

Supplementary Figure 1

Exclusion filters

- 3'and 5' UTR variant,
- downstream gene variant,
- intergenic region,
- Intronic region,
- no-coding exon variant,
- splice acceptor and donor variant,
- splice region variant,
- synonymous variant
- upstream gene variant

- Not Identified by at least two of POLYPHEN, POLYPHEN2 and PROVEAN as disease causing

78K-97K detected variants



50- 70 filtered variants

Inclusion filters

- Single nucleotide variants in the coding regions:
 - a- Missense
 - b- Stop codon gained and lost
 - Inframe deletions and insertions
 - Frameshift deletions and Insertions
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- Allele frequency (1000GP3_AF) to < 1%

Patient	# of SNPs	Missense Var	Stop gained	Stop lost	Frameshift Var	Het/Hom Ratio
13.1	78,595	10,294	90	38	271	1.3
13.2	96,911	11,274	110	46	333	1.5
14.1	81,277	10,549	94	34	275	1.4
14.2	82,234	10,626	94	35	290	1.5