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Letter to Editor

17q12 Deletion Syndrome with Convulsions and Family History of Increased Intracranial Pressure Syndrome

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To The Editor

We present a case of a 5.5-year-old boy diagnosed with 17q12 deletion syndrome, a rare genetic disorder that manifests in a variety of clinical symptoms, including neurological and renal abnormalities (1). We aim to underscore the importance of recognizing early clinical signs and implementing appropriate genetic testing and management.

The child experienced his first convulsion at 8 months, followed by additional seizures at 11 and 14 months, leading to the initiation of levetiracetam at a dosage of 10 mg/kg 2x1 daily (currently on 25 mg/kg 2x1). Despite these early seizures, his developmental milestones have been normal, and no developmental delay has been observed. At the age of 4, bilateral renal cysts were detected through routine imaging, a hallmark feature of 17q12 deletion syndrome due to the involvement of the HNF1B gene.

The clinical course includes an abnormal EEG conducted 1.5 years ago, revealing patterns suggestive of underlying neurological issues. This, coupled with the patient's history of seizures, prompted further investigations. Ultimately, the 17q12 deletion was confirmed through genetic testing, via chromosomal microarray analysis.

It was noted that the mother has been on acetazolamide therapy since the age of 18 due to 17q12 deletion syndrome with convulsions and family history of increased intracranial pressure syndrome syndrome and has intermittently received treatment for migraines associated with severe headaches. The parents and siblings were also evaluated through genetic testing, given the possibility of autosomal dominant inheritance, to identify asymptomatic carriers or those at risk for complications (test results are awaiting).

Key Clinical Features of This Case:

•Seizures: The child had three seizure episodes by the age of 14 months. Despite these, there have been no recurrent seizures since initiating levetiracetam levetiracetam.

•Abnormal EEG: The detection of an abnormal EEG pattern led to the decision to pursue genetic testing, which confirmed the presence of the 17q12 deletion.

•Renal Cysts: Bilateral renal cysts were discovered at age 4, consistent with the deletion's impact on the HNF1B gene, known for its role in renal development and function.

•No Developmental Delay: Despite the genetic diagnosis and seizure history, the child exhibits no signs of developmental delay, which is notable given the syndrome's variability in neurodevelopmental impact.

•Extrarenal monitoring: The patient is being closely monitored for possible future extrarenal manifestations, such as endocrine disorders like diabetes, that are associated with this genetic syndrome.

Management:

1.Neurological Care: The patient remains on Keppra to prevent seizure recurrence, and regular neurological assessments are advised to monitor for any changes in cognitive or motor function.

2.Renal Monitoring: The child is undergoing biannual renal evaluations, including ultrasound and serum creatinine tests, to track cyst progression. If the cysts significantly enlarge or impair renal function, future interventions like cyst drainage or nephrectomy may be considered.

3.Genetic Counseling: The family was informed about inheritance patterns and the potential for the condition to be passed on. Prenatal and preimplantation genetic testing options were discussed, and the family was offered psychological support for coping with the long-term implications

of the diagnosis.

This case demonstrates the importance of early recognition of the diverse manifestations of 17q12 deletion syndrome. Although the child has no developmental delays, his history of seizures, abnormal EEG findings, and renal cysts reflect the syndrome's broad clinical spectrum. Early diagnosis through genetic testing allows for timely interventions, particularly in managing seizure activity and monitoring renal health.

17q12 deletion syndrome is a chromosomal disorder resulting from the deletion of a segment on the long arm (q12) of chromosome 17. This deletion leads to a diverse spectrum of clinical manifestations that can vary significantly, even among affected individuals within the same family. A hallmark feature of this syndrome is the involvement of the renal and urinary systems, ranging from severe congenital malformations that can result in renal failure in utero, to milder or asymptomatic presentations. Renal cysts are particularly prevalent in affected individuals. The syndrome is also associated with the development of maturity-onset diabetes of the young type 5 (MODY5), which typically arises before the age of 25 and is attributed to pancreatic dysfunction. The combination of renal cysts and MODY5 is referred to as renal cysts and diabetes (RCAD) syndrome. Approximately 50% of individuals with 17q12 deletion syndrome exhibit developmental delays, particularly in speech and language, intellectual disabilities, or psychiatric conditions such as autism spectrum disorder, schizophrenia, anxiety, and bipolar disorder. Less commonly, abnormalities in other organ systems, including the eyes, liver, brain, and genitalia, are observed. In females, this may present as Mayer-Rokitansky-Küster-Hauser syndrome, characterized by underdevelopment or absence of the uterus and vagina. Additionally, subtle craniofacial dysmorphisms are sometimes noted. The phenotypic expression of 17q12 deletion syndrome is highly variable, underscoring the complexity of its clinical presentation (3).

We hope that this case adds to the growing body of literature on 17q12 deletion syndrome and encourages clinicians to consider genetic testing in cases of unexplained neurological and renal abnormalities in children.

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DECLARATIONS

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