DISCUSSION:

Patau's syndrome is a rare genetic disorder caused by having an additional copy of chromosome 13 in some or all of the body's cells. Patau's syndrome affects about 1 in every 5,000 births. The risk of having a baby with the syndrome increases with the mother's age. More than 9 out of 10 children born with Patau's syndrome die during the first year. About 1 in 10 babies with less severe forms of the syndrome, such as partial or mosaic trisomy 13, live for more than a year.

Trisomy 13 can affect facial features and cause defects such as: cleft lip and palate, microphthalmia, anophthalmia, hypotelorism. Other abnormalities of the face and head include: microcephaly, ear malformations and deafness and capillary hemangiomas.

Prevention: Trisomy 13 can be diagnosed before birth by amniocentesis with chromosome studies of the amniotic cells. Parents of infants with trisomy 13 that is caused by a translocation should have genetic testing and counselling. This may help them be aware of the chances of having another child with the condition. Cytogenetic testing is highly recommended in cases with dysmorphism and abnormal facial features to rule out chromosomal abnormalities and to provide genetic counselling.

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