An Aicardi-Goutieres Syndrome 2 Case

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INTRODUCTION

Aicardi-Goutieres Syndrome 2 (AGS2) is a rare disease characterized by progressive neurological dysfunction starting in the first 2 years of life, accompanyied by encephalopathy, spastic paraplegia and dystonia.

In this article, we report an AGS2 case who was admitted at the age of 4 months due to lack of weight gain, followed up with congenital glaucoma, febrile convulsion, psychomotor retardation, microcephaly, hypotonia and diffuse spasticity. Patient's definitive diagnosis was made by demonstrating the RNASEH2B gene mutation in the Whole Exome Sequencing (WES) analysis.

CASE

HISTORY: A 26-month-old female patient with psychomotor retardation, diffuse spasticity, and recurrent simple febrile convulsions. Patient was admitted at the age of 4 months due to lack of weight gain and developed congenital glaucoma, febrile convulsion, psychomotor growth retardation, microcephaly, hypotonia and diffuse spasticity in the follow-up

BACKROUND & FAMILY HISTORY: Parental consanguinity + (cousins)

PHYSICAL EXAMINATION: Head circumference: 37 cm (<3p), no eye contact, no object and light tracking, bilateral simian line in hands, axial hypotonia +, diffuse spasticity throughout the body +, bilateral DTR in upper extremities normactive, bilateral DTR in lower extremities +++/+++, bilateral Babinski +, no clonus (Figure 1).

LABORATORY

- Brain CT: Calcification in the cerebral and cerebellar hemispheres, basal ganglia (17 months) (Figure 2).
- Cranial MRI: Brain stem and vermis are hypoplasic, loss of white matter at centrum semiovale level, increased signal in the remaining white matter, white matter myelination backward for age (10 months) (Figure 3).
- Electroencephalography (EEG): Normal (due to suspected seizure at 10 months of age).
- Chromosome analysis: 46, XX.
- Sequence analysis of the SMN gene: Normal.
- Microarray: Normal.
- Whole exome sequencing: Homozygous c.511G>T mutation (OR) in the RNASEH2B (NM_024570.4) gene.

Figure 1.

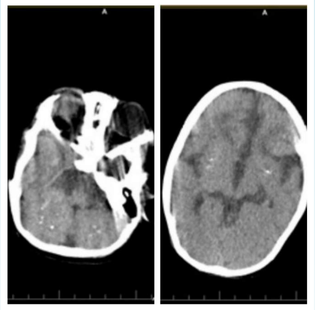

Figure 2.

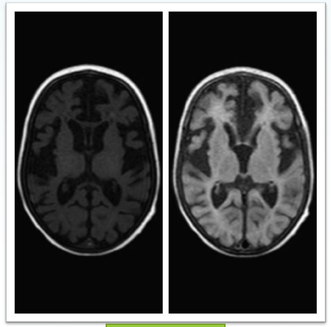

Figure 3.

CONCLUSIONS

AGS2 is characterized by progressive neurological dysfunction, encephalopathy, spastic paraplegia and dystonia. A small number of cases with normal cognitive development have also been reported. Cerebral and cerebellar calcifications, especially in the basal ganglia, are detected on brain computed tomography (CT) or brain magnetic resonance imaging (MRI). The disease is caused by homozygous or compound heterozygous mutations in the RNASEH2B gene region located in the 13q14 region and encoding the B subunit of ribonuclease H2 and is inherited as an autosomal recessive.

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