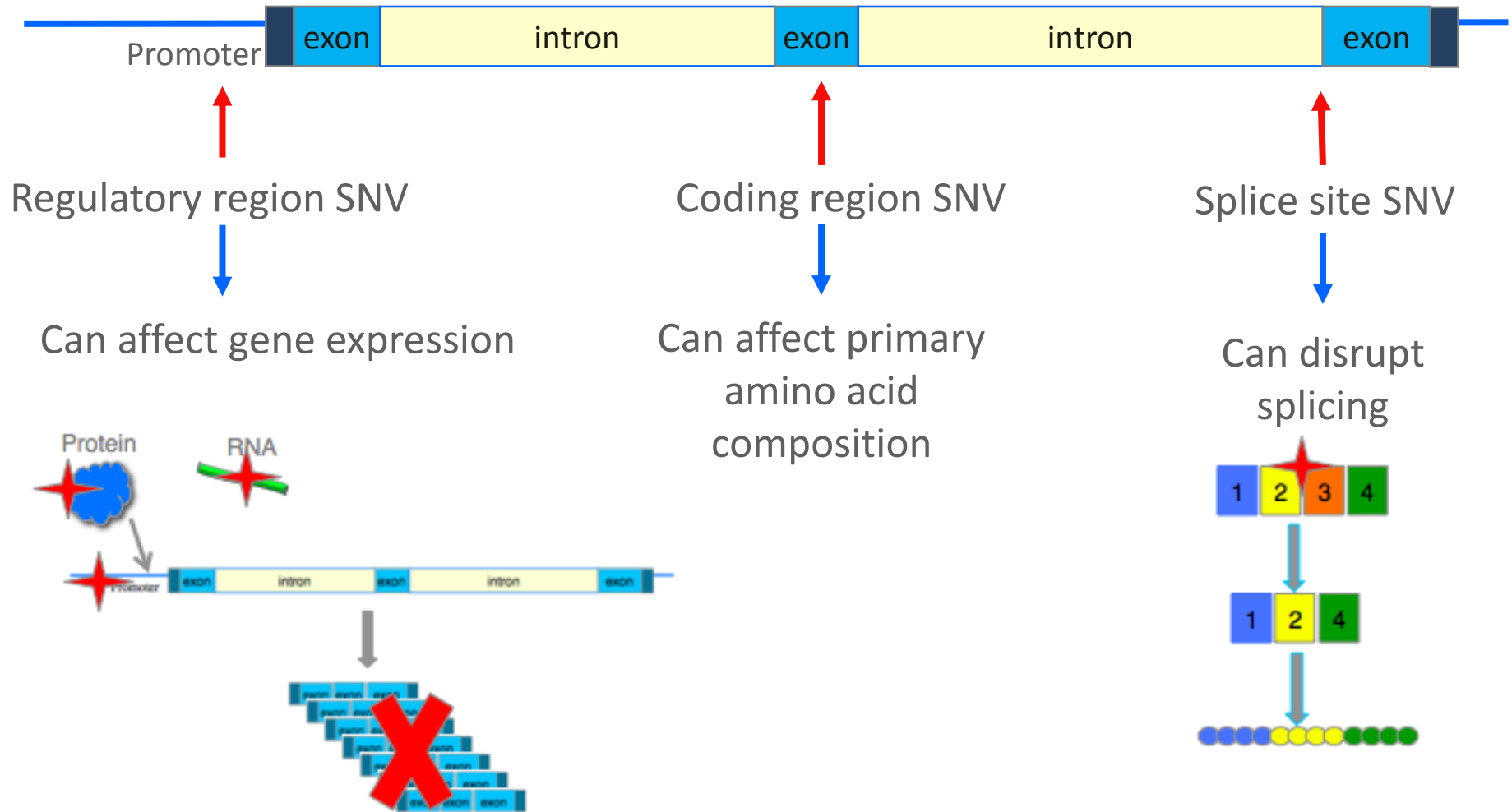


MODULE 4: Consequences of single nucleotide variants in genes

Location, location, location



Coding Region SNVs

Synonymous
(Silent)

A	T	G	C	T	T	C	A	A	C	A	G	C	G
MET			LEU			SER			THR		ALA		

A	T	G	C	T	C	T	C	A	A	C	A	G	C	G
MET			LEU			SER			THR		ALA			

A	T	G	C	T	T	C	A	A	C	A	G	C	G
MET			LEU			SER			THR		ALA		

A	T	G	C	C	T	T	C	A	A	C	A	G	C	G
MET			PRO			SER			THR		ALA			

A	T	G	C	T	T	C	A	A	C	A	G	C	G
MET			LEU			SER			THR		ALA		

A	T	G	C	T	T	A	A	A	C	A	G	C	G
MET			LEU			STOP							

Protein



Frameshift mutations due to InDels

Insertions/deletions of one or more nucleotides in coding region of gene can result in a shift in the reading frame that can dramatically alter the sequence of amino acids in the protein

Deletion of 'T'

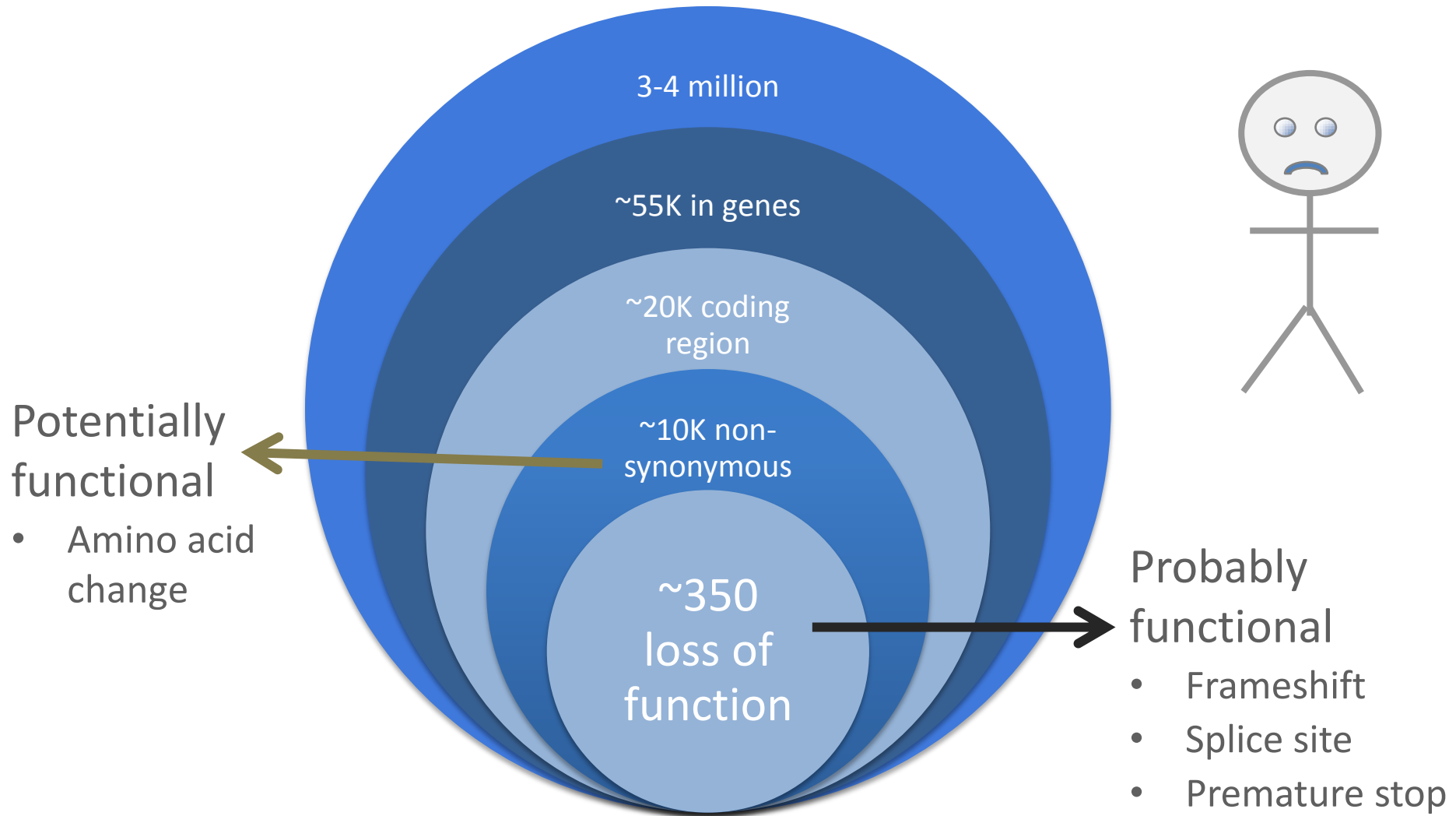


A	T	G	C	T	T	C	A	A	C	A	G	C	G
MET			LEU		SER	THR		ALA					

Frameshift



Load of SNVs in the average person



Question

TRUE or FALSE

Mutations in exons are always deleterious.

Answer

FALSE

Mutations in exons can be silent (i.e. not change the amino acid sequence) or missense (change amino acid) with minimum impact.