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Edwards Syndrome with Severe Anemia and Neurological Manifestations: A Case Report of a 13-Year-Old Female in Saudi Arabia

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Abstract

Edwards syndrome, also known as trisomy 18, is a rare chromosomal disorder characterized by multiple congenital anomalies and significant developmental delay. We present the case of a 13-year-old female diagnosed with Edwards syndrome, highlighting the intricate clinical course and management challenges encountered in her care. The patient exhibited dysmorphic facial features, severe anemia, and neurological manifestations, including seizures and abnormal movements. Diagnostic investigations revealed severe iron deficiency anemia necessitating packed red blood cell transfusion, alongside antiepileptic therapy for seizure management. A multidisciplinary approach involving specialists from various disciplines facilitated tailored interventions addressing the patient's medical, nutritional, and developmental needs. Reporting and documenting cases of Edwards syndrome are vital for advancing understanding of the condition and optimizing clinical care practices. This case underscores the importance of a comprehensive, multidisciplinary approach to care for individuals with rare genetic disorders like Edwards syndrome, aiming to optimize outcomes and enhance overall quality of life.

Keywords: Edwards Syndrome, Severe Anemia, Neurological Manifestations, Case Report, Old Female, Saudi Arabia.

BACKGROUND

Edwards syndrome, also known as trisomy 18, is a rare chromosomal disorder characterized by the presence of an extra copy of chromosome 18 in the cells of affected individuals [1]. It was first described by John Hilton Edwards in 1960, hence the eponymous name. Edwards syndrome is considered one of the most severe autosomal trisomies compatible with life, with a prevalence estimated to be approximately 1 in 5,000 live births, though this figure may vary due to underreporting and prenatal diagnosis resulting in terminations [2, 3].

The clinical features of Edwards syndrome are diverse and often involve multiple organ systems, leading to significant morbidity and mortality [2]. Common physical characteristics include craniofacial anomalies such as micrognathia, low-set ears, prominent occiput, and clenched fists with overlapping fingers. Other common features include cardiac anomalies, such as ventricular septal defects and atrial septal defects, as well as renal abnormalities, omphalocele, and musculoskeletal anomalies [4, 5].

Neurodevelopmental abnormalities are also prominent in individuals with Edwards syndrome, leading to severe intellectual disability and developmental delay. Hypotonia is a hallmark feature, contributing to feeding difficulties, delayed motor milestones, and impaired coordination [6]. Additionally, individuals with Edwards syndrome may experience seizures, ranging from focal seizures to generalized tonic-clonic seizures, further complicating their clinical course [6, 8].

Prenatal diagnosis of Edwards syndrome is often made through ultrasound findings suggestive of structural abnormalities, such as cardiac defects, omphalocele, or intrauterine growth restriction [9]. Confirmation of the diagnosis typically involves karyotyping or chromosomal microarray analysis, which reveals the presence of an extra chromosome 18 [9, 10]. Despite advances in prenatal screening and diagnosis, the prognosis for individuals with Edwards syndrome remains poor, with the majority of affected pregnancies resulting in fetal demise or neonatal death [11].

Management of Edwards syndrome focuses on supportive care and addressing the medical and developmental needs of affected individuals [12]. Given the complexity of the condition and the wide range of associated comorbidities, a multidisciplinary approach involving specialists from various disciplines is essential. Palliative care principles may be implemented to optimize comfort and quality of life, particularly in cases of severe congenital anomalies or life-limiting conditions [4, 12].

Prognosis for individuals with Edwards syndrome varies widely depending on the severity of associated anomalies and the presence of significant medical complications [13]. While some individuals may survive beyond infancy and childhood with appropriate medical interventions and supportive care, the majority experience significant morbidity and have a shortened lifespan. Long-term outcomes for survivors may include ongoing medical issues, developmental disabilities, and challenges in daily functioning [13, 14].

In recent years, there has been increasing recognition of the importance of holistic and family-centered care for individuals with Edwards syndrome [2]. Supportive services, including early intervention programs, genetic counseling, and psychosocial support for families, play a crucial role in navigating the complexities of the condition and optimizing outcomes for affected individuals and their caregivers [12].

Overall, Edwards syndrome represents a significant clinical challenge due to its complex array of clinical features and associated medical complications. Continued research efforts aimed at elucidating the underlying genetic and molecular mechanisms, improving diagnostic strategies, and developing targeted therapeutic interventions are essential for advancing our understanding of this rare chromosomal disorder and improving outcomes for affected individuals and their families.

CASE PRESENTATION

We present the case of a 13-year-old female diagnosed with Edwards syndrome, emphasizing the clinical complexity and management challenges encountered during her hospitalization.

Clinical History and Presentation

The patient, a known case of Edwards syndrome, was admitted due to abnormal movements and severe anemia. She exhibited multiple congenital anomalies and dysmorphic features, including a high arched palate, cataracts, hyperextended neck, left eye ptosis, micrognathia, and equinovarus deformity. The patient's medical history revealed difficulty swallowing, aphasia, mental retardation, and controlled fits.

Investigations

Upon admission, investigations revealed maternal complications during delivery, with the patient being born at 36 weeks gestation due to maternal bleeding and placental separation. Previous admissions were noted for cyanosis, chest infections, and seizures since infancy. Diagnostic tests confirmed the diagnosis of Edwards syndrome at 45 days old. Recent complaints of abnormal movements prompted rectal and IV diazepam administration for seizures. Laboratory investigations demonstrated severe iron deficiency anemia, prompting planned packed red blood cell transfusion and iron supplementation.

Clinical Examination and Management Plan

Clinical examination revealed severe pallor, generalized hypotonia, brisk reflexes, and inability to sit alone. Neurological findings included generalized spasticity and abnormal movements. A multidisciplinary management plan was devised, involving hematological and neurological consultations. The patient was scheduled for PRBC transfusion preceded by comprehensive investigations, including CBC, iron studies, and coagulation profile. Nutritional support and reassessment post-transfusion were planned to address the patient's anemia and nutritional deficiencies.

Hematology Consultation and Laboratory Results

Hematological consultation confirmed severe iron deficiency anemia based on CBC findings, including low hemoglobin levels, erythropenia, anisocytosis, hypochromia, and microcytosis. Blood grouping and crossmatching indicated AB positive blood type, with subsequent PRBC transfusion increasing hemoglobin levels to 8.1 g/dl. Oral iron and folic acid supplementation were initiated, with a plan for nutritional consultation and long-term follow-up in the hematology clinic.

Clinical Follow-up and Recommendations

Post-transfusion, the patient remained stable with ongoing antiepileptic therapy and observation for abnormal movements and convulsions. Follow-up recommendations included regular monitoring of vital signs and blood parameters, continued antiepileptic therapy, and observation for any recurrent seizures. Long-term management involved monthly follow-up in the hematology clinic for anemia management and nutritional support.

In conclusion, the presented case underscores the clinical complexity and management challenges encountered in individuals with Edwards syndrome. A multidisciplinary approach integrating hematological, neurological, and nutritional interventions is crucial for optimizing outcomes and enhancing quality of life for affected individuals. Continued research and reporting of cases are essential for advancing our understanding of Edwards syndrome and improving clinical care practices.

DISCUSSION

Edwards syndrome, also known as trisomy 18, is a rare chromosomal disorder characterized by multiple congenital anomalies and significant developmental delay [1, 3]. It is caused by the presence of an extra copy of chromosome 18, leading to a wide range of physical and cognitive impairments. With an estimated incidence of 1 in 5,000 live births, Edwards syndrome poses significant challenges in clinical management due to its complex array of symptoms and multisystem involvement [2].

The case presented here highlights the intricate clinical course and management challenges encountered in a 13-year-old female diagnosed with Edwards syndrome. The patient's medical history is emblematic of the characteristic features associated with the syndrome, including dysmorphic facial features, severe anemia, and neurological manifestations such as seizures and abnormal movements. These findings align with existing literature documenting the clinical spectrum of Edwards syndrome and underscore the importance of recognizing its varied presentations for timely diagnosis and intervention [15, 18].

Anemia is a common complication of Edwards syndrome, often stemming from abnormalities in hematopoietic development and chronic illness. The presentation with severe iron deficiency anemia necessitated prompt intervention with packed red blood cell transfusion, consistent with recommendations for managing anemia in individuals with trisomy 18 [16,17]. The profound pallor observed in the case underscores the importance of vigilant monitoring for hematological complications in patients with Edwards syndrome and the need for tailored therapeutic strategies to address their unique needs [2, 15, 17].

Neurological manifestations, including seizures and abnormal movements, are prevalent in individuals with Edwards syndrome and significantly impact their quality of life [4]. The history of convulsions and ongoing abnormal movements highlights the neurological burden associated with the syndrome and the challenges in managing seizure activity in this population. Antiepileptic therapy with Depakine was initiated to control her seizures, aligning with existing literature advocating for early and aggressive seizure management in individuals with trisomy 18 [4, 18].

The multidisciplinary approach to the patient's care, involving specialists from various disciplines, exemplifies the comprehensive management required for individuals with complex genetic disorders like Edwards syndrome [19]. Collaborative decision-making facilitated tailored interventions addressing the medical, nutritional, and developmental needs, thereby optimizing her overall quality of life. This integrated approach is consistent with current guidelines recommending multidisciplinary care for individuals with trisomy 18 to address the diverse array of medical and psychosocial issues they encounter [2, 18, 20].

Reporting and documenting cases of Edwards syndrome, such as the one presented here, are vital for advancing our understanding of the condition and optimizing clinical care practices [19, 20]. Each case provides insights into the natural history, clinical manifestations, and management strategies relevant to Edwards syndrome, contributing to the collective knowledge base and informing evidence-based practice.

CONCLUSION

In conclusion, the case presented here underscores the challenges and complexities inherent in the management of Edwards syndrome. Through a multidisciplinary approach encompassing medical intervention, nutritional support, and developmental care, we aim to optimize outcomes and enhance the quality of life for individuals with this rare chromosomal disorder. Continued research and reporting of cases are essential for advancing our understanding of Edwards syndrome and improving clinical outcomes for affected individuals.

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