

# Extremely Rare Syndrome: Pseudotrisomy 13

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## Abstract

Pseudotrisomy 13 syndrome is extremely rare, characterized by holoprosencephaly, microcephaly, anophthalmia, single atric nasi, cleft palate and extremity anomalies such as polydactyly and oligodactyly with a normal karyotype. We have report a newborn pseudotrisomy 13 patient, a normal karyotype 46 XY. Clinical and laboratory findings were diagnosed with pseudotrisomy 13 syndrome. The case died after 12 days due to cardio-respiratory failure.

**Keywords:** Pseudotrisomy 13 syndrome; Newborn; Olygodactyly

## Introduction

Pseudotrisomy 13 syndrome is extremely rare, autosomal recessive transmitted chromosomal disorder characterized by abnormal frontal cerebral development (holoprosencephaly), microcephaly, anophthalmia, middle facial defects such as single atric nasi, cleft palate and extremity anomalies. It is differentiated from trisomy 13 by normal karyotype.

We present a patient with phenotypic features highly suggestive of trisomy 13 with a normal karyotype.

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## Case Report

This male was born on term, by C-section, with a body weight of 2160 g (< 3rd centile) length of 47 cm (3 - 10th centile) and head circumference of 27 cm (< 3rd centile). The patient was immediately transferred to our neonatal intensive care unit because of facial dysmorphism and poor general condition (Fig. 1). He was the first child of non-consanguineous parents and the family history was unremarkable. The father was 30 and the mother 28 years old. Their karyotypes were normal. Clinical examination revealed microcephaly, a flat rudimentary nose with a single nasal cavity, hypotelorism, anophthalmia, cleft palate, olygodactyly and a short neck (Fig. 1, 2).

Cerebral ultrasonography revealed a lobar holoprosencephaly with an absence of midline structures of the brain. Echocardiogram and renal ultrasound were normal. Chromosome analysis from peripheral blood revealed a normal karyotype, 46 XY. Clinical and laboratory findings pointed out pseudotrisomy 13 syndrome. The case died at 12 days owing to cardio-respiratory failure.

## Discussion

Pseudotrisomy 13 syndrome was first suggested in 1991 by



**Figure 1.** The pseudotrisomy 13 syndrome was seen appearance of our case.



**Figure 2.** The patient was seen microcephaly, anophthalmia, hypotelorism, flat rudimentary nose with single nostril and short neck.

Cohen et al. It is defined in chromosomally normal patients with holoprosencephaly and associated features were suggestive of trisomy 13 such as microcephaly, hypotelorism, cleft palate, anophthalmia [1, 2]. An autosomal recessive pattern of inheritance for this situation seems most likely, but a gene has not yet been mapped.

Holoprosencephaly and polydactyly are major criteria in this anomaly but polydactyly may not coexist. The diagnostic findings are normal chromosomal pattern associated with holoprosencephaly and polydactyly but sometimes other characteristic anomalies without polydactyl [3]. The oligodactyly was present instead of polydactyly in our patient with other characteristic diagnostic findings. The autosomal recessive transmission is estimated in this patient which is noticeable point in usual relative marriages common countries.

The Meckel syndrome, Pallister-Hall syndrome, trisomy 13, hydrocephalus syndrome, and Smith-Lemli-Opitz syndrome must be excluded in differential diagnosis [4]. These syndromes were differentiated by specific characteristic findings. The microcephaly, anophthalmia, single atric nasal cavity, short neck, cleft palate, holoprosencephaly, patent foramen ovale and oligodactyly were observed in our patient. These finding are evaluated as trisomy 13. However

it is diagnosed as pseudotrisomy 13 finally because of the normal karyotype [5-7].

The diagnosis of pseudotrisomy 13 is an important one in view of the implications for genetic counseling. The family is referred to genetic consultation for further pregnancies.

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