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MOLECULAR ANALYSIS OF NCF1 GENE IN 42 IRANIAN PATIENTS WITH CHRONIC GRANULOMATOUS DISEASE (INTRODUCING 2 NOVEL MUTATIONS)

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Background:

Chronic granulomatous disease (CGD) is a rare primary immunodeficiency disorder of the innate immune system. The disease is classified by mutations in specific subunits of nicotinamide adenine dinucleotide phosphate (NADPH)-oxidase enzyme (membrane-bound subunits gp91-*phox*, p22-*phox*, and cytosolic subunits p47-*phox*, p67-*phox*). The defect leads to defective intracellular killing by phagocyte cells. One of the components of NADPH-oxidase is p47-*phox* encoded by *NCF1* gene. This study aims to identify new genetic changes in *NCF1* gene among 42 Iranian patients with CGD.

Methods:

Forty two CGD patients based on clinical findings, defect in NBT test, DHR-123 assay and loss of p47-phox in western blotting were entered this study. They were referred to Immunology, Asthma and Allergy Research Institute (IAARI) for diagnosis and treatment (between 2008- 2016). To identify mutations of *NCF1* gene, we used PCR amplification followed by direct sequencing for all exons and exon-intron boundaries.

Results:

Mutation analysis of *NCF1* gene in all patients identified six different mutations including 2 novel ones: a missense mutation c.328C>T (p.R110C), and a nine-nucleotide deletion, c.331_339TGTCCCCACdel (p.111_113CPHdel).

Conclusion:

Two novel mutations of *NCF1* gene were found in 42 Iranian patients with CGD. Genetic detection of these mutations results in early diagnosis and prevention of possible complications of disease. It will be helpful for carrier detection as well as prenatal diagnosis of next children in the affected families.