2011).

Case Report

The propositus (Figure 1A–E), a male born in 1994, was the second child of a normal 24-year-old woman and her 27-year-old husband after an unremarkable pregnancy (40 weeks gestation) and C-section. The infant's birth weight was 3000 g and his length was 48 cm. Neuropsychological development and language acquisition were normal. At a physical examination at age 16 years his height was 104 cm and weight was 20.8 kg (< 3rd centile).

Clinically he showed disproportionately short stature, rhizomeso-acromelic shortness of the extremities, short hands and feet, a peculiar distinctive high-pitched voice, peculiar facies, and other features already reported as characteristic of this syndrome. Radiographic findings (Figure 2A–G) included shape anomalies of the vertebral bodies, such as cuboid-shaped vertebral bodies, mild scoliosis, short and broad tubular bones, brachymetacarpalia, brachymetatarsalia, brachydactyly, lumbar hyperlordosis, generalized osteopenia, hypoplastic iliac wings, severe long bone shortening, small epiphyses and generalized delayed epiphyseal ossification. The bone age was delayed 7 years; at 15 years old it was compatible with an age of nearly 8 years for males. Ophthalmologic, cardiologic and karyotypic evaluations were normal. Table 1 shows all the clinical and radiological features of the propositus.

His father and 2 siblings (1 male and 1 female) also showed typical clinical features of SED-BDS (Figure 3).

Discussion

SED-BDS is a rare genetic syndrome. To our knowledge 14 cases have been described. Two unrelated patients were described by Cantú et al. (1991) [2], Garcia-Cruz et al. (2007) [4] described a mother and her son and 1 additional patient, and another individual was described by Nunez-Reveles et al. (2009) [5]. However, the first description is attributed to J.R. Gorling, referring to the character Tattoo in the television series "Fantasy Island" played by the actor Hervé

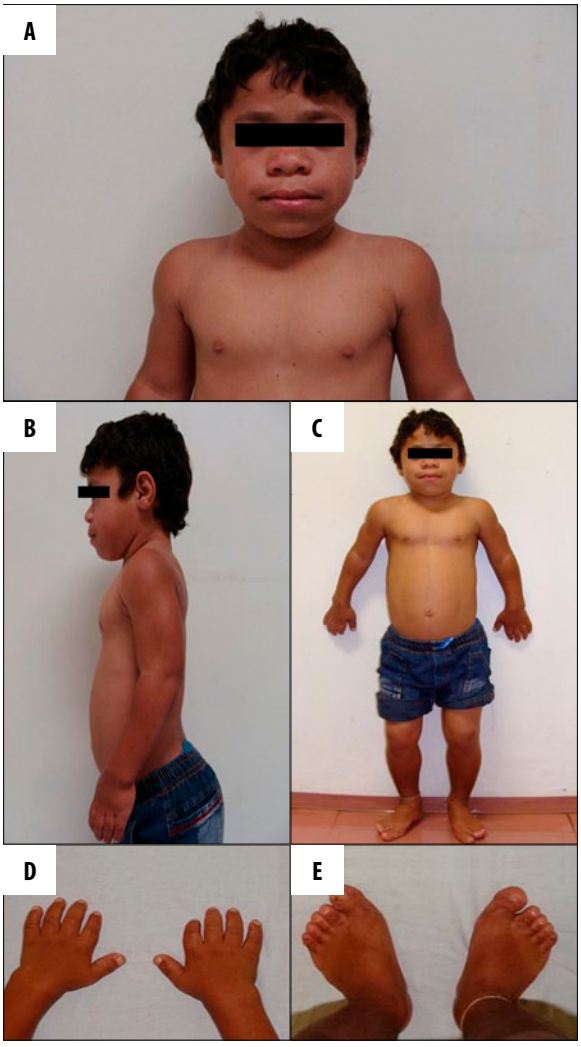

Figure 1. Frontal and lateral views of the patient. Note the peculiar round face with abundant eyebrows, upslanted palpebral fissures, short nose, long philtrum and round chin (**A**), lumbar hyperlordosis (**B**), short thorax (**C**), disproportionate short stature, rhizomesoacromelic shortness of the extremities and brachydactyly (**C–E**).

Villechaize. In addition, a father and son reported by J. Hall [3] are believed to have the same syndrome, as well as 5 other individuals reported by C. L. Johnson [6].

Here we report on a male with SED-BDS who presented nearly all the characteristics described by Cantu et al. (1995) [3]. Besides the abovementioned features, this case presented with scoliosis, spina bifida (L5 and S1) and necrosis of the femoral head. He did not present with hypoplastic 4th vertebra, bilateral coxa valga, spondylolisthesis, hypertrophy of the 1st ray, cervical kyphosis or restrictive lung disease.

The main differential diagnoses of SED-BDS are: acromicric dysplasia (OMIM 102370), geleophysic dysplasia (OMIM 231050), and Moore-Federman syndrome (OMIM 127200) [2,4,6].

Acromicric dysplasia is characterized by proportionate short stature, mild and variable facial anomalies which become

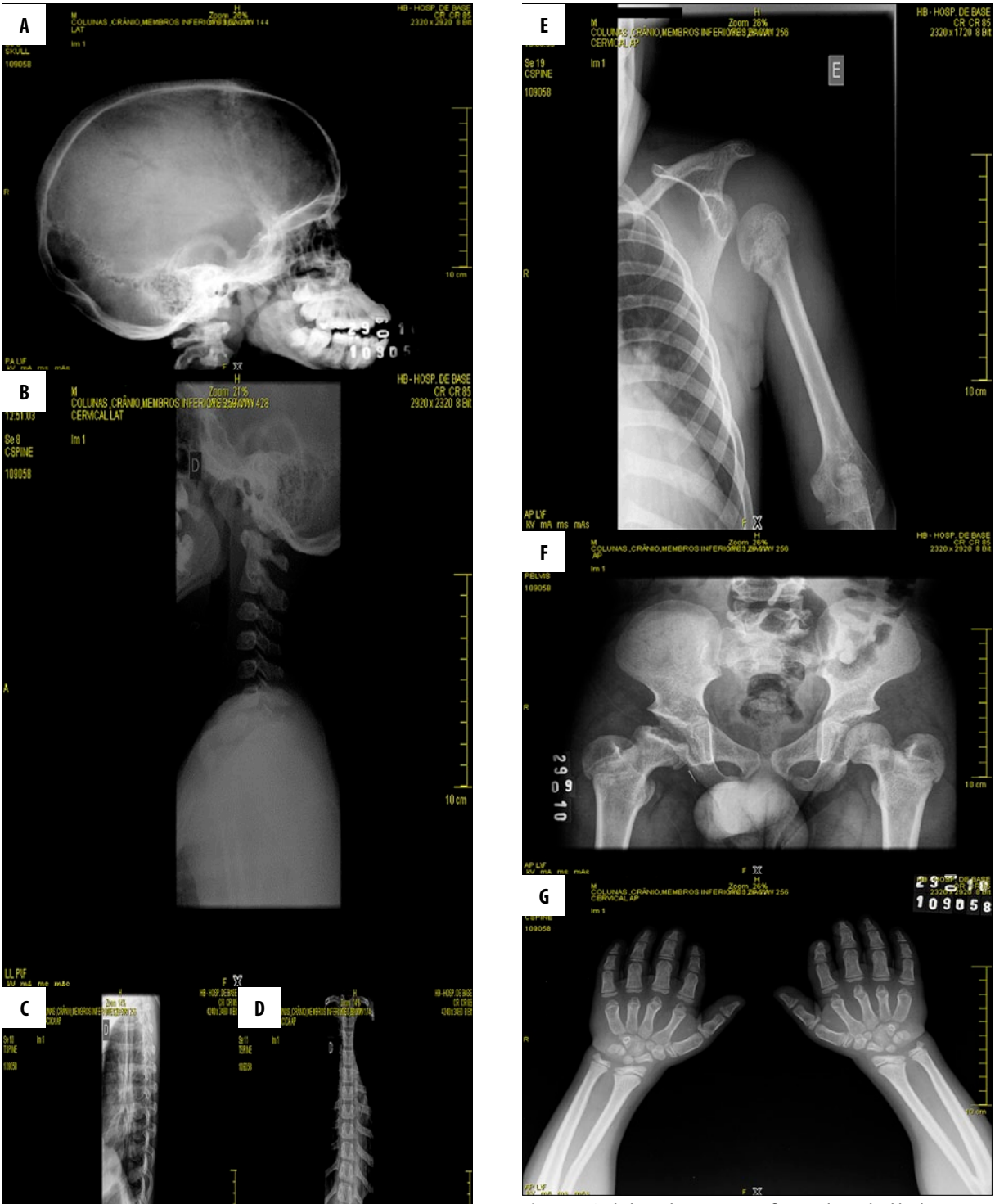

Figure 2. Radiological aspects. Note flattened vertebral bodies in c-spine (**A,B**), cuboid-shaped vertebral bodies, mild scoliosis, lumbar hyperlordosis and spina bifida L5-S1 (**C,D**), thickned clavicles (**E**), flared iliac wings and necrosis of the femoral head (**F**) and generalized brachydactily, more severe in the metacarpals (**G**).

The features of geleophysic dysplasia are proportionate short stature, 'pleasant, happy-nature' facial appearance, long, smooth philtrum, vascular, respiratory and heart alterations, *pectus excavatum*, hepatomegaly and thickened skin [9,10].

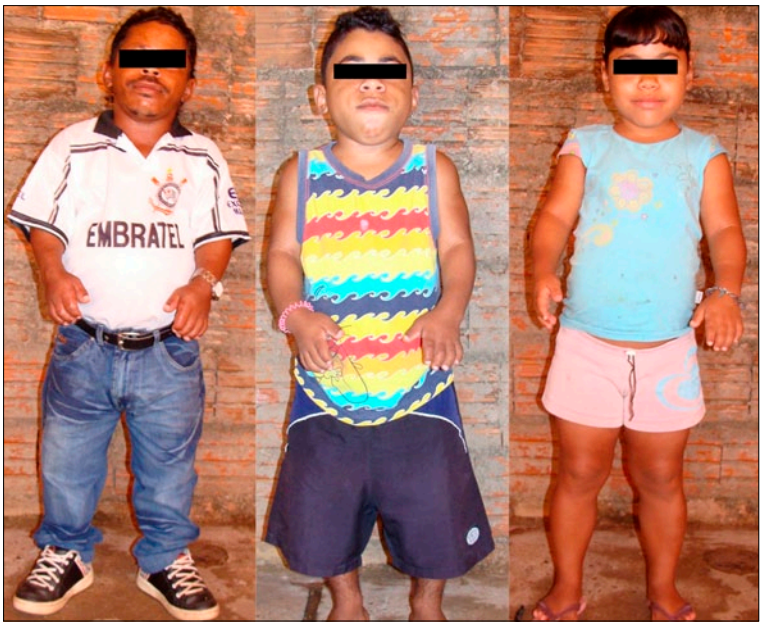

Figure 3. Frontal views of the patient's father, brother and sister presenting the same typical features of the syndrome.

Table 1. Clinical and radiological features of a current case by heading and subheading.

Heading	Subheading	Feature
Growth	Height	Disproportionate short stature
		Normal birth length
		Normal neuropsychomotor development
		Rhizo-meso-acromelic shortness
		Shortened limbs
	Other	Progressive growth redartadion
Head and neck	Face	Long philtrum
		Midface hypoplasia
		Peculiar facies
		Round face and chin
	Ears	Small pinnae
	Eyes	Abundant eyebrows and eyelashes
		Mild blepharophimosis
		Upslanted palpebral fissures
	Nose	Broad and depressed nasal bridge
	Mouth	Bulbous nose with everted nostrils
		Upturned nose
		Edge to edge bite
		Large mouth
		Thick lower lip
	Neck	Short neck
		Wide neck

Table 1 continued. Clinical and radiological features of a current case by heading and subheading.

Heading	Subheading	Feature
Chest	External features	Small thorax
		Pectus excavatum
		Thickened clavicles
Skeletal		Brachymetacarpalia, Brachymetatarsalia, Brachyphalangia
		Clinodactyly of the 5th fingers
		Cuboid vertebral bodies
		Generalized epiphyseal ossification delay
		Generalized osteopenia
		Hypoplastic iliac wings
		Joint limitations
		Limited pronosupination
		Lumbar hyperlordosis
		Necrosis of the femoral head
		Scoliosis
		Severe long bone shortening
		Shape anomalies of the vertebral bodies
		Short hands and feet
		Small epiphyses
		Spina bifida (L5 and S1)
		Spondyloepiphyseal dysplasia
Skin, nails, hair		Abundant and thick hair
		Dysplastic nails
		Hirsutism
		Lowset nuchal hair
Voice		Peculiar highpitched distinctive voice

Moore-Federman syndrome, among other things, is characterized by hypermetropia, glaucoma, asthma and hepatomegaly, cardiac anomalies, a hoarse voice and abnormal skin that feels firm and thickened [11,12]. These osteodysplasias have many clinical and radiological features in common with SED-BDS, but as the signs listed above were absent in our patients, these diagnoses were discarded.

Conclusions

The etiology of SED-BDS is still unknown. The genealogical study in this case showed that the father, brother and sister are affected too, which strongly supports the hypothesis of an autosomal dominant manner of inheritance with variable expressivity, as suggested by Garcia-Cruz et al. (2007) [4].

This is the 15th reported case and the first Brazilian case, which collaborates by delineating the clinical and radiological characterization of this rare skeletal genetic disorder that has an unknown molecular pathogenesis.

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