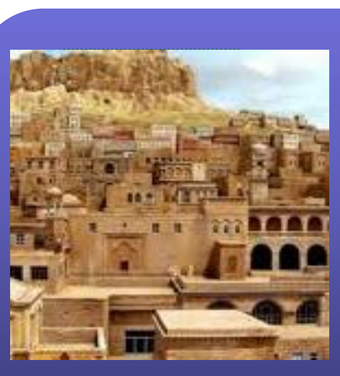

Identification of A Novel Homozygous Mutation in BBS10 in Five Children With Bardet-Biedl Syndrome

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Background

Bardet-Biedl syndrome (BBS) is a rare and multisystemic disorder. Characteristic features of the syndrome are rode-cone dystrophy, obesity, hypogonadism, postaxial polydactyly, learning difficulties and renal abnormalities. The disorder is genetically heterogeneous. To date, 21 genes on different chromosomes have been mapped. The most common genes are BBS1 (locus 11q13) and BBS10 (locus 12q21.2We aimed to report five children whose detected a homozygous mutation in the BBS10 gene.

Patients

Family 1,

The first family was Turkish. The parents were relatives. There were two affected girls in the turkish family, and they were 14 and 17 years old, respectively.

Family 2,

The second family was Syrian. As in the first family, there was a consanguineous marriage in this family. Two of the three affected children in the syrian family were boys and one was girl. Their ages were 4, 7 and 10.

All of the children had obesity, polydactyly and cognitive impairment. Rode-cone dystrophy and hepatic steatosis were more severe in Syrian families. They had no kidney disease. Whereas both children had pelvicalyceal ectasia in the first family. In addition, Elder girl in the first family had pulmonary hypertension. Cognitive impairment was mild in all.

Screening analysis was performed for BBS1, BBS2 and BBS10 genes. There were no changes that could be pathogenic in the BBS1 and BB2 genes.BBS10 gene 1-2. exons were amplified by PCR method and then DNA sequence analysis were done.

We identified a novel homozygous mutation in exon 2 in BSS10 gene. The change in pThr516Asnfs * 8 (c.1547 delC) detected in patients was not defined in HGMD. However, the "mutation taster bioinformatics program" predicts that this change is probably the cause of the disease. This change constitutes an early stop codon. It is thought to be pathogenic for this reason.

Picture 1: Patients with Bardet-Biedl syndrome in Syrian family

Picture 2: Patient with Bardet-Biedl syndrome in Turkish family

Conclusion

BBS has a rare and heterogeneous nature, almost 21 genes on the different choromosome have been mapped for the etiology. Even cardiovasculer disorders are rare in this syndrome we detected severe pulmoner hypertension in one patient. Its necessary to follow up other patients who have tis mutation for cardiovascular complications. Genetic counseling is necessary to confirm the diagnosis. Bardet biedl syndrome should be kept in mind in cases with obesity and cognitive impairment.