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What is Your Diagnosis?

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A six- year-old girl was seen because of failure to thrive, inability to talk, learning disabilities and behaviour disorders.

She had two brothers and one sister. Her parents and siblings were healthy.

Physical examination revealed growth retardation (11 kg 90 cm length and 44.5 cm head circumference; all at the 3 percentile). Her hearing was normal but speech was absent. The patient had a characteristic dysmorphic facial appearance (figure 1).

Laboratory findings including complete blood count, urinalysis and blood chemistry were normal.

Densoument:

Cornelia de Lange Syndrome (Brachmann-de Lange Syndrome)

The most characteristic features of the syndrome are: Shortness of stature, mental retardation, microbrachycephaly, bushy eyebrows and synophrys, long, curly eyelashes, small nose, anteverted nostrils, characteristic lips and mouth: Thin lips with small midline beak of the upper and corresponding notch in the lower lip; downward curving of the angle of the mouth, micrognathia, generalized hirsutism, hypoplastic nipples and umbilicus, micromelia, and failure to thrive (1).

The facial appearance of infants is quite characteristic. Retardation of growth is present at birth and continues in the following years. They are seldom able to talk. The IQ of these patients is generally less than 35.

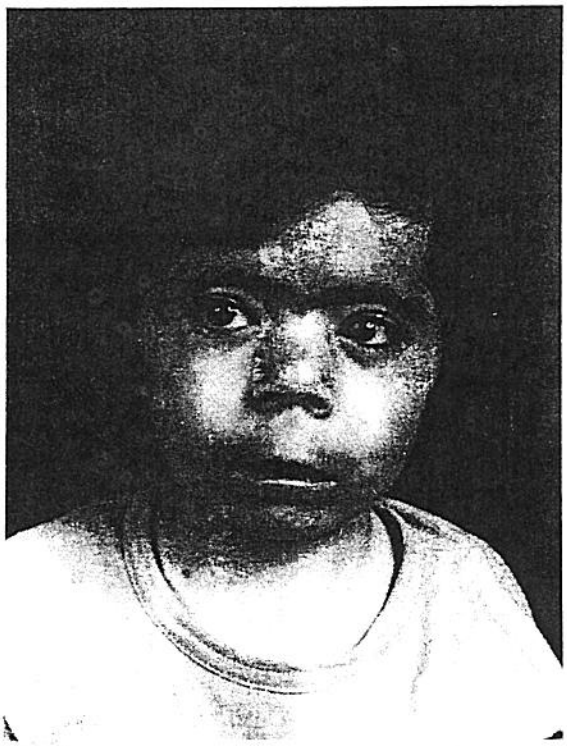

Figure 1. Characteristic facial feature of the patient.

The cause of this syndrome is unknown. Most cases are sporadic (2). Diagnosis dependends on the recognition of the distinctive facial features.

References

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