Mutation
And
Natural Selection

how genomes record a history of mutations and their effects on survival

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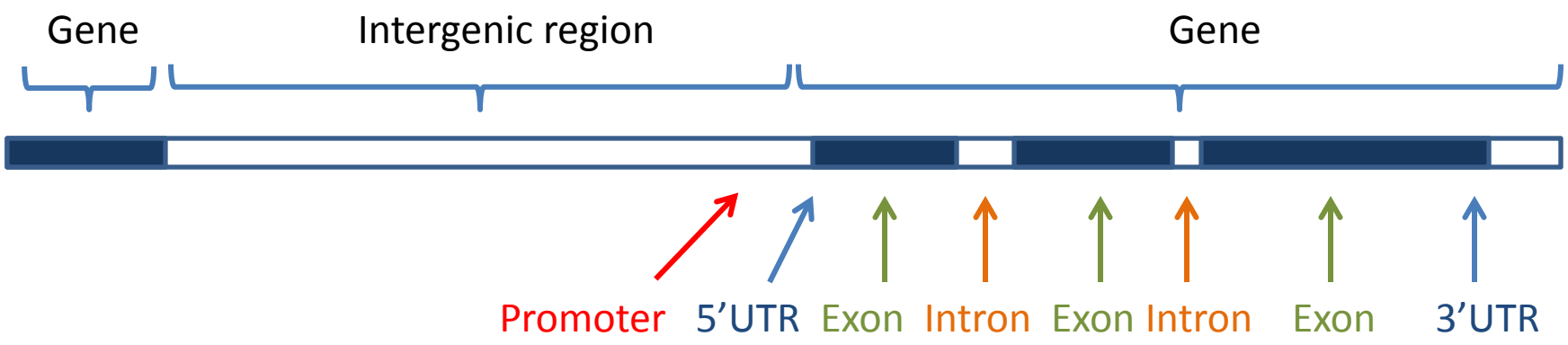
Gene Organization

Coding sequences are used during transcription to make RNAs that are translated into protein or RNAs that have a function (e.g. tRNA, rRNA, snRNA, microRNA). Coding sequences "code" for the production of a molecule. Includes exons of genes.

Noncoding sequences are not used as a code for production of a molecule. Includes promoters, introns, 5' and 3' untranslated regions and intergenic regions. They may have regulatory functions such as controlling gene expression or the half life of RNA. Many are presumed to have no function until a function is determined experimentally.

Gene Organization



DNA sequence classification

Functional sequences

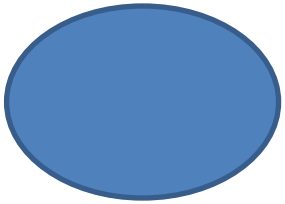
Coding sequences
Promoters
Other regulatory regions
- some 5’ UTRs
- some 3’ UTRs
- some introns
- some intergenic regions

Nonfunctional sequences

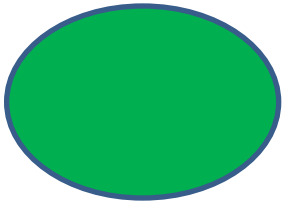
Some 5’ UTRs
Some 3’ UTRs
Some intronic regions
Most intergenic regions
Pseudogenes

Mutations

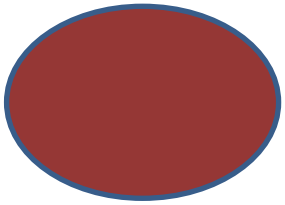
Occur every time DNA is replicated before cell division.
Occur throughout the genome in BOTH nonfunctional and functional sequences.
May be neutral, harmful or beneficial.
The following slides will use this color scheme:



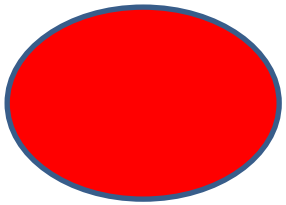
Organism with mutation in nonfunctional region



Organism with beneficial mutation in functional region



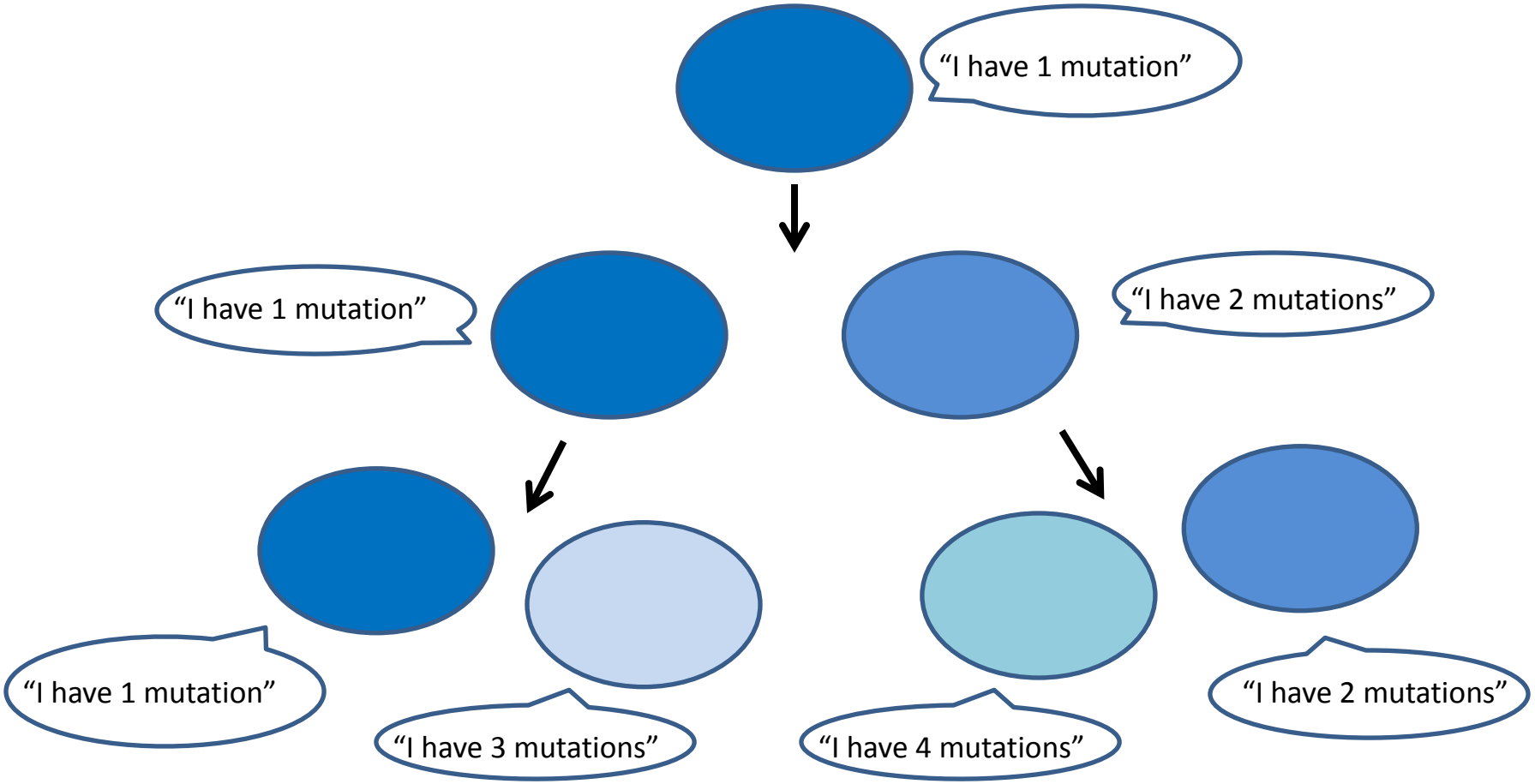
Organism with neutral mutation in functional region



Organism with harmful mutation in functional region

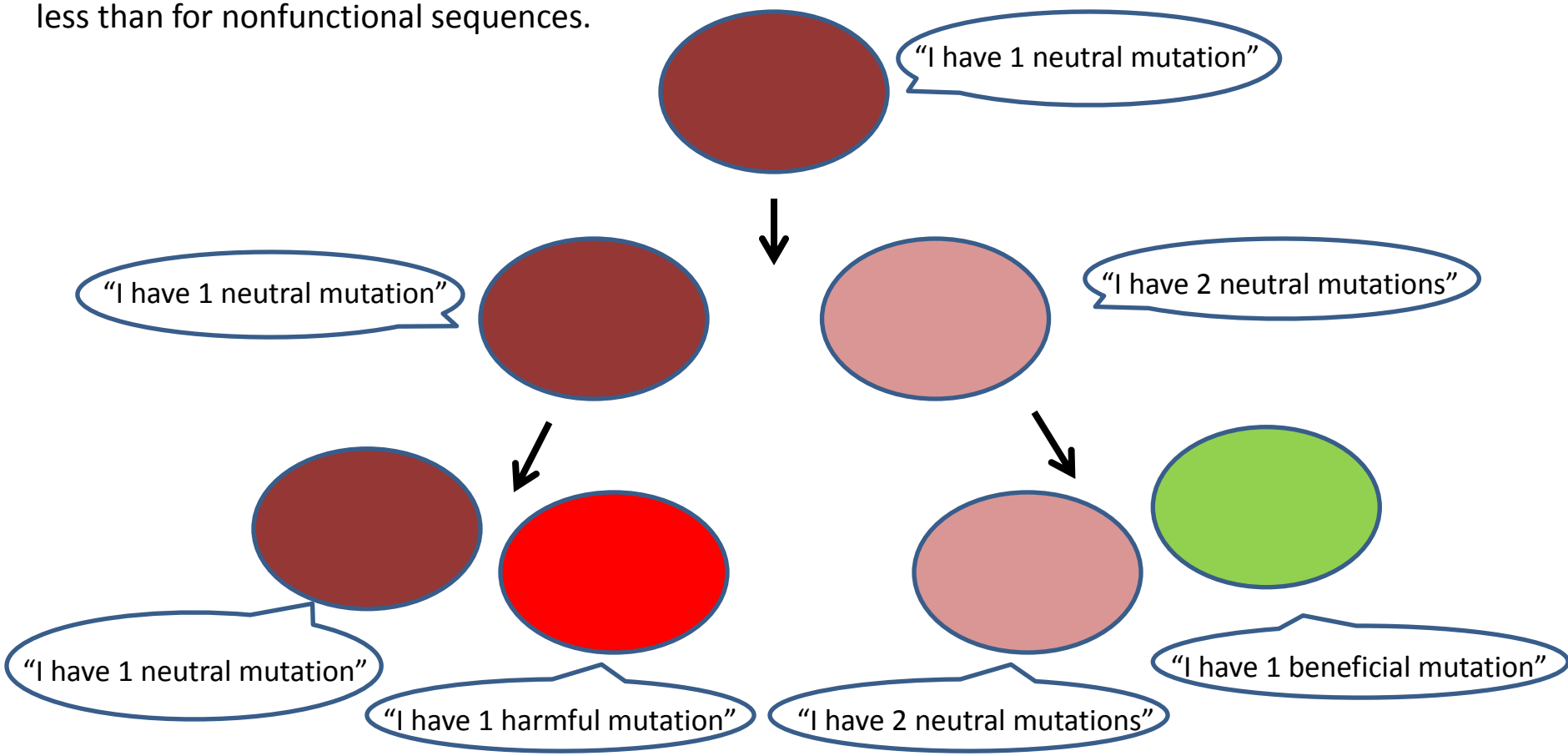
Selection, nonfunctional sequences

Mutations in **nonfunctional sequences** have no effect and organisms survive to pass mutated sequences on to offspring. So all mutations may be considered "neutral". Different shades of blue indicate additional accumulated mutations in nonfunctional regions as generations progress, resulting in highly variable sequences.



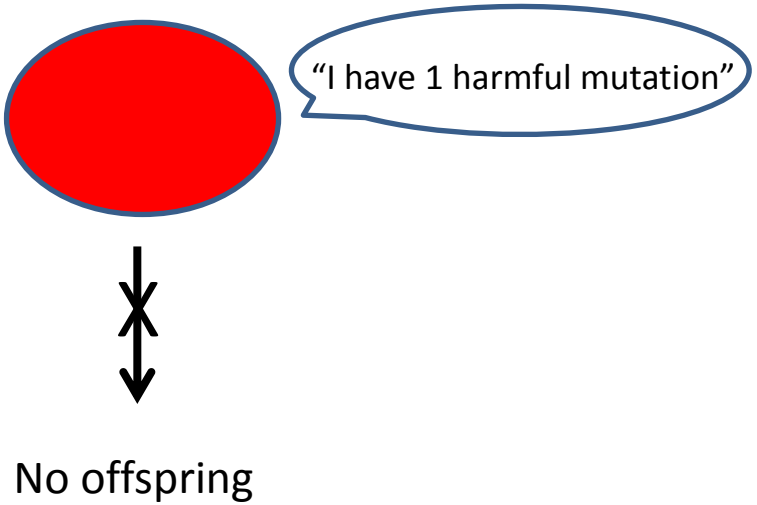
Selection, functional sequences (neutral mutations)

Mutations in **functional sequences** may be neutral, benefical or harmful. Neutral mutations have no effect (e.g. change one nucleotide and do not change the amino acids of a protein), therefore organisms survive to pass neutral mutations on to offspring. Different shades of burgundy/pink indicate additional accumulated neutral mutations, resulting in variable sequences that are often single nucleotide changes. Functional sequences may also acquire harmful (red) or beneficial (green) mutations that affect organisms' survival, so the chances of acquiring accumulated neutral mutations is less than for nonfunctional sequences.



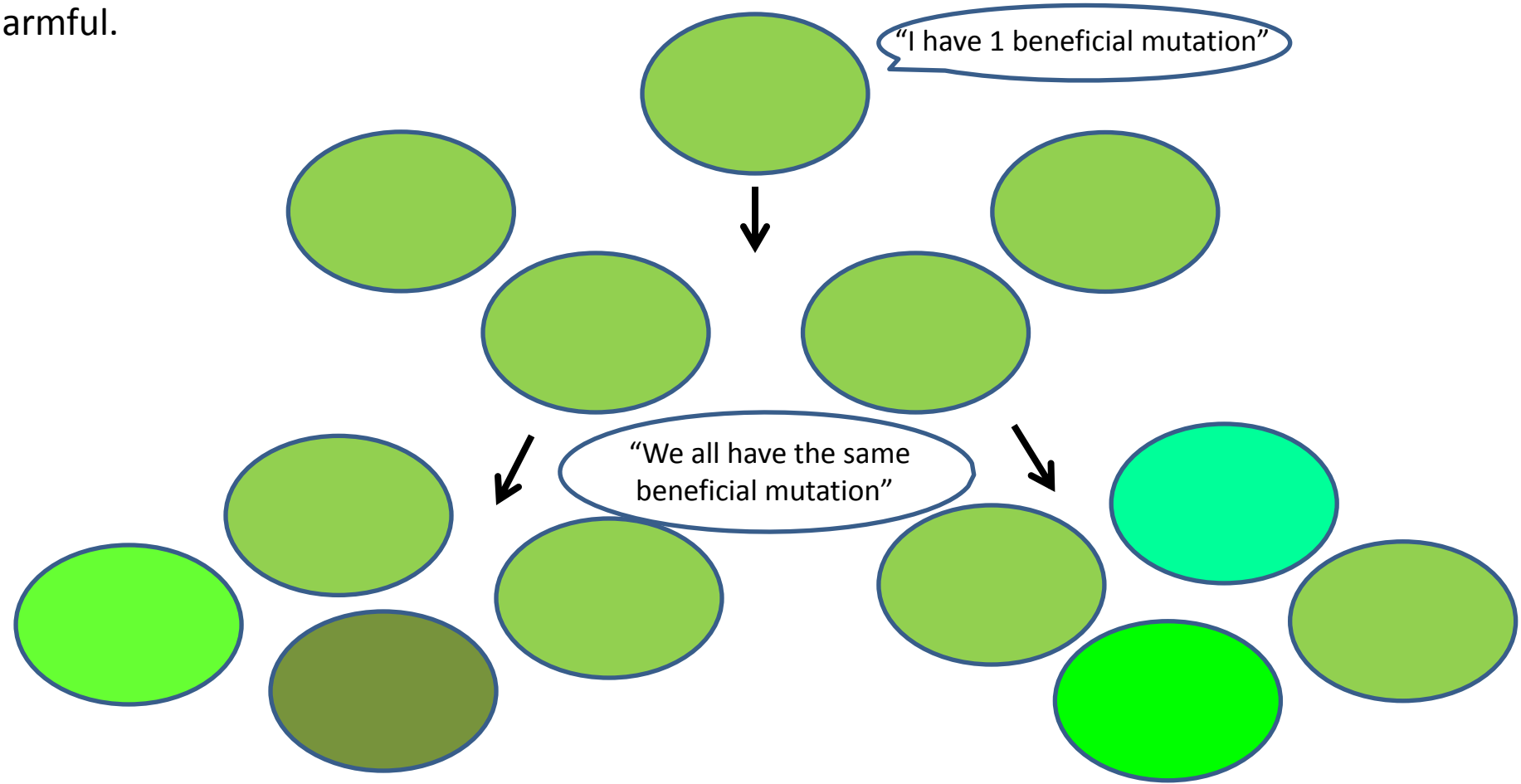
Selection, functional sequences (harmful mutations)

Mutations in **functional sequences** may be neutral, benefical or harmful. Harmful mutations have a negative effect (e.g. change one nucleotide that negatively impacts the structure and function of a protein). Organisms suffer and are less likely to survive to pass the mutations on to offspring. Therefore, harmful mutations often "disappear" from population gene pools.



Selection, functional sequences (beneficial mutations)

Mutations in **functional sequences** may be neutral, benefical or harmful. Benefical mutations have an advantageous effect (e.g. change one nucleotide and improve the structure and function of a protein). Organisms are more likely to survive, reproduce and pass this mutation to offspring. The same shade of green indicates a conserved sequence. Different shades of green indicate additional accumulated mutations that are not harmful.



!!! Summary !!!

Mutation location and type	Effect	Historical Record in Genome
Nonfunctional sequence	none	highly variable sequences
Functional sequence		
neutral	none	moderately variable sequences
harmful	deleterious	rarely observed in the genomes of surviving organisms
beneficial	advantageous	sequences conserved from generation to generation

The effect of selection on the number of surviving organisms carrying harmful (clear), neutral (grey) or beneficial (black) mutations