Introduction

Pseudo trisomy 13 syndrome is an extremely rare, autosomal recessive transmitted chromosomal disorder characterized by abnormal frontal cerebral development, microcephaly, anophthalmia, middle facial defects; single athresic nasi, cleft palate and abnormalities of the extremities. It is differentiated from trisomy 13 by normal karyotype.

We present a fetus with phenotypic features, highly suggestive of trisomy 13 however with a normal karyotype.

The Case:

A female patient - 25 years old, G3P2 was referred to the Cairo Fetal Medicine unit for an early 2nd trimester anomaly scan. The ultrasound scan revealed: hydrocephalus, occipital encephalocele, middle facial defects (hypospastic nasal bone, Median cleft lip and cleft palate, and defective soft tissue of the nose), post axial polydactylly. Trisomy13 was suspected however karyotyping was normal.

These finding are evaluated as trisomy 13. However it is diagnosed as pseudotrisomy 13 finally because of the normal karyotype.

Depending on the ultrasound finding the parents decided termination of pregnancy.

Postnatal examination after termination of pregnancy confirmed the prenatal ultrasound findings.

Discussion:

Pseudotrisomy 13 syndrome was first suggested in 1991 by 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).

Reference: