

The Outcome of Mutation

The table below summarizes the information in **Genetic Variation: The Outcome of Mutation**, available at learn.genetics.utah.edu/content/basics/outcomes/

Trait	Change in DNA	Protein Structure / Function
Thick, wrinkled skin in Shar Pei dogs	An extra "on" switch for the HAS2 gene makes it more active in skin.	<u>Structure</u> : No change. <u>Function</u> : More HAS2 protein is made, which makes more HA. HA makes skin grow more, becoming thick and wrinkled.
Double muscles in cattle	A deletion in the myostatin gene creates a premature stop codon.	<u>Structure</u> : Only a small piece of protein is made. <u>Function</u> : The protein cannot do its job, so muscle cells do not receive the signal to stop growing.
Extra toes in cats	A single base change creates a new switch in the Shh gene. The gene is turned "on" on the opposite side of the foot.	<u>Structure</u> : No change. <u>Function</u> : Protein is made on both sides of the foot, causing the cats to grow extra toes.
Curly hair in dogs	A two-base substitution in the keratin71 gene changes a single amino acid.	<u>Structure</u> : The keratin proteins cannot link together to form fibers. <u>Function</u> : The hair follicle is not supported by fibers, so it is misshapen.
Wrinkled pea seeds	A large insertion disrupts the coding sequence of the SBE1 gene.	<u>Structure</u> : The tail of the SBE1 protein is lost. <u>Function</u> : The protein is disabled. The peas can't make branched starch, so sugar accumulates. Sugar attracts water, causing peas to swell and then wrinkle as they dry.
Disease resistance/ Cystic fibrosis in people	Variations in the CFTR gene change the amino acid sequence.	<u>Structure</u> : The CFTR protein can't fold properly, so it is broken down by the cell. <u>Function</u> : (Disease Resistance) Half as many salt channels are made. Toxins from bacteria activate salt channels, leading to water loss. People with fewer channels have a lesser reaction and are less susceptible to dehydration. (Cystic Fibrosis) No salt channels are made, leading to a chloride imbalance and a thickening of fluid around cells.

Trait	Change in DNA	Protein Structure / Function
Red hair in people	Single-base variations (3 possibilities) in the MC1R gene change the amino acid sequence.	<p><u>Structure</u>: The changes decrease the amount or the activity of MC1R protein.</p> <p><u>Function</u>: Cells do not receive the signal to make black pigments, instead making only red.</p>
Song learning in male zebra finches	<p><i>[This information is not explicitly provided. Two situations are described: knocking out the gene, and (presumably through changes to switches) reducing the amount of protein that is made after the birds hatch.]</i></p>	<p><u>Structure</u>: <i>This information is not explicitly provided, but one could assume that the structure is not affected.</i></p> <p><u>Function</u>: When less FOXP2 protein is made, fewer new connections form between the neurons that perceive sound and those that control mouth movements and store memories. No learning takes place.</p>



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