

Examples of notable Mutations

ΔPSC deletion in cystic fibrosis

		2nd base			
		U	C	A	G
1st base	U	UUU (Phe) Phenylalanine	UCU (Ser) Serine	UAU (Tyr) Tyrosine	UGU (Cys) Cysteine
		UUC (Phe) Phenylalanine	UCC (Ser) Serine	UAC (Tyr) Tyrosine	UGC (Cys) Cysteine
		UUA (Leu) Leucine	UCA (Ser) Serine	UAA (Stop) (Stop)	UGA (Stop) (Stop)
		UUG (Leu) Leucine	UCG (Ser) Serine	UAG (Stop) (Stop)	UGG (Trp) Tryptophan
	C	CUU (Leu) Leucine	CCU (Pro) Proline	CAU (His) Histidine	CGU (Arg) Arginine
		CUC (Leu) Leucine	CCC (Pro) Proline	CAC (His) Histidine	CGC (Arg) Arginine
		CUA (Leu) Leucine	CCA (Pro) Proline	CAA (Gln) Glutamine	CGA (Arg) Arginine
		CUG (Leu) Leucine	CCG (Pro) Proline	CAG (Gln) Glutamine	CGG (Arg) Arginine
	A	AUU (Ile) Isoleucine	AUU (Thr) Threonine	AUU (Asn) Asparagine	AUU (Ser) Serine
		AUC (Ile) Isoleucine	AUC (Thr) Threonine	AAC (Asn) Asparagine	AUC (Ser) Serine
		AUA (Ile) Isoleucine	ACA (Thr) Threonine	AAA (Lys) Lysine	AUA (Arg) Arginine
		AUG (Met) Methionine	AAG (Thr) Threonine	AAG (Lys) Lysine	AUG (Arg) Arginine
	G	GUU (Val) Valine	GUU (Ala) Alanine	GAU (Asp) Aspartic acid	GGU (Gly) Glycine
		GUC (Val) Valine	GUC (Ala) Alanine	GAC (Asp) Aspartic acid	GGC (Gly) Glycine
		GUA (Val) Valine	GCA (Ala) Alanine	GAA (Glu) Glutamic acid	GGA (Gly) Glycine
		GUG (Val) Valine	GCG (Ala) Alanine	GAG (Glu) Glutamic acid	GGG (Gly) Glycine

Selection of notable mutations, ordered in a standard table of the genetic code of amino acids.

Crucially important missense mutations generally change the properties of the coded amino acid residue (between being basic, acidic, polar or nonpolar, when nonsense mutations result in a stop codon).

Amino acids

- Basic
- Acidic
- Polar
- Nonpolar (hydrophobic)

Fragile X Syndrome

Polyglutamine (PolyQ) Diseases

- Huntington's disease
- Spinocerebellar ataxia (SCA) (most types)
- Spinobulbar muscular atrophy (Kennedy disease)
- Dentatorubral-pallidum atrophy

Mutation type

- Trinucleotide repeat
- Deletion
- Missense
- Nonsense

Sickle-cell disease

Friedreich's ataxia

Mutations And Genetic Conditions Lesson 02

Brendan G. Carr



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