Barone's Amino Acid Code Cheatsheet

There are **20 amino acids** and 9 of them are essential.
Here are the one and three letter codes for the amino acids:

Amino acid	Three letter code	One letter code
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
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*Essential Amino Acids

The amino acid letter codes are often used when describing mutations.
Here are some key mutations that you should know for the boards!

Barone Rocks.com The Official Site of John Barone, M.D.	Gene	Mutation
Cystic Fibrosis	CFTR	• △**F508** Deletion of Phenylalanine at 508 • Results in a misfolded CFTR protein that is tagged with ubiquitin and destroyed by the proteosome complex.
Cystic Fibrosis	CFTR	• **G551D** Missense mutation • Glycine is replaced by Aspartic acid at position 551 • Results in a defective CFTR protein.
Hemochromatosis	HFE	• **C282Y** Missense mutation • Cystine is replaced by Tyrosine at position 282 • Results in decreased hepcidin and overabsorption of iron in the small intestine.
Hemochromatosis	HFE	• **H63D** Missense mutation • Histidine is replaced by Aspartic acid at position 63 • Results in decreased hepcidin and overabsorption of iron in the small intestine.
Sickle cell anemia	Beta globin	• **E6V** Missense mutation • Gluatamic acid is replaced by Valine at position 6 • Results in to formation of Hb S.
Melanoma Hairy Cell Leukemia Many other tumors	B-Raf	• **V600E** Missense mutation • Valine is replaced by Glutamic acid at position 600 • Results in activation of the BRAF serine-threonine kinase.
Hyperprothrombinemia (Hypercoaguability)	Prothrombin	• **G20210A** Missense mutation • Glycine is replaced by Alanine in the position 20210 • Results in increased plasma prothrombin levels.
Factor V Leiden (Hypercoaguability)	Factor V	• **G1691A** Missense mutation • Glycine is replaced by Alanine in the position 1691 • Results in loss of the Factor V cleavage site that activated protein C uses to breakdown Factor V.
Polycythemia vera Essential thrombocythemia Myelofibrosis	JAK2	• **V617F** Missense mutation • Valine is replaced by Phenylalanine in position 617 • Results in overactivity of the JAK2 tyrosine kinase causing EPO-independent myloproliferation and overproduction of RBCs (P.vera) or Platelets (ET).