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# CHARACTERISTIC FACIAL DYSMORPHISM, ARACHNODACTYLY AND MENTAL RETARDATION: ANOTHER CASE

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**Summary:** *Characteristic facial dysmorphism, arachnodactyly and mental retardation: another case:* A severely retarded girl is described with microcephaly and long face, hypertelorism, strabismus divergens, thin upper lip, slender extremities, resembling the cases reported by de Die-Smulders *et al.*

**Key-words:** Arachnodactyly – MCA/MR syndrome – Dysmorphology.

**Résumé:** *Dysmorphie faciale typique arachnodactylie et retard mental: Présentation d'une nouvelle observation:* Dans cet article nous présentons l'observation d'une fille âgée de 11 ans présentant un retard mental sévère associé à un syndrome dysmorphique typique (microcéphalie, facies long, hypertélorisme, strabisme et habitus longiligne) comme rapporté par de Die-Smulders et collaborateurs.

**Mots-clés:** Arachnodactylie – Retard mental – Dysmorphologie.

## CLINICAL REPORT

The proband, an 11-years-old girl, was presented because of developmental delay. She was the second child of non-consanguineous healthy parents. An older sister was healthy. A brother and a sister of the father were mentally retarded. Pregnancy was uneventful. Birth weight was 3080 g and length was 50 cm, both on the 50th centile. Birth was complicated by meconium aspiration. Because of severe feeding problems and upper respiratory tract infections, several hospitalisations were necessary. At the age of 3 years she was a floppy infant with severe psychomotor retardation, with a length of 90 cm, a weight of 11.5 kg and a head circumfe-

rence of 46 cm, all on the 10th centile for age.

Clinical examination at the age of 11 years showed a girl with a head circumference of 47.5 cm (2.5 cm < 3rd centile) and a weight of 25 kg (1 kg < 3rd centile). She had a long, narrow triangular face with an ICD of 32 mm (75th centile), telecanthus with strabismus divergens, a flat maxilla, a thin upper lip and a long flat philtrum (Fig. 1). The extremities were long and slender (Fig. 2), the hands appeared long although the total hand length was on the 3th percentile (Fig. 3). The finger joints were hyperextensible. There was clinodactyly of the 4th and 5th fingers. She had a triphalangeal thumb on the right hand. Her feet were long with extreme flatfeet. The external genitalia were normal. Neurologic examination showed spastic tetraplegia with distal hypertonia. She was severely mentally retarded and she had no speech. Technical investigations included X-rays of the total skeleton which revea-

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Figure 1: Face at the age of 11 years.

led no abnormalities except for the triphalangeal right thumb with ulnar deviation. EEG, EMG, and CT-scan of the brain were normal. Chromosomal investigation showed a normal 46,XX karyotype. Metabolic screening was normal.



Figure 3: Right hand with the triphalangeal thumb and clinodactyly of the 4th and 5th fingers.

## DISCUSSION

The present patient has an association of mental retardation with absent speech, a long face with a flat maxilla, hypertelorism, strabismus divergens, a thin upper lip, microcephaly, slender extremities and hyperextensible finger-joints. The combination of these clinical signs was reported by de Die-Smulders *et al.* (1) in two girls. This combination does not fit the common syndromes with mental retardation and marfanoid habitus

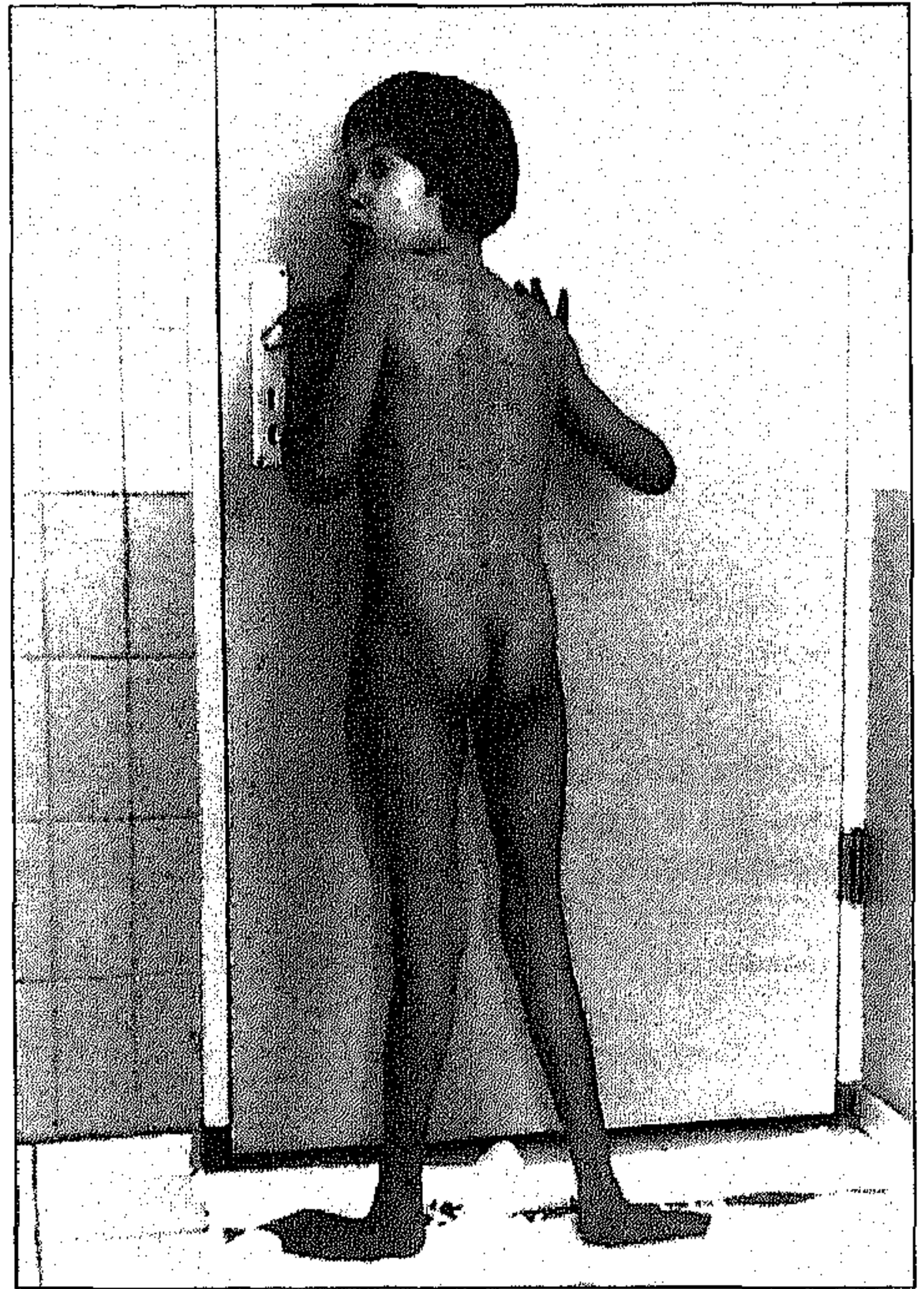


Figure 2: The long slender extremities and feet.

or Lujan-Fryns syndrome (2, 4, 5) and the family reported by Frago and Cantu (3).

Although our patient had a triphalangeal thumb, normal external genitalia, and no small mouth we believe that our patient shows a remarkable resemblance with the two patients of de Die-Smulders *et al.* (1). Description of other cases is necessary to delineate this as a distinct clinical entity and elucidate its possible etiology. Until now, only sporadic cases have been reported.

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