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Case Report

Early-Onset Epilepsy with Pontocerebellar Hypoplasia in a Consanguineous Pakistani Family

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Abstract: Epilepsy is a prevalent neurological condition marked by frequent, spontaneous seizures and symptoms which affect individuals of all ages worldwide (Leenen et al., 2016). Early-onset epilepsy frequently indicates an underlying genetic aetiology with autosomal recessive inheritance, especially when consanguinity and a favorable family history is present (Balestrini et al., 2021). According to estimates, there are 9.99 cases of epilepsy for every 1000 people in Pakistan. It is more common among younger people (those under 30) (Deng et al., 2014). The diagnosis of severe neurodevelopmental epileptic disorders is further supported by neuroimaging findings such as anomalies in the brainstem and cerebellum (Adamczyk et al., 2021).

Keywords: Epilepsy, Pontocerebellar hypoplasia, Consanguinity, Autosomal recessive inheritance, Myoclonic seizures, MRI

Case Presentation:

A consanguineous family from District Charsadda, Khyber Pakhtunkhwa, Pakistan was enlisted. The proband was a 6-year-old male who presented to the Neurology Department at Lady Reading Hospital (LRH), Peshawar with seizures that had started soon after birth. Pedigree analysis was done, and the parents provided written informed consent. Among those affected were the patient's sister, paternal cousin and paternal uncle; the uncle's death before to the examination made the family history crucial. The parents were clinically unaffected, but several first-cousin marriages were recorded, indicating an autosomal recessive route of inheritance. Generalized tonic–clonic seizures (GTCS) and young-onset multifocal myoclonic seizures were discovered during a neurological evaluation.

Investigations:

The laboratory and neurodiagnostic testing were done which included C-Reactive Protein (CRP), Complete Blood Count (CBC), urine examination, electroencephalography (EEG), Renal Function Testing (RFTs) and brain magnetic resonance imaging (MRI). All biochemical and metabolic tests were within normal norms, except for the CBC which revealed anemia (Hb: 9.2g/dL). The EEG showed normal electrical activity. An MRI of the brain showed pontocerebellar hypoplasia and flattened cerebellar hemisphere (the "Dragon sign"), which are signs of a significant neurodevelopmental disorder affecting the brainstem and cerebellum.

Conclusion:

This case highlights the need for detailed family history, pedigree analysis, and neuroimaging when evaluating early-onset epilepsy in consanguineous families. Pontocerebellar hypoplasia, autosomal recessive inheritance and distinctive MRI findings all strongly suggest a genetic origin (Namavar et al., 2011). Future molecular research, genetic counselling and effective therapeutic therapy all depend on early detection.

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