



Int J Hum Genet, 17(4): 145-150 (2017) DOI:10.31901/24566330.2017/17.04.01

## New Mutations in APC Gene Among Familial Adenomatous Polyposis (FAP) Patients in Iran

Payam Shahnazi Gerdehsang<sup>1</sup>, Najmeh Ranji<sup>2</sup>, Mojtaba Gorji<sup>3\*</sup>, Safoura Pakizehkar<sup>4</sup>, Ali Asghar Kiani<sup>5</sup> and Saeed Veysi<sup>6</sup>

<sup>1</sup>Department of Biology, Damghan Branch, Islamic Azad University, Damghan, Iran Phone: <sup>1</sup><+989128135903>, E-mail: <sup>1</sup><payampsq@yahoo.com> <sup>24</sup>Department of Biology, College of Science, Rasht Branch, Islamic Azad University, Rasht, Iran Phone: <sup>2</sup><+989112407063>,<sup>4</sup><+989121944463> E-mail: <sup>2</sup><najmehranji@gmail.com><sup>4</sup><PAKIZEHKAR\_S@yahoo.com> <sup>3</sup>Department of Hematology and Oncology, Lorestan University of Medical Sciences, School of Medicine, Khorramabad, Iran Phone: <sup>3</sup><+989121948358>, E-mail: <sup>3</sup><mogorji@yahoo.com> <sup>5</sup>Department of Immunology, Lorestan University of Medical Sciences, School of Medicine, Khorramabad, Iran Phone: <sup>5</sup><+989166638354>, E-mail: <sup>5</sup><aliasgharkianii@gmail.com> <sup>6</sup>Young Researchers and Elite Club, Rasht Branch, Islamic Azad University, Rasht, Iran Phone: <sup>6</sup><+989178714738> E-mail: <sup>6</sup><saeed.veysi@gmail.com>

KEYWORDS Adenomatous Polyposis Coli. Familial Adenomatous Polyposis. N862K. Small Deletion

**ABSTRACT** Familial adenomatous polyposis (FAP) is a disorder by autosomal dominant inheritance caused by mutations in adenomatous polyposis coli (APC) gene. The aim of this paper was the investigation of a part of exon 15 of the *APC* gene in FAP patients in several provinces of Iran. Blood sample was obtained from FAP patients. A part of exon 15 of *APC* gene was amplified by PCR and underwent direct sequencing. Researchers found 23 FAP patients by severe polyposis in colorectal and identified new mutations in the three patients, including c.2910deIT (17 year old) and c.3577-3578 deICA (30 and 34 year old) with severe polyposis and a substitution (N862K) in  $\alpha$  patients. In this paper, small deletions in *APC* gene led to produce truncated non-functional *APC* protein. N862K mutation appears to be important for developing the disease. The results of this paper confirmed that there is a correlation between age of onset and phenotype with the proximity of the mutation to 5'-end of gene.