MEDIBEATS
The Medical Bulletin

Clinical Scenario

Patau Syndrome / Trisomy 13

Definition :

✓ It is a genetic disorder caused by the presence of all or a portion of a third chromosome 13.

History :

✓ The syndrome was first described and reported in 1960 by a German-born American geneticist Klaus Patau.

Prevalence :

1/12,500 at birth.

Etiology :

- Meiotic non dysjunction - Most common cause, it occurs more frequently in mothers of advanced age (age greater than 35).
- Unbalanced Robertsonian translocation – It results in two normal copies of chromosome 13 and an additional long arm of chromosome 13.
- Mosaicism – It is least common cause & it results in 3 copies of chromosome 13 in some cells and two copies in the others. It is the outcome of a mitotic nondisjunction error and is unrelated to maternal age.

Clinical Features :

- Intrauterine growth restriction and microcephaly.
- Central nervous system abnormalities are usually midline with alobar holoprosencephaly.
- Facial defects are primarily midline - cyclopia, cleft lip and cleft palate.
- Facial features - sloping forehead, small malformed ears, anophthalmia or microphthalmia, micrognathia and pre-auricular tags.
- Cardiac disease – VSD, ASD, AVSD, TOF and double outlet right ventricle.
- Extremity defects - postaxial polydactyly, congenital talipes equinovarus or rocker-bottom feet.
- Others - omphalocele, incomplete rotation of the colon, Meckel diverticulum, polycystic kidney, hydronephrosis, horseshoe kidney, cryptorchidism, hypospadias, labia minora

hypoplasia & bicornuate uterus.

- Patients surviving past infancy have a severe psychomotor disorder, failure to thrive, intellectual disability, and seizures.

Investigations :

✓ Prenatal chorionic villus sampling and/or amniocentesis with karyotyping.

✓ Amniocentesis and CVS are quite reliable but offers risk of miscarriage of between 0.5 to 1%.

✓ Postnatal karyotyping (if prenatal karyotyping not done).

✓ Diagnosis of Patau syndrome may be suspected prenatally based on physical anomalies detected by

- Fetal ultrasonography
- Maternal serum screening
- Noninvasive prenatal screening (NIPT)

✓ Dual markers = hCG - , PAPP-A - .

✓ ***Prenatal USG diagnosis :***

- CNS anomalies (45%–55%): holoprosencephaly, agenesis of the corpus callosum and cerebellar malformations.
- Craniofacial anomalies (80%): bilateral cleft lip and palate, micro- and anophthalmia and micrognathia.
- Congenital heart disease (40%–50%): septation defects and absent pulmonary venous return.
- Abdominal wall anomalies (30%): exomphalos.
- Urinary tract anomalies (30%–35%): cystic renal dysplasia.
- Skeletal anomalies (20%–30%): postaxial polydactyly and clenched hands.
- FGR (45%–55%).
- Trisomy 13 is associated with an enlarged nuchal translucency at 12–14 weeks of gestation.
- Soft markers include echogenic intracardiac focus, short humerus & femur, renal pelvic dilatation and increased nuchal fold thickness.

Treatment :

- ✓ Syndromic presence of multiple organ dysfunctions in trisomy 13 are incompatible with life.
- ✓ The management of patients with Patau syndrome is multidisciplinary.
- ✓ Treatment is basically symptomatic and complete recovery is not possible.
- ✓ Despite aggressive management, median survival only extends to 733 days in the most recent cohorts of patients(Williams et al).

Prognosis :

- Lethal condition in most cases.
- 95% of the survivors die within 6 months.
- Very rare cases without severe malformations have survived for several years.
- Prognosis is better in patients with mosaicism and patients with unbalanced translocations.

Recurrence risk :

This is empirically estimated to comprise 1% of all autosomal trisomies.

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