

BRIEF CLINICAL REPORT: SPORADIC, IDIOPATHIC MCA/MR SYNDROME WITH THE TIBIAL APLASIA/FEMORAL BIFURCATION FIELD DEFECT

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ABSTRACT

This paper reports on an 8-year-old Brazilian boy with a previously undescribed, sporadic, idiopathic multiple congenital anomaly/mental retardation (MCA/MR) syndrome of microcephaly, mild malar hypoplasia, abnormal ears and nose, cleft lip, and the tibial hemimelia/femoral bifurcation field defect.

INTRODUCTION

The tibia represents a specific developmental field and its involvement in dysmorphogenetic abnormalities has been demonstrated in sporadic, aneuploid, autosomal dominant, and recessive syndromes (Cordeiro *et al.*, 1986; Richieri-Costa *et al.*, 1987a,b; Richieri-Costa, 1990; Richieri-Costa *et al.*, 1990).

Here we report on a boy with a sporadic MCA/MR Syndrome with severe involvement of the tibial developmental field, which seems to be different from the other known syndromes involving this field.

CLINICAL REPORT

CCS (Figures 1 and 2), the propositus, was the second child of non consanguineous Brazilian parents. The 26-year-old mother, the 27-year-old father and a 12-year-old brother were normal. There were no similar cases among close relatives.

Gestation was normal, there was no known exposure to toxic or infectious agents or to X-rays and no trauma was reported. Delivery was normal, at term. Birth weight, length and OFC were not recorded. Cleft lip and lower limb anomalies were noticed at birth. Neuropsychological development was delayed.

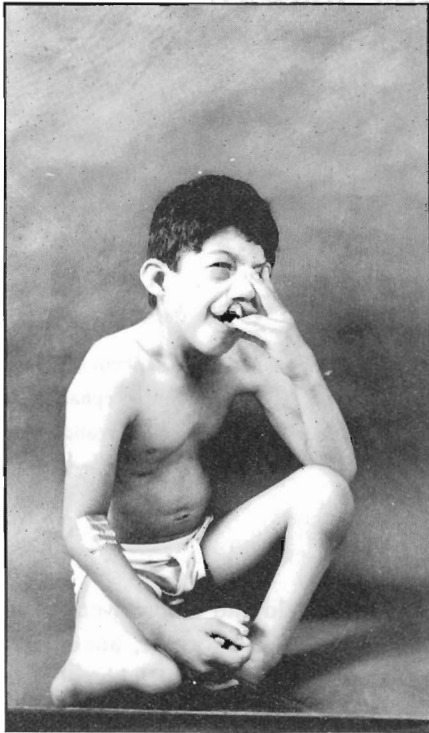


Figure 1 - Clinical aspects of the propositus.



Figure 2 - Clinical aspects of the face of the propositus.

Examination at age 8 years showed: weight 22.4 kg, OFC 47 cm. Severe mental retardation, microcephaly, flat occiput, large ears with a prominent tag at left, hypoplastic tragus, prominent anti-helix, mild antimongoloid slant of palpebral fissures, broad and asymmetric nose, right cleft lip, short lingual frenulum. The lower limbs were grossly abnormal with shortness of the legs, markedly at right. In the distal portion of the right thigh there was a medial prominence. The right leg assumed an abnormal posture, with its proximal portion overlapped by the distal portion of the right thigh. Bilateral 5-toe varus-adductus feet (Figure 3). The upper limbs were normal.



Figure 3 - Clinical aspects of the lower limbs of the propositus.

Results of laboratory blood tests were normal.

G-banded chromosomes (peripheral lymphocytes) were normal.

Roentgenograms showed: distal bifurcation of the right tibia, hypoplastic and abnormally modeled distal portion of the left tibia, bilaterally abnormally modeled astragalus, calcaneus, tarsal bones and 5-toe varus-adductus feet (Figures 4 and 5).

DISCUSSION

At least 12 well known inherited clinical conditions involve tibial aplasia (Majewski *et al.*, 1985; Richieri-Costa, 1987; Richieri-Costa, 1988; Richieri-Costa, 1990; Richieri-Costa *et al.*, 1987a,b; Prescott *et al.*, 1989; Richieri-Costa *et al.*, 1990). In spite of variations concerning expressivity and penetrance in these conditions, involvement of the central nervous system and the oro-facial structures is exceedingly rare.

Cleft lip/palate-cleft palate associated with tibial hemimelia has been reported in only very few instances (Ho *et al.*, 1975; Richieri-Costa, 1987), while mental retardation in the presence of a complex multiple congenital abnormalities syndrome has not been reported previously in association with tibial aplasia.

Among the reported patients with oro-facial involvement and tibial aplasia, the patient of Ho *et al.* (1975), a baby girl, had a distinct combination of anomalies (tibial hemimelia, cleft palate, preaxial polydactyly of the feet, congenital heart defect



Figure 4 - Radiological aspects of the right lower limb of the propositus.



Figure 5 - Radiological aspects of the left lower limb of the propositus.

and wormian bones) and in her antecedents there were references to intrauterine exposure to multiple medications, suggesting an environmental cause. In a Brazilian boy, born to consanguineous parents and reported by Richieri-Costa (1987), tibial aplasia-cleft lip/palate were the only signs observed, and autosomal recessive inheritance was suggested.

In the patient reported here, the cluster of anomalies is unique and can be clearly differentiated from the above mentioned conditions, since the occurrence of microcephaly and mental retardation, among other signs, associated with tibial hemimelia has, to our knowledge, not been reported till now.

The present report stresses the heterogeneity of tibial hemimelia conditions, especially when one accepts the hypothesis that femoral bifurcation, tibial hemimelia/ectrodactyly, (the Gollop-Wolfgang complex) and the limb defect in the present patient are variants of the involvement of the same developmental field - namely - the tibial developmental field.

It is highly desirable to report new cases concerning tibial aplasia conditions, in order to widen the knowledge of the involvement of this field, to delineate the phenotypic spectrum of these conditions, and to provide reliable data for genetic counselling.

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RESUMO

Os autores relatam sobre um menino de 8 anos de idade, com uma síndrome malformativa, provavelmente ainda não descrita, associada à retardo mental, microcefalia, hipoplasia malar, orelhas e nariz anômalos, fissura labial, hemimelia tibial e bifurcação femoral.

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