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Poster Presentation

A Case Report on a New Aicardi-Goutieres Syndrome Inducing Gene

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Abstract

Aicardi-Goutieres syndrome (AGS) is an inflammatory genetic disease inherited in an autosomal recessive manner. Common features of this disease are encephalopathy, splenomegaly and hepatomegaly, muscle stiffness, irritability, unstoppable crying, seizures and dilation in growth. According to previous studies, primary genes responsible for this Syndromes are as followed: TREX 1, RNASEH2A, RNASEH2B and RNASEH2C. Moreover, mutation in ADAR and SAMHD1 genes are assumed to play part in AGS. In this case we found a new gene mutation probably responsible for this syndrome. A 4 years old female with encephalopathy, one ear hearing impairment, strabismus, and hypertonic upper and lower limbs with tapering fingers was admitted to our genetic clinic. According to these clinical features and supplementary testing, she was diagnosed with AGS. Further molecular genetic testing indicated no homozygous mutations in common genes responsible for AGS in despite that both of her parents had a mutation in RNASEH2C and she carries only one mutated copy of RNASEH2C but a homozygous mutation in LAMA 1 gene, which encodes Laminin alpha 1 chain, was found. Previous studies demonstrated that LAMA 1 mutation could lead to motor neuron impairment and optical defections. It is necessary to emphasize that both of her parents had abnormal LAMA1 gene. With regard to our testing results, RNASEH2C mutation and LAMA 1 c.1957C>T mutation was concluded to be responsible for AGS in this case as a compound heterozygote. As a result, LAMA 1 can be introduced as a new AGS inducer. The findings of present research suggest the family should be subjected to PND in any pregnancy. After this report it is recommended to check LAMA 1 gene along with other responsible genes as a candidate for this syndrome.

Keywords: Aicardi-Goutieres Syndrome, LAMA 1, RNASEH2C

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