SPONDYLO-META-EPIPHYSEAL DYSPLASIA SHORT LIMB-HAND TYPE
A CASE REPORT IN A PALESTINIAN CHILD

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ABSTRACT

A 13 month-old male, a result of a consanguineous marriage was diagnosed to have spondylo-meta epiphyseal dysplasia SMED short limb-hand type. The case is the first to be reported among Palestinians and the 12 th on the disease record. The patient death was due to spinal cord compression.

INTRODUCTION

A new form of generalized autosomal recessive bone dysplasia has been recently delineated. Eleven cases of spondylo-meta epiphyseal dysplasia short limb-hand type were reported mainly among Sepharadic Jews and Puerto Ricans. We report below a new case born to a Palestinian consanguineous couple.

CASE REPORT

A male boy, born on 21-8-1993 was referred to our hospital at the age of 14 months due to progressive hypotonia, inability to stand or sit and difficult breathing since 1-2 weeks prior to admission. He was a result of caesarean section delivery, at term due to fetal distress; his birth weight was 3.500gm. Parents were first degree cousins aged 30 years, both are healthy and the patient was their first baby.

Family history was negative for abnormal babies or dwarfism. Examination revealed: short stature, length 64cm (-4 SD), head circumference 44 cm (-1 SD), and weight 7.370 kg (-3 SD). He had prominent forehead, wide nostrils, depressed nasal bridge, long philtrum, hypertelorism, retrognathia, short limbs, short and wide hands, fingers, and toes.

The chest was narrow with pectus excavatum. No excessive joint laxity was noted (Figures 1, 2, 3).

Developmental history revealed that he smiled at the age of two months, sat without support at 7 months, was able to stand with support at 11 months. At the age of 13 months, he had only 2 lower incisors, he could hold and move objects from one hand to the other, had no hearing deficit and was able to say mama and dada.

X-rays revealed: platyspondyly, irregularity of epiphyses and metaphyses, wide metaphyses, stippling and flattening of different epiphyses , short ilium with a prominent sclerotic sciatic spur, short and cupped ribs, short phalanges of hands and feet (Figures 4,5,6,7,8).

Hours after admission, the baby developed severe respiratory distress and respiratory failure mainly due to hypotonia and diaphragmatic paralysis. He was started on mechanical ventilation.

Upon admission blood culture was taken and the patient was started on antibiotics, initial chest x-ray was normal and all blood cultures remained negative.

The referral diagnosis was a case of achondroplasia and respiratory infection, but with these clinical and radiographic features this diagnosis was doubted.

Neurologically the baby remained hypotonic with total areflexia the first 4 days. Gradual reappearance of reflexes, hyperreflexia and clonus were then noted. No deficit to painful stimuli was stated. The diagnosis of spinal transection at the level C1-C2 was advanced. No notion or signs of trauma were found.

No cervical fracture was detected on plane x-rays or cervical CT scan. Brain CT Scan was normal and the cervical spinal canal was judged narrower than expected.

EMG and nerve conduction velocities in lower and upper limbs were within normal limits for age.

Lumbar puncture done on the third day of admission showed CSF protein of 20mg/dl. Liver function tests, CPK and LDH were normal.
The family refused bone and muscle biopsies. Cervical myelogram was planned but could not be done due to technical problems.

The clinical picture and the x-rays of this patient were suggestive of spondylo-meta epiphyseal dysplasia short limb-hand type.

The patient remained ventilator dependent, was never able to breath by himself and died on 29.10.1994.

DISCUSSION

Two recent reports had identified a generalized bone dysplasia with a rather typical clinical appearance. Borochowitz et al., (1) reported three children with spondylo-meta-epiphysial dysplasia, short limb-hand type. Langer et al., (2) reported further eight cases.

Affected individuals have short limbs with brachydactyly, a relatively large head with a prominent forehead, a depressed nasal bridge, hypertelorism, micrognathia and a long philtrum. The chest is narrow and the joints can be lax.

Radiographs reveal shortening of the long bones with wide metaphyses. There is stippling of some of the epiphyses, which can be flattened. There may be abnormal lucencies in the metaphyses giving a 'flocked' appearance. The basilar portion of the ilium is short and there may be a very prominent sclerotic sciatic spur. The ribs are short and cupped and the vertebral bodies are pear-shaped with progressive flattening.

Langer et al., (2) emphasized the abnormal stippled calcification of the epiphyses and suggested that this should be in the title of the syndrome.

Consanguinity was reported in one case by Brochowitz et al. (1) in 2 cases and suspected in another by Langer et al. (2). Most of cases were of Puerto Rican or Sepharadic Jew origin.

Parents of our case were first cousins Palestinian Moslems from Nablus area.

Atlantoaxial instability is a danger. Death was explained by cord compression, as in our case, in 4 cases of Langer et al., (2)

We agree with the conclusion of Langer et al. (2) that this condition has many abnormal radiographic "and clinical" findings and the combination of these findings is diagnostic. It can be easily distinguished from conditions with disproportionate short limb and platyspondyly described earlier , Spranger and Maroteaux (3) Kozlowski et al (4), Borochowitz et al.,1988 (5), Peri (6) Torrington and Beighton (7).

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