Table 2 | Estimated numbers of potentially functional variants in genes

Class	Combined total	Combined novel	Low coverage		High-coverage trio		Exon capture		
			Total	Interquartile*	Total	Individual range	Total	Interquartile*	GENCODE extrapolation
Synonymous SNPs	60,157	23,498	55,217	10,572–12,126	21,410	9,193–12,500	5,708	461–532	11,553–13,333
Non-synonymous SNPs	68,300	34,161	61,284	9,966–10,819	19,824	8,299–10,866	7,063	396–441	9,924–11,052
Small in-frame indels	714	383	666	198–205	289	130–178	59	1–3	~25–75
Stop losses	77	40	71	9–11	22	4–14	6	0–0	~0–0
Stop-introducing SNPs	1,057	755	951	88–101	192	67–100	82	2–3	~50–75
Splice-site-disrupting SNPs	517	399	500	41–49	82	28–45	3	1–1	~50
Small frameshift indels	954	551	890	227–242	433	192–280	37	0–1	~0–25
Genes disrupted by large deletions	147	71	143	28–36	82	33–49	ND	ND	ND
Total genes containing LOF variants	2,304	NA	1,795	272–297	483	240–345	77	3–4	~75–100
HGMD 'damaging mutation' SNPs	671	NA	578	57–80	161	48–82	99	2–4	~50–100

NA, not applicable; ND, not determined.
*Interquartile range of the number of variants of specified type per individual.