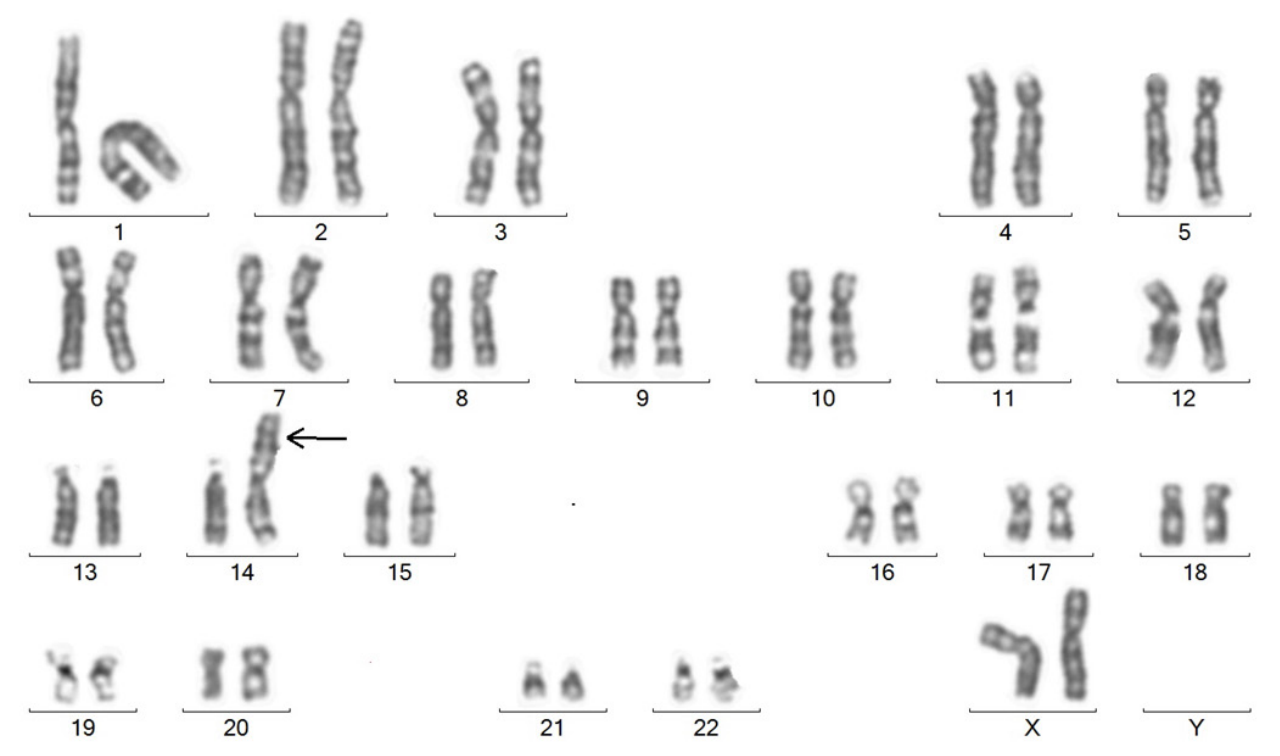

PATHOGENESIS OF PATAU:

Free trisomy 13 is found in about 75 percent of the cases due to error in the meiotic division. In 20% of cases, trisomy 13 is associated with a Robertsonian translocation in which the supernumerary chromosome 13 becomes attached to another acrocentric chromosome (chromosomes 13, 14, 15, 21 or 22). In rare cases, the syndrome is caused by reciprocal translocation between chromosome 13 and a non-acrocentric chromosome. Another type is mosaic trisomy 13 in which both trisomic and normal cell types are present in the patient. The clinical picture of mosaic trisomy patients varies between a normal phenotype and that of classical trisomy 13 according to the number of trisomic cells present in the tissues. The risk of recurrence of trisomy 13 in families of an index case with trisomy 13 is around 1%. However, in families in which trisomy 13 is associated with translocation (Robertsonian or balanced) the risk of recurrence is higher if one of the parents is a carrier of a balanced translocation.