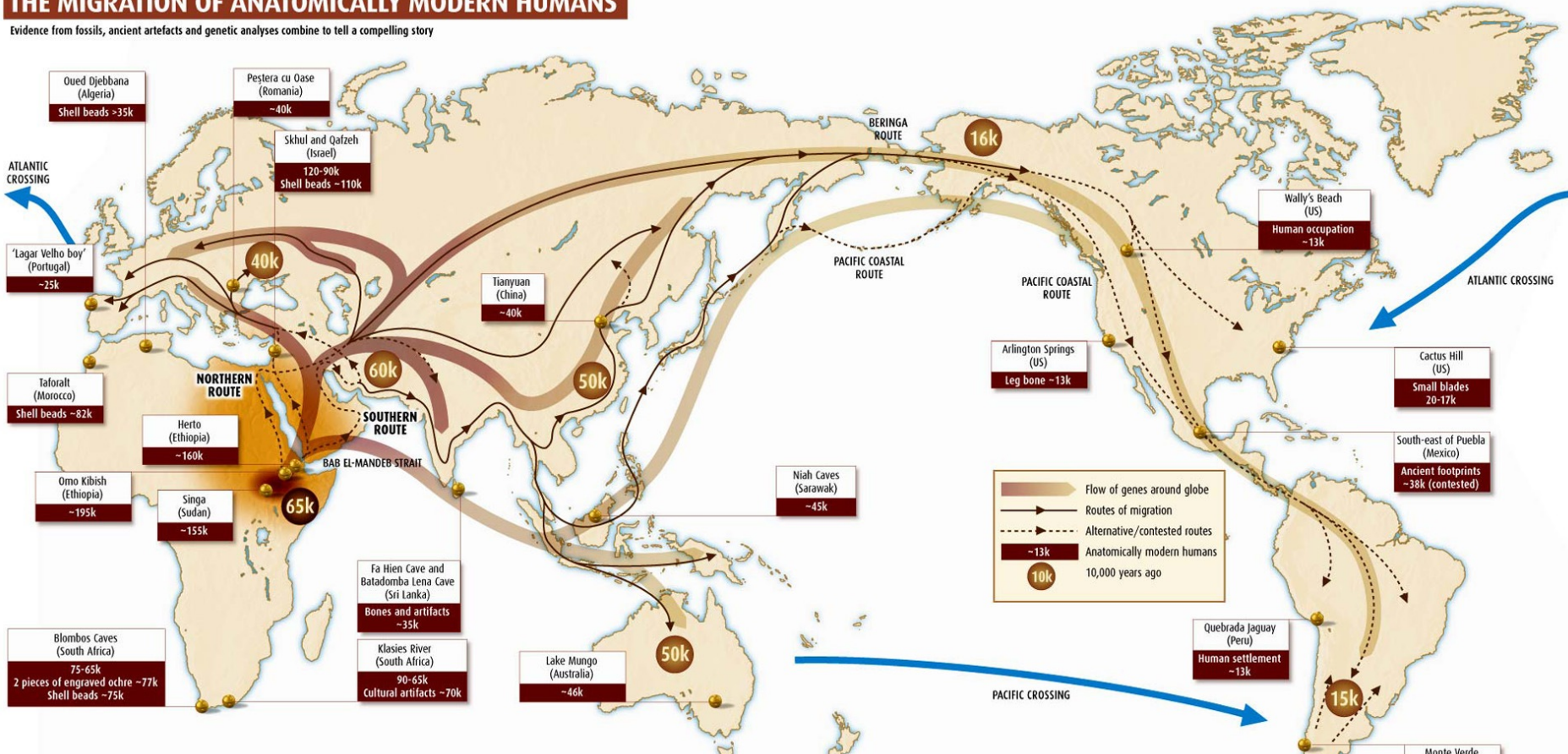


Genetic Mutations



THE MIGRATION OF ANATOMICALLY MODERN HUMANS

Evidence from fossils, ancient artefacts and genetic analyses combine to tell a compelling story



Two routes jump out as prime candidates for the human exodus out of Africa. A northern route would have taken our ancestors from their base in eastern sub-Saharan Africa across the Sahara desert, then through Sinai and into the Levant. An alternative southern route may have charted a path from Djibouti or Eritrea in the Horn of Africa across the Bab el-Mandeb strait and into Yemen and around the Arabian peninsula. The plausibility of these two routes as gateways out of Africa has been studied as part of the UK's Natural Environment Research Council's

programme "Environmental Factors in the Chronology of Human Evolution & Dispersal" (ECHED).
 During the last ice age, from about 80,000 to 11,000 years ago, sea levels dropped as the ice sheets grew, exposing large swathes of land now submerged under water and connecting regions now separated by the sea. By reconstructing ancient shorelines, the ECHED team found that the Bab el-Mandeb strait, now around 30 kilometres wide and one of the world's busiest shipping lanes, was then a narrow, shallow channel.

Early humans may have taken this southern route out of Africa. The northern route appears easier, especially given the team's finding that the Suez basin was dry during the last ice age. But crossing the Sahara desert is no small matter. ECHED scientist Simon Armitage of the Royal Holloway University of London has found some clues as to how this might have been possible. During the past 150,000 years, North Africa has experienced abrupt switches between dry, arid conditions and a humid climate. During the longer wetter periods huge lakes existed in both Chad and Libya, which would have

provided a "humid corridor" across the Sahara.
 Armitage has discovered that these lakes were present around 10,000 years ago, when there is abundant evidence for human occupation of the Sahara, as well as around 115,000 years ago, when our ancestors first made forays into Israel. It is unknown whether another humid corridor appeared between about 65,000 and 50,000 years ago, the most likely time frame for the human exodus. Moreover, accumulating evidence is pointing to the southern route as the most likely jumping-off point.





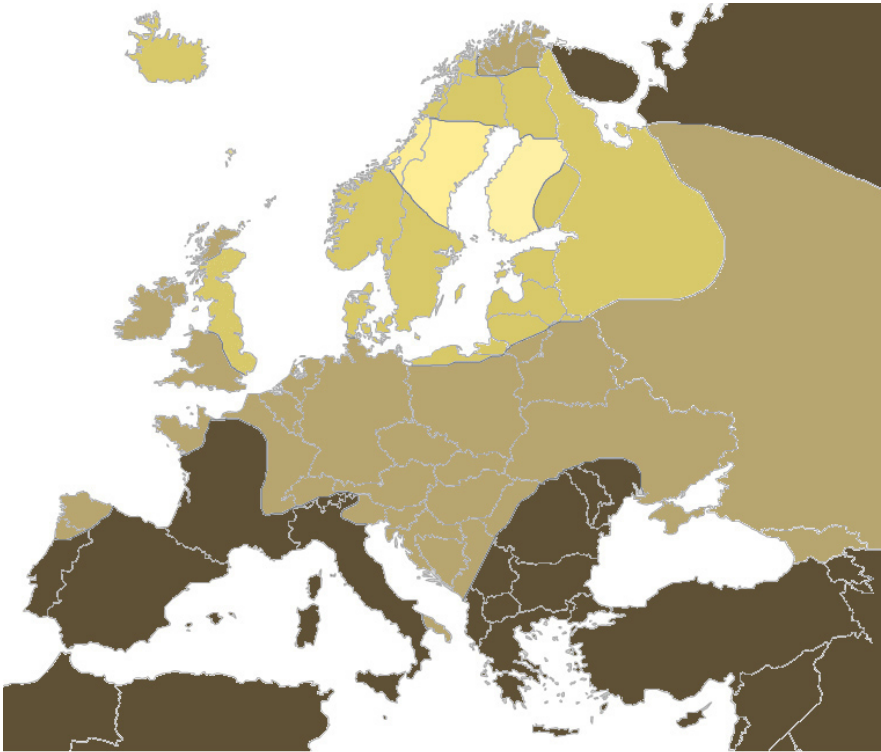
Mutations- changes in the DNA code

+ changes results in improved success to the individual

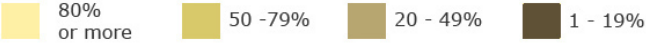
neutral(\emptyset) changes results has no effect

- changes results in creating a disadvantage

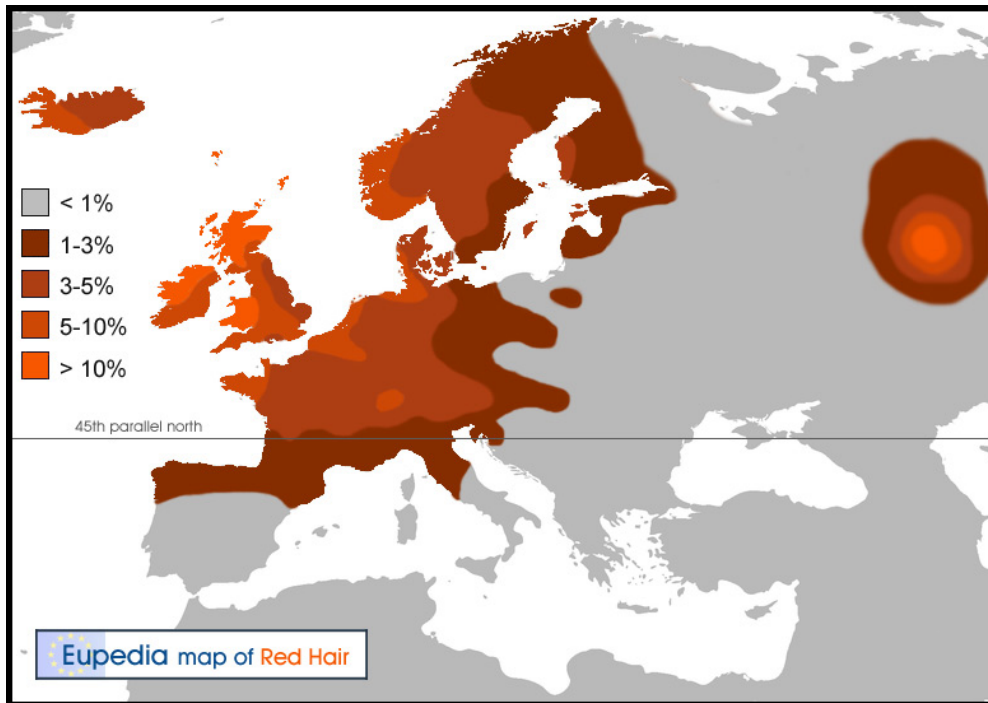
Neutral effects-Alleles



Percentage of light hair in Europe



Neutral Effects-Alleles



[Asp294His](#)



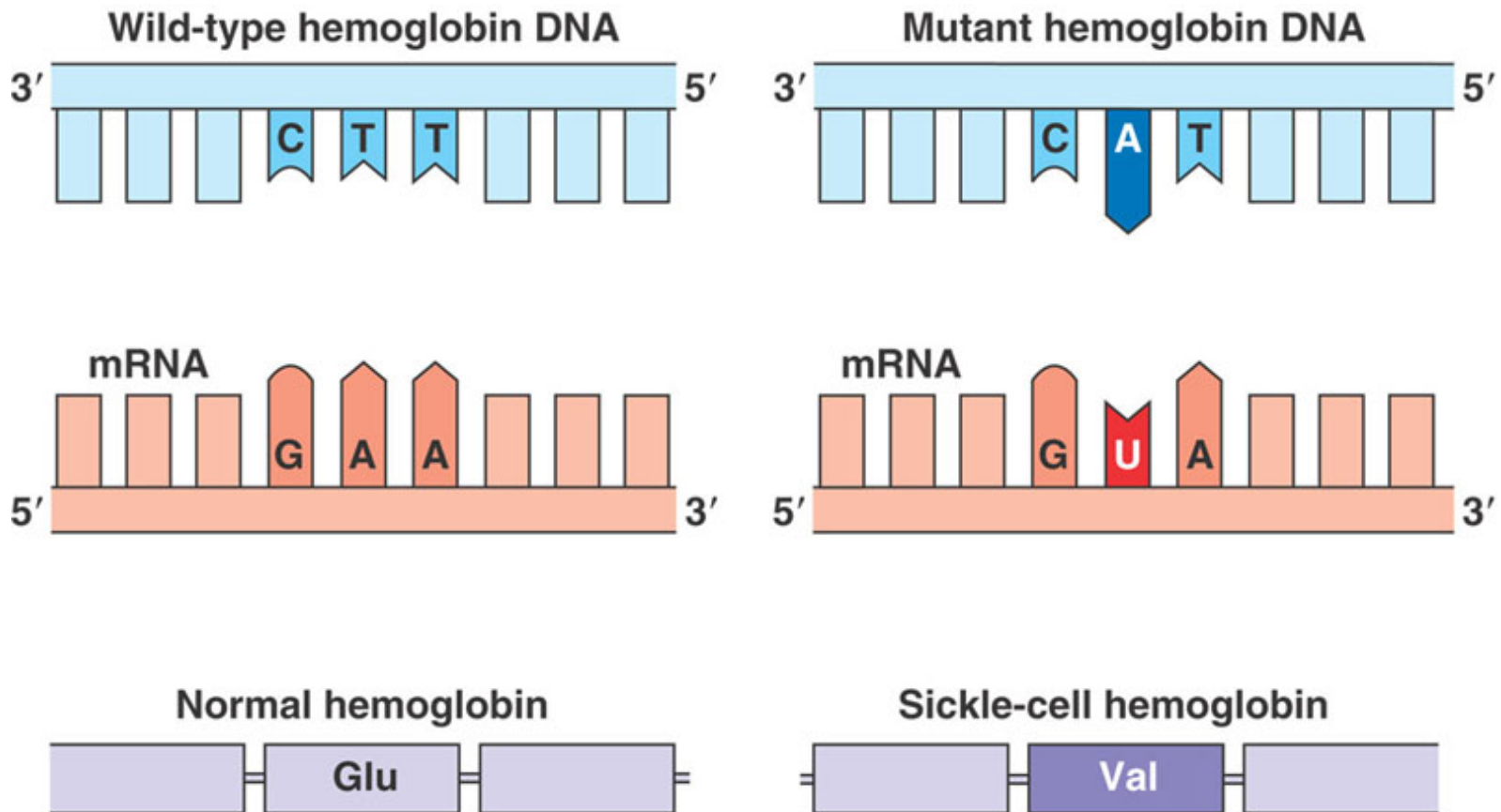


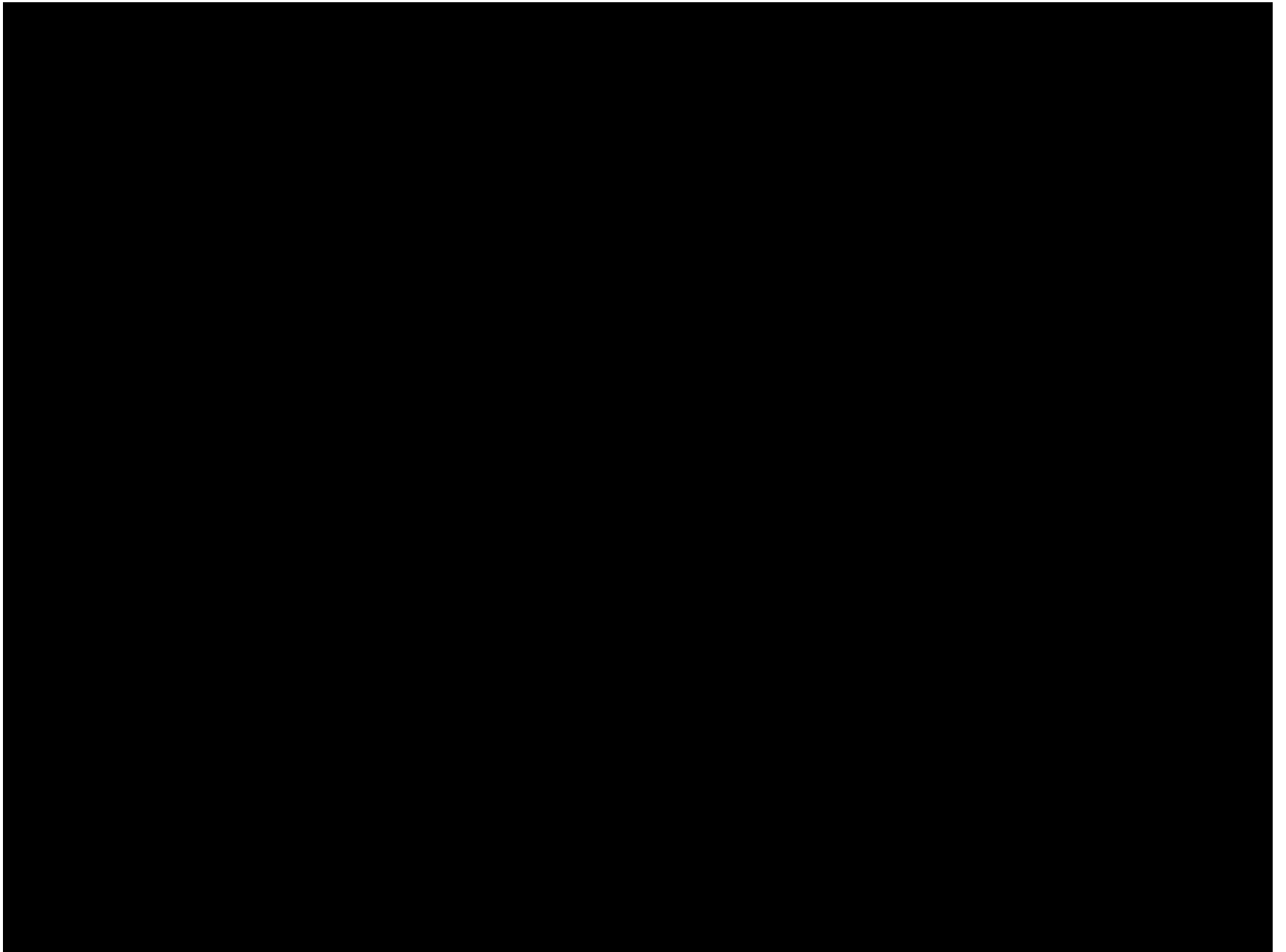
Small-Scale - Mutations

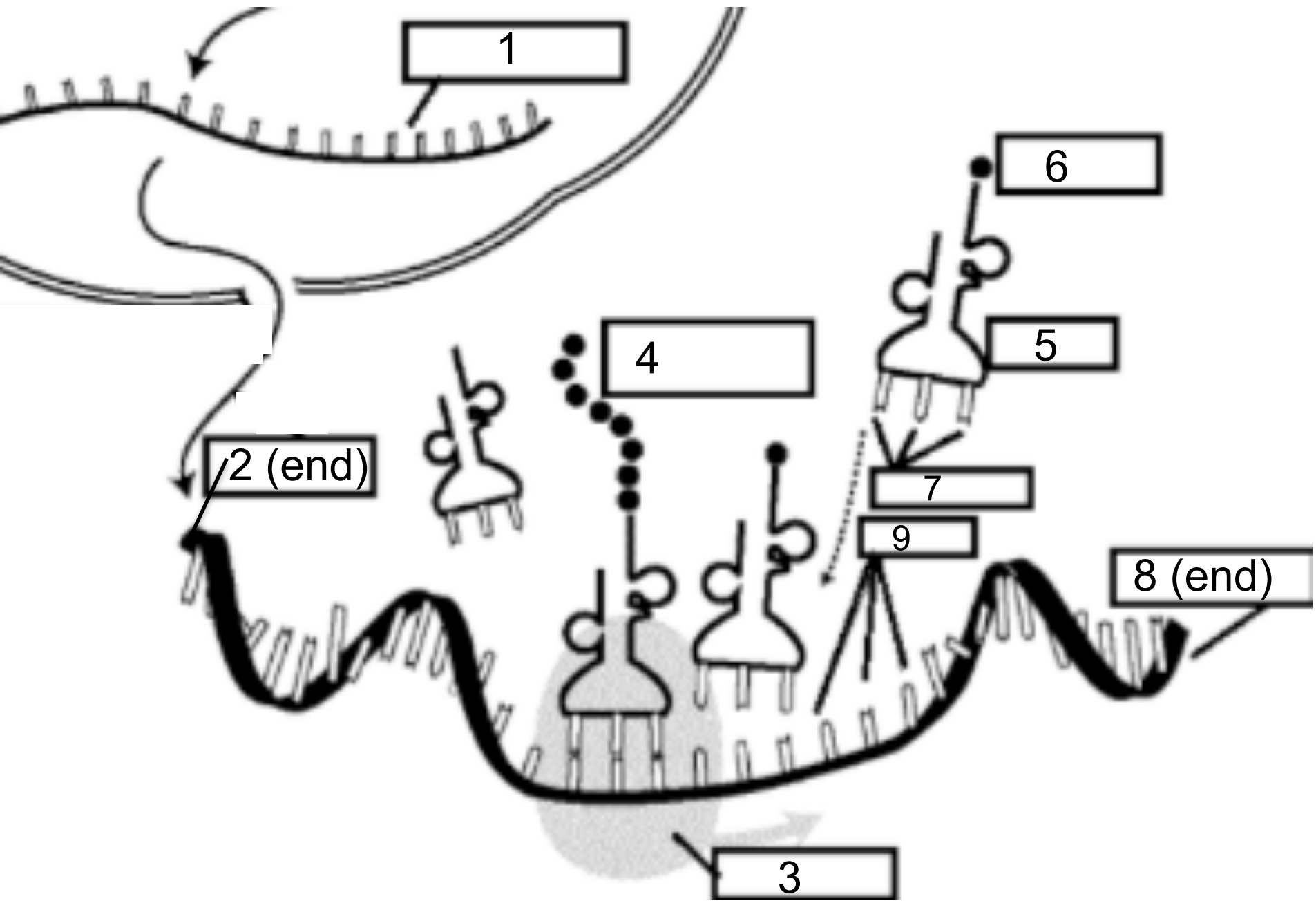
- **point mutations** (single base pair)
 - Substitution
 - Insertion/deletion
 - Inversion (switch two adjacent bases)



Point mutation (One letter wrong)













Tripthalangeal Thumb (TPT)

Point Mutation (one letter wrong)



Effects of Point Mutations

(substitution)

- silent mutations (no effect)
- missense mutations (one amino acid change)
- nonsense mutations (one point mutation that can terminate the translation)

(deletion/insertion)

- frameshift mutations - (affects every codon down the line)

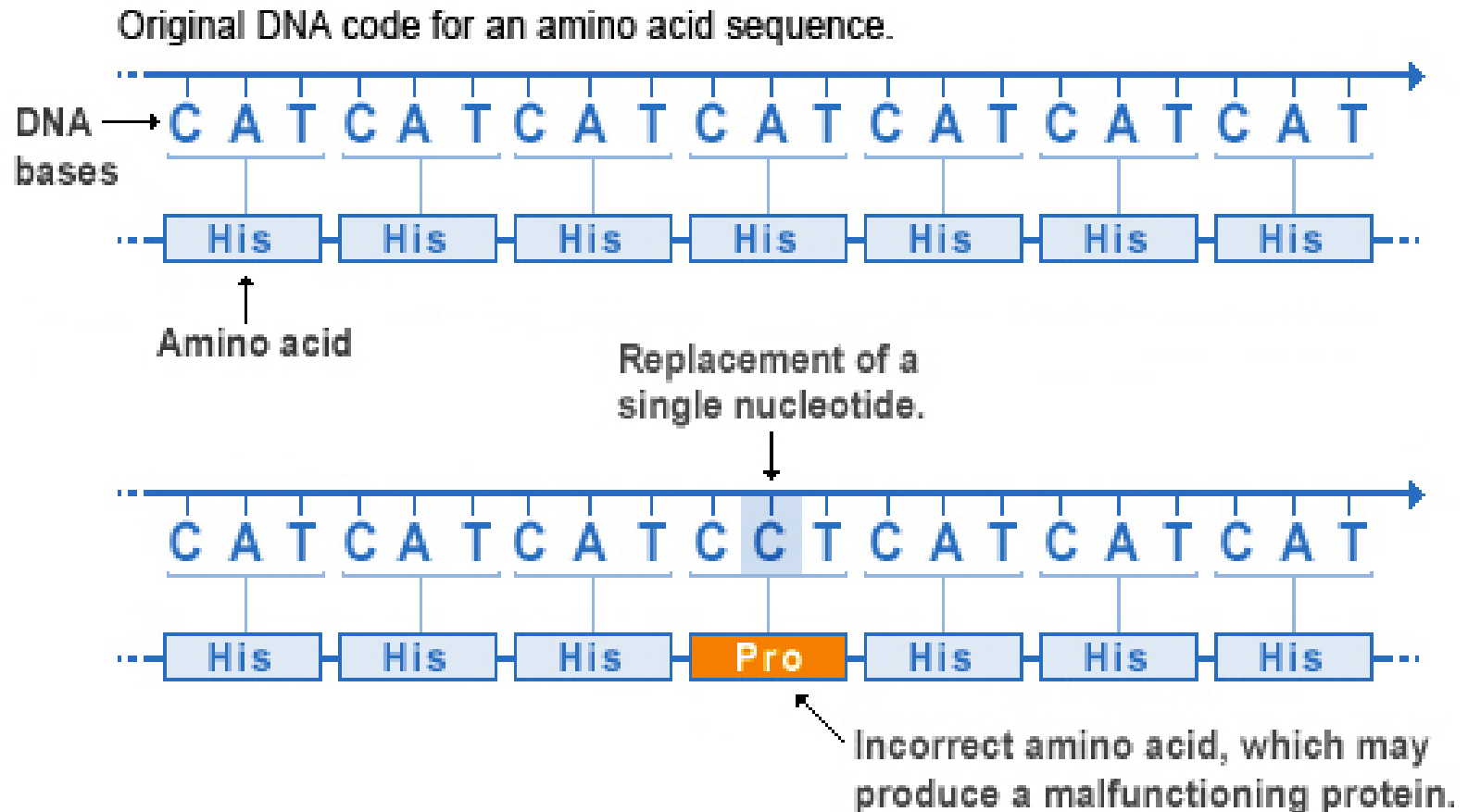
Silent Mutations

ATG	GAA	GCA	CGT
Met	Glu	Ala	Gly

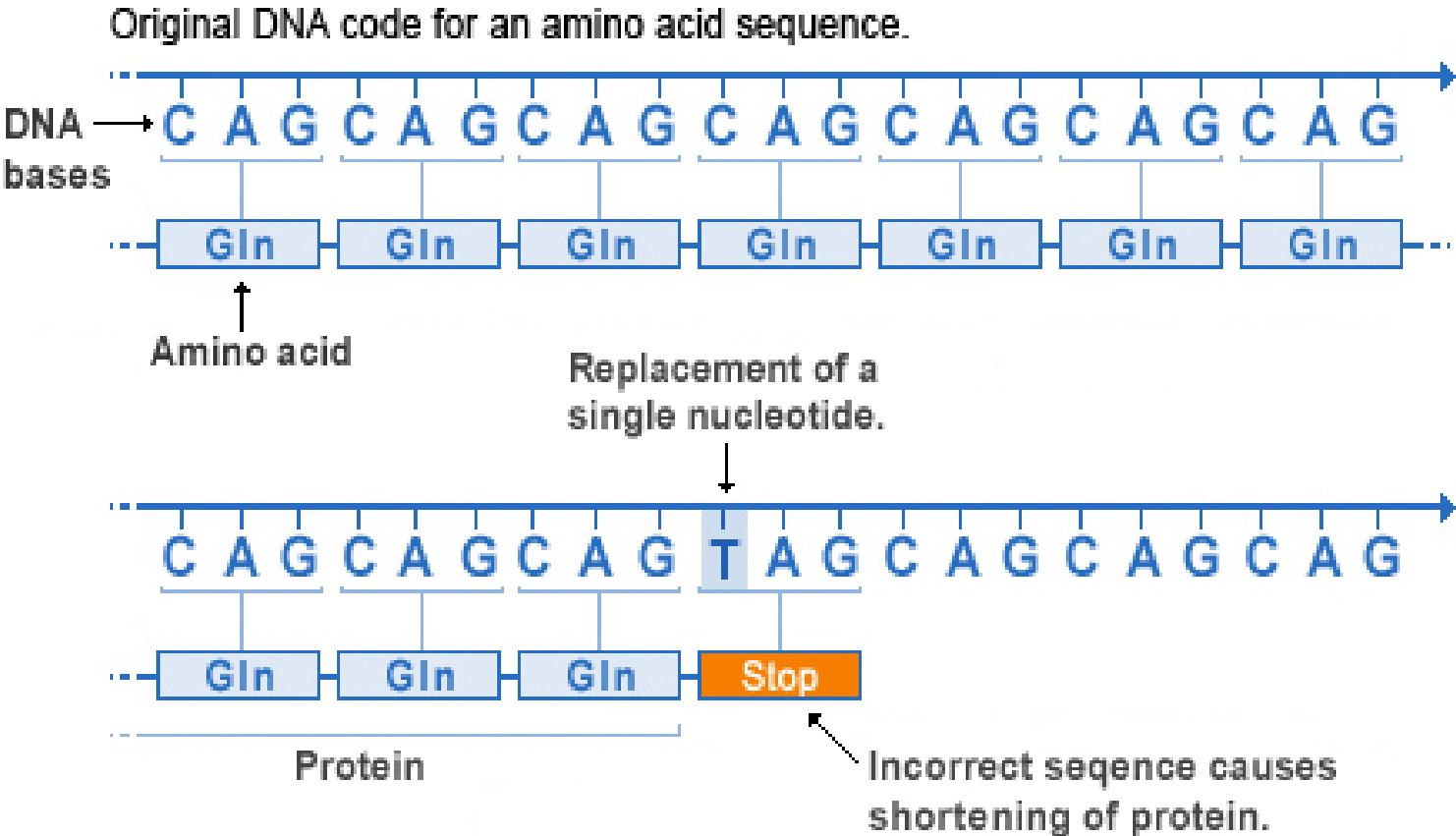


ATG	GAG	GCA	CGT
Met	Glu	Ala	Gly

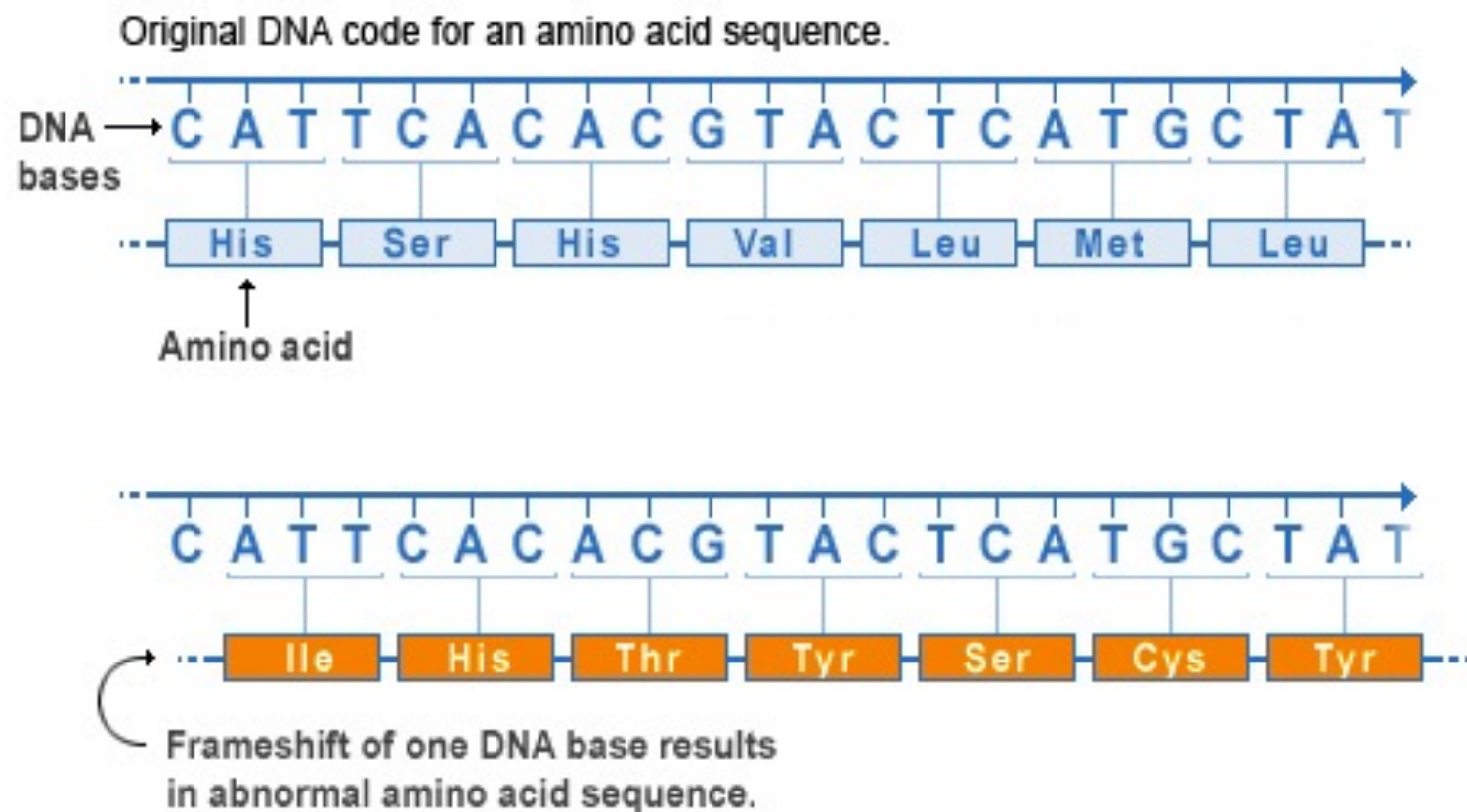
Missense mutation

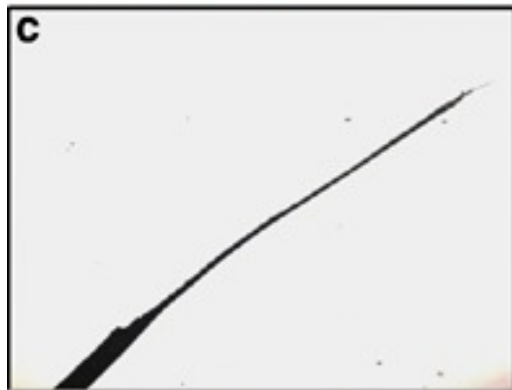


Nonsense mutation



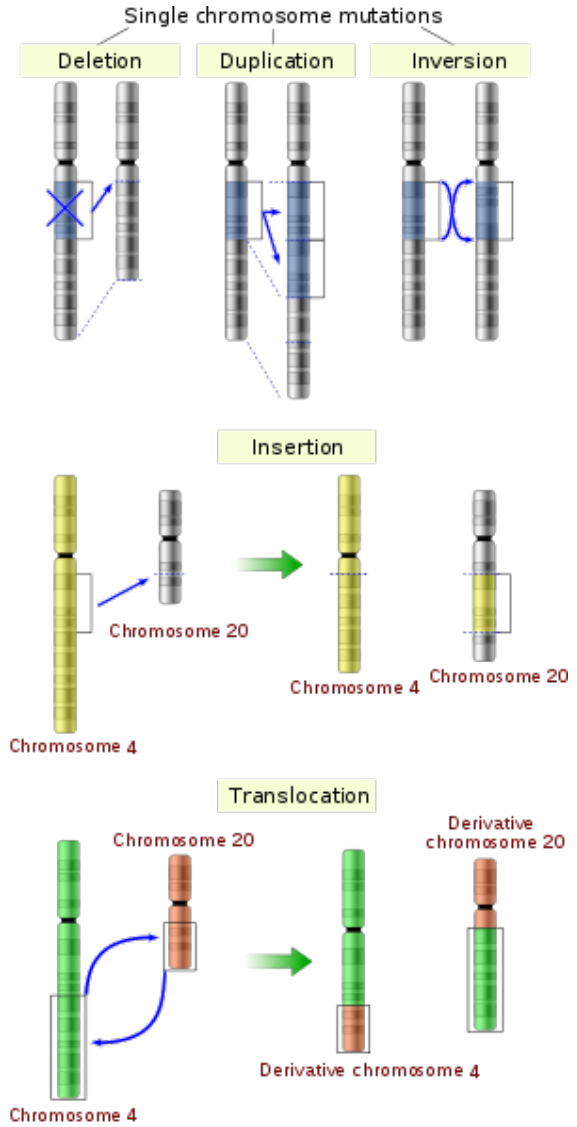
Frameshift mutation

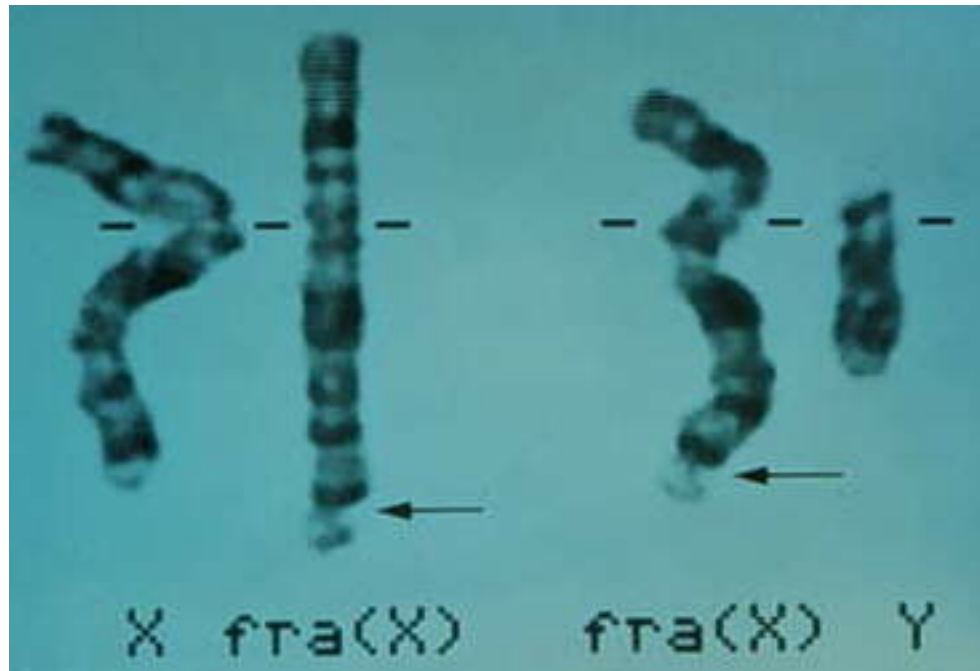




Large-Scale Mutations

- involve multiple nucleotides, entire genes, or whole regions of chromosomes

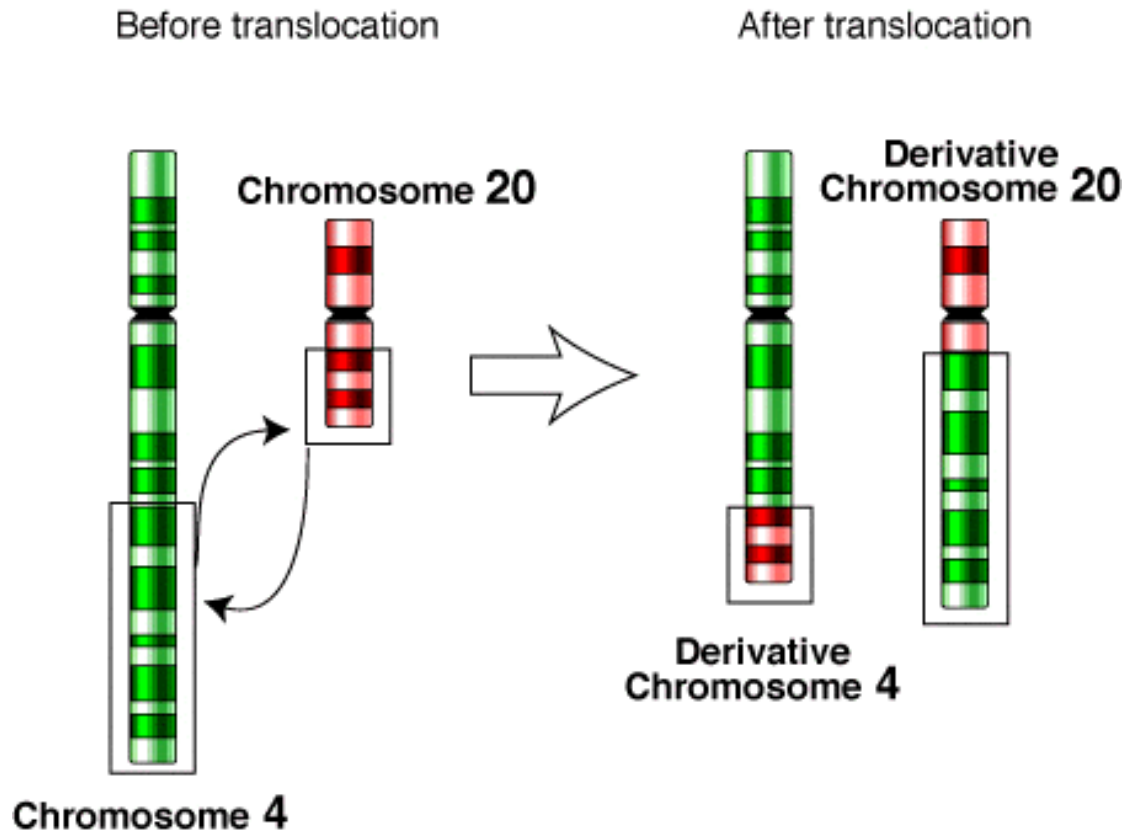




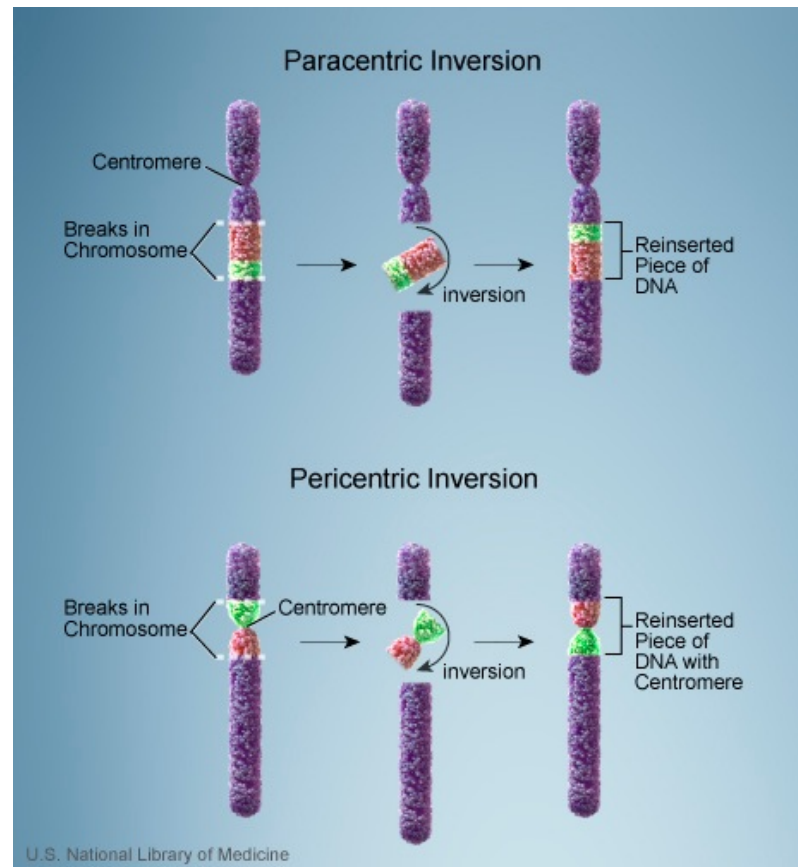
Large-Scale Mutations

- **translocations** are the movement of entire genes or DNA sequences from one chromosome to another
- **inversions**

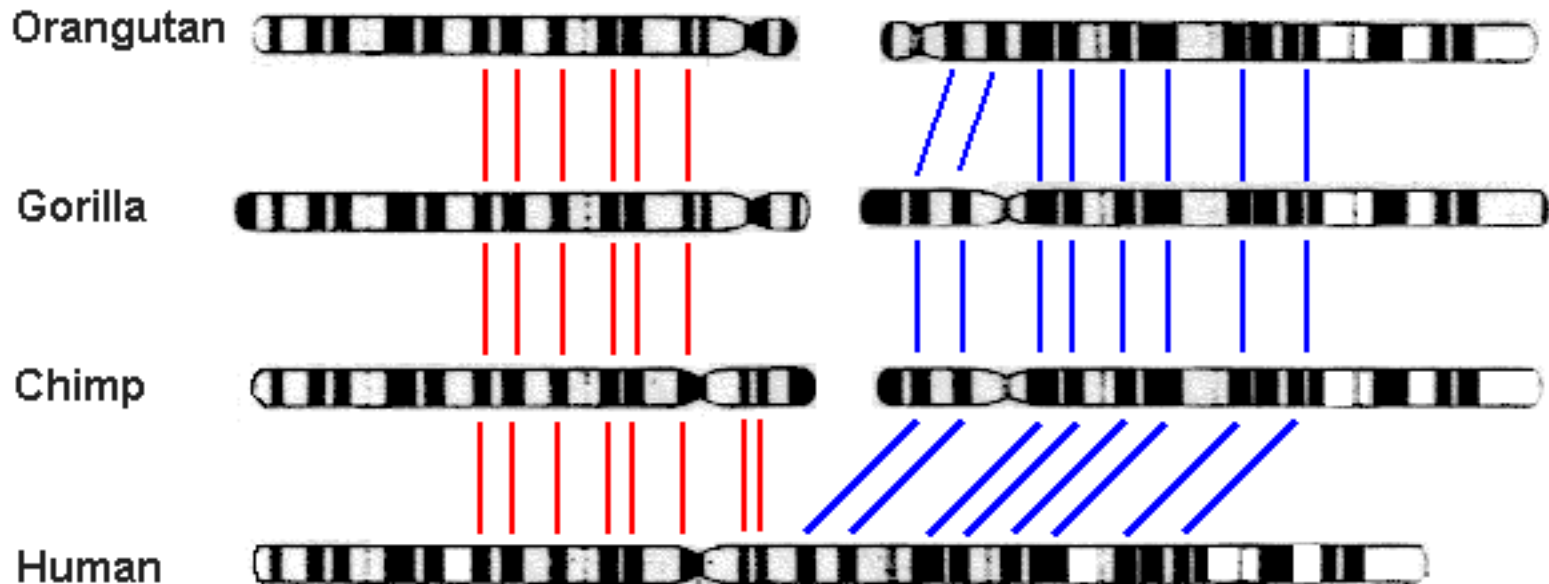
Translocation



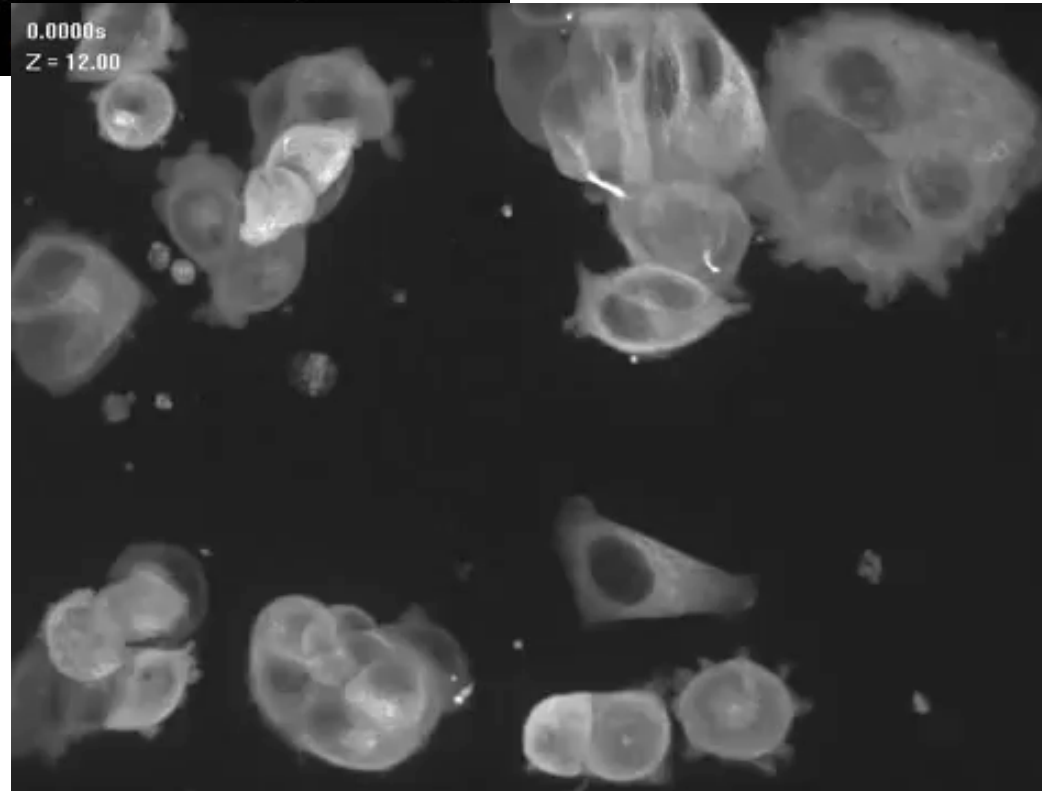
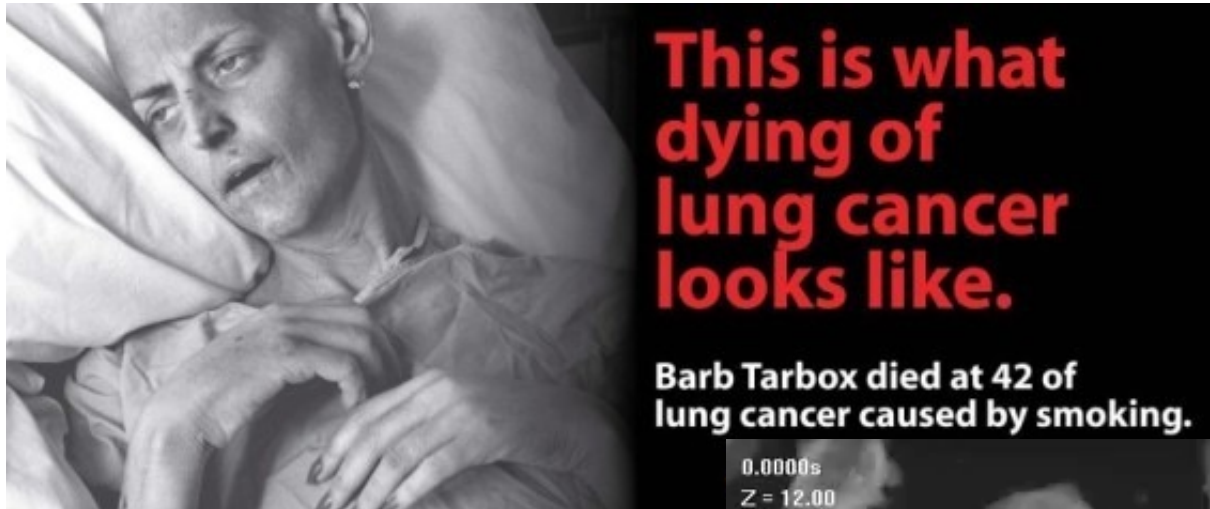
Inversion



» eg. Large scale mutation occurred in the evolution of human chromosome 2. It appears to be the result of a fusion of two smaller chromosomes that appears in our closes genetic relatives



Warning: Mutations



Warning: Mutations



UV exposure

Before

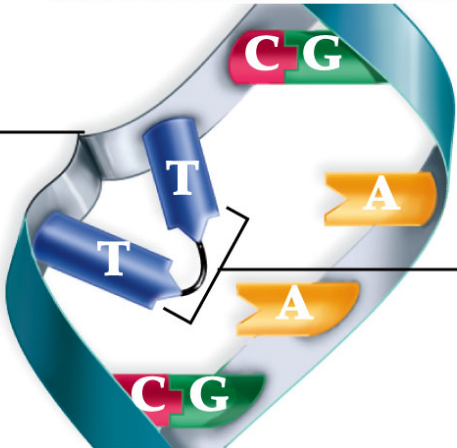


After



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kink



thymine dimer

Causes of Mutations

- **spontaneous mutations** can be caused by errors in replication
- **induced mutations** are caused by environmental agents (**mutagens**)