**Finding a very rare mutation in non‐Caucasian LCAT patients from Southwest Asia for the first time**

[Farshid Oliaei](https://onlinelibrary.wiley.com/action/doSearch?ContribAuthorStored=Oliaei%2C+Farshid)

[Behdokht Batebi](https://onlinelibrary.wiley.com/action/doSearch?ContribAuthorStored=Batebi%2C+Behdokht)

[Reza Tabaripour](https://onlinelibrary.wiley.com/action/doSearch?ContribAuthorStored=Tabaripour%2C+Reza)

[Haleh Akhavan Niaki](https://onlinelibrary.wiley.com/action/doSearch?ContribAuthorStored=Akhavan+Niaki%2C+Haleh)

First published: 02 December 2018

[**https://doi.org/10.1002/jcb.27981**](https://doi.org/10.1002/jcb.27981)

Citations: [2](https://onlinelibrary.wiley.com/doi/abs/10.1002/jcb.27981#citedby-section)

[**Read the full text**](https://onlinelibrary.wiley.com/doi/full/10.1002/jcb.27981)

[PDF](https://onlinelibrary.wiley.com/doi/epdf/10.1002/jcb.27981)

[TOOLS](https://onlinelibrary.wiley.com/doi/abs/10.1002/jcb.27981)

[SHARE](https://onlinelibrary.wiley.com/doi/abs/10.1002/jcb.27981)

Abstract

Introduction

Lecithin cholesterol acyltransferase (LCAT) deficiency is an autosomal recessive disorder occurred by different mutations in the LCAT gene that cause two extremely rare syndromes including familial LCAT deficiency (FLD) and fish‐eye disease (FED). Unlike FED in FLD renal failure is the most important defect due to deposition of abnormal lipoproteins in the renal stroma. In this study, FLD patients from the North of Iran were investigated for mutations in the LCAT gene.

Materials and Methods

Eight patients with corneal opacification and renal defect were analyzed for mutations in the LCAT gene by PCR sequencing.

Results

Sequencing analysis revealed a missense pathogenic variation c.301 G>A in exon 2 of LCAT gene in all patients changing the amino acid aspartate to asparagine at the conserved position of amino acid 101 of LCAT protein.

Conclusion

In this study, a very rare variation was reported for the first time in this part of the world. Investigation of a larger number of LCAT patients in different parts of Iran can provide availability of mutations panel that is useful for phenotype prediction and also prenatal diagnosis programming in families with a previous history of the disease.