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Mutation spectrum of Mucopolysaccharidosis type III in Iranian patients

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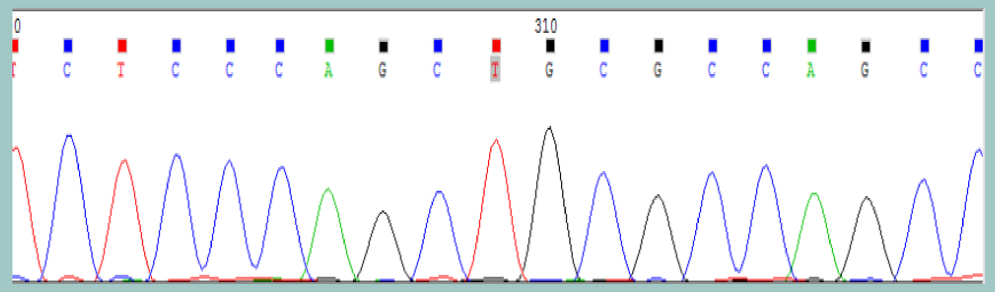
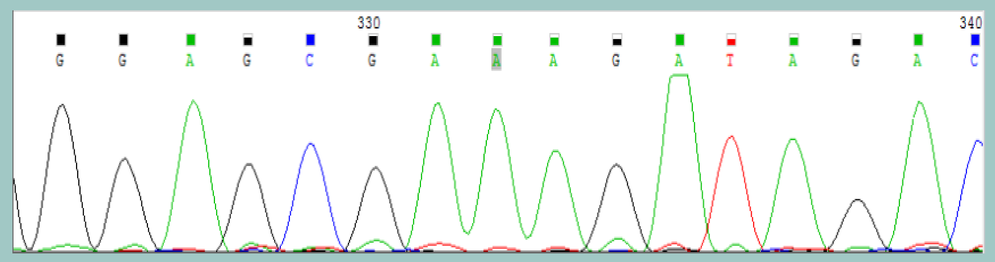
INTRODUCTION

Mucopolysaccharidosis type III (MPS III, Sanfilippo disease) is characterized by delayed speech development, sleep disturbance, hyperactivity and aggressiveness. It results from deficiency of one of the four enzymes normally involved in degradation of heparin and heparin sulphate. These enzymes are heparan sulfate sulfamidase (SGSH), α-N-acetylglucosaminidase (NAGLU), heparan sulfate acetyl-CoA: α-glucosaminide N-acetyltransferase (HGSNAT), and N-acetylglucosamine-6-sulfatase (GNS).



SGSH gene
c.220>T



NAGLU gene
c.457G>A

Material and methods

4 Iranian patients were referred to Medical Genetics lab of Dr.Zeinali. DNA was extracted using salting out procedure. There were analyzed for MPSIII genes using Sanger sequencing.

Result and Discussion

Three different mutations were identified in SGSH gene (c.220>T, c.1079 Del C and c.1298G>A) that have been previously described. In one of the families, c.457G>A mutation was located at NAGLU gene. These mutations influenced the structure and function of the protein. The results obtained from the present study increased our understanding from the molecular basis of MPS III in Iranian patients.

Keywords

Mucopolysaccharidosis type III, Iranian population, SGSH gene, NAGLU gene

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