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Case Report

Severe Intellectual Disability in a Child Born to Consanguineous Parents

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Background

Intellectual disability (ID) is a neurodevelopmental disorder characterized by significant limitations in intellectual functioning and adaptive behavior, with onset during the developmental period (Schalock, 2011). Consanguinity is a known risk factor for inherited neurodevelopmental disorders (Mokhtari & Bagga, 2003).

Case Presentation

The proband is a 9-year-old female child with severe intellectual disability, born to consanguineous parents who are first cousins. The family has two children, of whom only the proband is affected. According to the mother's history, there were no reported illnesses, infections, trauma, or medication used during pregnancy. Antenatal history was unremarkable, and the pregnancy progressed normally. The child was delivered at 38 weeks of gestation via normal vaginal delivery. At birth, the proband had a poor Apgar score and an extremely low birth weight. She required oxygen supplementation immediately after delivery. No congenital anomalies were noted at birth.

Developmental History

The child exhibited significant global developmental delay. Milestone achievements were markedly delayed: Sitting without support at approximately 10 months of age, independent walking at around 2 years of age, First meaningful words at approximately 5 years of age, at present, the proband has severe speech impairment. She can speak only a few words and has difficulty with clear articulation.

Neurological and Behavioral History

The child began experiencing frequent seizures at the age of five years. These seizures are currently managed with antiepileptic medication. In addition, she displays aggressive and violent behavior, for which she is receiving behavioral-modulating medication. She demonstrates poor eye contact and exhibits behaviors consistent with autism spectrum disorder, including social withdrawal and limited communication.

Clinical Examination

Clinical examination revealed normal limbs with no evidence of motor deformities. There were no dysmorphic facial features observed. General physical examination did not reveal any significant abnormalities.

Clinical Assessment

The patient was examined at Lady Ridgeway Hospital (LRH), where clinical findings were consistent with severe intellectual disability associated with epilepsy and autistic features, in the context of parental consanguinity.

Conclusion

This case highlights severe intellectual disability with associated epilepsy and autistic behaviors in a child born to first-cousin parents, emphasizing the importance of genetic counseling and early neurodevelopmental assessment in consanguineous families. Children who have ID frequently also have additional disabilities, and the frequency and quantity of concurrent neurosensory problems rise as the severity of ID increases. In particular, it has been discovered that 4-7% of kids with mild ID and 20-30% of kids with severe ID had epilepsy (Kiely, 1987). Up to 30% of children with severe ID and 6-8% of children with mild ID develop cerebral palsy. Less frequently, sensory problems affect 2% and 11% of children with mild and severe ID, respectively (Murphy et al., 1995). 9-20% of children with ID have been shown to have autism or pervasive developmental disorders, which are more common in children with severe ID. Additionally, 75% of children with pervasive developmental disorders also, have ID (Potgieter & Fryns, 1999).

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