



Human Gene Nomenclature Quiz by Laura King, MA, ELS

Directions: Edit the following sentences based on section [15.6.2](#) of the [AMA Manual of Style](#), keeping in mind that some examples may be correct as is. Below is a brief list of how to edit gene symbols, gene names, alleles, genotypes, and phenotypes. All this information can be found in section [15.6.2](#).

Gene Symbols: A gene symbol is a short term, typically 3 to 7 characters long, that conveys in abbreviated form the name or other attribute of a gene. Human gene symbols usually consist of uppercase letters and may also contain (but never begin with) numerals. Approved gene symbols do not contain Greek letters, roman numerals, superscripts, or subscripts and usually contain no punctuation. In *JAMA* and the *Archives Journals*, gene symbols are italicized, per official recommendations, for example, *AFB* for the α -fetoprotein gene.

Gene Names: Genes are usually named for the molecular product of the gene, the function of the gene, or the condition associated with the gene, if known. Gene names are not italicized, for example, the β_2 -microglobulin gene.

Alleles: Alleles denote alternative forms of a gene. Alleles are often characterized by particular variant sequences (mutations). Because alleles are alternative forms of a particular gene, they are expressed by means of both the gene name or symbol and an appendage that indicates the specific allele. Classically, allele symbols consist of the gene symbol plus an asterisk plus the italicized allele designation, for example, *HBB***S* for the *S* allele of the *HBB* gene.

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Genotypes: Genotype comprises the set of alleles in an individual. Genotypes are italicized, for example, *CYP2D6*4A/*5*. When a genotype is being expressed in terms of nucleotides (eg, a polymorphism), italics and other punctuation are not needed, for example, *MTHFR677* TT genotype. When the individual is being described in terms of the 2 possible amino acids at 1 position in the protein owing to a single-nucleotide polymorphism (nonsynonymous mutation), the corresponding amino acids are separated by a virgule, for example, Met/Val (heterozygous). Such terms should be explained at first mention with the amino acid terms expanded, for example, the common methionine/valine (Met/Val) polymorphism at codon 129.

Phenotypes: Phenotype is the collection of traits in an individual resulting from his or her genotype. When phenotypes are expressed in terms of the specific alleles, the phenotype term derives from the genotype term, but no italics are used, and, instead of asterisks, spaces are used. For example, for genotype *ABO*A1/ABO*O*, the phenotype is ABO A1.

1. Women who have inherited mutations in the breast cancer genes BRCA1 or BRCA2 have substantially elevated risks of breast cancer and ovarian cancer, with a lifetime risk of breast cancer of 56% to 84%.

2. We measured HER2/neu protein expression and HER2/neu gene amplification by means of immunohistochemical assessment and fluorescence in situ hybridization, respectively.

3. We examined measures that may be associated with disease in individuals carrying the gene expansion for Huntington disease (HD). Our study included 505 at-risk individuals who had previously undergone elective DNA analyses for the CAG expansion in the HD gene (predictive testing) and did not currently have a clinical diagnosis of HD.



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4. The cause of the faulty enzyme was found to be a defect in the glucose-6-phosphate dehydrogenase (G6PD) gene.

5. On the basis of our biochemical results and in agreement with previously published criteria, the m.5728A>G mutation qualifies as being pathogenic.

6. Discordant phenotypes resulted from the same mutation in exon ORF15 of the retinitis pigmentosa GTPase regulator gene (RPGR) in 2 presumed dizygotic twin brothers with X-linked retinal disease. An identical mutation in RPGR-ORF15 manifested distinct clinical phenotypes in individuals of the same family.

7. The apolipoprotein E ϵ 4 allele (*APOE** ϵ 4) is associated with an increased risk of developing Alzheimer disease.

8. The HLA-DQB1*0503 allele is significantly associated with susceptibility to tuberculosis in Cambodian patients.

9. We investigated the characterization of mutations of the ATP7B gene and the correlation between the ATP7B genotype and the ATP7B phenotype in the Chinese population.

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- 10.** Our study found that valine/valine (val/val) individuals were susceptible to Alzheimer disease in late life and methionine/valine (met/val) allele carriers were susceptible to Alzheimer disease in early adult life.
- 11.** The ADRB2 gene Gln27Glu (79 CG) G allele carriers with heart failure were significantly more likely to demonstrate an improved ejection fraction with carvedilol therapy than were patients homozygous for the C allele.
- 12.** The distribution of the ADH3 genotypes AA, GA, and GG was similar in the groups with and without cancer.