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Pataus Syndrome

Ida Divya Sherly E.[1]; Dr. Sheeba Chellappan[2]; Bruelin Melshia M.[3]; Aslin Johnsi. L.[4]; Kavitha S.[5]; Rajeshwari. G.[6]

[1]MSc(N), Associate Professor/PhD Research Scholar, Child Health Nursing, Dr Kumaraswami Health Centre College of Nursing, Perumalpuram.

[2]Msc(N), PhD(N), Professor/ Research Guide, Medical Surgical Nursing, Christian College of Nursing.

[3]Assistant Professor, Child Health Nursing, C.S.I St.Luke's College of Nursing, Nazareth.

[4]Associate Professor, Medical Surgical Nursing, Ave maria College of Nursing, Attoor.

[5]Associate Professor, Mental Health Nursing, Sardar Raja College of Nursing, Kavalkinaru.

[6]Professor/ Research Scholar, Mental Health Nursing, Dr Kumaraswami Health Centre College of Nursing, Perumalpuram.

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Abstract: Patau syndrome, also known as Trisomy 13, is a rare chromosomal disorder caused by the presence of an additional copy of chromosome 13, resulting in multiple congenital anomalies and severe neurodevelopmental impairment. The condition arises due to meiotic nondisjunction, mosaicism, or chromosomal translocation, with full trisomy being the most common form. The prognosis is extremely poor, as the majority of affected infants do not survive beyond the first year of life due to severe systemic complications. Diagnosis is typically established prenatally through ultrasonography and confirmed by cytogenetic analysis using procedures such as amniocentesis or chorionic villus sampling. Management is primarily supportive and focuses on symptom relief, family counselling, and palliative care. Understanding the clinical features, diagnosis, and outcomes of Patau syndrome is essential for healthcare professionals to provide appropriate care and informed decision-making support to affected families.

Keywords: *Patau Syndrome, Trisomy 13.*

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I. INTRODUCTION

Patau syndrome, also known as Trisomy 13, is a rare and severe chromosomal disorder caused by the presence of an extra copy of chromosome 13 in all or some of the body's cells. This genetic abnormality results in multiple congenital malformations that affect several organ systems, including the brain, heart, eyes, and limbs. Common clinical features include severe intellectual disability, cleft lip and palate, microcephaly, polydactyly, congenital heart defects, and abnormalities of the central nervous system.

Patau syndrome occurs in approximately 1 in 10,000 – 20,000 live births and is associated with a high rate of prenatal loss. The prognosis is poor, with many affected infants not surviving beyond the first year of life due to life-threatening complications. Early diagnosis through prenatal screening and confirmatory genetic testing allows for informed parental counselling and multidisciplinary planning of care. Despite its rarity, Patau syndrome represents a significant clinical and ethical challenge because of its severity, high mortality, and the need for comprehensive supportive and palliative care.

II. DEFINITION

Patau syndrome, also known as Trisomy 13, is a rare genetic disorder caused by the presence of an extra copy of chromosome 13 in all or some of the body's cells, leading to multiple congenital anomalies, severe intellectual disability, and a high rate of infant mortality.

➢ *Incidence*
- Trisomy 13 occurs in an estimated 1 out of 10,000 to 20,000 live births.
- Only 5% to 10% of babies born with trisomy 13 survive past their first year.

III. ETO PATHOGENIC FACTORS

The etiopathogenesis of Patau syndrome involves genetic abnormalities that lead to the presence of extra chromosome 13 material, resulting in abnormal development of multiple organ systems. The main etiopathogenic factors include:

➢ *Meiotic Nondisjunction (Full Trisomy 13)*
- The most common cause.
- Failure of chromosome 13 to separate properly during meiosis results in an extra chromosome 13 in all body cells.
- Strongly associated with advanced maternal age.

➢ *Mosaic Trisomy 13*
- Occurs due to mitotic nondisjunction after fertilisation.
- Some cells have the normal chromosome number, while others have trisomy 13.
- Clinical features may be milder depending on the proportion of affected cells.

➢ *Robertsonian Translocation*
- Part or all of chromosome 13 becomes attached to another chromosome (commonly chromosome 14 or 15).
- Can be inherited from a parent who is a balanced translocation carrier.
- Important for genetic counselling due to increased recurrence risk.

➢ *Advanced Maternal Age*
- Increases the risk of chromosomal nondisjunction during oocyte meiosis.
- A well-recognised risk factor in full trisomy 13 cases.

➢ *Genetic Imbalance and Gene Dosage Effect*
- The extra chromosome 13 leads to overexpression of genes critical for embryonic development.
- Results in abnormal organogenesis, especially of the brain, heart, and craniofacial structures.

➢ *Prenatal Environmental Interaction (Indirect Role)*
- While not a direct cause, environmental factors may influence pregnancy outcomes in genetically affected foetuses.

IV. CLINICAL FEATURES

Patau syndrome presents with multiple, severe congenital anomalies affecting almost every organ system. The clinical manifestations are evident at birth or detected prenatally and vary in severity depending on the type of trisomy.

➢ *Craniofacial Abnormalities*
- Microcephaly
- Cleft lip and/or cleft palate
- Low-set, malformed ears
- Flat nasal bridge
- Microphthalmia or anophthalmia
- Scalp defects (aplasia cutis)

➢ *Central Nervous System Defects*
- Severe intellectual disability
- Holoprosencephaly
- Seizures
- Hypotonia
- Developmental delay

➢ *Musculoskeletal Abnormalities*
- Polydactyly (extra fingers or toes)
- Overlapping fingers
- Rocker-bottom feet
- Short neck

➢ *Cardiovascular Anomalies*
- Congenital heart defects (very common), such as:
✓ Ventricular septal defect (VSD)
✓ Atrial septal defect (ASD)
✓ Patent ductus arteriosus (PDA)

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➢ *Ophthalmic Manifestations*
- Small or absent eyes
- Coloboma
- Visual impairment or blindness

➢ *Genitourinary Abnormalities*
- Polycystic kidneys
- Hydronephrosis
- Cryptorchidism in males
- Abnormal external genitalia

➢ *Gastrointestinal Abnormalities*
- Omphalocele
- Inguinal or umbilical hernia
- Feeding difficulties

➢ *Growth and Development*
- Intrauterine growth restriction (IUGR)
- Low birth weight
- Failure to thrive

➢ *Other Features*
- Respiratory distress
- Frequent infections
- Severe feeding and swallowing difficulties

V. DIAGNOSTIC CRITERIA

➢ *Prenatal Screening Criteria*
- Ultrasonography findings, including:
✓ Holoprosencephaly
✓ Cleft lip and/or palate
✓ Congenital heart defects
✓ Polydactyly
✓ Omphalocele
✓ Intrauterine growth restriction

- *Maternal serum screening:*
✓ *Abnormal levels in first- or second-trimester screening (e.g., low PAPP-A, abnormal β-hCG)*

- *Non-invasive prenatal testing (NIPT):*
✓ *Detection of increased chromosome 13 material in cell-free fetal DNA*

➢ *Confirmatory Genetic Testing (Definitive Criteria)*

Diagnosis is confirmed by demonstrating extra chromosome 13 material through:
- Karyotyping (47, XX,+13 or 47, XY,+13)
- Fluorescence in situ hybridization (FISH)
- Chromosomal microarray analysis (CMA)
- Prenatal procedures:
✓ Chorionic villus sampling (CVS)
✓ Amniocentesis

➢ *Postnatal Clinical Criteria*

Presence of multiple characteristic features, such as:
- Severe intellectual disability
- Microcephaly
- Cleft lip and/or palate
- Microphthalmia or anophthalmia
- Polydactyly
- Congenital heart defects
- Scalp defects (aplasia cutis)

VI. MANAGEMENT

There is no curative treatment for Patau syndrome. Management is primarily supportive and palliative, focusing on improving quality of life, treating complications, and supporting the family through informed decision-making.

➢ *Prenatal Management*
- Early diagnosis through prenatal screening and genetic testing
- Genetic counseling for parents regarding prognosis, outcomes, and recurrence risk
- Planned delivery at a tertiary care center with neonatal intensive care facilities

➢ *Immediate Neonatal Care*
- Airway and breathing support (oxygen therapy, CPAP, or mechanical ventilation if required)
- Temperature regulation and monitoring of vital signs
- Management of feeding difficulties using nasogastric or gastrostomy feeding
- Treatment of hypoglycemia and electrolyte imbalance

➢ *Management of Congenital Anomalies*
- Cardiac defects: medical management of heart failure; surgical correction in selected cases
- Cleft lip and palate: surgical repair if survival permits
- Polydactyly: surgical correction (optional and usually delayed)
- Omphalocele and other GI defects: surgical intervention when appropriate
- Renal anomalies: monitoring renal function and managing complications

➢ *Neurological Care*
- Seizure control with antiepileptic medications
- Management of hypotonia and developmental delays

➢ *Supportive and Palliative Care*
- Focus on comfort care, pain relief, and minimizing invasive procedures
- Decision-making guided by family preferences and ethical considerations
- End-of-life care planning when prognosis is poor

➢ *Multidisciplinary Approach*
- Neonatologist
- Pediatric cardiologist
- Pediatric neurologist
- Pediatric surgeon
- Geneticist
- Nurses, nutritionists, and social workers

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➢ *Long-Term Care (for Survivors)*
- Ongoing medical surveillance
- Physiotherapy, occupational therapy, and speech therapy
- Psychosocial support for family members

VII. PROGNOSIS OF PATAU SYNDROME (TRISOMY 13)

The prognosis of Patau syndrome is poor due to severe multisystem congenital anomalies and profound neurological impairment.
- A high proportion of affected foetuses result in spontaneous abortion or stillbirth.
- Among live-born infants, more than 80–90% do not survive beyond the first year of life, with many dying within the first days or weeks.
- Early mortality is mainly due to congenital heart defects, central nervous system malformations, respiratory failure, and recurrent infections.
- Infants with mosaic trisomy 13 or translocation forms may have a slightly better survival and milder clinical manifestations compared to full trisomy 13.
- Survivors beyond infancy have severe intellectual disability, significant developmental delay, and require lifelong medical and supportive care.

Overall, Patau syndrome carries a grave prognosis, and management focuses on supportive, palliative, and family-centred care rather than curative treatment.

VIII. PREVENTION OF PATAU SYNDROME (TRISOMY 13)

Patau syndrome cannot be completely prevented because it is caused by a spontaneous chromosomal abnormality. However, certain measures can help in risk reduction, early detection, and informed reproductive decision-making.

➢ *Genetic Counselling*
- Recommended for couples with:
✓ Previous child with Patau syndrome
✓ History of chromosomal abnormalities
✓ Advanced maternal age
- Helps assess recurrence risk and guide future pregnancies.

➢ *Prenatal Screening*
- First-trimester screening (nuchal translucency, PAPP-A, β-hCG)
- Second-trimester anomaly scan to detect structural defects
- Non-invasive prenatal testing (NIPT) using cell-free fetal DNA

➢ *Prenatal Diagnostic Testing*
- Chorionic villus sampling (CVS)
- Amniocentesis
- Confirms trisomy 13 through karyotyping or chromosomal microarray analysis.

➢ *Preconception Measures*
- Parental karyotyping if one parent is a balanced translocation carrier
- Planning pregnancy at a younger maternal age, when possible
- Avoidance of harmful environmental exposures during pregnancy

➢ *Assisted Reproductive Technology (ART)*
- Preimplantation genetic testing (PGT) for couples with known chromosomal rearrangements
- Helps select embryos without trisomy 13.

➢ *Informed Decision-Making*
- Early diagnosis allows families to make informed choices regarding pregnancy continuation and perinatal care planning.

IX. HEALTH EDUCATION FOR PATAU SYNDROME (TRISOMY 13)

Health education plays a vital role in supporting families affected by Patau syndrome by promoting understanding, informed decision-making, and appropriate care.

➢ *Education for Parents and Family*
- Explain the genetic nature of Patau syndrome and that it is not caused by parental actions.
- Discuss the poor prognosis and expected clinical outcomes in simple, compassionate language.
- Provide information on treatment options, emphasizing supportive and palliative care.
- Encourage parents to participate actively in care planning and decision-making.

➢ *Prenatal Education*
- Importance of early antenatal registration and regular check-ups.
- Awareness of prenatal screening and diagnostic tests for chromosomal abnormalities.
- Counseling regarding genetic testing and its implications.

➢ *Education on Newborn Care*
- Feeding techniques for infants with poor suck or cleft palate.
- Recognition of danger signs such as breathing difficulty, cyanosis, and seizures.
- Importance of maintaining warmth, hygiene, and infection prevention.

➢ *Psychosocial Support*
- Address emotional stress, grief, and anxiety in parents.
- Provide information about support groups, counseling services, and palliative care teams.
- Encourage family and community support.

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➢ *Education on Future Pregnancies*
- Importance of genetic counseling before planning another pregnancy.
- Discuss recurrence risk and available prenatal and preimplantation screening options.

➢ *Role of Healthcare Providers*
- Nurses and healthcare professionals should provide continuous education, emotional support, and follow-up guidance.
- Use clear, culturally sensitive communication tailored to the family's educational level.

X. CONCLUSION

Patau syndrome is a rare and severe chromosomal disorder caused by the presence of an extra copy of chromosome 13, leading to profound multisystem congenital anomalies and severe neurodevelopmental impairment. The condition is associated with high prenatal and infant mortality, and survivors experience significant physical and intellectual disabilities. Early prenatal screening and confirmatory genetic testing are essential for timely diagnosis and informed parental counselling. As there is no curative treatment, management primarily focuses on supportive, palliative, and family-centred care delivered through a multidisciplinary approach. Comprehensive health education and genetic counselling play a crucial role in helping families understand the condition, cope with its outcomes, and make informed decisions regarding current and future pregnancies.

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