ORIGINAL RESEARCH PAPER

Paediatrics

"A RARE CASE REPORT OF C SYNDROME ."

KEY WORDS: C syndrome, trigonocephaly, dysmorphism, rare disorder.genetic testing.

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ABSTRACT

C Syndrome or Opitz C syndrome (Trigonocephaly-C syndrome) is a very rare genetic disorder characterized by trigonocephaly, facial dysmorphism, hypotonia, and multisystem anomalies. We report a term infant presenting with trigonocephaly,mid facialhypoplasiaand moderate develpoemental delay diagnosed with C syndrome based on clinical feature and genetic tesing .

INTRODUCTION

Opitz C Syndrome, also known as Trigonocephaly C Syndrome, is a rare genetic disorder characterized by premature fusion of the metopic suture (trigonocephaly), distinctive craniofacial features, severe developmental delay, and multiple congenital anomalies. First described in 1969, fewer than 100 cases have been reported worldwide, with very limited data from India. The condition is associated with high neonatal mortality due to complications like central apnea,feeding difficulties, and multisystem involvement. Diagnosis is primarily clinical, often supported by imaging and, when available, genetic testing. We report a rare neonatal case with classical features of C syndrome to emphasize the importance of early recognition, supportive care, and genetic counseling in such lethal syndromes.

Etiopathogenesis -

C Syndrome (Opitz C Syndrome) is a rare, primarily sporadic genetic disorder with an unclear but presumed autosomal recessive or de novo mutational origin. Though no consistent gene has been identified, some cases have shown chromosomal abnormalities such as deletions on chromosomes 13q, 3q, or 12p. The underlying molecular defect likely disrupts early embryogenesis, particularly affecting neural crest cell migration and midline developmental pathways. This results in the hallmark cranial anomaly—trigonocephaly due to premature fusion of the metopic suture—along with other craniofacial and systemic malformations.

The pathophysiology is multisystemic. Abnormal brain development, including ventriculomegaly, corpus callosum agenesis, and cortical dysplasia, contributes to hypotonia, seizures, and global developmental delay. Dysmorphic facial features and systemic anomalies such as congenital heart defects (e.g., ASD, VSD), renal abnormalities, and central respiratory dysfunction further complicate the clinical picture. High neonatal mortality is often due to central apnea, feeding difficulties, and multiorgan involvement, underscoring the syndrome's severe and often lethal nature.

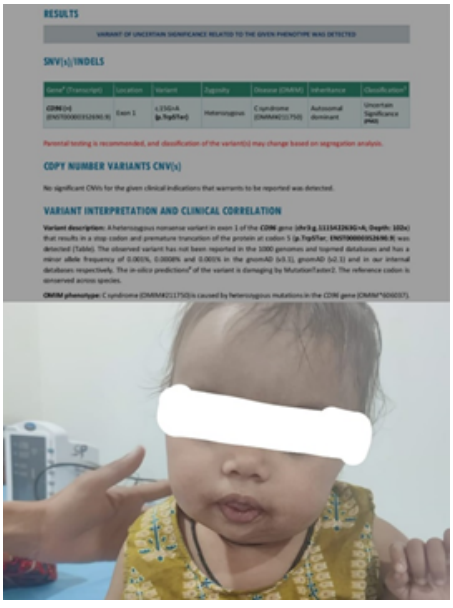
Clinical Feature And Diagnosis

It is charachterised by trigonocephaly, distinct facial dysmorphism, hypotonia, developmental delay, and multisystem anomalies.

Diagnosis is clinical, supported by neuroimaging and genetic studies. Early identification is vital due to high neonatal mortality and absence of definitive treatment.

Case Report-

8 month old female child born to a consanguinously married couple brought by her parents with complain of not able to hold neck and sit with no significant perinatal history,no history of seizures or respiratory distress. On examination patient had axial and appendicular hypotonia and moderate developmental delay with facial dysmorphisim in form of mid facial hypoplasia, triangular shape skull. frontal bossing, depressed nasal bridge, hypertelorism, low set ears ,high arch palate and small chin. Neuroimaging was normal however clinical features were suggestive of craniofacial midline malformation. So supportive care started and genetic testing was done ,which confirmed the diagnosis as " C syndrome". Hence neurodevelopmental therapy continued and genetic counseling was done to the parents .



Treatment And Prognosis-

Treatment of C Syndrome is primarily supportive and multidisciplinary. It includes seizure control, respiratory support for central apnea, feeding assistance, and management of cardiac and renal anomalies. Surgical correction of trigonocephaly is rarely feasible. Early neurodevelopmental therapy and genetic counseling are essential. Prognosis remains poor, with most infants succumbing in the neonatal period due to central apnea, seizures, or multisystem failure.