

In vitro transcription of compound heterozygous hypofibrinogenemia Matsumoto IX; first identification of FGB IVS6 deletion of 4 nucleotides and FGG IVS3-2A > G causing abnormal RNA splicing

著者	Terasawa Fumiko, Kamiyjo Yuka, Fujihara Noriko, Yamauchi Kazuyoshi, Kumagai Toshiko, Honda Takayuki, Shigematsu Satoshi, Okumura Nobuo
journal or publication title	Clinica chimica acta
volume	411
number	17-18
page range	1325-1329
year	2010-09
権利	(C) 2010 Elsevier B.V.
URL	<a href="http://hdl.handle.net/2241/106057">http://hdl.handle.net/2241/106057</a>

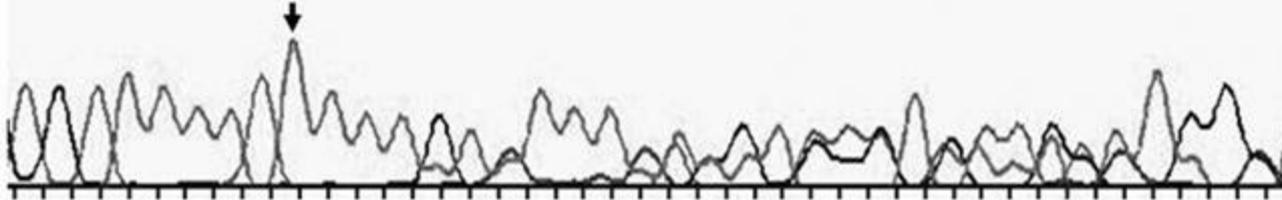
doi: 10.1016/j.cca.2010.05.030

**Genomic DNA**

AGATTTTATTTTGTCTTTCNNGTNANTGAAGNNTGGC

6489

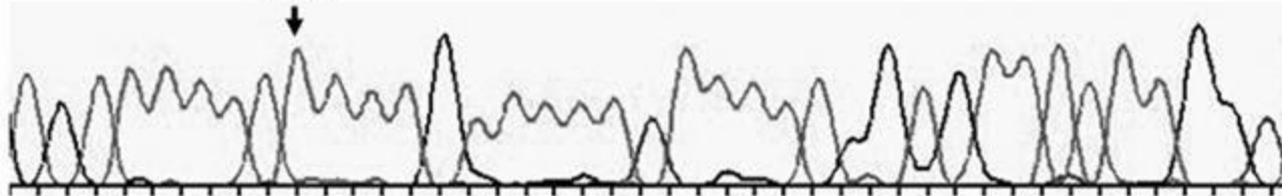
MIX

**Subcloning DNA**B $\beta$ -intron6B $\beta$ -exon7

AGATTTTATTTTGTTTTCTTTTAGGTGAATATTGGC

6489

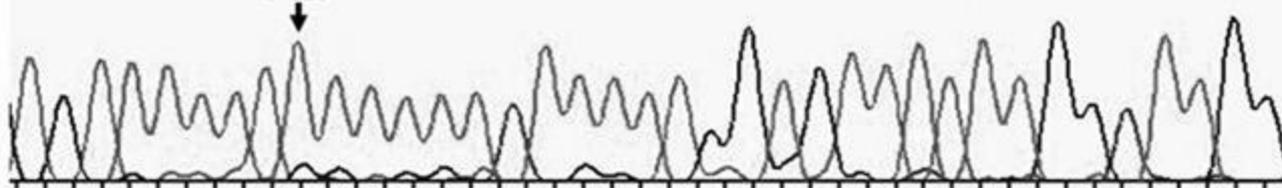
Wild type

TTT,G deletion

AGATTTTATTTTTTTCTTTTAGGTGAATATTGGCTTGG

6489

MIX

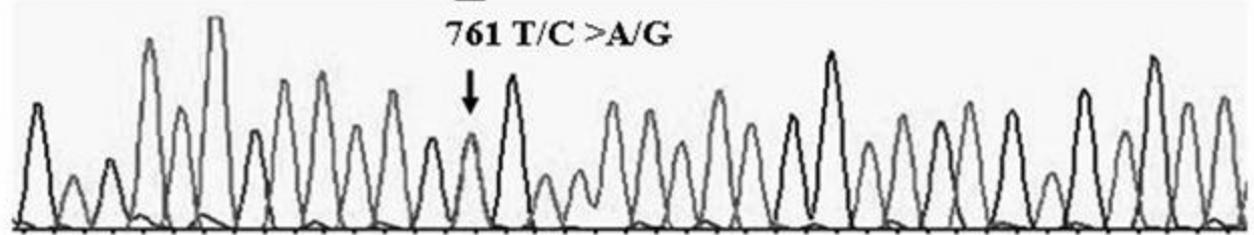


**Genomic DNA**

GTCTATCATATC N GTAATATA GGATCAGAGACAT

761 T/C > A/G

MIX

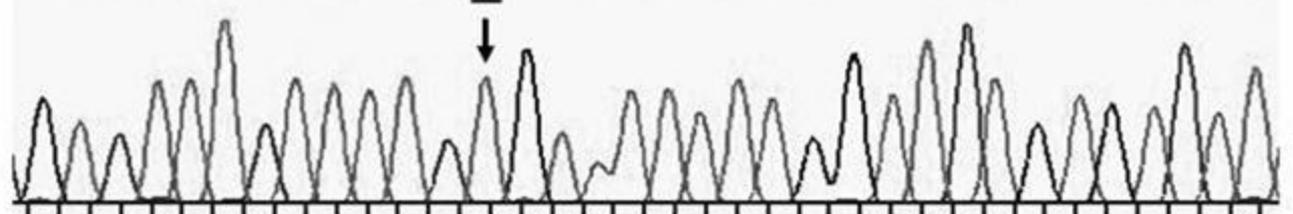
**Subcloning DNA**

$\gamma$ -exon4

$\gamma$ -intron3

GTCTATCATATC T GTAATATA GGATCAGAGACAT

Wild type

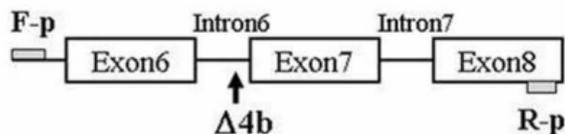


GTCTATCATATC C GTAATATA GGATCAGAGACAT

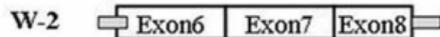
MIX



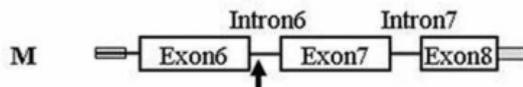
A

Amplified PCR products of the *FGB*

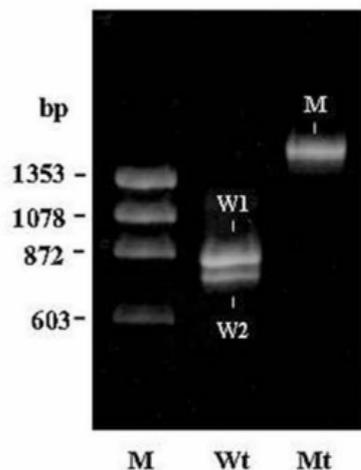
## Wild type transcript

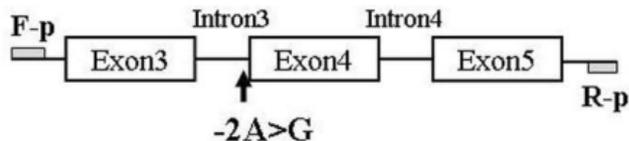
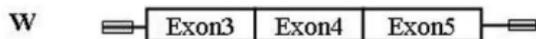


## IVS6 Δ4b transcript



+17 novel amino acids, and stop codon



**B****Amplified PCR products of the *FGG*****Wild type transcript****IVS3 -2G transcript**