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Deletion of 15q26.1 region with absence epilepsy respond to valproic acid: A Literature Overview and A Case Report from Qatar.

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ABSTRACT

Chromosomal abnormalities involving deletions and duplications are known to cause severe developmental disorders, including mental retardation, dysmorphism, and seizures, in children. As the technique of array-based comparative genomic hybridization is being applied more frequently in the diagnostic evaluation of children with developmental disorders; novel pathologic chromosomal abnormalities are being identified in relation to various type of epilepsies in childhood.

We report the case of a 4-year-old girl with a history of speech delay and communication disorder, mild dysmorphic features, and absence epilepsy with a de novo microdeletion 15q26.1. A much larger (5 Mb) but overlapping microdeletion has been previously reported in similar several cases with similar phenotype including intractable myoclonic and absence epilepsy, growth delay, and dysmorphic features. This leads us to propose that a potential candidate gene or genes within the deleted region involved in the pathogenesis of some forms of generalized intractable epilepsy, previously considered idiopathic should consider genetic study for childhood epilepsies especially if it was associated with underlying developmental delay in any particular aspect as speech delay in our case.

Keywords: epilepsy/seizures, Chromosomes.

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CASE REPORT

We report a 4 years old girl who is referred to the pediatric neurology clinic at Sidra Medical Center for evaluation for her recurrent attacks of staring with occasional up rolling of eyes for seconds

She had history of uneventful perinatal period with normal developmental progress apart of speech delay. Her neurological examination was unremarkable with subtle dysmorphic features.

EEG showed evidence of generalized epilepsy as below:

Awake: During wakefulness, the background characterized by moderate amounts of bilaterally symmetrical 20-30 microvolts rhythmic 8-9 Hz alpha activity seen over the parietal-occipital head region(s). Reactivity to eye opening is symmetrical.

Sleep: Sleep not recorded.

Interictal Activity: There was a frequent multifocal epileptic abnormality recorded in the form of medium amplitude spike and slow waves seen in the left fronto-centro-parietal (F4, C4, P4), left parietal (P3) and left temporo-occipital (P7, O1) regions. In addition, generalized irregular bursts of spike and slow waves were also recorded, with shifting predominance between the right and left hemisphere.

Electro-Clinical Events: There were no clinical events during this recording.

Intermittent Photic Stimulation: was carried out between 3 Hz to 30 Hz and resulted in a good and symmetric driving response bilaterally.

Hyperventilation: was performed for 3 minutes with good effort and resulted in increase in the frequency of the interictal discharges.

Valproic acid started on 40 mg/kg/day and she had both clinical and electroencephalographic improvement concerning her absence seizures in addition to his speech.

Her genetic which was sent as part of evaluation to her speech delay showed evidence of deletion of 15 q 26 .1 which could be associated with epileptic encephalopathy as the result below Interpretation:

The aCGH analysis revealed a loss of ~2 Megabases (Mb) in the long arm of chromosome 15 within cytogenetic band 15q26.1. This copy number change results in deletion of CHD2 gene. Heterozygous mutations and distruption of this gene have been reported in patients with Epileptic encephalopathy, childhood-onset (OMIM 615369).

DISCUSSION:

Microarray analysis has been shown to be of crucial importance for the understanding of the genetic etiology of intellectual disability (ID). With this technique, numerous (de novo) copy number variations have been discovered leading to several circumscribed microdeletion syndromes.1, 2 especially in patients in whom neuropsychiatric disorders or congenital anomalies are present, in addition to ID, extensive genetic analysis is warranted.

A 15q26 deletion is a genetic condition that occurs when there is a small piece of genetic material (DNA) missing from one of the 46 chromosomes – chromosome 15. Deletions of the 15q26 region encompassing the chromodomain helicase DNA binding domain 2 (CHD2) gene have been associated with intellectual disability, behavioral problems, and several types of epilepsy.1.

Partial deletions of the long arm of chromosome 15, particularly in the 15q26 region, have been typically associated with somatic anomalies, especially congenital diaphragmatic hernia.2

In addition, ID, epilepsy, and behavioral problems are frequently mentioned. Both microdeletions encompassing this gene and de novo loss of function mutations lead to a syndrome characterized by ID and epileptic encephalopathy with generalized seizures.4-6 febrile seizures starting at early age followed by various epileptic features, myoclonic as well as tonic–clonic and absence seizures were reported in several cases.10

Conclusion

Potential candidate gene or genes within the deleted region involved in the pathogenesis of some forms of generalized intractable epilepsy, previously considered idiopathic should consider genetic study for childhood epilepsies especially if it was associated with underlying developmental delay in any particular aspect as speech delay in our case. Absence epilepsy can be associated with deletion of 15q26.1 region with good response to valproic acid.

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