Online journal ISSN - 1676-5680
GMR Genetics and Molecular Research
EVOLUTION AND TECHNOLOGY

The G1138A mutation rate in the *fibroblast growth factor receptor 3* (*FGFR3*) gene is increased in cells carrying the t (4; 14) translocation

P.L. Reddy and R.P. Grewal

Laboratory of Neurogenetics, New Jersey Neuroscience Institute,
JFK Medical Center, Edison, NJ, USA

Corresponding author: R.P. Grewal
E-mail: rgrewal@solarishs.org

Genet. Mol. Res. 8 (2): 435-439 (2009)
Received December 5, 2008
Accepted March 9, 2009
Published April 22, 2009

ABSTRACT. Spontaneous mutations are a common phenomenon, occurring in both germ-line and somatic genomes. They may have deleterious consequences including the development of genetic disorders or, when occurring in somatic tissues, may participate in the process of carcinogenesis. Similar to many mutational hotspots, the G1138A mutation in the *fibroblast growth factor receptor 3* (*FGFR3*) gene occurs at a CpG site. In germ-line tissues, the G1138A mutation results in achondroplasia and has one of the highest spontaneous mutation rates in the human genome. Although not at the G1138A site, there are increased rates of other somatic mutations in the *FGFR3* gene that have been reported in multiple myeloma cases associated with a translocation, t (4; 14). The chromosome-4 break points in this translocation are clustered in a 70-kb region centromeric to the *FGFR3* gene. We hypothesized that this translocation may impact the mutation rate at the G1138A site. We employed a semi-quantitative polymerase chain reaction-based assay to measure the frequency of this mutation in multiple myeloma cell lines carrying t (4; 14) translocation. Analysis of these cell lines varied from no change to a 10-fold increase in the mutation frequency compared with normal controls. In general, there was an increase in the G1138A mutational frequency suggesting that chro-

mosomal rearrangement can affect the stability of the CpG hotspots.

Key words: Spontaneous mutation; Translocation; G1138A mutation; *Homo sapiens*; Multiple myeloma; Semi-quantitative assay

