2nd International & 14th Iranian Genetics Congress
دومین کنگره بین المللی و چهاردهمین کنگره ملی ژنتیک ایران

Detection of A New Mutation in TREX1 Gene Associated with Aicardi-Goutieres Syndrome

Mohammad Hasan Samiee Aref[1], Azadeh Hadadianpour[2], Pardis Ghazizadeh Tehrani[2], Samira dabbagh Bagheri[2], Masoumeh Amini[2], Hamideh Bagherian[2], Sirous Zeinali[2,3]

1,2- Kawsar Human Genetics Research Center
3- Department of Molecular Medicine, Biotechnology Research Center, Pasteur Institute of Iran, Tehran. Iran

Introduction

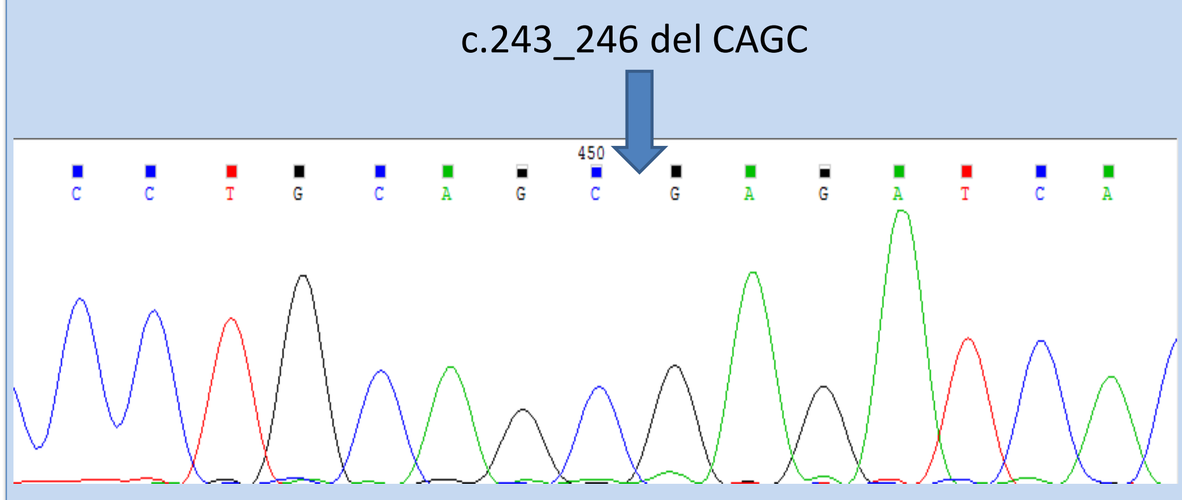
Aicardi-Goutieres syndrome (AGS) is a rare inherited encephalopathy with a clinical phenotypes mimicking congenital viral infection resulting from homozygous or compound heterozygous mutations in the TREX1 gene. AGS is mostly inherited in an autosomal recessive pattern, while autosomal dominant pattern of inheritance is also reported. The brain, the immune system, and the skin are the main target organs of this disease. Besides, cerebral calcifications, white matter abnormalities and cerebral atrophy are the main symptoms of this disease.

Method

A couple referred to our PGD center because their first child was affected with AGS according to the clinical symptoms presented. A pedigree was drawn and general screening questions were asked about each relative. Then, The TREX1 gene was analyzed using whole exome sequencing (NGS) which suggested a homozygous deletion in exon 1 of this gene. Besides, Sanger sequencing was used for backfilling NGS data. Forward and reverse primers were designed for flanking regions of the mutation site. The entire TREX1 gene was sequenced for parents and the affected child. The affected child showed a homozygous deletion and the parents were heterozygous for that deletion.

Results

A novel pathogenic mutation was observed in the first exon of TREX1 gene. This mutation is c.243_246 del CAGC (p.Ala135Alafs*5) which has not been reported yet.



References

1. Goutières, Françoise. "Aicardi–Goutières syndrome." Brain and Development 27.3 (2005): 201-206.
2. Rice, Gillian, et al. "Clinical and molecular phenotype of Aicardi-Goutieres syndrome." The American Journal of Human Genetics 81.4 (2007): 713-725.
3. Orcesi, S., R. La Piana, and E. Fazzi. "Aicardi–Goutières syndrome." British medical bulletin 89.1 (2009): 183-201.