Medical Genetics Laboratory of Dr.Zeinali

He is Allah, the Creator, the Inventor, the Fashioner; to Him belong the best names.

Hurler syndrome study in Iran with reporting two novel mutations

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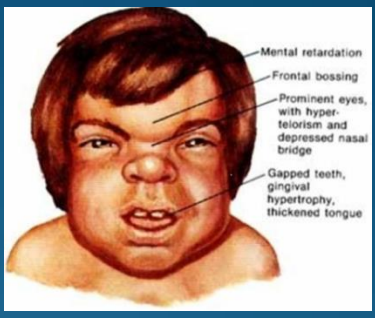
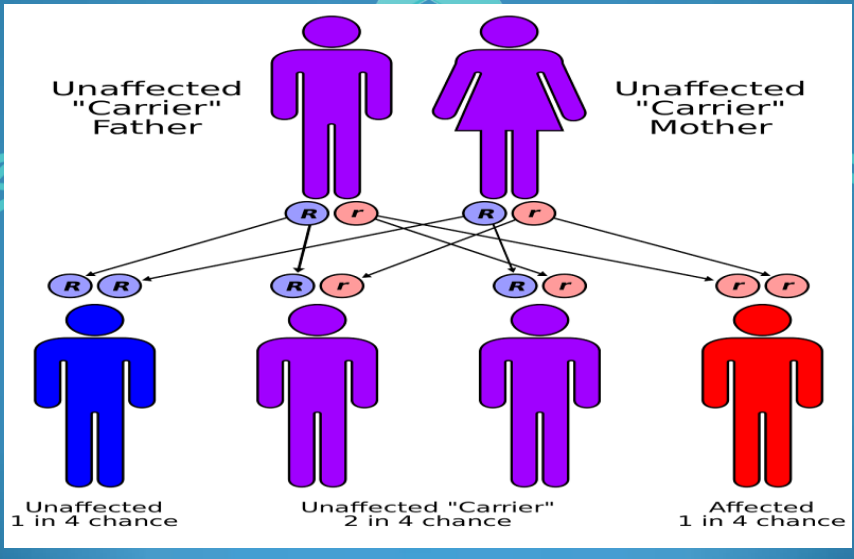


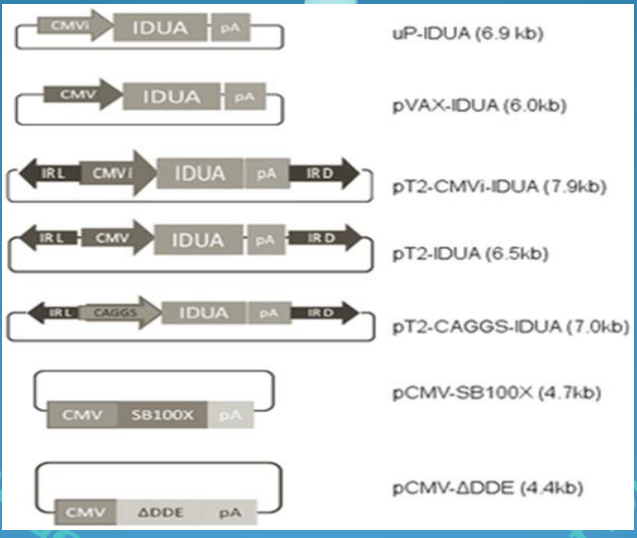
Introduction

Hurler syndrome is the most severe form of mucopolysaccharidoses which is a kind of lysosomal storage disorders. Hurler syndrome is one of the autosomal recessive inborn errors of metabolism with the incidence of 1:100000 in every live birth. Patients show different symptoms like cloudy cornea, scoliosis, hearing impairment, facial anomalies, Joint stiffness, liver and splenomegaly. This syndrome is caused by mutations in α-L-iduronidase (IDUA) gene which is located on 4p16.3 and has 15 exons encoding Iduronate Sulfatase.In this study we investigate IDUA gene mutation in 7 affected Iranian Families using short tandem repeat (STR) markers and Sanger sequencing.



Material and methods

In the present study, mutations in **IDUA** gene were analyzed in a total of 7 Iranian families referred to Kawsar Human Genetic Research Center. Informed consent forms were obtained and DNA extraction was performed using salting out procedure. Haplotype analyses were done using STR markers. These 7 families showed segregation of the disease with the **IDUA** gene. Subsequently all exons and introns boundaries of the **IDUA** gene were sequenced by Sanger sequencing. Potential pathogenicity of the novel variants were evaluated by on line softwares such as Fathmm, Polyphen-2, Hope.



Results

We identified 3 missense mutations which 2 of them were novel. The mentioned soft wares all revealed that the novel mutations could be pathogenic ones.

Conclusion

The obtained results can increase our understanding about etiology of Hurler syndrome in Iranian population and it is helpful in genetic counseling and genetic diagnosis of this disease In Iran.

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