Human genes associated with disease / syndromes by missense mutations

In the list below, some variants are known to cause detrimental phenotypes, others are known to be neutral, or a phenotype is currently not known (neutral or deleterious). All the proteins below are known to be associated with disesase / syndromes when mutations disrupt their function. The condition is followed by the protein name; in parenthesis is the official human gene abbreviation; then the accession number for the version of the human protein to use.

Nomenclature example for AA variants: I65T – at position 65 (65th amino acid in the protein), there is usually or normally an I (isoleucine); in the variant, there is a T (threonine). Note that this may or may not affect the function of the protein.

Condition/Syndrome: Protein (enzyme) name (Human gene abbreviation) – Accession No. List of variants

Phenylketonuria: Phenylalanine hydroxylase (PAH) - P00439

D59Y

165T

R158Q

Q160P

L255V

R261Q

M276K

P281L

P314S

Y343F

S349P

K363N

R408W

Segawa Syndrome, Dopa-responsive Dystonia, Infantile Parkinsonism: Tyrosine Hydroxylase (TH) - P07101

R233H

L236P

T276P

G280R

E284V

V306I

T314M

R337H

C359F

I440T

Tay Sachs disease: Beta-Hexosaminidase A (HEXA) - P06865

L39R

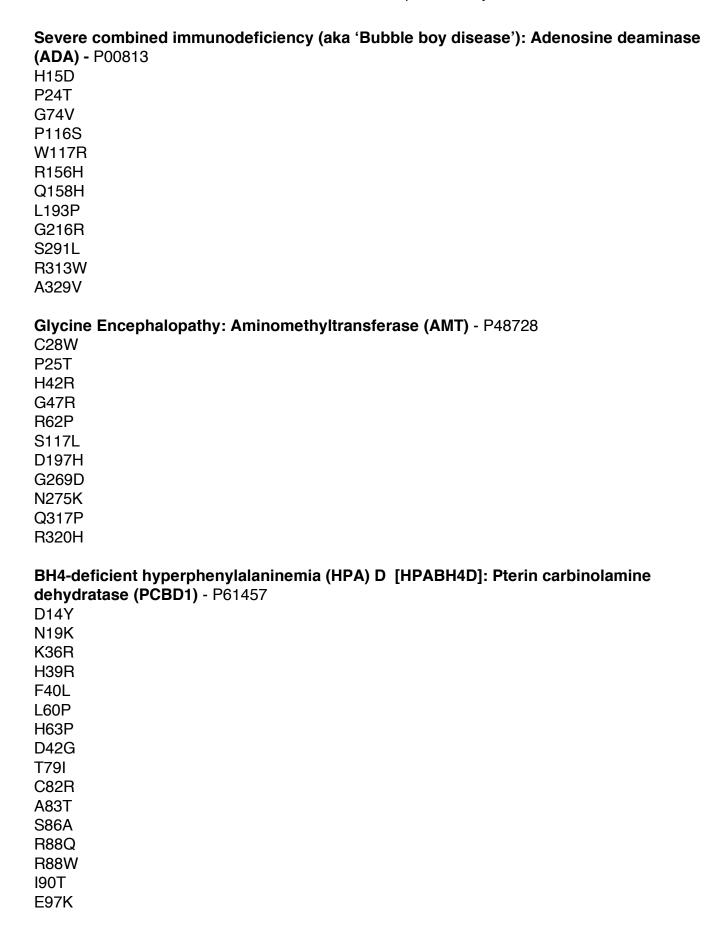
S59L

L127R

R170Q

R178H

V192A



Bio / Chem 330 Bioinformatics Exercise: Protein Sequence Analysis of Human Disease Genes

BH4-deficient hyperphenylalaninemia (HPA) C [HPABH4C]: Quinoid dihydropteridine reductase (QDPR) - P09417

G23D

W36R

K55T

A91T

W108G

H120R

A139S

Y150C

H158Y

L164V

S193P