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CASE REPORT

New Case of an EEC-Like Syndrome in Twins

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Abstract. A patient wrongly referred as a possible victim of thalidomide showed the three classical cardinal features of the EEC syndrome, plus severe mental retardation, an unusual finding in this condition. His twin brother was similarly affected, and died at four months of age due to complications caused by the malformations. Their normal parents were first cousins. The concordance of the manifestation in the twins and the parents' consanguinity suggest that they had the recessive form of the EEC syndrome.

Key words: EEC-like syndrome, Mental retardation, Consanguinity, Twins

INTRODUCTION

Ectodermal dysplasia, ectrodactyly, and cleft lip/palate are the cardinal signs of the EEC syndrome. They occur, however, singly or combined, as parts of other clinical entities. Richieri-Costa et al [1] have thoroughly reviewed the literature on these conditions, and the reader is referred to their paper for details. They also presented apparently the first report of a pair of MZ twins concordant for ectodermal dysplasia and ectrodactyly. We herewith describe a second case of twins similarly affected with the triad, ectodermal dysplasia, ectrodactyly, and cleft lip/palate.

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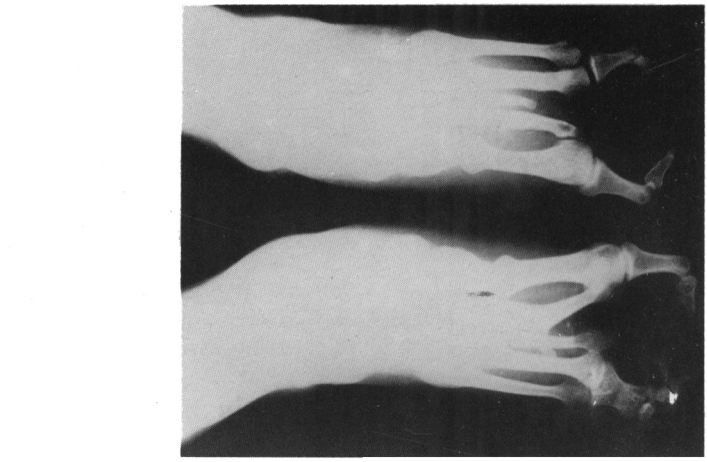

Fig. 3. X-rays of P.S.'s feet.

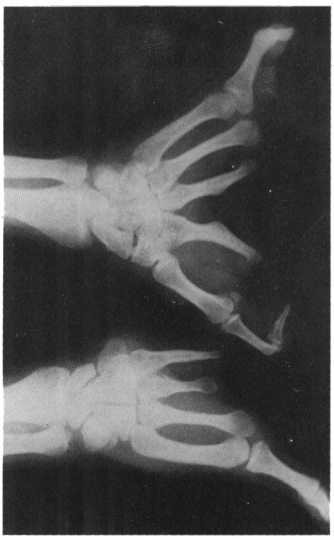

Fig. 2. X-rays of P.S.'s hands.

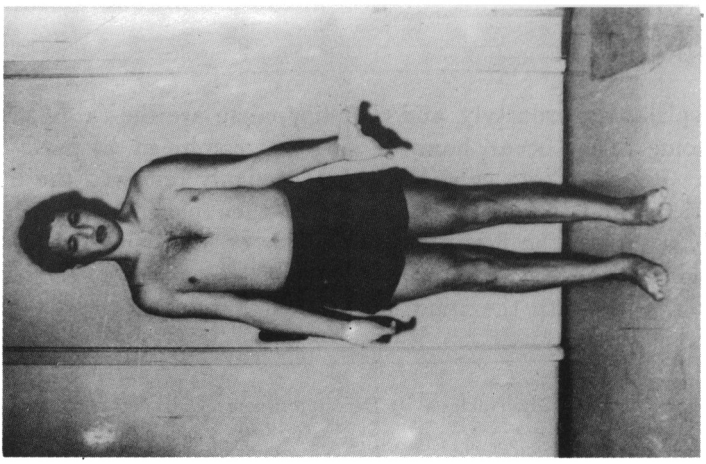

Fig. 1. General view of P.S., the surviving twin.

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P.S., male, Caucasoid, born at Porto Alegre, Brazil, was 26-year-old at the time of the study, and was ascertained during a previous investigation about the effects of thalidomide in our country [2,3]. He was born from a twin pregnancy, but his brother died at four months of age due to suckling difficulties and other complications. According to the mother, the dead child had cleft lip and palate plus ectrodactyly in both hands and feet. She also reported exposition to X-rays and infections during the pregnancy. Her husband seems to have had syphilis before the birth of the twins. At the time of the exam she showed difficulties in word articulation, bad diction, and difficulties of understanding. In the recent past she had one of her breasts extirped due to cancer.

Examination of P.S. (Fig. 1) disclosed blindness in his right eye and severe (8/10) vision impairment in his left eye. The cleft lip had been surgically corrected, but he presented a longitudinal fissure in the hard palate. Head X-rays indicated a shortening of the upper jaw in its anteroposterior diameter and absence of the anterior nasale spine. There was slight opacity in the maxillary sinuses and absence of upper and lower teeth (anodontia). At his right hand we noticed absence of four fingers (1-4), and absence of fingers 2, 3 and 4 at his left hand. In the feet there was absence of toes 2 and 3 and syndactyly of toes 4 and 5 bilaterally. His skin was dry, with a suggestion of keratosis at hands and feet. X-rays (Figs. 2 and 3) showed, at the right hand, aplasia of fingers 1-4 and of the first metacarpal, with marked dysplasia in the others. At the left hand, aplasia of fingers 2-4 and of the fourth metacarpal, the others showing dysplasia. First and 5th fingers with the proximal articulations twisted 90°. Right foot: agenesis of toes 2 and 3 and of the third metacarpal, with dysplasia of the second metacarpal. Syndactyly of toes 4 and 5. Left foot: first toe flexed, with abduction of the interphalangeal joints. Agenesis of toes 2 and 3 with severe dysplasia of the corresponding metatarsals. P.S. showed severe mental retardation; he could move around independently, but depended heavily on outside help for his hygiene, food intake and dressing.

P.S.'s parents, who are biologically related as first cousins, are apparently normal, despite the difficulties of communication of his mother. Their first son is also normal, and had two normal daughters. No indications of the occurrence of the syndrome in the two sisters of P.S.'s father or the seven sibs of his mother could be obtained, the same being true in relation to their ascendants and descendants. But there is a high prevalence of neurological and psychiatric problems in the proband's maternal side, with episodes of amnesia, deep depressions and hospitalization in psychiatric clinics. A grandson of one of P.S.'s maternal sisters has severe mental retardation.

COMMENTS

P.S. shows all the classical signs of the EEC syndrome, with the possible exception of his hair, that looked normal. The presence of consanguinity among his parents suggests that he may have the recessive form of the syndrome [1], an interpreta-

tion that is strengthened by the concordance of its manifestation in the twins. An unusual finding is his severe mental retardation, present in 15% only of 41 patients with this condition [1]. But the presence of mental problems among his mother's relatives without the syndrome may indicate that this could be a secondary association.

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