



# Genetics

(in a medical context)















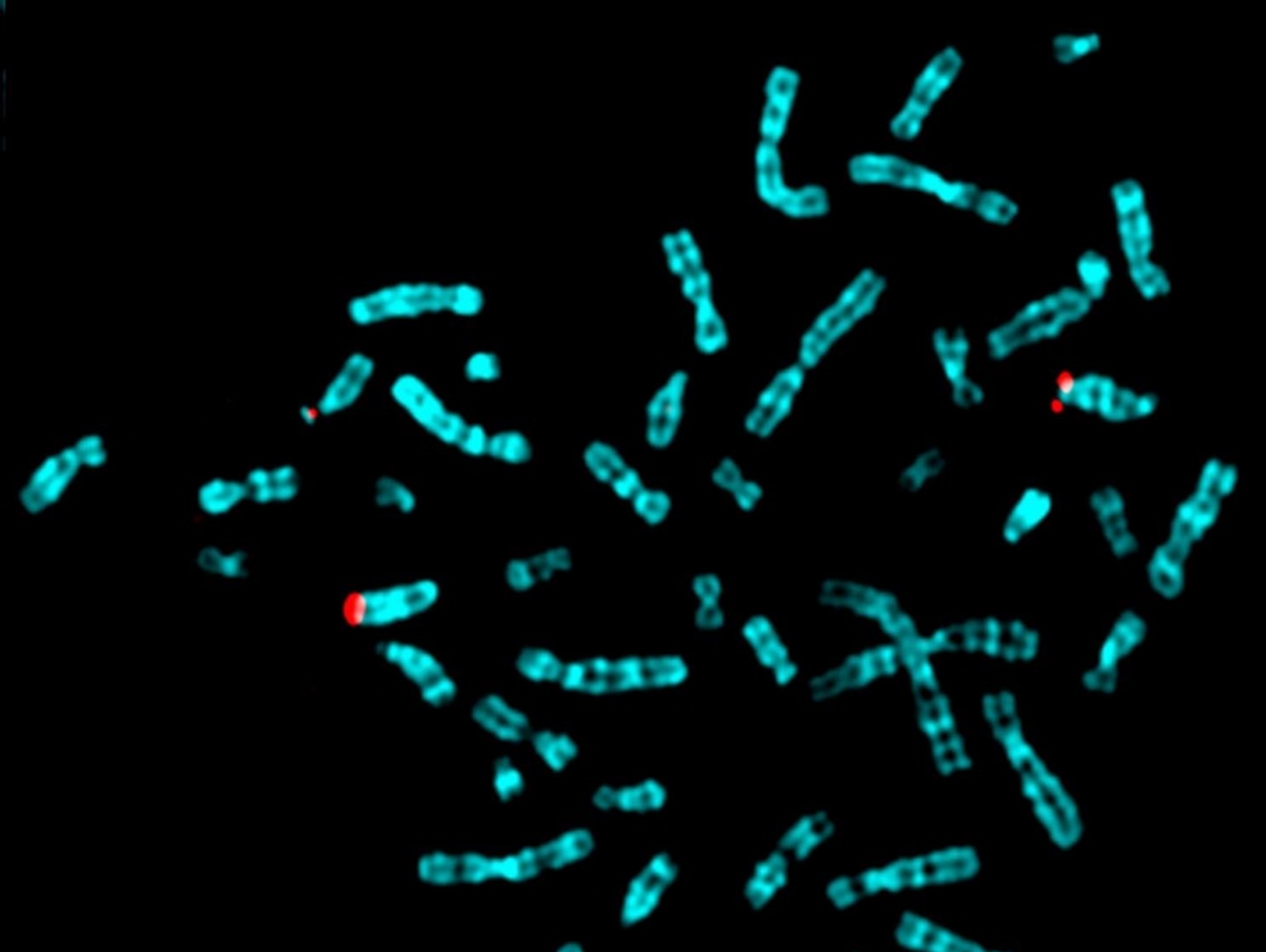
















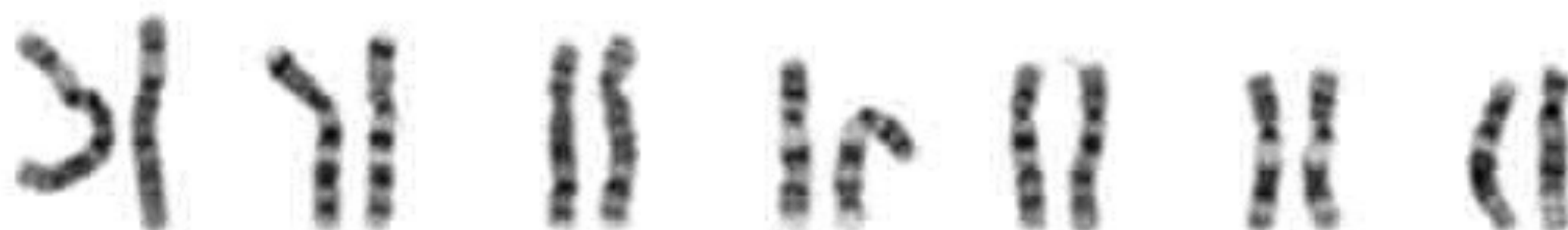
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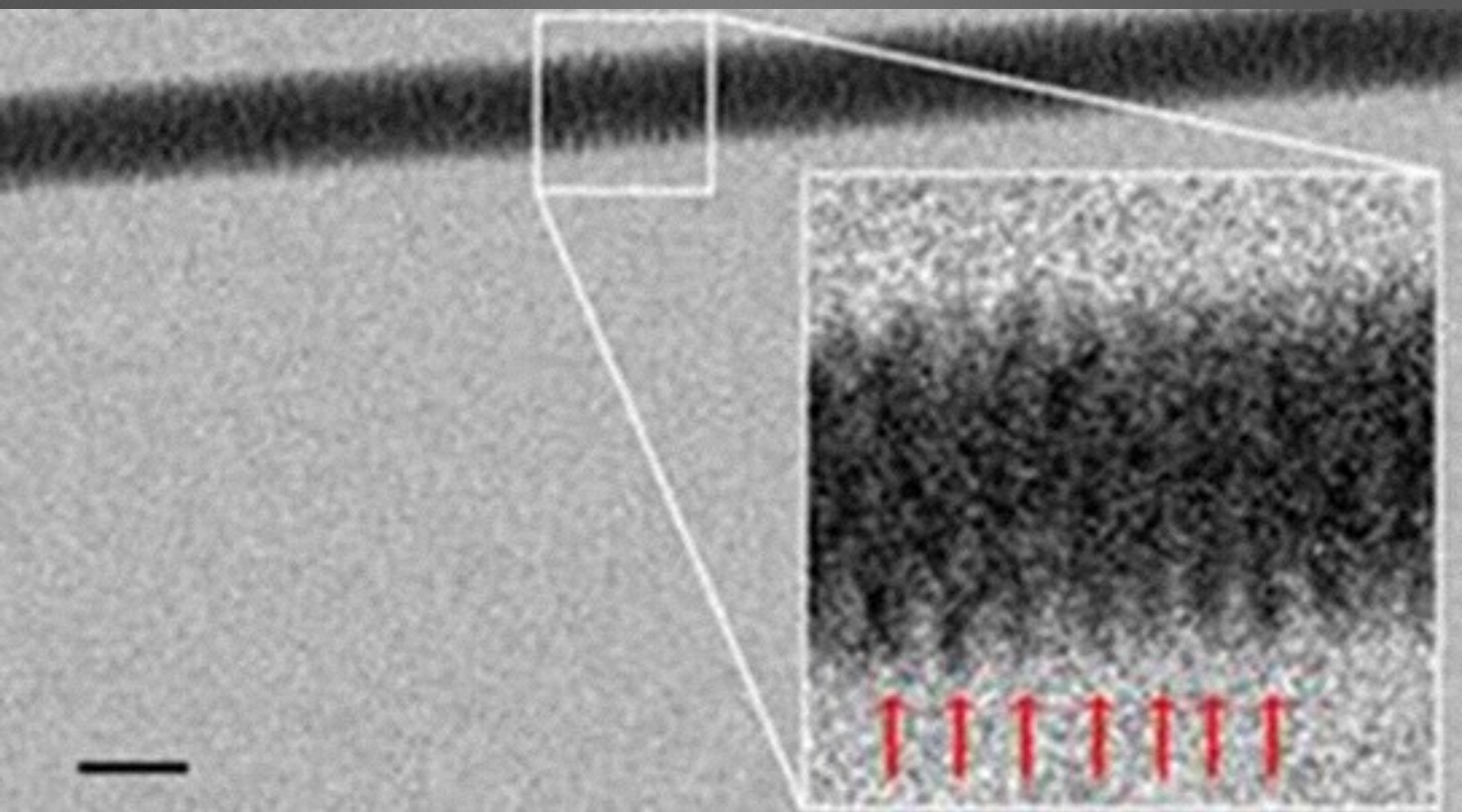
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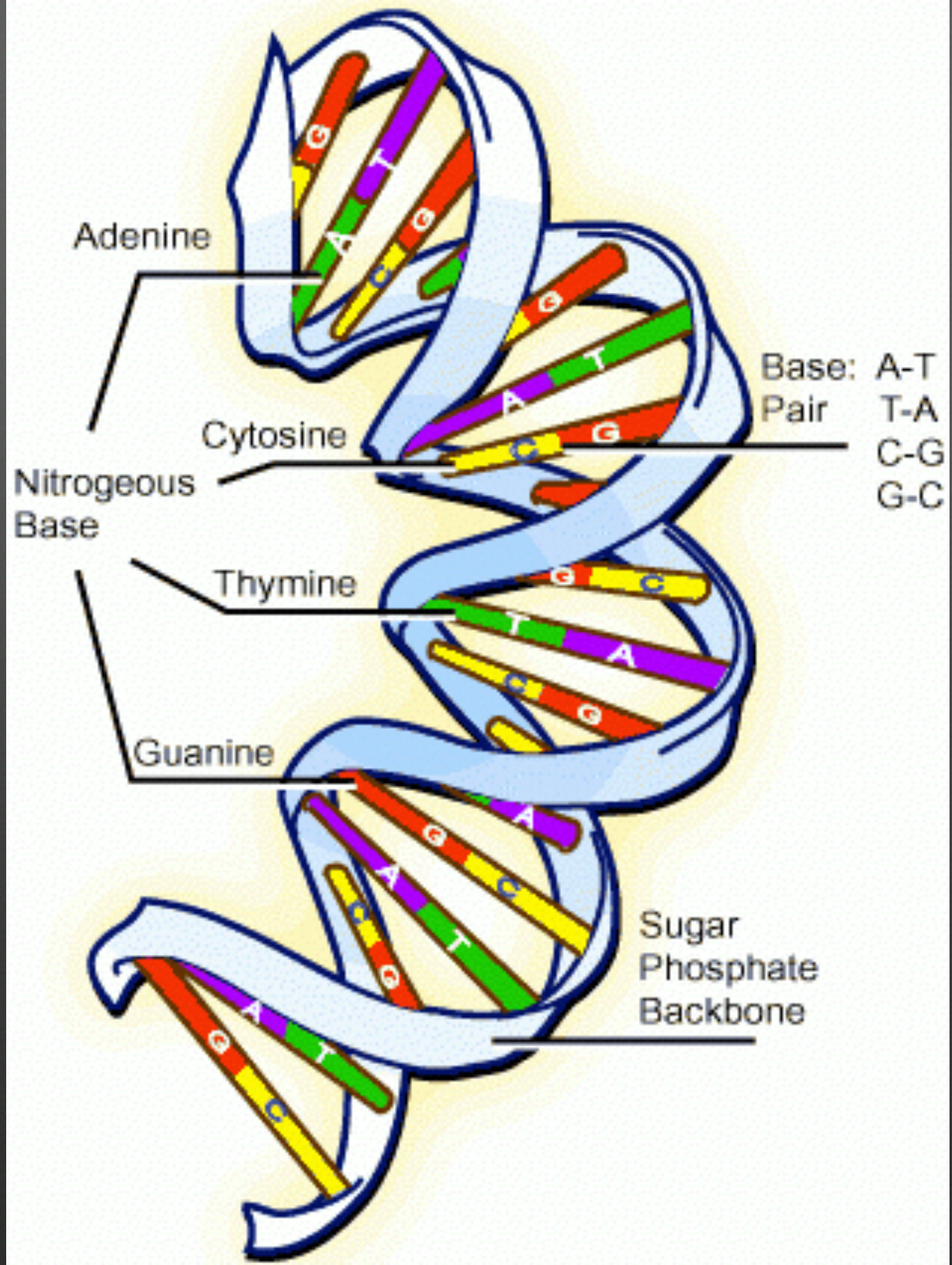
y

x









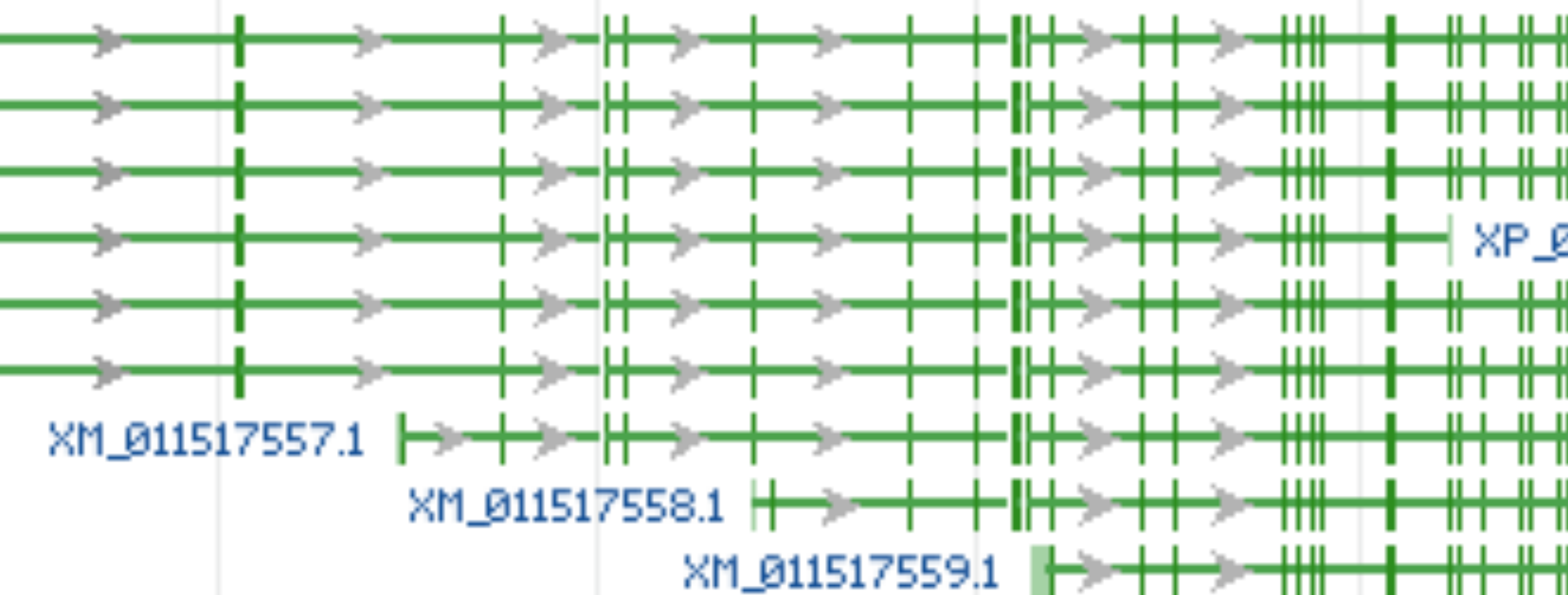






K 60,780 K 60,800 K 60,820 K 60,840 K

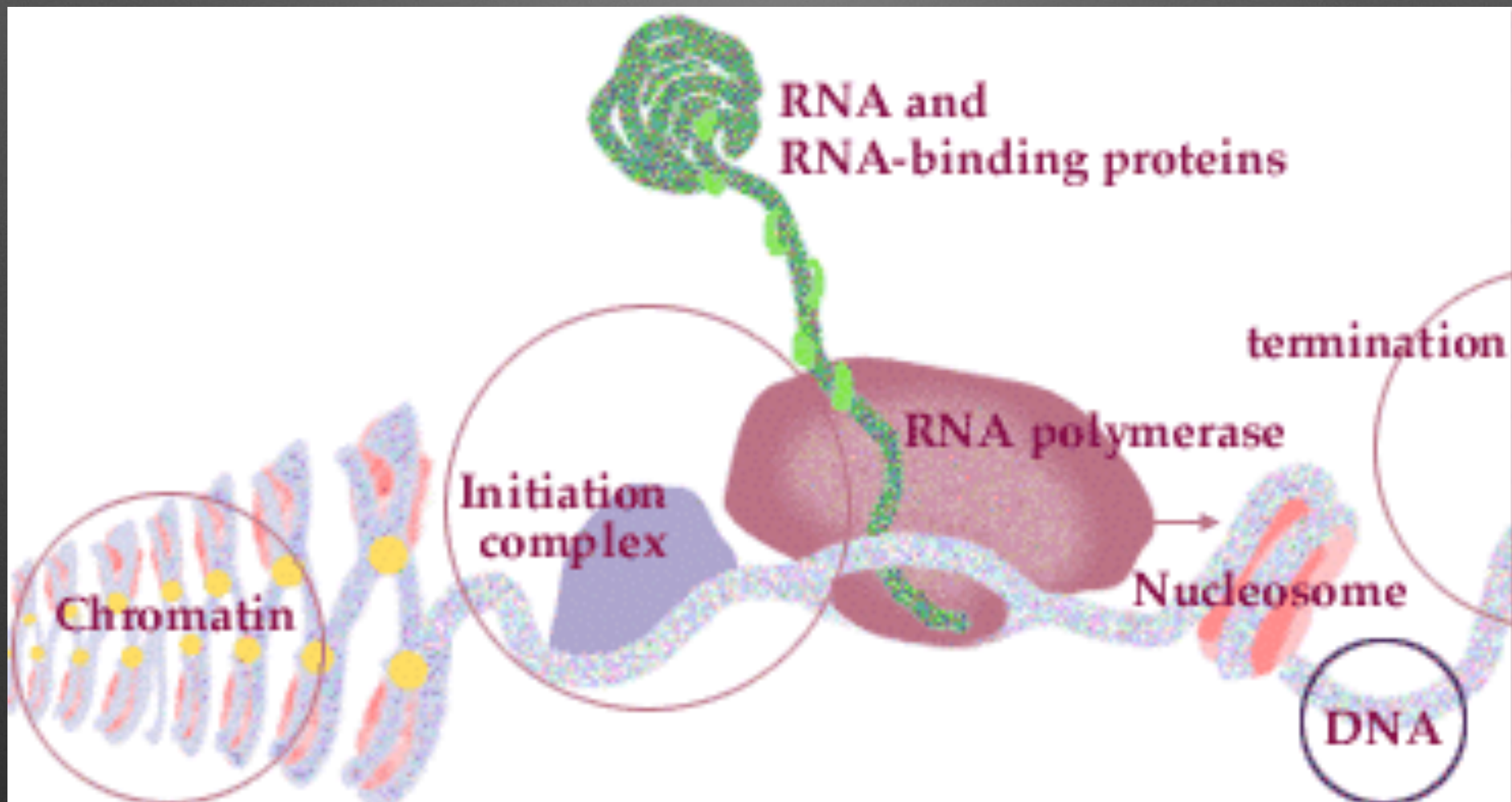
CHD7



LOC105375868

XR\_928948.1

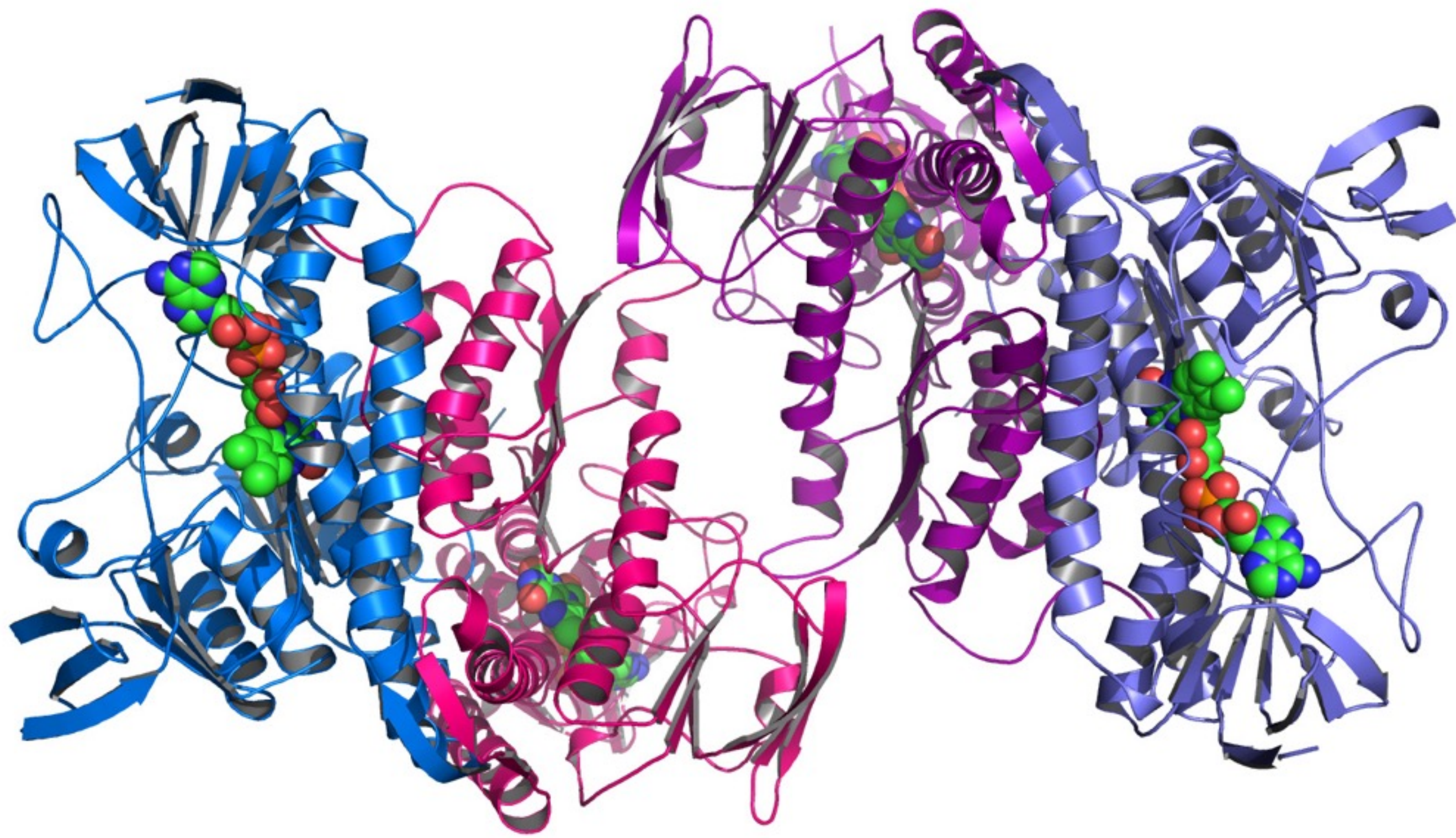




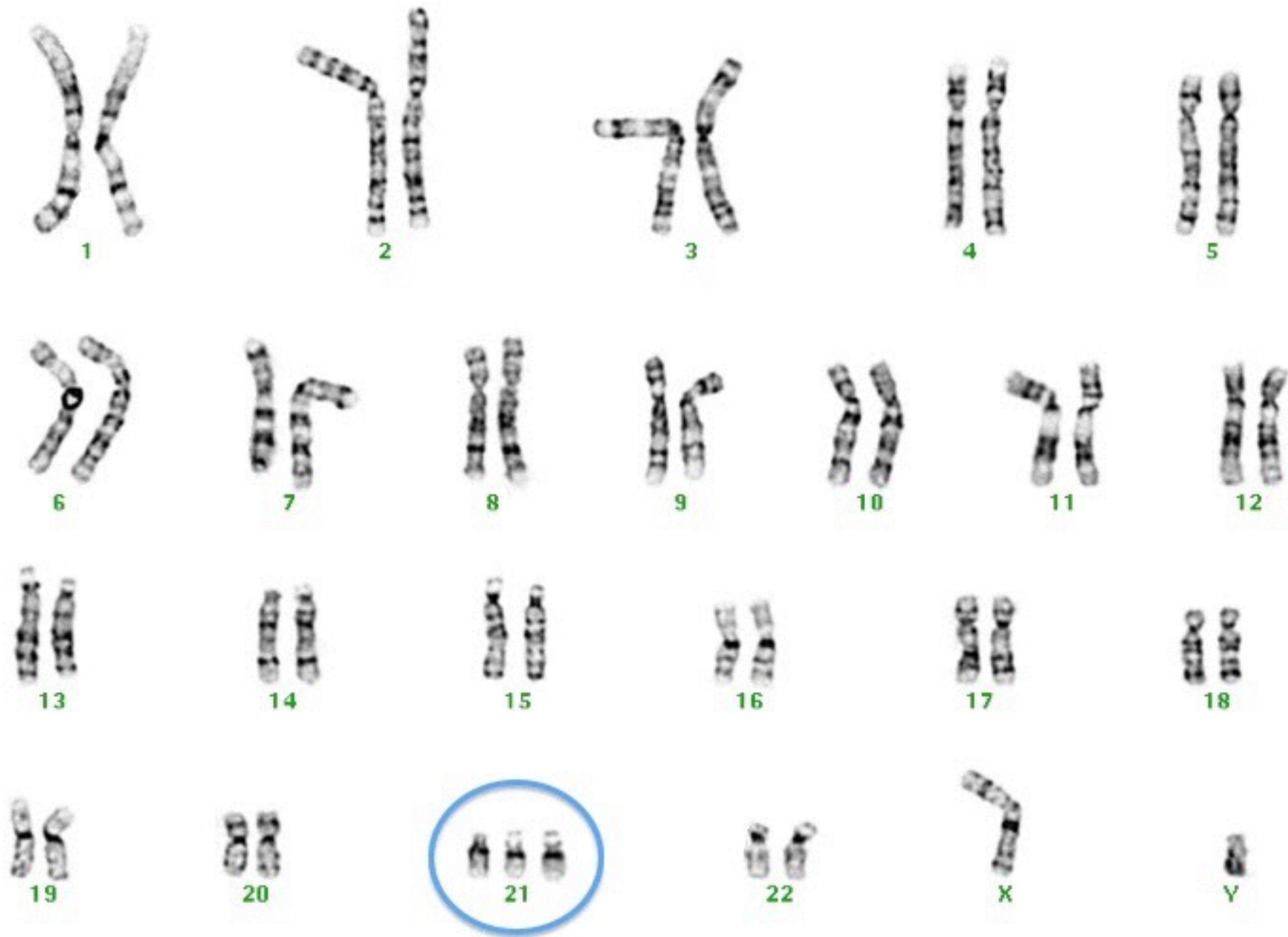


		Second letter				Third letter
		U	C	A	G	
First letter	U	UUU } Phe UUC } UUA } Leu UUG }	UCU } UCC } Ser UCA } UCG }	UAU } Tyr UAC } UAA Stop UAG Stop	UGU } Cys UGC } UGA Stop UGG Trp	
	C	CUU } CUC } Leu CUA } CUG }	CCU } CCC } Pro CCA } CCG }	CAU } His CAC } CAA } Gln CAG }	CGU } CGC } Arg CGA } CGG }	
	A	AUU } AUC } Ile AUA } AUG Met	ACU } ACC } Thr ACA } ACG }	AAU } Asn AAC } AAA } Lys AAG }	AGU } Ser AGC } AGA } Arg AGG }	
	G	GUU } GUC } Val GUA } GUG }	GCU } GCC } Ala GCA } GCG }	GAU } Asp GAC } GAA } Glu GAG }	GGU } GGC } Gly GGA } GGG }	



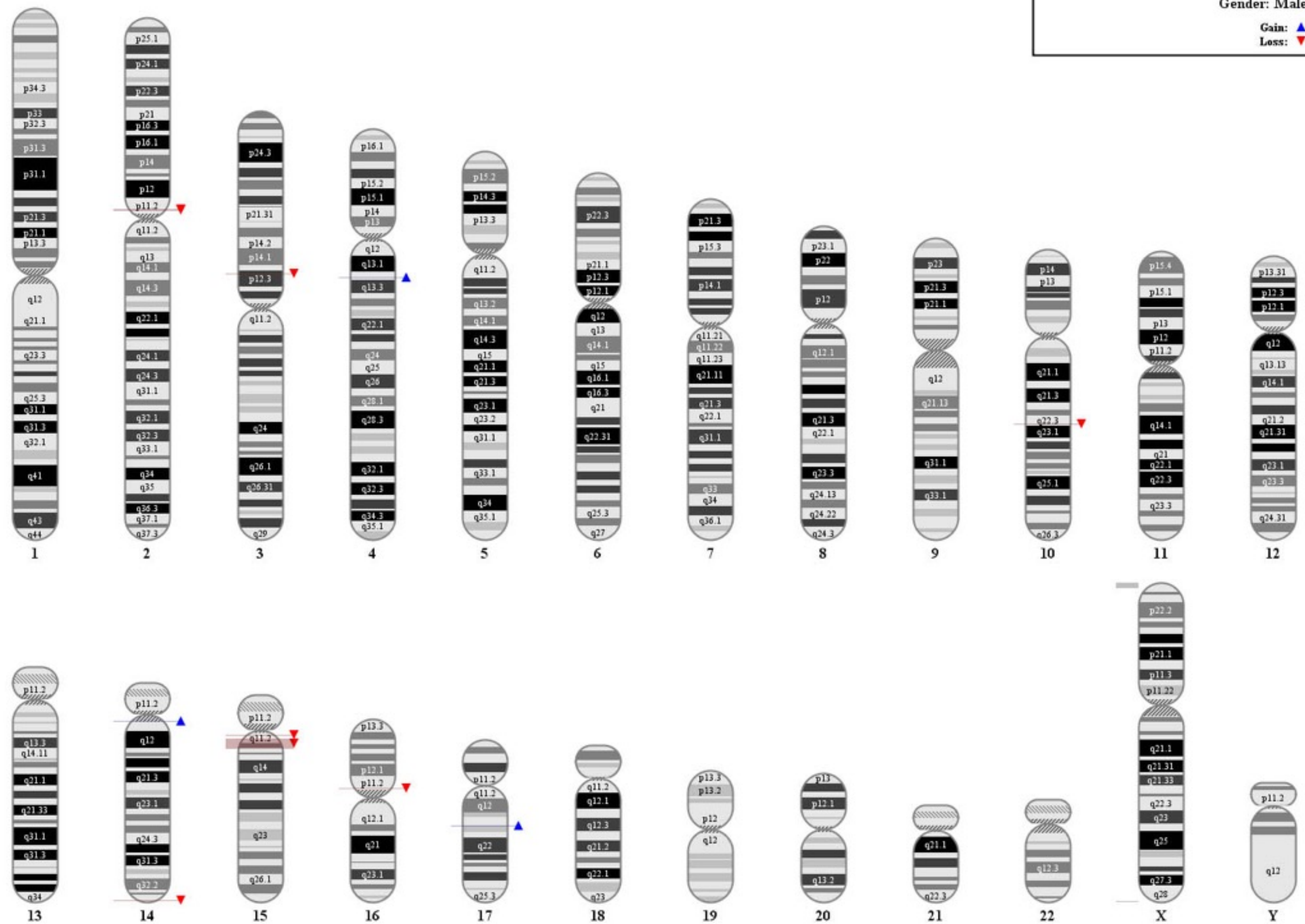






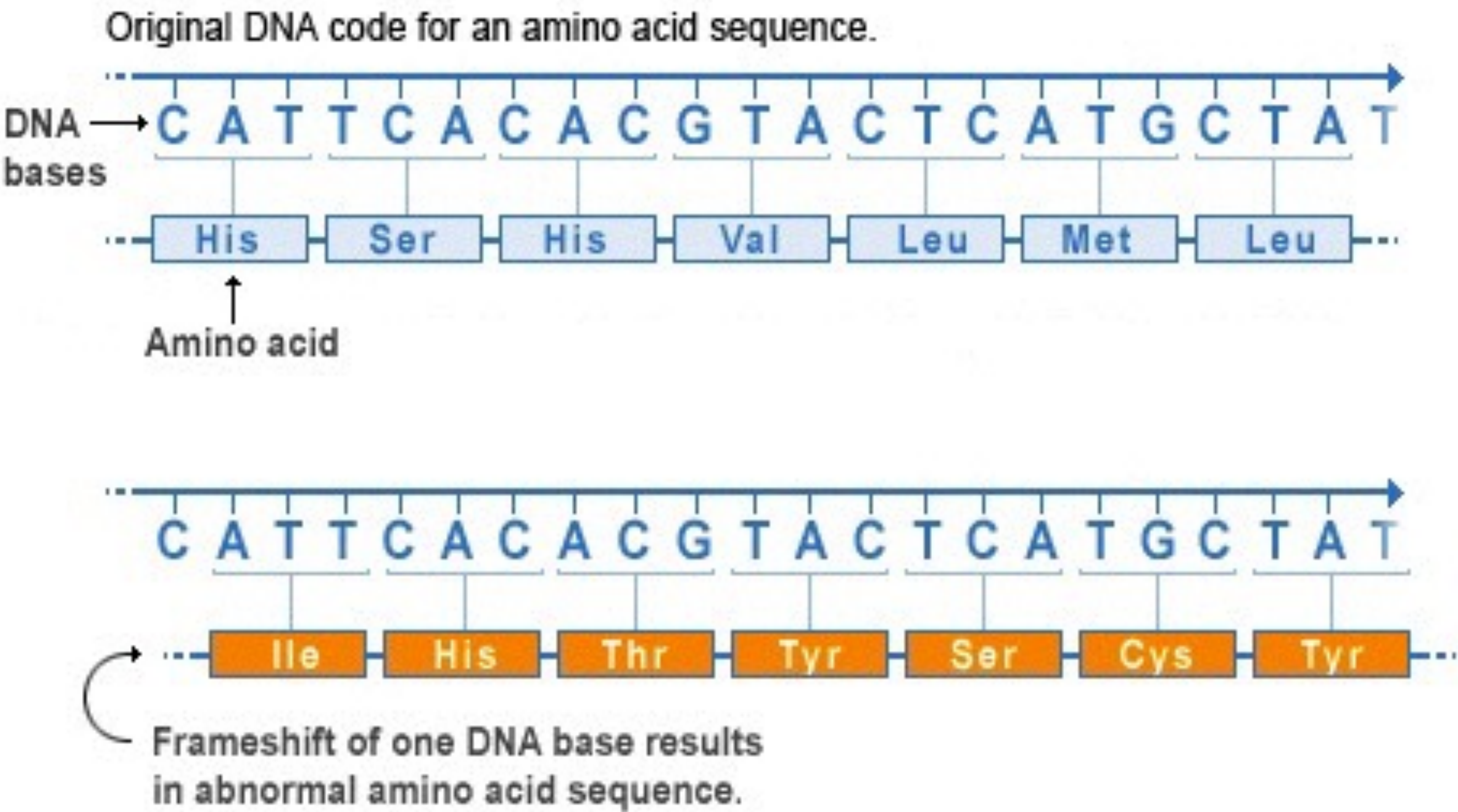
Down Syndrome

47,XY,+21

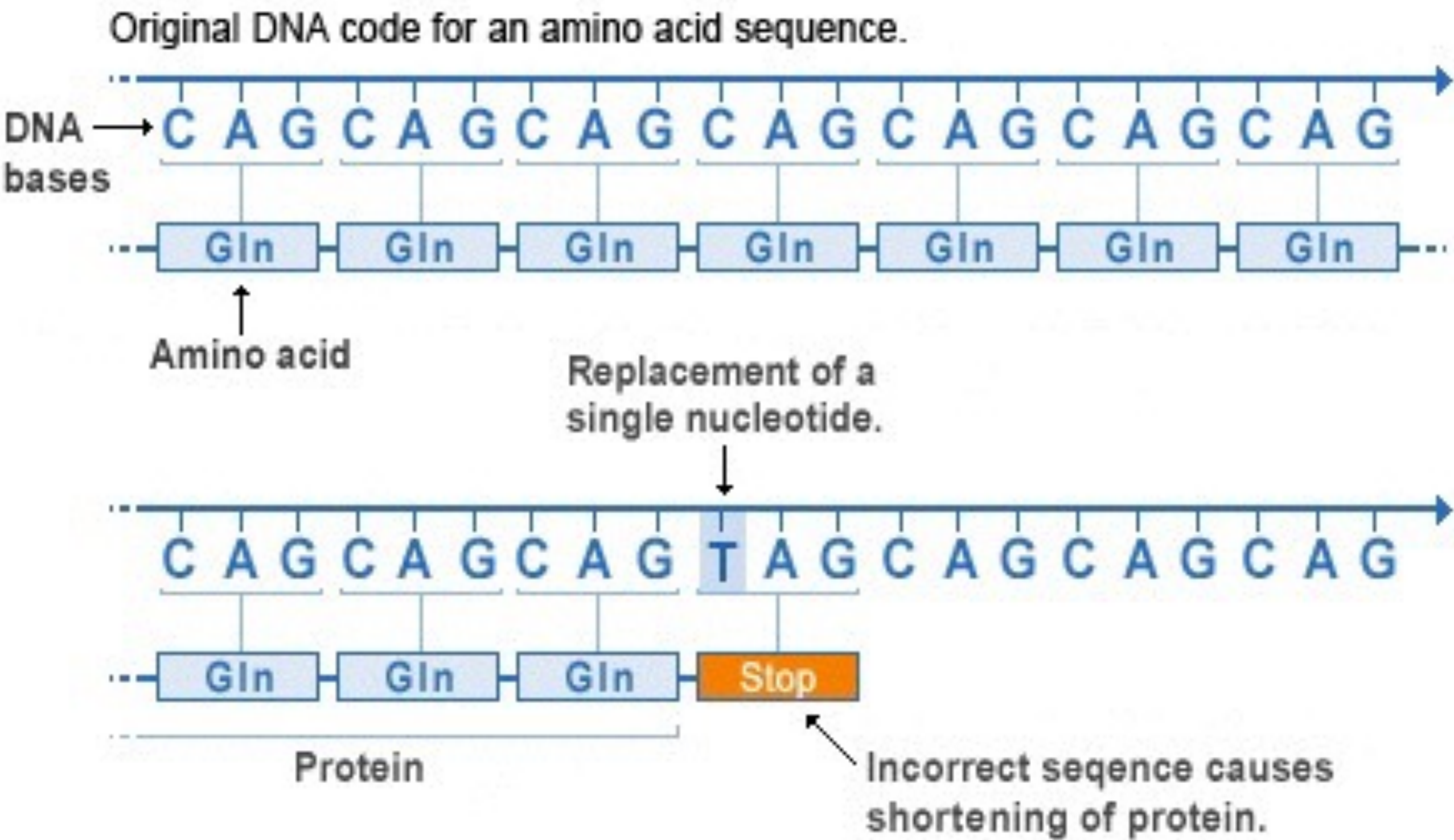




# Frameshift mutation

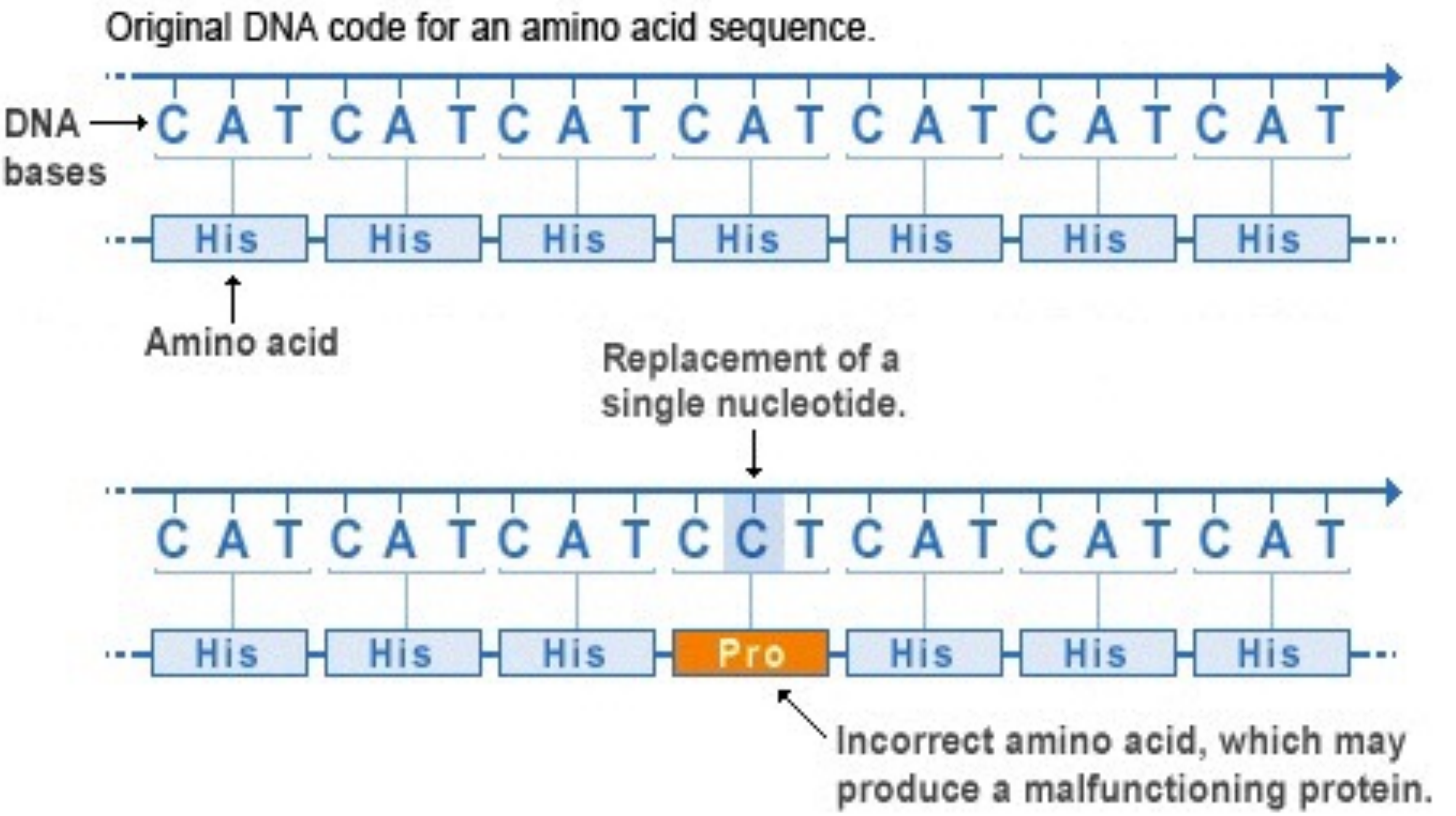


# Nonsense mutation





# Missense mutation



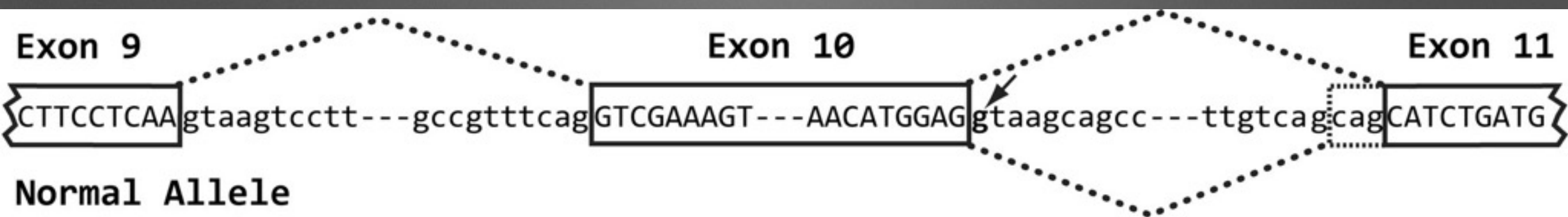
																								wild type		
G	U	C	A	U	G	U	U	U	A	G	C	U	C	A	A	U	C	A	G	G	A	A	G	U	G	U
Val			Met			Phe			Ser			Ser			Ile			Arg			Lys			Cys		
G	U	C	A	U	G	U	U	C	A	G	C	U	C	A	A	U	C	A	G	G	A	A	G	U	G	U
Val			Met			Phe			Ser			Ser			Ile			Arg			Lys			Cys		



Exon 9 Exon 10 Exon 11

CTTCCTCAA gtaagtcctt---gccgtttcag GTCGAAAGT---AACATGGAG gtaagcagcc---ttgtcagcag CATCTGATG

Normal Allele



CTTCCTCAA gtaagtcctt---gccgtttcag GTCGAAAGT---AACATGGAG ataagcagcc---ttgtcagcag CATCTGATG

Mutant Allele

