

# INTRO TO MUTATIONS: GENE MUTATION

1/29/93, rvsd 1/26/94, 1/27/95, 1/28/98, 26 Jan 00, 31 Jan 03, 28 Jan 04, 26Jan05, 1Feb08, 27Jan10, 28Jan11  
 [SGML P 151-GMSLG: p. 178-201, GMSLG 7<sup>th</sup>: 464-487, GWLC, 9<sup>th</sup>: 515-530

Discovery of mutagenesis in 1947 by Auerbach and Robson, working for British military with mustard gas  
 See table 15-6 and 15-7 for list of mutagens (p. 485)

**Variants** are a manifestation of **mutations**.

Two general classes: **gene mutation** point mutations, deletions  
**chromosome mutation** gross alteration in structure or number

Can be used to 1) **genetic dissection** of biological function  
 2) understand **mechanisms of mutagenesis**

**forward mutation:** alteration from wild type to variant

KNOW THESE FOUR TERMS:

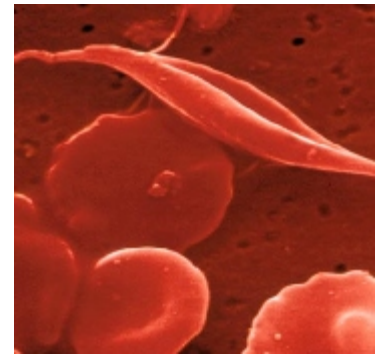
**Mutant** is individual carrying **mutation**, caused by **mutagen**, process **mutagenesis** or mutational event  
 change *back* to the wild type allele is: *reverse mutation* (individual = revertant)  
 or reversion  
 or back mutation

**Somatic mutations** lead to cell clones, the mechanism for carcinogenesis.  
**Germinal mutation** in germ line lead to mutant progeny (inborn error, birth defect)

Location of mutation in gene affects the effect:  
 enzyme's active site, edge of active site, away from active site

**mutation effect on phenotype:** 1) morphological (often affect development)  
 2) conditional (temperature sensitivity)  
 3) lethal (as in Manx cat)

**Conditional:** example **restrictive** **permissive**  
 siamese cat, temp sens pigment synthesis warm cool  
 sickle cell carriers hypoxic adequate oxygenation



Biochemical mutations: for nutritional requirement: from **prototrophic (wild type)** to **auxotrophic** ("help nourish")

**Resistant mutation:** resistance to some poison or inhibitor, usually by altering a preexisting protein - new function

## DETECTION SYSTEMS FOR MUTAGENESIS: (p. 473)

**Tradescantia:** stamen color of anther hair cells in blue with P. pp is pink. line 02 is Pp, mutation in hair cell P gene shows up as pink cell. Can scan millions of cells easily. (P 474)

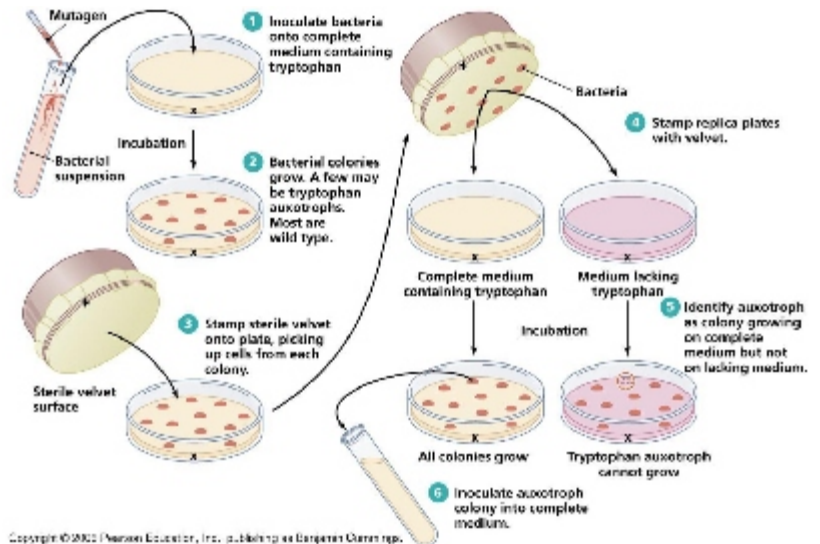
**X-linked recessives** show up immediately in males (hemizygous), screening males easiest, most sensitive test

Mutation **rate:** **mutations/time** usually per generation or 1/x generations  
 Mutation **frequency:** **mutations/population** population size X. (I.e., 1-500/ million)

## Microbial Selective systems:

to select auxotrophs (p 231 p. 477)

Fungi: **Filtration enrichment**  
 Penicillin enrichment: **replicate plating** on minimal vs enriched medium (p 519)



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