Neonate with Microphthalmia, Cleft Lip and Palate, Omphalocele, Polydactyly, Atrial Septal Defect and Patent Ductus Arteriosus: A Likely Case of Patau Syndrome

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Case Report

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Neonate with Microphthalmia, Cleft Lip and Palate, Omphalocele, Polydactyly, Atrial Septal Defect and Patent Ductus Arteriosus: A Likely Case of Patau Syndrome

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Abstract

This report describes a one-day-old female neonate presenting with multiple congenital anomalies, including bilateral microphthalmia, cleft lip and palate, omphalocele, unilateral polydactyly, Atrial Septal Defect (ASD), and Patent Ductus Arteriosus (PDA). The omphalocele was surgically repaired shortly after birth. These features are characteristic of Patau syndrome (Trisomy 13), a rare chromosomal disorder caused by an extra copy of chromosome 13. Genetic testing is ongoing to confirm the diagnosis. This case underscores the importance of early recognition, multidisciplinary care, and family counseling for neonates with severe congenital malformations.

Keywords: Patau syndrome, Trisomy 13, Congenital anomalies, Cleft lip and palate, Omphalocele, Postaxial polydactyly, Atrial Septal Defect (ASD), Patent Ductus Arteriosus (PDA), Neonatal surgery, Genetic disorders, Chromosomal abnormalities, Craniofacial anomalies, Multidisciplinary care, Neonatal outcomes, Family counseling, Pediatric congenital disorders, Abdominal wall defects, Microphthalmia, Cardiac anomalies, Rural healthcare, Yemen case report

Introduction

Patau syndrome, also known as Trisomy 13, is a rare and severe chromosomal disorder caused by the presence of an extra chromosome 13 in all or some cells of the body [1]. This condition disrupts normal embryonic and fetal development, leading to multisystem malformations that are often incompatible with long-term survival. Patau syndrome occurs with an incidence of approximately 1 in 10,000 to 20,000 live births, making it one of the least common trisomies observed in live births, after Trisomy 21 (Down syndrome) and Trisomy 18 (Edwards syndrome) [2].

The clinical manifestations of Patau syndrome include a spectrum of craniofacial anomalies, such as microphthalmia or anophthalmia, cleft lip and palate, and midline facial defects. Cardiac anomalies, such as Patent Ductus Arteriosus (PDA) and Atrial Septal Defects (ASD), are frequently observed and contribute significantly to morbidity and mortality. Additionally, abdominal wall defects, such as omphalocele, and limb anomalies, such as postaxial polydactyly, are common features [3]. Neurological abnormalities, including holoprosencephaly and profound intellectual disability, further complicate the prognosis for affected neonates [1].

The prognosis for infants with Patau syndrome remains exceedingly poor, with most succumbing to complications within the first year of life. Survival beyond infancy is rare, and long-term outcomes are characterized by significant developmental and functional impairments. The most common causes of mortality are cardiorespiratory failure and severe congenital anomalies that are incompatible with survival. Early recognition of the clinical features, genetic confirmation, and multidisciplinary management are critical to optimizing outcomes and supporting affected families [3].

This case report describes a neonate presenting with characteristic features of Patau syndrome, including bilateral microphthalmia, cleft lip and palate, omphalocele, unilateral polydactyly, Atrial Septal Defect (ASD), and Patent Ductus Arteriosus (PDA). The presentation underscores the need for prompt diagnosis, surgical intervention for life-threatening anomalies, and genetic counseling for the family.

Case Presentation

Patient information

A one-day-old female neonate, born at term via normal vaginal delivery to a 29-year-old mother and a 33-year-old father, presented with multiple congenital anomalies.

Family and prenatal history:

- The mother is from Badan, a rural area in Ibb, Yemen, and has four healthy offspring: three girls and one boy.

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- Both parents are healthy with no known medical conditions.
- There is no consanguinity between the parents, and no family history of genetic or congenital disorders.
- Routine prenatal ultrasounds detected an omphalocele but failed to identify other anomalies.
- The pregnancy was uneventful, with no known teratogenic exposures.

Laboratory investigations:

- Hemoglobin: 13.3 g/dL
- Creatinine: 1.2 mg/dL
- Sodium: 117 mmol/L (indicative of hyponatremia)
- Potassium: 4.2 mmol/L
- Chloride: 93 mmol/L
- Ionized calcium: 1.13 mmol/L
- Random blood sugar: 72 mg/dL
- Coagulation studies: Clotting time: 2:00 minutes, Bleeding time: 4:00 minutes (within normal range for neonate)
- Serological tests (HBsAg, HCV, HIV 1,2) negative.

Arterial Blood Gas (ABG):

- pH: 7.49 (elevated, indicative of respiratory alkalosis)
- PCO_2: 27.3 mmHg (low)
- PO_2: 94.7 mmHg (normal)
- Bicarbonate: 20.6 mmol/L (low)
- SO_2: 97.7% (normal)
- Sodium: 115 mmol/L (severe hyponatremia)
- Potassium: 3.9 mmol/L (normal)

Clinical features:

1. Craniofacial anomalies:
   - Bilateral microphthalmia.
   - Cleft lip and cleft palate.
2. Abdominal wall defect:
   - Omphalocele containing intestinal contents, surgically repaired on day 1 of life.
3. Limb anomalies:
   - Unilateral polydactyly of the hand.
4. Cardiac defects:
   - Atrial Septal Defect (ASD) identified on echocardiography.
   - Patent Ductus Arteriosus (PDA) with mild left-to-right shunting.

Investigations:

Genetic testing: Karyotyping and Fluorescence *In-Situ* Hybridization (FISH) for Trisomy 13 are pending.

Echocardiogram: Demonstrated ASD and PDA with no immediate hemodynamic compromise.

Brain imaging: Planned evaluation for associated neurological abnormalities, including holoprosencephaly.

Differential diagnosis:

The constellation of anomalies cleft lip and palate, microphthalmia, omphalocele, polydactyly, ASD, and PDA is highly suggestive of Trisomy 13 (Patau syndrome).

Discussion

Patau syndrome results from an extra chromosome 13, disrupting normal embryonic development. Craniofacial anomalies, including cleft lip and palate, are frequently seen and may contribute to feeding difficulties and speech impairments if the neonate survives [2]. Cardiac anomalies, such as ASD and PDA, significantly contribute to early mortality in Patau syndrome (Figure 1-3).

Figure 1: Neonate with Clinical Features of Patau Syndrome. **Note:** The image depicts a female neonate with characteristic features of Patau syndrome (Trisomy 13), including bilateral microphthalmia, cleft lip, and an omphalocele containing a portion of the intestines. The patient also presents with polydactyly and other dysmorphic features consistent with the syndrome. These findings highlight the multisystem malformations typically associated with this rare chromosomal disorder.

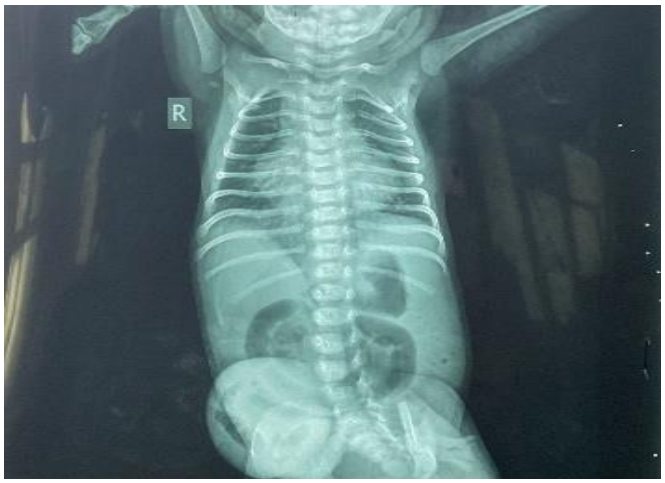

Figure 2: Radiographic findings in a neonate with patau syndrome. **Note:** An Anteroposterior (AP) chest and abdominal X-ray of the neonate demonstrates normal bony thoracic structures and vertebral alignment. The abdominal contents appear consistent with post-operative changes following surgical repair of an omphalocele. No obvious pneumoperitoneum, gross organomegaly, or abnormal calcifications are seen. The radiograph supports the diagnosis and management of abdominal wall defects characteristic of Patau syndrome.

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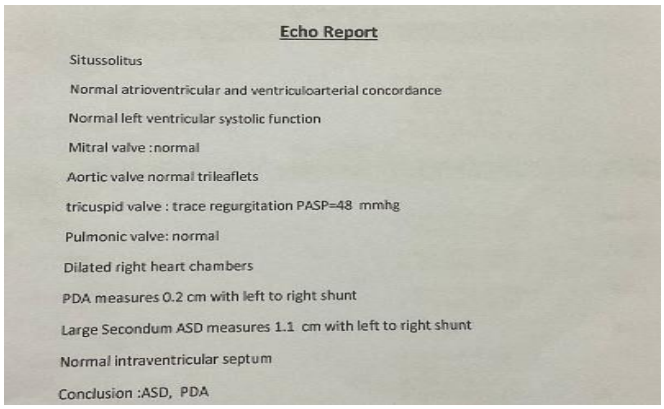

Figure 3: Echocardiographic findings in a neonate with patau syndrome. **Note:** The echocardiography report reveals structural and functional cardiac abnormalities. Notable findings include dilated right heart chambers, a Patent Ductus Arteriosus (PDA) measuring 0.2 cm with a left-to-right shunt, and a large secundum Atrial Septal Defect (ASD) measuring 1.1 cm with a left-to-right shunt. The tricuspid valve shows trace regurgitation with a Pulmonary Artery Systolic Pressure (PASP) of 48 mmHg. Other cardiac structures, including the mitral and aortic valves, show no abnormalities, and the left ventricular systolic function remains normal. These findings confirm the presence of congenital cardiac defects commonly associated with Patau syndrome.

Management overview

Craniofacial anomalies: Early consultation with a craniofacial team and feeding specialists is planned to address cleft lip and palate. Surgical repair is deferred until the neonate's condition stabilizes.

Omphalocele: Successfully repaired surgically shortly after birth. Postoperative care is ongoing.

Cardiac defects: ASD and PDA require continued monitoring. Intervention will depend on the neonate's hemodynamic status and clinical course.

Feeding and nutrition: Feeding assistance with specialized equipment is in place due to cleft palate.

Conclusion

This case illustrates the classical features of Patau syndrome, with multiple congenital anomalies, including cleft lip and palate, omphalocele, polydactyly, and congenital heart defects. While surgical repair of the omphalocele was successful, the overall prognosis remains guarded due to the severity of associated anomalies. Multidisciplinary care involving neonatology, surgery, cardiology, and genetics is critical to optimize immediate outcomes and provide comprehensive support for the family.

Ethical Considerations

Informed consent was obtained from the parents for the publication of this case report and accompanying images. All patient details were anonymized to protect confidentiality.

Competing Interests

The authors report no conflicts of interest in this work.

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