2nd International and 10th National Biotechnology Congress of Islamic Republic of Iran
August 29-31, 2017
Seed and plant Improvement Institute, Karaj, Iran
دومین همایش بین المللی و دهمین همایش ملی بیوتکنولوژی جمهوری اسلامی ایران
۷-۹ شهریور ماه ۱۳۹۶
کرج، سالن همایش های موسسه تحقیقات اصلاح و تهیه نهال و بذر

# A novel mutation in the *LYST* gene caused Chediak Higashi syndrome in an Iranian family

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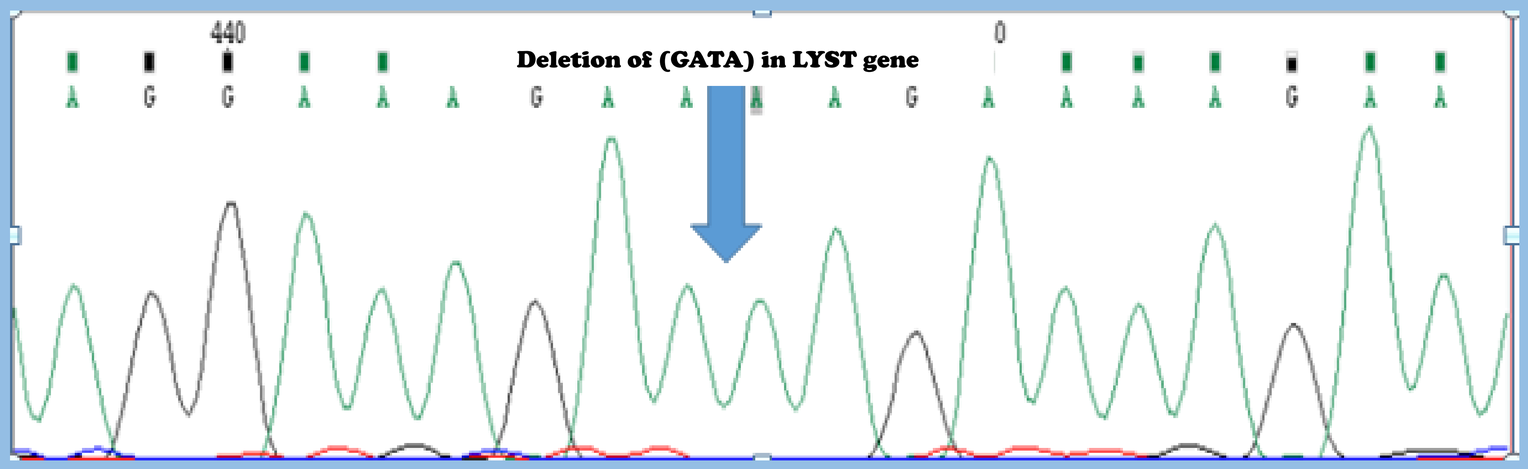
## Introduction

Chediak-Higashi syndrome (CHS) is a rare autosomal recessive immunodeficiency disorder caused by mutations in LYST gene encoding lysosomal trafficking regulator. It is characterized by oculocutaneous albinism, frequent infections, hypopigmentation, neurological dysfunction and susceptibility to infections of the skin. To the best of our knowledge, there are a few reports from CHS in Iran. Herein, we present a case with continuous high fever, hypopigmentation of the skin and long infection

## Materials and methods

A 3-y-old boy suspicious to CHS with no family history of the disease was referred to Dr.Zeinali's Medical Genetics laboratory for mutation detection and confirmation of clinical diagnosis. The parents were first cousins. LYST gene sequencing was performed to identify the causative mutation. Segregation analysis of the identified variant was performed in family. The mutation was evaluate by ACMG guideline to assess the pathogenicity.




## Result

*Targeted* gene sequencing revealed a novel homozygous mutation of c.4488-4491del GATA *(p.Lys1496LysfsX8)* in *LYST gene. Sanger sequencing confirmed the heterozygosity of the identified variants in the parents.*

## Discussion

Identification of novel mutations in *LYST gene will expand the databases presenting the causative mutations of* CHS. It also helps prenatal diagnosis (PND) and preimplantation genetic diagnosis (PGD) for at risk families. *The mutation presented here is pathogenic according to ACMG guidelines. The resulting protein is 2297 residues smaller than the wild type protein. This protein cannot be active properly and has the potential to be disease causing.*

**Key words:** *LYST, next generation sequencing, novel mutation, Chediak-Higashi syndrome*

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