



Appendix: Quick Reference

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Naming Conventions

- **Gene:** HUGO Gene Nomenclature Committee (www.genenames.org)
- **Sequence variants / pathogenic variants:** Human Gene Variation Society (varnomen.hgvs.org)

Standard nomenclature for DNA and protein amino acid sequence. *GeneReviews* uses the nomenclature guidelines at varnomen.hgvs.org. If the standard nomenclature for a sequence variant cannot be determined, the variant is designated as an **alias** and a reference to the earliest reported description of the variant is provided.

Table 1. Basic structure for DNA and Protein Amino Acid Sequence Nomenclature

Prefix	Type of Sequence from which the Number(s) are Derived	Example	Interpretation	Commonly Describes
c.	coding DNA sequence, where nucleotide #1 is the first nucleotide of the first codon	c.535T>C	The T at nucleotide number 535 of the coding region changed to a C.	Nucleotide changes in genes (exons, splice sites, untranslated regions)
p.	protein amino acid sequence, where #1 is the first amino acid residue of the protein	p.Cys179Arg	The cysteine amino acid at residue 179 changed to an arginine.	Amino acid changes in a protein
g.	genomic DNA sequence, where #1 is the first nucleotide in a given reference sequence	g.66781T>C	The T at nucleotide 66781 in the genomic reference sequence changed to a C.	Nucleotide changes in introns, regulatory, and intergenic regions
m.	mitochondrial DNA sequence, where #1 is the first nucleotide in the sequence of the entire mitochondrion genome	m.8994T>C	The T at nucleotide 8994 of the mitochondrial genome changed to a C.	Coding and non-coding variants of mitochondrial genome

varnomen.hgvs.org

Reference sequence. For the variant nomenclature to be valid, a reference sequence is required. Reference sequences are found at www.ncbi.nlm.nih.gov. Note:

- Reference sequences beginning with "NM" refer to coding nucleotide sequences and those beginning with "NP" refer to protein sequences.
- Genes may have multiple reference sequences. The reference sequence shown in the *GeneReview* matches the exon structure and reference used by the majority of citations.

Abbreviations for amino acids. *GeneReviews* standard is use of the 3-letter symbol for amino acids.

Table 2. Amino Acid Abbreviations

Name	Symbol	
	3-Letter	1-Letter
Alanine	Ala	A
Arginine	Arg	R
Asparagine	Asn	N
Aspartic acid	Asp	D
Cysteine	Cys	C
Glutamic acid	Glu	E
Glutamine	Gln	Q
Glycine	Gly	G
Histidine	His	H
Isoleucine	Ile	I
Leucine	Leu	L
Lysine	Lys	K
Methionine	Met	M
Phenylalanine	Phe	F
Proline	Pro	P
Serine	Ser	S
Threonine	Thr	T
Tryptophan	Trp	W
Tyrosine	Tyr	Y
Valine	Val	V
stop codon	Ter ¹	* ²

1. Previously designated as X or *

2. Previously designated as X

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