



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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