SPOT DIAGNOSIS (IMAGE GALLERY)

WHICH SYNDROME? - PATAU SYNDROME

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This newborn has bulbous nose, micrognathia, ocular hypotelorism, cutis aplasia over parieto-occipital region, polydactyly, left sided inguinal hernia, head circumference of 33cm, chest circumference of 31cm and 44cm length with birth weight of 1.535kg. MRI brain showed hypointensities with prominence of occipital horn. Echocardiography showed ventricular septal defect, atrial septal defect and patent ductus arteriosus of 3mm.

What is the diagnosis?

Karyotype of the baby was suggestive of Trisomy 13. Trisomy 13 or Patau syndrome was first observed by Erasmus Bartholin in 1657 but the chromosomal nature of the disease was ascertained by Klaus Patau in 1960. Incidence is seen more among females. (1) The frequency of this syndrome is 1:10000-15000 live births. (2) Median survival is fewer than 3 days. It is due to a non-disjunction of chromosomes during meiosis. Some are caused by Robertsonian translocations. The risk of this syndrome in the offspring increases with maternal age at pregnancy with about 31years being the average. (3) The presence of holoprosencephaly may cause early death (4). Eighty-five percent do not survive beyond one year of life and most die before completing six months. (4) Patau syndrome is characterized by the following triad : microphthalmia, cleft lip and palate and polydactyly. The face may also have features like ocular hypotelorism, strabismus, iris coloboma, bulbous nose, micrognathia, hemangiomas. In our patient there was polydactyly, ocular hypotelorism, bulbous nose and micrognathia. (5) Structural limb abnormalities like polydactyly in the hand, rocker bottom feet and convex soles may be present. In our patient polydactyly and rocker bottom feet were present. Capillary hemangiomatas and polycystic kidneys or other renal malformations have been reported. Some patients of Patau syndrome may show cicatrical alopecia at occipital region representing scar due to imperfect closure of neural tube, best described as cutis aplasia. The observed cardiac malformations are ventricular septal defect, atrial septal defect and patent ductus arteriosus. Survivors with Patau syndrome exhibit severe mental retardation and developmental delays and are at increased risk for malignancy. (5)

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