A Child with 5q Deletion and Accompanying Chiari 1 Malformation

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Received: 4 February 2020 / Accepted: 9 July 2020 / Published online: 16 July 2020
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To the Editor: We present a 1-y-old girl who had been detected to have a ~13 Mb deletion on chromosome 5q11.2, and was referred to from pediatrics with Chiari 1 malformation. Prescott et al. have firstly described 5q11.2 microdeletion syndrome [1].

The patient was the second healthy child of non-consanguineous parents. She was the 3rd pregnancy of the mother; 2nd pregnancy resulted in abortion. She had a healthy elder brother of 5 y. She was born by cesarean section at the 39th wk of pregnancy. Birth parameters were within normal range. Choanal atresia was noted at birth. She had nasal surgery at 8 d of age. She was found to have nystagmus and hypermetropia. Prominent nasal root/broad nasal bridge, micrognathia, retrognathia and long filtrum and a high/narrow palate were detected. She had bilateral dysplastic ears. Her second toe was overlapping with third toe, on the left foot. Developmental Quotient (DQ) was calculated as >85. Brain MRI showed, mild ventricular dilatation without parenchymal loss with Chiari 1 malformation [2].

The karyotype analysis revealed 46,XX,del(5)(q11.2-q12.1). Whole genome array comparative genomic hybridization (aCGH) was performed on an Agilent 4x180K (CGH + SNP) array slide. It revealed a clinically significant ~13 Mb deletion at 5q11.2q12.1 starting from 50.288.355 to 63.149.770 bp. Parents' karyotypes and aCGH results were normal. So the deletion was specified as de novo. We used a next-generation sequencing (NGS) panel, targeting 49 genes mutated in autism spectrum disorder but it did not reveal any pathogenic variants.

To date, only 13 cases of 5q11.2 microdeletion have been reported [2]. Although no different phenotype has been identified for this microdeletion syndrome, published cases share many common clinical features [3, 4]. Dysplastic-ears,

micrognathia, hypermetropia, central nervous system (CNS) malformations were evaluated as concurrent findings with the literature in our patient. Among the cases in the literature, development delay (DD) in ~91%, intellectual disability (ID) in ~67%, CNS anomalies in ~64% and facial dysmorphism was reported in almost all patients. However, Chiari 1 malformation has been reported for the first time in our case. Some genes that are included in the deleted region are known to be expressed in the CNS and that could potentially cause the anomalies [5]. More studies are needed to demonstrate the effects of the other genes in 5q11.

Compliance with Ethical Standards

Conflict of Interest None.

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