Trisomy 13 and Rubinstein–Taybi syndrome*

Summary. Initial diagnosis of Rubinstein–Taybi syndrome was made in an infant with a prominent nose and broad thumbs and first toes. However, due to the presence of other anomalies such as low-set, malformed ears, antimongoloid slant of the eyes, colobomata of the iris, and cleft palate, cytogenetic studies were carried out and the diagnosis of trisomy 13 was confirmed. Since, occasionally, trisomy 13 syndrome may mimic the Rubinstein–Taybi syndrome, cytogenetic studies should be considered in all patients with clinical diagnosis of Rubinstein–Taybi syndrome.

The clinical diagnosis of trisomy 13 syndrome is based upon the findings of major congenital defects of eye, nose, lip, forebrain of holoprosencephaly type, skin, hands, and feet (Smith, 1969). The syndrome of Rubinstein–Taybi is identified clinically by recognition of a constellation of abnormal features which include broad thumbs and toes, characteristic facies with beaked or straight nose, antimongoloid slant of palpebral fissures, hypoplastic maxilla, and other somatic abnormalities, ie, short stature, microcrania, mental and motor retardation (Rubinstein, 1969). Theoretically, these two syndromes are two distinct and different clinical entities. The diagnosis of trisomy 13 can be confirmed cytogenetically, but the diagnosis of Rubinstein–Taybi syndrome is entirely a clinical one. Wilson (1968) reported three infants with trisomy 13 all of whom had broad thumbs and first toes; two of these patients were initially diagnosed as having Rubinstein–Taybi syndrome. We wish to report an additional infant with trisomy 13 who was also initially diagnosed as having Rubinstein–Taybi syndrome.

Case report

The patient, a full-term female infant, was born on 10 September 1970 to a 21-year-old mother. The father was 37 years of age. Both parents and one two-year-old sister are of normal phenotype. There were no abortions or stillbirths. The family history showed no consanguinity and no affected member with any congenital abnormality.

The pregnancy and delivery were uncomplicated. Birth weight was 2 800 g, birth length was 50 cm, head circumference was 33-5 cm, and chest circumference 31-0 cm. However, at birth, the baby had Apgar score of 3 at 1 min and 5 at 5 min. Immediately after birth, the following abnormalities were noted: narrow forehead; antimongoloid slant of the eyes; bilateral colobomata of iris; very large and broad-bridged nose, malformed and slightly low-set ears, cleft soft palate, broad and malopposed thumbs, broad big toes, and rocker-bottom feet (Fig. 1). Frequent seizure-like movements and apneic episodes were noted after 2 days of age. At 6 days, an eye examination showed lens opacity (left greater than right) and retinal haemorrhages in addition to iris colobomas. At 12 days, although there was no cardiac murmur, chest radiography showed a globular heart suggestive of congenital heart disease.

The dermatoglyphic analysis showed distally located axial triradii (at r°), one arch (right thumb), two radial loops (left fourth and fifth fingers), and seven ulnar loops of the other fingers. Both hallucal areas showed distal loop patterns.

The patient expired at age 33 days despite all supportive measures. The necropsy showed, in addition to the congenital abnormalities noted at birth, many other anomalies. These include autolysis of the nervous system, interatrial septal defect, absence of interlobar fissures of the left lung, and absence of minor fissures of the right lung, ectopic pancreatic tissue in the duodenum, cystic dysplasia of kidneys, hydronephrosis and hydro-
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urter (right), stenosis of ureteral ostia, lack of involution of fetal adrenal cortex, and accessory spleens.

![Image of partial karyotype](image)

**Partial 12p deletion: a cause for a mental retardation, multiple congenital abnormality syndrome**

**Summary.** A severely mentally retarded man displayed the following main symptoms: short stature, microcephaly, antimongoloid slant of palpebral fissures, big ears with hyperplastic helices, imperfect dental enamel, short and webbed neck, short arms, short hands, brachy-metaphalangy, short second fingers, broad thumbs, short metatarsal bones, and unusually big first toes. It seems almost certain that the syndrome was caused by a chromosome deletion involving about half of 12p which was present in all of the lymphocytes examined.

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