

Pharmacogenomics Terminology in *AJHP*

AJHP supports efforts by the HUGO Gene Nomenclature Committee (a committee of the Human Genome Organisation [HUGO]) to promote consistent use of current approved gene nomenclature to facilitate literature indexing and retrieval. Approved symbols and names are listed on the HGNC website (www.genenames.org). Authors should adhere to the following conventions in all *AJHP* articles:

1. Take care to use the correct font style to differentiate gene symbols, which are italicized (e.g., *EGFR*, *CYP2C19*), from abbreviations for encoded proteins, which are denoted in Roman type (*EGFR*, *CYP2C19*); this is particularly important in the reference list.
2. Specific mutations are denoted in Roman type (e.g., the T790M mutation of *EGFR*, the *BRAF* V600E mutation).
3. Allelic variants of genes are denoted in italics using “star-allele” nomenclature (e.g., *CYP2C19*1A*), with the exception of alleles of the major histocompatibility locus, which are denoted in Roman type (e.g., HLA-B*1502).
4. For proteins involved in cell-signaling pathways (e.g., the Raf-MEK-MAPK and PI3K-AKT-mTOR pathways), it is usually not necessary to give the full name of each component protein; these proteins are denoted in Roman type and separated by hyphens.
5. ***Even if a gene is mentioned only once, introduce the approved symbol and full HUGO-approved name or a contextually appropriate functional descriptor***; a descriptor is preferable when the full name would be disruptive to readability. For instance, in referring to *CYP2D6*, a gene whose full name is “cytochrome P450, family 2, subfamily D, poly-peptide 6,” better flow is achieved by using a descriptor (see example 2 below).

Example 1 (use of full HGNC-approved gene names):

Patients were tested for mutations of *JAK2*, the gene coding for Janus kinase 2; and *EGFR*, which codes for epidermal growth factor receptor.

Example 2 (use of functional descriptors):

Two major foci of pharmacogenomics research are *CYP2D6*, the gene that codes for a cytochrome P-450 isozyme critical to drug metabolism; and *BCL2*, mutations of which are linked to chronic lymphoid leukemia.

6. In rare situations—for example, in referring to a gene whose previous approved symbol is still very widely used—it may be appropriate to use a previously approved symbol instead of or in addition to the current symbol (e.g., “. . . mutations of *ERBB2* [also known as HER2] . . .”), but such usages should be avoided.