EthniVar: a source-stratified catalogue of BRCA1/2 germline variants quantifies the classification gap for variants reported in Indian patients

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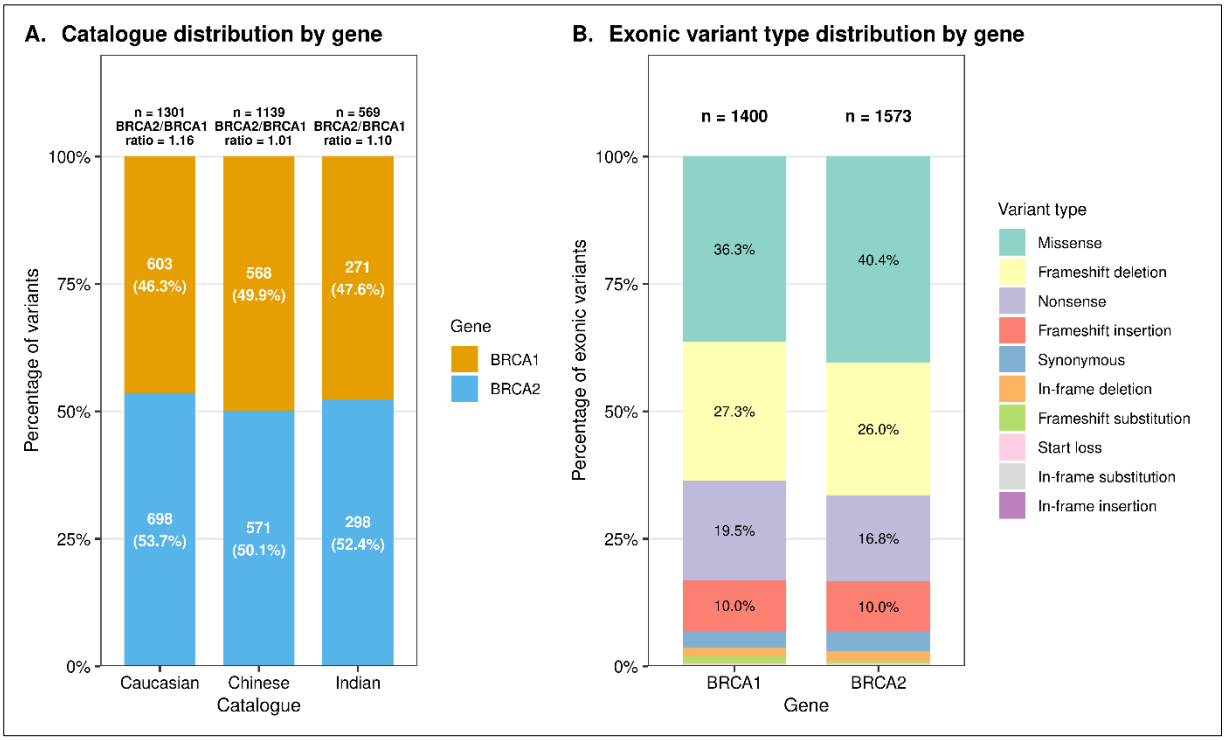


FIGURE S1. BRCA1/2 specific variants across three populations and their variant type for exonic variants. Panel A) shows population distribution by gene, stacked to 100% for individual populations. Panel B) shows variant type distribution by gene, where stacked percentage bars show exonic variant composition.

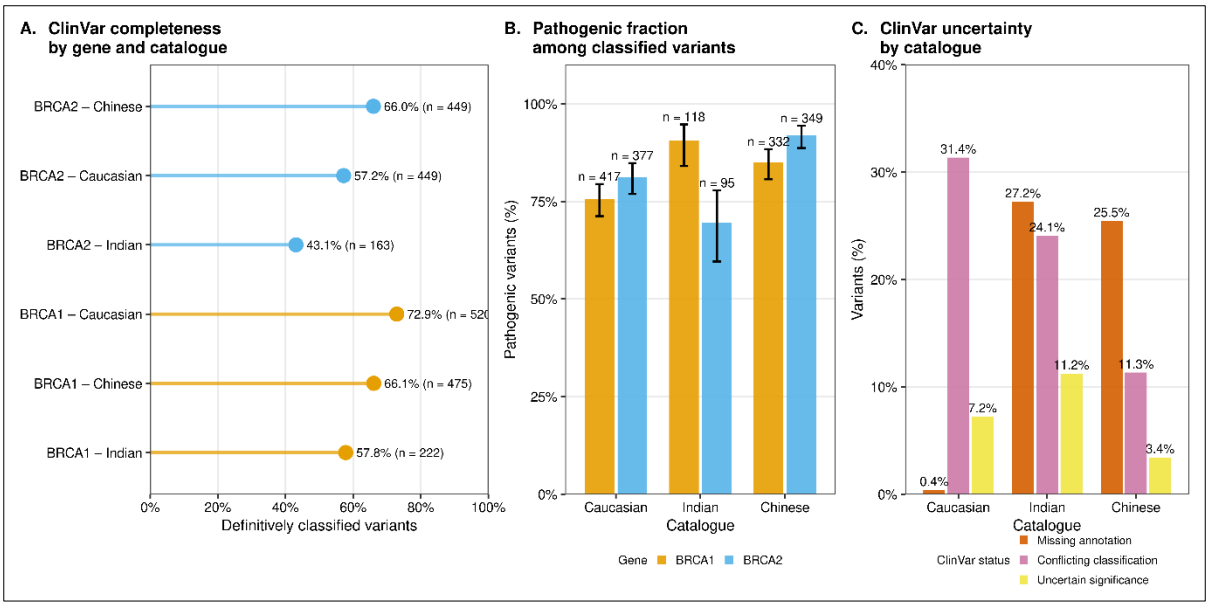


FIGURE S2. **Variant curation, pathogenicity patterns and ascertainment bias.** Panel A shows ClinVar definitive annotation completeness by gene and population, where lollipop plot horizontal lines reflect the trend (BRCA2: blue lines and BRCA1 yellow lines). Panel B shows pathogenic ratios grouped by gene and population, where error bars reflect 95% Wilson confidence intervals for each gene-population combination. Panel C show grouped bar chart with percentage of variants lacking definitive interpretation (divided by subtypes, 'Missing' ClinVar annotation (no submission), 'Conflicting' classifications (discordant expert opinions), and VUS (explicitly uncertain).

Manual variant curation and reasoning: Our systematic validation pipeline using VariantValidator successfully standardized the majority of input variants while identifying and correcting numerous annotation errors. Largely the annotation errors comprised of typographical errors **(**Missing delimiters, spacing errors, incorrect HGVS syntax), transcript mismatches (Variants reported with outdated transcript versions or wrong transcript assignments), assembly ambiguity (genomic coordinates provided without assembly specification) and tough to resolve (Insufficient information or conflicting data).

Detailed error logs and correction rationale for majority of manually curated variants are provided in Supplementary Texts S1-S4. These represent changes from source files to that of the GitHub (data/Phase1_dataCollect)

Text S1: Updating variants, validation failure and mismatch in Verma et al (India)

- In pubmed '30555256', refGene seq was specified without accession version, considering its published year, appropriate accessions were manually added. (NM_007300 ->NM_007300.4 and NM_000059 -> NM_000059.3)
- For variants where Ref to Alt sequence were co-specifed with cDNA change, they were stripped from cDNA to improve detection in variantValidator. E.g. C>CAc.956_957insA -> c.956_957insA and GA/GC.3860_DELA -> c.3860_delA.

1. BRCA1	c.10082A>C	NM_007294.3	GRCh38
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh38/NM_007294.3:c.10082A>C/NM_007294.3?content-type=application/json		

	The given coordinate is outside the bounds of the reference sequence. (NM_007294.3)			
	LovdSyntaxcheckValid: NM_007294.3:c.10082A>C is syntactically correct			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.10082A>C			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
CORRECTION:	[Yes][actually BRCA2, outcome matches with protein change (supp), NP_000050.3:p.(Gln3361Pro)]			
2. BRCA1	c.5278-1G>T	NM_007300.4	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007300.4:c.5278-1G>T/NM_007300.4?content-type=application/json			
	NM_007300.4:c.5277G>T: Variant reference (G) does not agree with reference sequence (A)			
	LovdSyntaxcheckInvalid: NM_007300.4:c.5278-1G>T is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007300.4:c.5278-1G>T, probability = 0.1]			
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007300.4:c.5278-1G>T			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
CORRECTION	Use genomic HGVS			
3. BRCA1	c.80+5G>A^{ψ}	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.80+5G>A^{ψ}/NM_007294.3?content-type=application/json			
	VariantSyntaxError: Submitted variant description contains an invalid character(s) [ψ] at position(s) [24]: Please remove this character and re-submit: A useful search function for Unicode characters can be found at https://unicode-search.net/			
	LovdSyntaxcheckInvalid: NM_007294.3:c.80+5G>A^{ψ} is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.80+5G>A^{ψ}, probability = 0.1]			
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.80+5G>A^{ψ}			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
CORRECTION	working without the ^{ψ}			
4. BRCA2	c.316+5G>A^{φ}	NM_000059.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.316+5G>A^{φ}/NM_000059.3?content-type=application/json			
	VariantSyntaxError: Submitted variant description contains an invalid character(s) [φ] at position(s) [25]: Please remove this character and re-submit: A			

	useful search function for Unicode characters can be found at https://unicode-search.net/			
	LovdSyntaxcheckInvalid: NM_000059.3:c.316+5G>A^{φ} is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.316+5G>A^{φ}, probability = 0.1]			
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.316+5G>A^{φ}			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
CORRECTION	working without the ^{ψ}			
5. BRCA1	c.5634_5642del	NM_007294.3	GRCh38	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh38/NM_007294.3:c.5634_5642del/NM_007294.3?content-type=application/json			
	Variant coordinate is out of the bound of CDS region (CDS length : 5592)			
	LovdSyntaxcheckValid: NM_007294.3:c.5634_5642del is syntactically correct			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5634_5642del			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
CORRECTION	Correct seq to NM_007300.3			
6. BRCA1	c.4165_1466del	NM_007294.3	GRCh38	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh38/NM_007294.3:c.4165_1466del/NM_007294.3?content-type=application/json			
	Interval end position 1466 < interval start position 4165			
	LovdSyntaxcheckInvalid: NM_007294.3:c.4165_1466del is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.1466_4165del, probability = 0.9]			
	LovdSyntaxcheckWarning: This variant description contains positions not given in the correct order.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.4165_1466del			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
CORRECTION	correct typo in variant to c.4165_4166			
7. BRCA1	c.4548-G>T	NM_007294.3	GRCh38	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh38/NM_007294.3:c.4548-G>T/NM_007294.3?content-type=application/json			
	HgvsSyntaxError: 4548-G>T: char 5: expected a digit			
	LovdSyntaxcheckInvalid: NM_007294.3:c.4548-G>T is not syntactically correct, see LovdSyntaxcheckSuggestions for details			

	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3, probability = 0.1]				
	LovdSyntaxcheckWarning: We stopped reading past NM_007294.3. We could not interpret :c.4548-G>T.				
	LovdSyntaxcheckError: This input requires a variant description; reference sequence given.				
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.4548-G>T				
	LovdSyntaxcheckLibraryVersion: 0.5.7				
CORRECTION	correct typo in variant, to c.4548-1G>T and update seq to NM_007300.3]				
8. BRCA1	c.4738G>C	NM_007294.3	GRCh38		
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh38/NM_007294.3:c.4738G>C/NM_007294.3?content-type=application/json				
	NM_007294.3:c.4738G>C: Variant reference (G) does not agree with reference sequence (T)				
	LovdSyntaxcheckValid: NM_007294.3:c.4738G>C is syntactically correct				
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.4738G>C				
	LovdSyntaxcheckLibraryVersion: 0.5.7				
CORRECTION	Matches for seq NM_007300.3				
9. BRCA1	c.5137+1G>A	NM_007294.3	GRCh38		
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh38/NM_007294.3:c.5137+1G>A/NM_007294.3?content-type=application/json				
	NM_007294.3:c.5138G>A: Variant reference (G) does not agree with reference sequence (T)				
	LovdSyntaxcheckInvalid: NM_007294.3:c.5137+1G>A is not syntactically correct, see LovdSyntaxcheckSuggestions for details				
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.5137+1G>A, probability = 0.1]				
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.				
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5137+1G>A				
	LovdSyntaxcheckLibraryVersion: 0.5.7				
CORRECTION	Matches for seq NM_007300.3				
10. BRCA1	c.5314C>T	NM_007294.3	GRCh38		
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh38/NM_007294.3:c.5314C>T/NM_007294.3?content-type=application/json				
	NM_007294.3:c.5314C>T: Variant reference (C) does not agree with reference sequence (T)				
	LovdSyntaxcheckValid: NM_007294.3:c.5314C>T is syntactically correct				
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5314C>T				
	LovdSyntaxcheckLibraryVersion: 0.5.7				

CORRECTION	Matches for seq NM_007300.3			
11. BRCA1	c.5369A>G	NM_007294.3	GRCh38	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh38/NM_007294.3:c.5369A>G/NM_007294.3?content-type=application/json			
	NM_007294.3:c.5369A>G: Variant reference (A) does not agree with reference sequence (C)			
	LovdSyntaxcheckValid: NM_007294.3:c.5369A>G is syntactically correct			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5369A>G			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
CORRECTION	Matches for seq NM_007300.3, [Variant matches rs397509257, but resulting HGVS belongs to another seq, "NM_007300.4:c.5369A>G", https://www.ncbi.nlm.nih.gov/snp/rs397509257#variant_details]			
12. BRCA1	c.5429C>T	NM_007294.3	GRCh38	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh38/NM_007294.3:c.5429C>T/NM_007294.3?content-type=application/json			
	NM_007294.3:c.5429C>T: Variant reference (C) does not agree with reference sequence (T)			
	LovdSyntaxcheckValid: NM_007294.3:c.5429C>T is syntactically correct			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5429C>T			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh38/NM_007300.3:c.5429C>T/NM_007300.3?content-type=application/json			
CORRECTION	Matches for seq NM_007300.3			
13. BRCA1	c.5536A>G	NM_007294.3	GRCh38	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh38/NM_007294.3:c.5536A>G/NM_007294.3?content-type=application/json			
	NM_007294.3:c.5536A>G: Variant reference (A) does not agree with reference sequence (C)			
	LovdSyntaxcheckValid: NM_007294.3:c.5536A>G is syntactically correct			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5536A>G			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
CORRECTION	Known variant in BRCA2 (so change gene name, protein description also matches), https://www.ncbi.nlm.nih.gov/clinvar/variation/141804/, with HGVS: NM_001407581.1:c.5536A>G, (OR NM_000059.3)			
14. BRCA1	c.5572T>C	NM_007294.3	GRCh38	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh38/NM_007294.3:c.5572T>C/NM_007294.3?content-type=application/json			
	NM_007294.3:c.5572T>C: Variant reference (T) does not agree with reference sequence (A)			
	LovdSyntaxcheckValid: NM_007294.3:c.5572T>C is syntactically correct			

	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5572T>C				
	LovdSyntaxcheckLibraryVersion: 0.5.7				
CORRECTION	Change seq to NM_007300.4				
15. BRCA1	c.5620T>A	NM_007294.3	GRCh38 [Yes*][Another seq, NM_007300.4, this also matches consequence p.Tyr1874Asn]		
	#PMCID: PMC12008341 PMID: 39733403				
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh38/NM_007294.3:c.5620T>A/NM_007294.3?content-type=application/json				
	NM_007294.3:c.5620T>A: Variant reference (T) does not agree with reference sequence (C)				
	LovdSyntaxcheckValid: NM_007294.3:c.5620T>A is syntactically correct				
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5620T>A				
	LovdSyntaxcheckLibraryVersion: 0.5.7				
CORRECTION	Change seq to NM_007300.4				
16. BRCA1	c.(?_-232)_(134+1_135-1)del	NM_007294.3	GRCh37		
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.(?_-232)_(134+1_135-1)del/NM_007294.3?content-type=application/json				
CORRECTION	Can’t be corrected				
17. BRCA1	c.(?_-232)_(80+1_81-1)del	NM_007294.3	GRCh37		
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.(?_-232)_(80+1_81-1)del/NM_007294.3?content-type=application/json				
CORRECTION	Can’t be corrected				
18. BRCA1	c.(5340+1_5341-1)_(*1_?)del	NM_007294.3	GRCh38		
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh38/NM_007294.3:c.(5340+1_5341-1)_(*1_?)del/NM_007294.3?content-type=application/json				
CORRECTION	Can’t be corrected				

19. BRCA1	c.(?_-232)_(5277+1_5278-1)del	NM_007294.4	GRCh38		
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh38/NM_007294.4:c.(?_-232)_(5277+1_5278-1)del/NM_007294.4?content-type=application/json				
CORRECTION	Can't be corrected				
20. BRCA1	Splicesite_3(G>T)	NM_007300.4	GRCh37 [*Pending]		
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007300.4:Splicesite_3(G>T)/NM_007300.4?content-type=application/json				
	HgvsSyntaxError: Splicesite_3(G>T): char 0: expected one of (, *, or a digit				
	LovdSyntaxcheckInvalid: NM_007300.4:Splicesite_3(G>T) is not syntactically correct, see LovdSyntaxcheckSuggestions for details				
	LovdSyntaxcheckSuggestions: [suggestion = NM_007300.4, probability = 0.1]				
	LovdSyntaxcheckWarning: We stopped reading past NM_007300.4. We could not interpret :Splicesite_3(G>T).				
	LovdSyntaxcheckError: This input requires a variant description; reference sequence given.				
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007300.4:Splicesite_3(G>T)				
	LovdSyntaxcheckLibraryVersion: 0.5.7				
CORRECTION	Use genomic convention with mrna seq (https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NC_000017.10:g.41226347C%3EA/NM_007294.3?content-type=application/json)				
21. BRCA1	c.5644_7151del	NM_007300.4	GRCh37 [No] [*Pending]		
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007300.4:c.5644_7151del/NM_007300.4?content-type=application/json				
	The given coordinate is outside the bounds of the reference sequence. (NM_007300.4)				
	LovdSyntaxcheckValid: NM_007300.4:c.5644_7151del is syntactically correct				
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007300.4:c.5644_7151del				
	LovdSyntaxcheckLibraryVersion: 0.5.7				
CORRECTION	Difficult to correct as nontrivial to remove entire exon from cDNA convention, as if force on cDNA, can't remove region beyond the translation end site from cDNA HGVS.				

Text S2: Validation failure and mismatch in Bhaskaran et al 2021 (India)

The initial supplementary file contains 260 BRCA1 and 220 BRCA2 variants. Our set (Supplementary File: "P_Mutants.xlsx", Spreadsheet: "Bhaskaran et al PMID 32963034") contains 256 BRCA1 variants where following 5 variants are updated.

Present in source	Status in our set	Reason
c.2458G>A	Absent	Validation error, ref sequence mismatch

c.301-41T>C	Replaced with c.260T>C	After reading, from similar genomic hgvs, we sought to replace "https://www.ncbi.nlm.nih.gov/clinvar/variation/865611/?oq=%22g.41256926A%3EG%22[ALL+FIELDS]&m=NM_007294.4(BRCA1):c.260T%3EC%20(p.Leu87Ser)%3Fterm=g.41256926A%3EG"
c.3548G>A	Absent	Validation error, ref sequence mismatch
c.4915C>T	Absent	Validation error, ref sequence mismatch
c.5465G>A	Absent	Validation error, ref sequence mismatch

Description of other failed and mismatched variants

0. BRCA2	c.1114C>A	NM_000059.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.1114C>A/NM_000059.3?content-type=application/json		
	NM_000059.3:c.1114C>A: Variant reference (C) does not agree with reference sequence (A)		
	LovdSyntaxcheckValid: NM_000059.3:c.1114C>A is syntactically correct		
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.1114C>A		
	LovdSyntaxcheckLibraryVersion: 0.5.7		
1. BRCA2	c.2621_2627delAACTGTC	NM_000059.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.2621_2627delAACTGTC/NM_000059.3?content-type=application/json		
	VariantSyntaxError: Removing redundant reference bases from variant description		
	NM_000059.3:c.2621_2627del: Variant reference (AACTGTC) does not agree with reference sequence (CTGTCAA)		
	LovdSyntaxcheckInvalid: NM_000059.3:c.2621_2627del is not syntactically correct, see LovdSyntaxcheckSuggestions for details		
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.2621_2627del, probability = 1]		
	LovdSyntaxcheckWarning: The deleted sequence is redundant and should be removed.		
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.2621_2627del		
	LovdSyntaxcheckLibraryVersion: 0.5.7		
2. BRCA2	c.4698insGACC	NM_000059.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.4698insGACC/NM_000059.3?content-type=application/json		
	Insertion length must be 1 e.g. 4698_4699insGACC		
	LovdSyntaxcheckInvalid: NM_000059.3:c.4698insGACC is not syntactically correct, see LovdSyntaxcheckSuggestions for details		
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.4697_4698insGACC, probability = 0.7]		
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.4698_4699insGACC, probability = 0.3]		
	LovdSyntaxcheckWarning: An insertion must be provided with the two positions between which the insertion has taken place.		
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.4698insGACC		

	LovdSyntaxcheckLibraryVersion: 0.5.7		
3. BRCA2	c.6148delAA	NM_000059.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.6148delAA/NM_000059.3?content-type=application/json		
	VariantSyntaxError: Removing redundant reference bases from variant description		
	Length implied by coordinates must equal sequence deletion length		
	LovdSyntaxcheckInvalid: NM_000059.3:c.6148delAA is not syntactically correct, see LovdSyntaxcheckSuggestions for details		
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.6148_6149del, probability = 0.1]		
	LovdSyntaxcheckWarning: The variants positions have been corrected.		
	LovdSyntaxcheckError: The variants positions indicate a sequence thats shorter than the given deleted sequence. Please adjust either the variants positions or the given deleted sequence.		
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.6148delAA		
	LovdSyntaxcheckLibraryVersion: 0.5.7		
4. BRCA2	c.6214_6218delCTTAA	NM_000059.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.6214_6218delCTTAA/NM_000059.3?content-type=application/json		
	VariantSyntaxError: Removing redundant reference bases from variant description		
	NM_000059.3:c.6214_6218del: Variant reference (CTTAA) does not agree with reference sequence (TCCTT)		
	LovdSyntaxcheckInvalid: NM_000059.3:c.6214_6218del is not syntactically correct, see LovdSyntaxcheckSuggestions for details		
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.6214_6218del, probability = 1]		
	LovdSyntaxcheckWarning: The deleted sequence is redundant and should be removed.		
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.6214_6218del		
	LovdSyntaxcheckLibraryVersion: 0.5.7		
5. BRCA2	c.6841+79delTTAA	NM_000059.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.6841+79delTTAA/NM_000059.3?content-type=application/json		
	VariantSyntaxError: Removing redundant reference bases from variant description		
	Required information for NM_000059.3 is missing from the Universal Transcript Archive		
	Query gene2transcripts with search term NM_000059 for available transcripts		
	LovdSyntaxcheckInvalid: NM_000059.3:c.6841+79del is not syntactically correct, see LovdSyntaxcheckSuggestions for details		
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.6841+79_6841+82del, probability = 0.1]		
	LovdSyntaxcheckWarning: The variants positions have been corrected.		
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.		
	LovdSyntaxcheckError: The variants positions indicate a sequence thats shorter than the given deleted sequence. Please adjust either the variants positions or the given deleted sequence.		

	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.6841+79del		
	LovdSyntaxcheckLibraryVersion: 0.5.7		
6. BRCA2	c.7804-12T>C	NM_000059.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.7804-12T>C/NM_000059.3?content-type=application/json		
	NM_000059.3:c.7792T>C: Variant reference (T) does not agree with reference sequence (G)		
	LovdSyntaxcheckInvalid: NM_000059.3:c.7804-12T>C is not syntactically correct, see LovdSyntaxcheckSuggestions for details		
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.7804-12T>C, probability = 0.1]		
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.		
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.7804-12T>C		
	LovdSyntaxcheckLibraryVersion: 0.5.7		
7. BRCA2	c.7807-14T>C	NM_000059.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.7807-14T>C/NM_000059.3?content-type=application/json		
	ExonBoundaryError: Position c.7807-14 does not correspond with an exon boundary for transcript NM_000059.3		
	LovdSyntaxcheckInvalid: NM_000059.3:c.7807-14T>C is not syntactically correct, see LovdSyntaxcheckSuggestions for details		
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.7807-14T>C, probability = 0.1]		
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.		
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.7807-14T>C		
	LovdSyntaxcheckLibraryVersion: 0.5.7		
8. BRCA2	c.9501-17T>C	NM_000059.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.9501-17T>C/NM_000059.3?content-type=application/json		
	NM_000059.3:c.9484T>C: Variant reference (T) does not agree with reference sequence (A)		
	LovdSyntaxcheckInvalid: NM_000059.3:c.9501-17T>C is not syntactically correct, see LovdSyntaxcheckSuggestions for details		
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.9501-17T>C, probability = 0.1]		
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.		
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.9501-17T>C		
	LovdSyntaxcheckLibraryVersion: 0.5.7		
9. BRCA2	c.9501+58InsG	NM_000059.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.9501+58InsG/NM_000059.3?content-type=application/json		
	Edit type Ins should be in the lower case, i.e. ins		
	Insertion length must be 1 e.g. 9501+58_9501+59insG		

	LovdSyntaxcheckInvalid: NM_000059.3:c.9501+58InsG is not syntactically correct, see LovdSyntaxcheckSuggestions for details		
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.9501+57_9501+58insG, probability = 0.07]		
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.9501+58_9501+59insG, probability = 0.03]		
	LovdSyntaxcheckWarning: An insertion must be provided with the two positions between which the insertion has taken place.		
	LovdSyntaxcheckWarning: This is not a valid HGVS description, due to characters being in the wrong case.		
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.		
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.9501+58InsG		
	LovdSyntaxcheckLibraryVersion: 0.5.7		
10. BRCA1	c.176delCA	NM_007294.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.176delCA/NM_007294.3?content-type=application/json		
	VariantSyntaxError: Removing redundant reference bases from variant description		
	Length implied by coordinates must equal sequence deletion length		
	LovdSyntaxcheckInvalid: NM_007294.3:c.176delCA is not syntactically correct, see LovdSyntaxcheckSuggestions for details		
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.176_177del, probability = 0.1]		
	LovdSyntaxcheckWarning: The variants positions have been corrected.		
	LovdSyntaxcheckError: The variants positions indicate a sequence thats shorter than the given deleted sequence. Please adjust either the variants positions or the given deleted sequence.		
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.176delCA		
	LovdSyntaxcheckLibraryVersion: 0.5.7		
11. BRCA1	c.4156_5160delCTCTC	NM_007294.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.4156_5160delCTCTC/NM_007294.3?content-type=application/json		
	VariantSyntaxError: Removing redundant reference bases from variant description		
	Length implied by coordinates must equal sequence deletion length		
	LovdSyntaxcheckInvalid: NM_007294.3:c.4156_5160del is not syntactically correct, see LovdSyntaxcheckSuggestions for details		
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.(4156_5160)del, probability = 0.07]		
	LovdSyntaxcheckError: The variants positions indicate a sequence thats longer than the given deleted sequence. Please adjust either the variants positions or the given deleted sequence.		
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.4156_5160del		
	LovdSyntaxcheckLibraryVersion: 0.5.7		
12. BRCA1	c.5263insC dupder	NM_007294.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.5263insC dupder/NM_007294.3?content-type=application/json		

	VariantSyntaxError: Whitespace removed from variant description NM_007294.3:c.5263insC dupder
	HgvsSyntaxError: 5263insCdupder: char 10: expected EOF
	LovdSyntaxcheckInvalid: NM_007294.3:c.5263insC dupder is not syntactically correct, see LovdSyntaxcheckSuggestions for details
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.5262_5263insC, probability = 0.7]
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.5263_5264insC, probability = 0.3]
	LovdSyntaxcheckWarning: An insertion must be provided with the two positions between which the insertion has taken place.
	LovdSyntaxcheckWarning: We stopped reading past NM_007294.3:c.5263insC. We could not interpret dupder.
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5263insC dupder
	LovdSyntaxcheckLibraryVersion: 0.5.7

Text S3: Failures Caucasian

0. BRCA1	NC_000017.10	c.464_465delA	NM_007294.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.464_465delA/NM_007294.3?content-type=application/json			
	VariantSyntaxError: Removing redundant reference bases from variant description			
	Length implied by coordinates must equal sequence deletion length			
	LovdSyntaxcheckInvalid: NM_007294.3:c.464_465delA is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.(464_465)delA, probability = 0.07]			
	LovdSyntaxcheckError: The variants positions indicate a sequence thats longer than the given deleted sequence. Please adjust either the variants positions or the given deleted sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.464_465delA			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
1. BRCA1	NC_000017.10	c.5333-4_5333-2del3	NM_007294.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.5333-4_5333-2del3/NM_007294.3?content-type=application/json			
	Cannot validate sequence of an intronic variant (NM_007294.3:c.5333-4_5333-2del)			
	Trailing digits are not permitted in HGVS variant descriptions			
	Refer to http://varnomen.hgvs.org/recommendations/DNA/variant/			
	LovdSyntaxcheckInvalid: NM_007294.3:c.5333-4_5333-2del3 is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.5333-4_5333-2del, probability = 0.1]			
	LovdSyntaxcheckWarning: The part after del does not follow HGVS guidelines.			
	LovdSyntaxcheckWarning: The deleted sequence is redundant and should be removed.			

	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5333-4_5333-2del3			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
2. BRCA1	NC_000017.10	c.876+30G>A	NM_007294.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.876+30G>A/NM_007294.3?content-type=application/json			
	NM_007294.3:c.906G>A: Variant reference (G) does not agree with reference sequence (T)			
	LovdSyntaxcheckInvalid: NM_007294.3:c.876+30G>A is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.876+30G>A, probability = 0.1]			
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.876+30G>A			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
3. BRCA1	NC_000017.10	c.2488_2489dupAAGTATCCAT	NM_007294.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.2488_2489dupAAGTATCCAT/NM_007294.3?content-type=application/json			
	VariantSyntaxError: Removing redundant reference bases from variant description			
	NM_007294.3:c.2488_2489dupAAGTATCCAT: Variant reference (AAGTATCCAT) does not agree with reference sequence (AA)			
	LovdSyntaxcheckInvalid: NM_007294.3:c.2488_2489dupAAGTATCCAT is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.2488_2489dupAAGTATCCAT, probability = 0.1]			
	LovdSyntaxcheckError: The variants positions indicate a sequence thats shorter than the given duplicated sequence. Please adjust either the variants positions or the given duplicated sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.2488_2489dupAAGTATCCAT			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
4. BRCA1	NC_000017.10	c.952_1015delCATAACAGATGGGCTGGAAGTAAGGAAACATGTAATGATAGGCGGACTCCCAGCACA	NM_007294.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.952_1015delCATAACAGATGGGCTGGAAGTAAGGAAACATGTAATGATAGGCGGACTCCCAGCACA/NM_007294.3?content-type=application/json			
	VariantSyntaxError: Removing redundant reference bases from variant description			
	Length implied by coordinates must equal sequence deletion length			
	LovdSyntaxcheckInvalid: NM_007294.3:c.952_1015del is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.(952_1015)del, probability = 0.07]			

	LovdSyntaxcheckError: The variants positions indicate a sequence thats longer than the given deleted sequence. Please adjust either the variants positions or the given deleted sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.952_1015del			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
5. BRCA1	NC_000017.10	c.4065_4068delTCA	NM_007294.3	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.4065_4068delTCA/NM_007294.3?content-type=application/json			
	VariantSyntaxError: Removing redundant reference bases from variant description			
	Length implied by coordinates must equal sequence deletion length			
	LovdSyntaxcheckInvalid: NM_007294.3:c.4065_4068delTCA is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.(4065_4068)delTCA, probability = 0.07]			
	LovdSyntaxcheckError: The variants positions indicate a sequence thats longer than the given deleted sequence. Please adjust either the variants positions or the given deleted sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.4065_4068delTCA			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
6. BRCA2	NC_000013.10	c.1796_1800delTTTAT	NM_000059.4	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.4:c.1796_1800delTTTAT/NM_000059.4?content-type=application/json			
	VariantSyntaxError: Removing redundant reference bases from variant description			
	NM_000059.4:c.1796_1800del: Variant reference (TTTAT) does not agree with reference sequence (CTTAT)			
	LovdSyntaxcheckInvalid: NM_000059.4:c.1796_1800del is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.4:c.1796_1800del, probability = 1]			
	LovdSyntaxcheckWarning: The deleted sequence is redundant and should be removed.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.4:c.1796_1800del			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
7. BRCA2	NC_000013.10	c.6754_6755dupT	NM_000059.4	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.4:c.6754_6755dupT/NM_000059.4?content-type=application/json			
	VariantSyntaxError: Removing redundant reference bases from variant description			
	NM_000059.4:c.6754_6755dupT: Variant reference (T) does not agree with reference sequence (TC)			
	LovdSyntaxcheckInvalid: NM_000059.4:c.6754_6755dupT is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.4:c.(6754_6755)dupT, probability = 0.07]			

	LovdSyntaxcheckError: The variants positions indicate a sequence thats longer than the given duplicated sequence. Please adjust either the variants positions or the given duplicated sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.4:c.6754_6755dupT			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
8. BRCA2	NC_000013.10	c.7397C>T	NM_000059.4	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.4:c.7397C>T/NM_000059.4?content-type=application/json			
	NM_000059.4:c.7397C>T: Variant reference (C) does not agree with reference sequence (T)			
	LovdSyntaxcheckValid: NM_000059.4:c.7397C>T is syntactically correct			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.4:c.7397C>T			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
9. BRCA2	NC_000013.10	c.8169_8170dupTGGG	NM_000059.4	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.4:c.8169_8170dupTGGG/NM_000059.4?content-type=application/json			
	VariantSyntaxError: Removing redundant reference bases from variant description			
	NM_000059.4:c.8169_8170dupTGGG: Variant reference (TGGG) does not agree with reference sequence (TG)			
	LovdSyntaxcheckInvalid: NM_000059.4:c.8169_8170dupTGGG is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.4:c.8169_8170dupTGGG, probability = 0.1]			
	LovdSyntaxcheckError: The variants positions indicate a sequence thats shorter than the given duplicated sequence. Please adjust either the variants positions or the given duplicated sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.4:c.8169_8170dupTGGG			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
10. BRCA2	NC_000013.10	c.426-12_426-8del5	NM_000059.4	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.4:c.426-12_426-8del5/NM_000059.4?content-type=application/json			
	Cannot validate sequence of an intronic variant (NM_000059.4:c.426-12_426-8del)			
	Trailing digits are not permitted in HGVS variant descriptions			
	Refer to http://varnomen.hgvs.org/recommendations/DNA/variant/			
	LovdSyntaxcheckInvalid: NM_000059.4:c.426-12_426-8del5 is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.4:c.426-12_426-8del, probability = 0.1]			
	LovdSyntaxcheckWarning: The part after del does not follow HGVS guidelines.			
	LovdSyntaxcheckWarning: The deleted sequence is redundant and should be removed.			
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.			

	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.4:c.426-12_426-8del5			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
11. BRCA2	NC_000013.10	c.7008-20_7008-17del4	NM_000059.4	GRCh37
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.4:c.7008-20_7008-17del4/NM_000059.4?content-type=application/json			
	Cannot validate sequence of an intronic variant (NM_000059.4:c.7008-20_7008-17del)			
	Trailing digits are not permitted in HGVS variant descriptions			
	Refer to http://varnomen.hgvs.org/recommendations/DNA/variant/			
	LovdSyntaxcheckInvalid: NM_000059.4:c.7008-20_7008-17del4 is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.4:c.7008-20_7008-17del, probability = 0.1]			
	LovdSyntaxcheckWarning: The part after del does not follow HGVS guidelines.			
	LovdSyntaxcheckWarning: The deleted sequence is redundant and should be removed.			
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.4:c.7008-20_7008-17del4			
	LovdSyntaxcheckLibraryVersion: 0.5.7			

Text S4: Failures China

0. BRCA1	c.66insA	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.66insA/NM_007294.3?content-type=application/json			
	Insertion length must be 1 e.g. 66_67insA			
	LovdSyntaxcheckInvalid: NM_007294.3:c.66insA is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.65_66insA, probability = 0.7]			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.66_67insA, probability = 0.3]			
	LovdSyntaxcheckWarning: An insertion must be provided with the two positions between which the insertion has taken place.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.66insA			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
1. BRCA1	c.442-15del10	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.442-15del10/NM_007294.3?content-type=application/json			
	Length implied by coordinates must equal sequence deletion length			
	Trailing digits are not permitted in HGVS variant descriptions			
	Refer to http://varnomen.hgvs.org/recommendations/DNA/variant/			
	LovdSyntaxcheckInvalid: NM_007294.3:c.442-15del10 is not syntactically correct, see LovdSyntaxcheckSuggestions for details			

	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.442-15delN[10], probability = 0.1]			
	LovdSyntaxcheckWarning: The part after del does not follow HGVS guidelines.			
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.			
	LovdSyntaxcheckError: The variants positions indicate a sequence thats shorter than the given deleted sequence. Please adjust either the variants positions or the given deleted sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.442-15del10			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
2. BRCA1	c.470_477del8/c.470_477delCTAACCTT	NM_007294.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.470_477del8/c.470_477delCTAACCTT/NM_007294.3?content-type=application/json			
3. BRCA1	c.4548-2A>C	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.4548-2A>C/NM_007294.3?content-type=application/json			
	ExonBoundaryError: Position c.4548-2 does not correspond with an exon boundary for transcript NM_007294.3			
	LovdSyntaxcheckInvalid: NM_007294.3:c.4548-2A>C is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.4548-2A>C, probability = 0.1]			
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.4548-2A>C			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
4. BRCA1	c.4636C>T/c.4636G>T	NM_007294.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.4636C%3ET/c.4636G%3ET/NM_007294.3?content-type=application/json			
5. BRCA1	c.4956A>G	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.4956A>G/NM_007294.3?content-type=application/json			
	NM_007294.3:c.4956A>G: Variant reference (A) does not agree with reference sequence (G)			
	LovdSyntaxcheckValid: NM_007294.3:c.4956A>G is syntactically correct			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.4956A>G			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
6. BRCA1	c.5113T>A/c.5114T>A	NM_007294.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.5113T%3EA/c.5114T%3EA/NM_007294.3?content-type=application/json			

7. BRCA1	c.5173insA	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.5173insA/NM_007294.3?content-type=application/json			
	Insertion length must be 1 e.g. 5173_5174insA			
	LovdSyntaxcheckInvalid: NM_007294.3:c.5173insA is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.5172_5173insA, probability = 0.7]			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.5173_5174insA, probability = 0.3]			
	LovdSyntaxcheckWarning: An insertion must be provided with the two positions between which the insertion has taken place.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5173insA			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
8. BRCA1	c.5215+268G>A	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.5215+268G>A/NM_007294.3?content-type=application/json			
	ExonBoundaryError: Position c.5215+268 does not correspond with an exon boundary for transcript NM_007294.3			
	LovdSyntaxcheckInvalid: NM_007294.3:c.5215+268G>A is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.5215+268G>A, probability = 0.1]			
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5215+268G>A			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
9. BRCA1	c.5127delT/c.5127delA	NM_007294.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.5127delT/c.5127delA/NM_007294.3?content-type=application/json			
10. BRCA1	c.5093_5096delCTAA	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.5093_5096delCTAA/NM_007294.3?content-type=application/json			
	VariantSyntaxError: Removing redundant reference bases from variant description			
	NM_007294.3:c.5093_5096del: Variant reference (CTAA) does not agree with reference sequence (AACG)			
	LovdSyntaxcheckInvalid: NM_007294.3:c.5093_5096del is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.5093_5096del, probability = 1]			
	LovdSyntaxcheckWarning: The deleted sequence is redundant and should be removed.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5093_5096del			
	LovdSyntaxcheckLibraryVersion: 0.5.7			

11. BRCA1	c.5194−2A>G	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.5194−2A>G/NM_007294.3?content-type=application/json			
	VariantSyntaxError: Submitted variant description contains an invalid character(s) [−] at position(s) [19]: Please remove this character and re-submit: A useful search function for Unicode characters can be found at https://unicode-search.net/			
	LovdSyntaxcheckInvalid: NM_007294.3:c.5194−2A>G is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.5194-2A>G, probability = 0.1]			
	LovdSyntaxcheckWarning: Invalid character − found in variant position; only regular hyphens are allowed to be used in the HGVS nomenclature.			
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5194−2A>G			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
12. BRCA1	c.5278-68insA	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.5278-68insA/NM_007294.3?content-type=application/json			
	Insertion length must be 1 e.g. 5278-69_5278-68insA			
	LovdSyntaxcheckInvalid: NM_007294.3:c.5278-68insA is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.5278-69_5278-68insA, probability = 0.07]			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.5278-68_5278-67insA, probability = 0.03]			
	LovdSyntaxcheckWarning: An insertion must be provided with the two positions between which the insertion has taken place.			
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5278-68insA			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
13. BRCA1	c.5396−2A>G	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.5396−2A>G/NM_007294.3?content-type=application/json			
	VariantSyntaxError: Submitted variant description contains an invalid character(s) [−] at position(s) [19]: Please remove this character and re-submit: A useful search function for Unicode characters can be found at https://unicode-search.net/			
	LovdSyntaxcheckInvalid: NM_007294.3:c.5396−2A>G is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.5396-2A>G, probability = 0.1]			
	LovdSyntaxcheckWarning: Invalid character − found in variant position; only regular hyphens are allowed to be used in the HGVS nomenclature.			
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5396−2A>G			

	LovdSyntaxcheckLibraryVersion: 0.5.7			
14. BRCA1	c.5533_5540delATTGGGCA/delTACCAGTG	NM_007294.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.5533_5540delATTGGGCA/delTACCAGTG/NM_007294.3?content-type=application/json			
15. BRCA1	c.5521A>C	NM_007294.3	GRCh37	
	Exception:429 Client Error: TOO MANY REQUESTS for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.5521A%3EC/NM_007294.3?content-type=application/json			
16. BRCA1	c.5587-1del81	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.5587-1del81/NM_007294.3?content-type=application/json			
	Length implied by coordinates must equal sequence deletion length			
	Trailing digits are not permitted in HGVS variant descriptions			
	Refer to http://varnomen.hgvs.org/recommendations/DNA/variant/			
	LovdSyntaxcheckInvalid: NM_007294.3:c.5587-1del81 is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.5587-1delN[81], probability = 0.1]			
	LovdSyntaxcheckWarning: The part after del does not follow HGVS guidelines.			
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.			
	LovdSyntaxcheckError: The variants positions indicate a sequence thats shorter than the given deleted sequence. Please adjust either the variants positions or the given deleted sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5587-1del81			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
17. BRCA1	c.5592*31G>A	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.5592*31G>A/NM_007294.3?content-type=application/json			
	HgvsSyntaxError: 5592*31G>A: char 4: expected one of =, _, con, copy, del, dup, ins, inv, or a digit			
	LovdSyntaxcheckInvalid: NM_007294.3:c.5592*31G>A is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3, probability = 0.1]			
	LovdSyntaxcheckWarning: We stopped reading past NM_007294.3. We could not interpret :c.5592*31G>A.			
	LovdSyntaxcheckError: This input requires a variant description; reference sequence given.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.5592*31G>A			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
18. BRCA1	c.1352_1364del13/c.1352_1364delCAGTAGAGAGTAA	NM_007294.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_00729			

	4.3:c.1352_1364del13/c.1352_1364delCAGTAGAGAGTAA/NM_007294.3?content-type=application/json			
19. BRCA1	c.2061dellG	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.2061dellG/NM_007294.3?content-type=application/json			
	HgvsSyntaxError: 2061dellG: char 8: expected EOF			
	LovdSyntaxcheckInvalid: NM_007294.3:c.2061dellG is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.2061delLG, probability = 0.1]			
	LovdSyntaxcheckWarning: This is not a valid HGVS description, due to characters being in the wrong case.			
	LovdSyntaxcheckError: This variant description contains one or more invalid nucleotides: L.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.2061dellG			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
20. BRCA1	c.3076_3077ins76	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.3076_3077ins76/NM_007294.3?content-type=application/json			
	The length of the variant is not formatted following the HGVS guidelines. Please rewrite e.g. 10 to N[10](where N is an unknown nucleotide)			
	LovdSyntaxcheckInvalid: NM_007294.3:c.3076_3077ins76 is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.3076_3077insN[76], probability = 1]			
	LovdSyntaxcheckWarning: The part after ins does not follow HGVS guidelines.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.3076_3077ins76			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
21. BRCA1	c.3180insA	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.3180insA/NM_007294.3?content-type=application/json			
	Insertion length must be 1 e.g. 3180_3181insA			
	LovdSyntaxcheckInvalid: NM_007294.3:c.3180insA is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.3179_3180insA, probability = 0.7]			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.3180_3181insA, probability = 0.3]			
	LovdSyntaxcheckWarning: An insertion must be provided with the two positions between which the insertion has taken place.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.3180insA			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
22. BRCA1	c.3288_3289delTT	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.3288_3289delTT/NM_007294.3?content-type=application/json			

	VariantSyntaxError: Removing redundant reference bases from variant description			
	NM_007294.3:c.3288_3289delTT: Variant reference (TT) does not agree with reference sequence (AA)			
	LovdSyntaxcheckInvalid: NM_007294.3:c.3288_3289delTT is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.3288_3289del, probability = 1]			
	LovdSyntaxcheckWarning: The deleted sequence is redundant and should be removed.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.3288_3289delTT			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
23. BRCA1	c.3329insA	NM_007294.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.3329insA/NM_007294.3?content-type=application/json			
	Insertion length must be 1 e.g. 3329_3330insA			
	LovdSyntaxcheckInvalid: NM_007294.3:c.3329insA is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.3328_3329insA, probability = 0.7]			
	LovdSyntaxcheckSuggestions: [suggestion = NM_007294.3:c.3329_3330insA, probability = 0.3]			
	LovdSyntaxcheckWarning: An insertion must be provided with the two positions between which the insertion has taken place.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_007294.3:c.3329insA			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
24. BRCA1	c.3780_3781delAG/c.3780_3781delAT	NM_007294.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_007294.3:c.3780_3781delAG/c.3780_3781delAT/NM_007294.3?content-type=application/json			
25. BRCA2	c.1-40delGA	NM_000059.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.1-40delGA/NM_000059.3?content-type=application/json			
	VariantSyntaxError: Removing redundant reference bases from variant description			
	Required information for NM_000059.3 is missing from the Universal Transcript Archive			
	Query gene2transcripts with search term NM_000059 for available transcripts			
	LovdSyntaxcheckInvalid: NM_000059.3:c.1-40delGA is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.1-40_1-39del, probability = 0.1]			
	LovdSyntaxcheckWarning: The variants positions have been corrected.			
	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.			
	LovdSyntaxcheckError: The variants positions indicate a sequence thats shorter than the given deleted sequence. Please adjust either the variants positions or the given deleted sequence.			

	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.1-40delGA			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
26. BRCA2	c.260_261delCT	NM_000059.3	GRCh37	
	Exception:HTTPSConnectionPool(host='rest.variantvalidator.org', port=443): Max retries exceeded with url: /VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.260_261delCT/NM_000059.3?content-type=application/json (Caused by NewConnectionError('<urllib3.connection.HTTPSConnection object at 0x7f09c2d6ed50>: Failed to establish a new connection: [Errno 111] Connection refused'))			
27. BRCA2	c.262_263delCT	NM_000059.3	GRCh37	
	Exception:HTTPSConnectionPool(host='rest.variantvalidator.org', port=443): Max retries exceeded with url: /VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.262_263delCT/NM_000059.3?content-type=application/json (Caused by NewConnectionError('<urllib3.connection.HTTPSConnection object at 0x7f09c2d6f610>: Failed to establish a new connection: [Errno 111] Connection refused'))			
28. BRCA2	c.915_918delAGAT	NM_000059.3	GRCh37	
	Exception:HTTPSConnectionPool(host='rest.variantvalidator.org', port=443): Max retries exceeded with url: /VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.915_918delAGAT/NM_000059.3?content-type=application/json (Caused by NewConnectionError('<urllib3.connection.HTTPSConnection object at 0x7f09c2d6f110>: Failed to establish a new connection: [Errno 111] Connection refused'))			
29. BRCA2	c.993_994dupA	NM_000059.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.993_994dupA/NM_000059.3?content-type=application/json			
	VariantSyntaxError: Removing redundant reference bases from variant description			
	NM_000059.3:c.993_994dupA: Variant reference (A) does not agree with reference sequence (AA)			
	LovdSyntaxcheckInvalid: NM_000059.3:c.993_994dupA is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.(993_994)dupA, probability = 0.07]			
	LovdSyntaxcheckError: The variants positions indicate a sequence thats longer than the given duplicated sequence. Please adjust either the variants positions or the given duplicated sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.993_994dupA			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
30. BRCA2	c.1036delAA/c.1036_1037delAA	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.1036delAA/c.1036_1037delAA/NM_000059.3?content-type=application/json			

31. BRCA2	c.1759_1763delAAAAC/c.1759_1763delACAAA	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.1759_1763delAAAAC/c.1759_1763delACAAA/NM_000059.3?content-type=application/json			
32. BRCA2	c.1794_1798delATTTT/c.1794_1798delATCTT	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.1794_1798delATTTT/c.1794_1798delATCTT/NM_000059.3?content-type=application/json			
33. BRCA2	c.1796_1800delATCTT/c.1796_1800delCTTAT	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.1796_1800delATCTT/c.1796_1800delCTTAT/NM_000059.3?content-type=application/json			
34. BRCA2	c.1847_1848delTG/c.1847_1848delGT	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.1847_1848delTG/c.1847_1848delGT/NM_000059.3?content-type=application/json			
35. BRCA2	c.2017_2023delAAATGAA/c.2017_2023delAATGAAA	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.2017_2023delAAATGAA/c.2017_2023delAATGAAA/NM_000059.3?content-type=application/json			
36. BRCA2	c.8400_8401del4ins5	NM_000059.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.8400_8401del4ins5/NM_000059.3?content-type=application/json			
	The length of the variant is not formatted following the HGVS guidelines. Please rewrite e.g. 10 to N[10](where N is an unknown nucleotide)			
	LovdSyntaxcheckInvalid: NM_000059.3:c.8400_8401del4ins5 is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.8400_8401delN[4]insN[5], probability = 0.1]			
	LovdSyntaxcheckWarning: The part after ins does not follow HGVS guidelines.			
	LovdSyntaxcheckError: The variants positions indicate a sequence thats shorter than the given deleted sequence. Please adjust either the variants positions or the given deleted sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.8400_8401del4ins5			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
37. BRCA2	c.8400_8402delTTTins4	NM_000059.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.8400_8402delTTTins4/NM_000059.3?content-type=application/json			
	The length of the variant is not formatted following the HGVS guidelines. Please rewrite e.g. 10 to N[10](where N is an unknown nucleotide)			

	LovdSyntaxcheckInvalid: NM_000059.3:c.8400_8402delTTTins4 is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.8400_8402delinsNNNN, probability = 1]			
	LovdSyntaxcheckWarning: The deleted sequence is redundant and should be removed.			
	LovdSyntaxcheckWarning: The part after ins does not follow HGVS guidelines.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.8400_8402delTTTins4			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
38. BRCA2	c.8400_8403del4ins5	NM_000059.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.8400_8403del4ins5/NM_000059.3?content-type=application/json			
	The length of the variant is not formatted following the HGVS guidelines. Please rewrite e.g. 10 to N[10](where N is an unknown nucleotide)			
	LovdSyntaxcheckInvalid: NM_000059.3:c.8400_8403del4ins5 is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.8403dup, probability = 1]			
	LovdSyntaxcheckWarning: The part after ins does not follow HGVS guidelines.			
	LovdSyntaxcheckWarning: Based on the given sequences, this deletion-insertion should be described as a duplication.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.8400_8403del4ins5			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
39. BRCA2	c.8820_8823delAAAC	NM_000059.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.8820_8823delAAAC/NM_000059.3?content-type=application/json			
	VariantSyntaxError: Removing redundant reference bases from variant description			
	NM_000059.3:c.8820_8823del: Variant reference (AAAC) does not agree with reference sequence (ACAA)			
	LovdSyntaxcheckInvalid: NM_000059.3:c.8820_8823del is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.8820_8823del, probability = 1]			
	LovdSyntaxcheckWarning: The deleted sequence is redundant and should be removed.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.8820_8823del			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
40. BRCA2	c.8941_8942delAG/c.8941_8942delGA	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.8941_8942delAG/c.8941_8942delGA/NM_000059.3?content-type=application/json			
41. BRCA2	c.8955_8956insA/c.8956dupA	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.8955_8956insA/c.8956dupA/NM_000059.3?content-type=application/json			

42. BRCA2	c.9841delA/c.9841delC	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.9841delA/c.9841delC/NM_000059.3?content-type=application/json			
43. BRCA2	c.10462A>G/c.10462T>G	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.10462A%3EG/c.10462T%3EG/NM_000059.3?content-type=application/json			
44. BRCA2	c.3036delA/c.3036delT	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.3036delA/c.3036delT/NM_000059.3?content-type=application/json			
45. BRCA2	c.3267_3268delAG/c.3267_3268delGA	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.3267_3268delAG/c.3267_3268delGA/NM_000059.3?content-type=application/json			
46. BRCA2	c.4563_4564delGT	NM_000059.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.4563_4564delGT/NM_000059.3?content-type=application/json			
	VariantSyntaxError: Removing redundant reference bases from variant description			
	NM_000059.3:c.4563_4564delGT: Variant reference (GT) does not agree with reference sequence (AT)			
	LovdSyntaxcheckInvalid: NM_000059.3:c.4563_4564delGT is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.4563_4564del, probability = 1]			
	LovdSyntaxcheckWarning: The deleted sequence is redundant and should be removed.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.4563_4564delGT			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
47. BRCA2	c.4409_4413delATAAG/c.4408_4412delATAAG	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.4409_4413delATAAG/c.4408_4412delATAAG/NM_000059.3?content-type=application/json			
48. BRCA2	c.5074+1G>A	NM_000059.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.5074+1G>A/NM_000059.3?content-type=application/json			
	ExonBoundaryError: Position c.5074+1 does not correspond with an exon boundary for transcript NM_000059.3			
	LovdSyntaxcheckInvalid: NM_000059.3:c.5074+1G>A is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.5074+1G>A, probability = 0.1]			

	LovdSyntaxcheckError: To verify intronic positions, add a genomic context to the transcript reference sequence.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.5074+1G>A			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
49. BRCA2	c.5101insT	NM_000059.3	GRCh37	
	https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.5101insT/NM_000059.3?content-type=application/json			
	Insertion length must be 1 e.g. 5101_5102insT			
	LovdSyntaxcheckInvalid: NM_000059.3:c.5101insT is not syntactically correct, see LovdSyntaxcheckSuggestions for details			
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.5100_5101insT, probability = 0.7]			
	LovdSyntaxcheckSuggestions: [suggestion = NM_000059.3:c.5101_5102insT, probability = 0.3]			
	LovdSyntaxcheckWarning: An insertion must be provided with the two positions between which the insertion has taken place.			
	LovdSyntaxcheckSource: https://api.lovd.nl/v2/checkHGVS/NM_000059.3:c.5101insT			
	LovdSyntaxcheckLibraryVersion: 0.5.7			
50. BRCA2	c.5154_5158del5/c.5154_5158delTAATT	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.5154_5158del5/c.5154_5158delTAATT/NM_000059.3?content-type=application/json			
51. BRCA2	c.5217_5221delACAA/c.5217_5221delTTTAA	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.5217_5221delACAA/c.5217_5221delTTTAA/NM_000059.3?content-type=application/json			
52. BRCA2	c.5795_5799delACAA/c.5795_5799delATAAC	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.5795_5799delACAA/c.5795_5799delATAAC/NM_000059.3?content-type=application/json			
53. BRCA2	c.6359C>G	NM_000059.3	GRCh37	
	Exception:('Connection aborted.', RemoteDisconnected('Remote end closed connection without response'))			
54. BRCA2	c.6368dupA	NM_000059.3	GRCh37	
	Exception:HTTPSConnectionPool(host='rest.variantvalidator.org', port=443): Max retries exceeded with url: /VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.6368dupA/NM_000059.3?content-type=application/json (Caused by NewConnectionError('<urllib3.connection.HTTPSConnection object at 0x7f09c2d6efd0>: Failed to establish a new connection: [Errno 111] Connection refused'))			
55. BRCA2	c.6696_6697delTC/c.6696_6697delAG	NM_000059.3	GRCh37	

	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.6696_6697delTC/c.6696_6697delAG/NM_000059.3?content-type=application/json			
56. BRCA2	c.6873_6876delCTCC/c.6873_6876delTGAA	NM_000059.3	GRCh37	
	Exception:404 Client Error: NOT FOUND for url: https://rest.variantvalidator.org/VariantValidator/variantvalidator/GRCh37/NM_000059.3:c.6873_6876delCTCC/c.6873_6876delTGAA/NM_000059.3?content-type=application/json			