Mechanics of mutation

Types of mutation

Mutation: Ultimate source of variation

Nucleotide sequence in a gene changes, so allele is changed

- Point mutations: change nucleotides
- Block mutations: change segments of chromosome

Purines

- Adenine
- Guanine

Pyrimidines

- Cytosine
- Thymine

Point mutations: Substitution (2 types)

Transition: purine to purine, or pyrimidine to pyrimidine

- $A \rightarrow G$ or $G \rightarrow A$
- $C \rightarrow T$ or $T \rightarrow C$

Transversion: purine to pyrimidine, or vice versa

Point mutations: Substitution (2 types)

Transition: purine to purine, or pyrimidine to pyrimidine

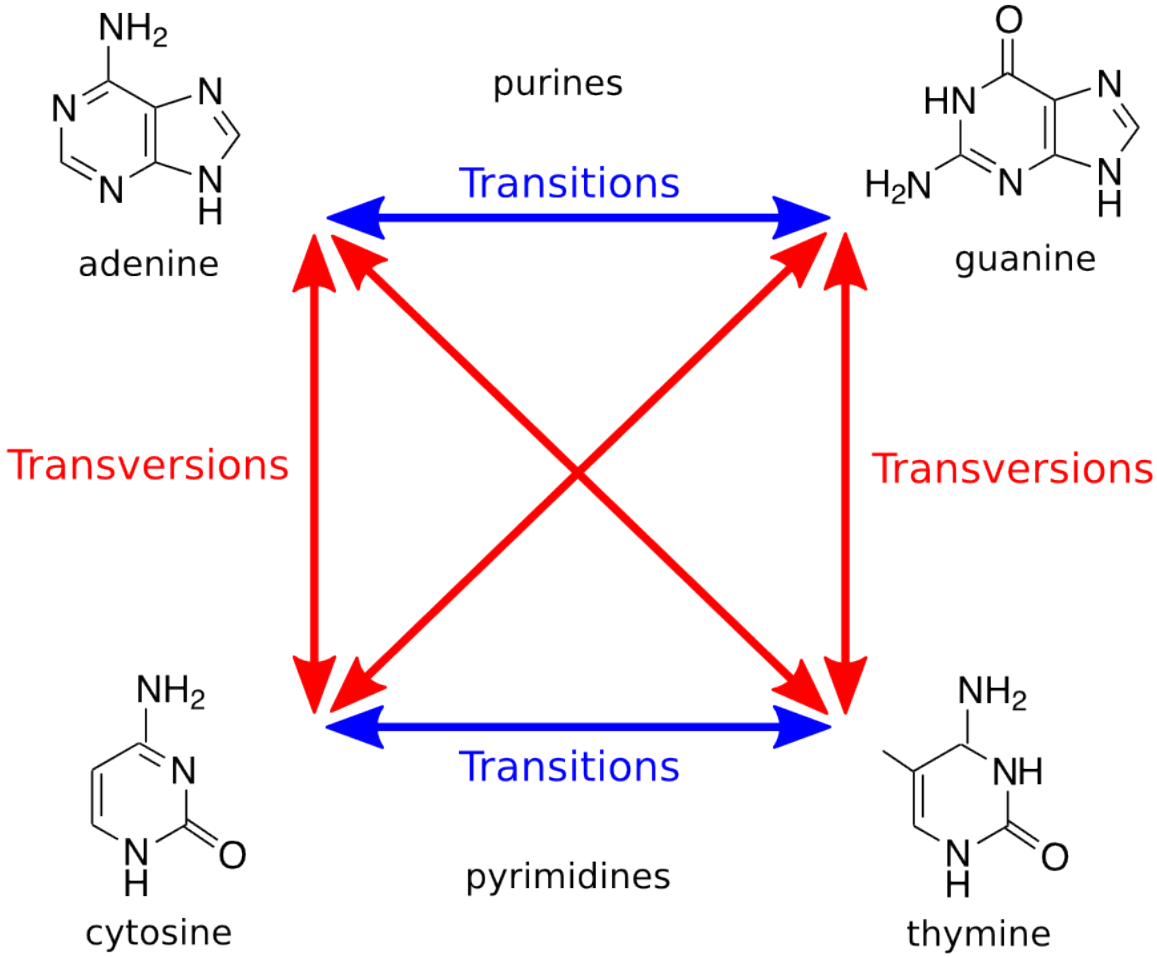
- $A \rightarrow G$ or $G \rightarrow A$
- $C \rightarrow T$ or $T \rightarrow C$

Transversion: purine to pyrimidine, or vice versa

Do or do not change amino acid sequence

- **Synonymous**: Do not change sequence
- **Non-synonymous**: Do change sequence

Point mutations: transitions and translations



[1] Image: Public Domain

BEAST

Sequence	Type
FEAST	Substitution

BEAST

Sequence	**Type**
FEAST	Substitution
B**R**EAST	Insertion

BEAST

Sequence	Type
FEAST	Substitution
B**R**EAST	Insertion
BEST	Deletion

BEAST

Sequence	Type
FEAST	Substitution
B**R**EAST	Insertion
BEST	Deletion
BEA**TS**	Inversion

Point mutations: types

CTAGC

Sequence	Type
GTAGC	Substitution
C**C**TAGC	Insertion
CAGC	Deletion
CTA**CG**	Inversion

Block mutations

- Changes large chunks of DNA
- Often caused by **transposable elements**
- Transposons are sections of DNA that can change position within the genome[1,2]
  - Selection within a genome[3]
  - Selfish genetic elements

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[1]McClintock, B. (1950). The origin and behavior of mutable loci in maize. *Proceedings of the National Academy of Sciences*, 36:344-355.

[2]Ravindran, S. (2012). Barbara McClintock and the discovery of jumping genes. *Proceedings of the National Academy of Sciences*, 109:20198-20199.

[3]Burt, A., & Trivers, R. (2006). *Genes in conflict: the biology of selfish genetic elements*. Harvard University Press.

Mutation rates vary by organism and locus

Species	**Tissue**	**Mutation rate**
Humans	Germline	0.06
	Retina	0.99
	Intestine	0.27
Mice	Germline	0.96
Fruit flies	Germline	0.13
Escherichia coli		0.26

Mutation rate here is mutations per cell division.

[1] Lynch, M. (2010). Evolution of the mutation rate. *TRENDS in Genetics*, 26:345-352.

Effects of mutations

- Most mutations are mildly deleterious
- Some mutations are strongly deleterious
- Very few mutations are beneficial

Effects of mutations

- Not all types of mutation are equally likely
- Not all loci are equally mutable
- Transitions more likely than transversions

Karyotype alterations

[1] Image: Public Domain

Karyotype alterations

- Change in chromosomes
- Can cause reproductive incompatibility and speciation
- **Autopolyploid**: Multiples of same genome
- **Allopolyploid**: Hybrids of 2 whole genomes

Karyotype alterations: Polyploidisation

- Polyploidisation adds complete sets of chromosomes
- One of the most dramatic mutations
- Occurs in many groups of eukaryotes[1,2] (especially flowering plants)

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[1] Otto, S. P. (2007). The evolutionary consequences of polyploidy. *Cell* 131:452-462.

[2] Logsdon, J., et al. (2017). A very recent whole genome duplication in *Potamopyrgus antipodarum* predates multiple origins of asexuality & associated polyploidy. *PeerJ preprints*. https://peerj.com/preprints/3046/