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# Lecture – 11

## Mutation

by

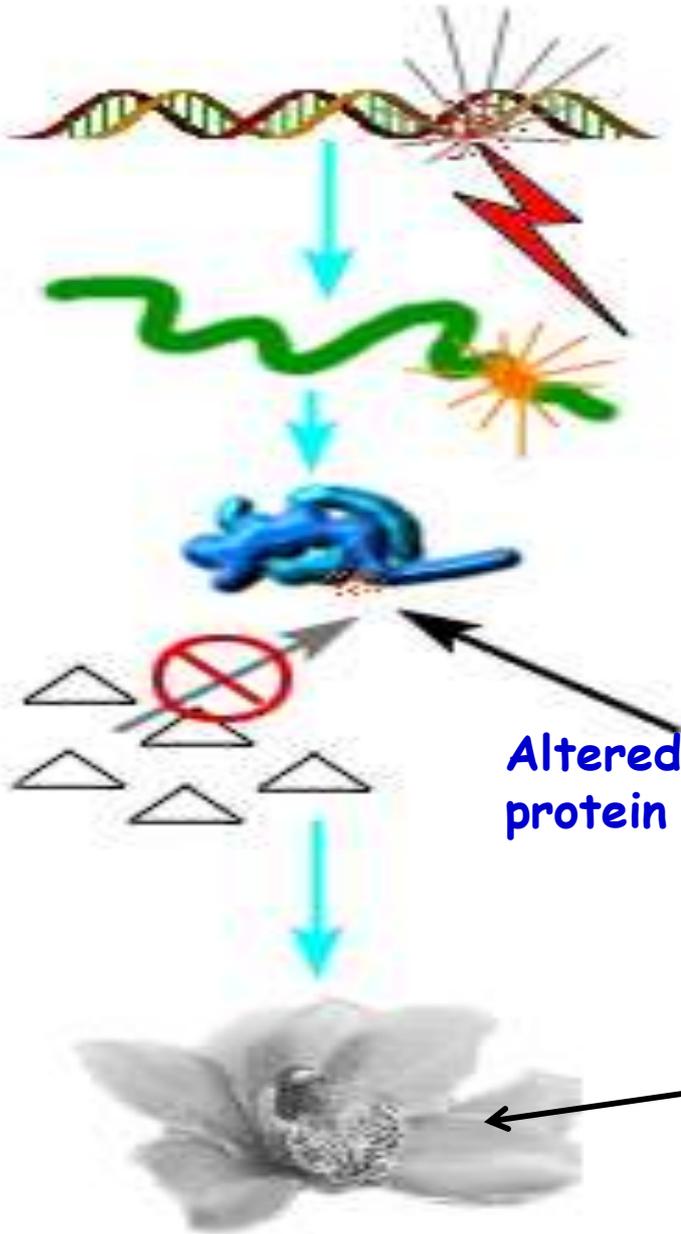
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# Definitions



- sudden heritable change in genetic material, that is not due to recombination and segregation. Which includes chromosomal aberration as well as change in individual gene or even a base pair.
- **Mutations** are changes in individual gene.
- **Mutagen:** Any substance that can bring about the mutation.
- **Mutant:** The organism exhibiting a novel phenotype as a result of mutation.



# Historical Background

- ❖ Term mutation introduced by **Hugo de Veris (1900)** while studying evening primrose (*Oenothera lamarckiana*)
- ❖ But the earliest record of mutations dates to 1791 when S Wright noticed a male lamb with unusually short legs in his flock of sheep. This lamb served as a source of short leg trait for the development of Ancon breed of sheep.
- ❖ Systematic studies on mutation began in 1910 with the discovery of **white eye mutants of *Drosophola* by T. H. Morgan.**
- ❖ Mutagenic action of X-rays discovered by **H. J. mulier (1927) in *Drosophola*.**
- ❖ **Stadler (1929)** described the mutagenic effect of X-rays in barley.
- ❖ Mutagenic effect of mustard gas and some other chemical compound discovered by **Auerbach and Robson in 1946 in *Drosophila*.**



# Characteristics of mutation

- 1) Mutations are mostly **recessive** and very rarely dominant e.g. epiloia in human, *Notch* wing in *Drosophila* etc.
- 2) Most mutations have **harmful effects** and very few (less than 0.1 %) are beneficial.
- 3) Mutations are **random event** in term of time of their occurrence and the gene in which they occur.
- 4) Mutations may be due to a change in a gene, a group of genes or in entire chromosome.
- 5) If gene mutations are not lethal, the mutant individuals may survive. However, chromosomal mutations are generally lethal and such mutants do not survive.
- 6) If mutation occur at both loci simultaneously, the mutants can be identified in  $M_1$  generation. However, if it is restricted to one locus only, (dominant to recessive) the effect can be seen only in  $M_2$  generation.

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- 7) **Macro-mutations** are visible and can be easily identified, while **micro-mutations** cannot be seen with naked eye and need special statistical tests (or statistical analysis).
- 8) Many of the mutants **show sterility**.
- 9) Most mutants are of **negative selection value**.
- 10) Mutations are random i.e. they can occur in any tissue or cell of an organism. However, some genes show higher mutation rate than others.
- 11) Mutations **can be sectorial**. The branches arising from mutated sector show mutant characters.
- 12) Mutations are **recurrent** i.e. the same mutation may occur again and again.
- 13) Induced mutations commonly show pleiotropy often due mutation in closely linked genes.
- 14) Mutations occur in both **forward** (from wild type allele to mutant allele) and **reverse** (from mutant allele to wild type allele) direction. Generally, the rate of forward mutations are much higher than those for reverse mutations



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# Terminology

**Muton:** The smallest unit of gene capable of undergoing mutation and it is represented by a nucleotide.

**Mutator gene:** A gene which causes another gene or genes to undergo spontaneous mutation.

**Mutable genes:** Genes which show very high rates of mutation as compared to other genes.

**Mutant:** An organism or cell showing a mutant phenotype due to mutant allele of a gene. **Mutagen:** A physical or chemical agent which induces mutation.

**Hot spots:** Highly mutable sites within a gene.

**Gene mutations or point mutations:** The changes which alter the chemical structure of a gene at molecular level.



# Classification of mutations:

## 1. Based on direction of mutations:

a) *Forward mutation*: Any change from wild type allele to mutant allele

b) *Backward mutation or reverse mutation*: A change from mutant allele to wild type

## 2. Based on source / cause of mutations:

a) *Spontaneous mutation*: Mutation that occur naturally

b) *Induced mutation*: Mutation that originates in response to mutagenic treatment

## 3. Based on tissue of origin:

a) *Somatic mutation*: A mutation in somatic tissue

b) *Germinal mutation*: A mutation in germ cells or in reproductive tissues

## 4. Based on trait or character effected:

a) *Morphological mutation*: A mutation that alters the morphological features of an individual.

b) *Biochemical mutation*: A mutation that alters the biochemical function of an individual.



**5. Based on effect on survival:**

- a) **Lethal mutation:** Mutation which kills the individual that carries it. (survival 0%)
- b) **Sub-lethal mutation:** When mortality is more than 50% of individuals that carry mutation
- c) **Sub-vital mutation:** When mortality is less than 50% of individual that carry mutation.
- d) **Vital mutation:** When all the mutant individuals survive (survival-100%)

**6. Based on visibility or quantum of morphological effect produced:**

- a) **Macro-mutations:** Produce a distinct morphological change in phenotype (which can be detected easily without any confusion due to environmental effects). Generally found in qualitative characters. Eg : colour of flowers, height of plant etc.
- b) **Micro-mutations:** Mutations with invisible phenotypic changes, (which can be easily confused with effects produced due to environment). Generally observed in quantitative characters.



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## 7. Based on the site of mutation or on cytological basis:

a) **Chromosomal mutations:** Mutations associated with detectable changes in either chromosome number or structure.

b) **Gene or point mutations:** Mutations produced by alterations in base sequences of concerned genes.

c) **Cytoplasmic mutations:** Mutations associated with the changes in chloroplast DNA (cpDNA) and mitochondrial DNA (mtDNA).

## 8. Molecular basis: point or gene mutations can be subdivided as

(a) **Base substitution** (b) **Deletion** (c) **Addition** (d) **Transposition**

(e) **Trinucleotide expansion**

## 9. Based on type of Amino Acid replacement in the polypeptide

(a) **Missense**

(b) **Nonsense**

(c) **Frame-shift**



## 10. Effect on the expression of neighbouring genes:

a) ***Polar mutations***: some gene mutations suppress the expression of other genes located next to them on one side (downstream side).

b) ***Nonpolar mutations***: affect the expression of only those genes within they are located.

## 11. Intensity of characters expression:

a) ***Amorphic mutations***: almost total loss of the expression of the affected phenotype as they produce totally non-functional gene products.

b) ***Hypomorphic mutations***: when a mutation leads to partial loss in the expression of a trait.

c) ***Hypermorphic mutation***: the mutant alleles lead to an increased expression than that of wild type allele,  $r^{\text{II}}$  mutants of phage  $T_4$  of *E. coli* (in terms of plaque size).



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## Molecular basis of mutations:

- ❖ The term mutation is presently used to cover only those changes which alter the chemical structure of the gene at molecular level. Such changes are commonly referred to as “**point mutations**”.
- ❖ Point mutations involve a change in the base sequence of a gene which results in the production of a mutant phenotype.
- ❖ Point mutations can be subdivided into the following three classes based on molecular change associated with them.

1. Base substitution
2. Base deletion
3. Base addition



# MOLECULAR BASIS OF MUTATION

## 1. Base Substitution:

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When a single base in a DNA molecule is replaced by another base it is known as base substitution. This can be of two types.

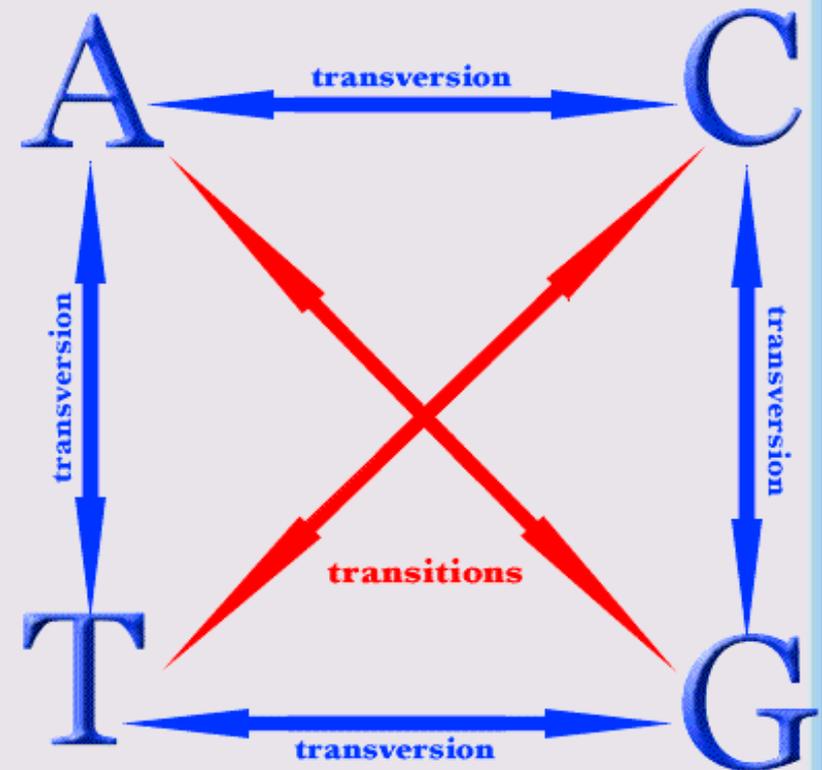
**Transition:** Replacement of a purine by another purine or a pyrimidine by another pyrimidine.

**Transversion:** Replacement of a purine by a pyrimidine and vice versa.

❖ Transitions are actually more common than transversion.

❖ Main cause of base substitution is

**TAUTOMERIC SHIFT**



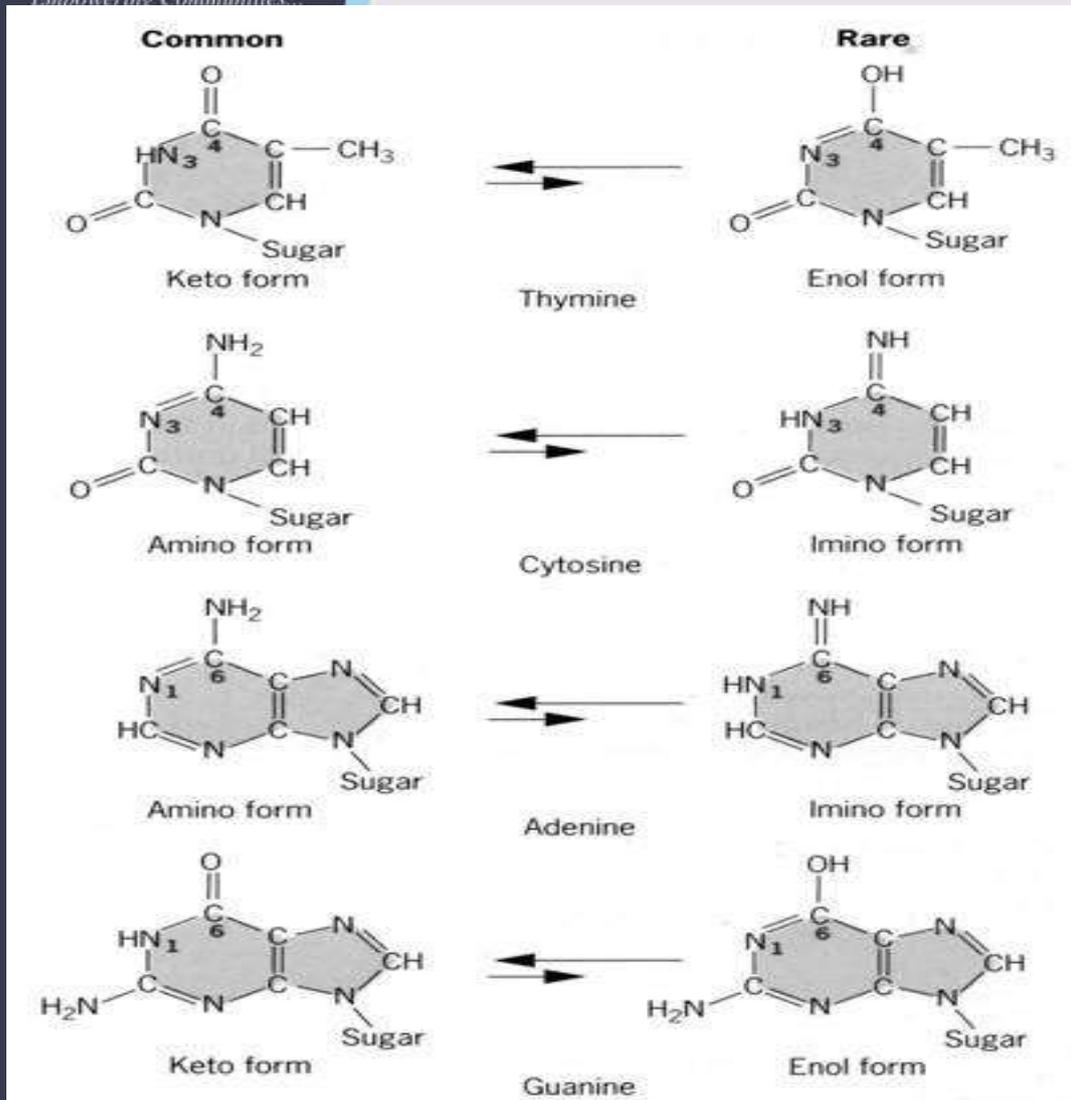
4 different types of transition and 8 different types of transversion is possible



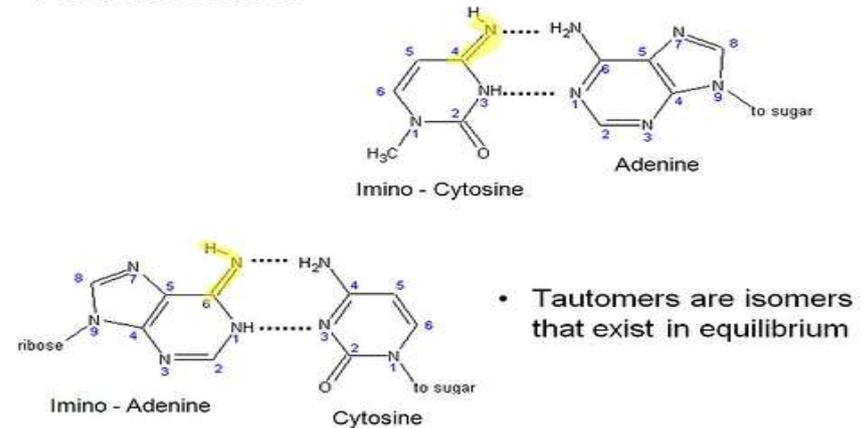
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# Tautomerisation:

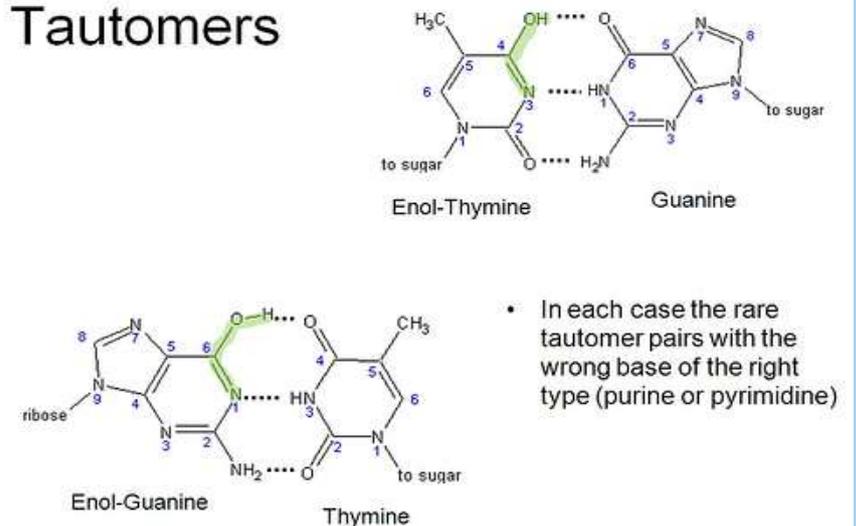
is the shifting of hydrogen (H) from one position in a purine or pyrimidine ring to other position



## Tautomers



## Tautomers





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# A base substitution may causes Neutral, Sense (silent), Missense or Nonsense mutation

normal

AUG GCC TGC AAA CGC TGG  
met ala cys lys arg trp

silent

↓  
AUG GCT TGC AAA CGC TGG  
met ala cys lys arg trp

nonsense

↓  
AUG GCC TGA AAA CGC TGG  
met ala --- --- --- ---

missense

↓  
AUG GCC GGC AAA CGC TGG  
met ala arg lys arg trp

## Neutral mutation

A change in a base pair results in an amino acid change but the new amino acid has the same chemical properties as the old amino acid.



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## 2. Base addition and deletion

- Addition or loss of bases in multiple of three, added or delete one to several amino acids from concerned protein, which may or may not give profound effect on activity of polypeptide.
- If insertion or deletion of not in multiple of 3, the base sequence of all the codon beyond the point of insertion or deletion shifted which causes **Frame-shift mutation**.

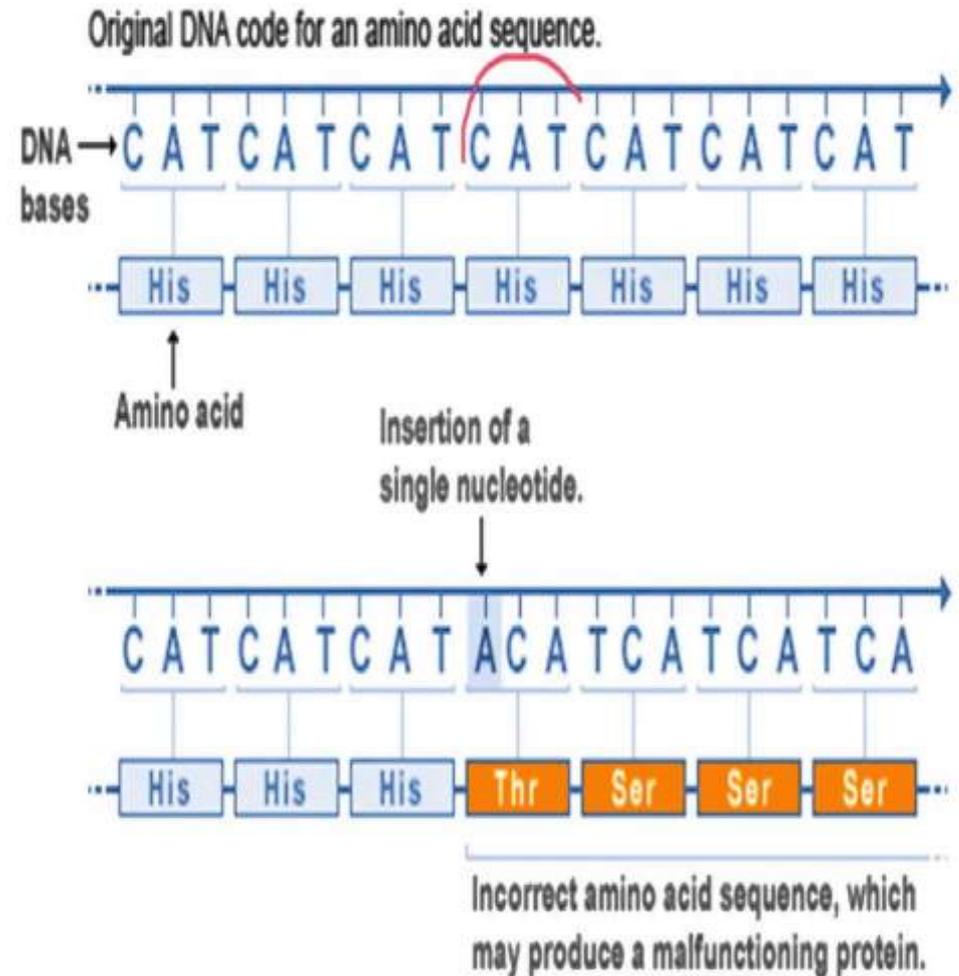


# Frameshift mutations:

## Frameshift Mutation

ATG	GAA	GCA	CGT
Met	Glu	Ala	Gly

ATG	AAG	CAC	GT
Met	Lys	His	





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# Spontaneous mutations:

Spontaneous mutations occur naturally without any apparent cause. There are two possible sources of origin of these mutations.

1. Due to error during DNA replication.
  2. Due to mutagenic effect of natural environment Eg. UV rays from sunlight
- ❖ The rate of spontaneous mutations is very low. 1 in 10 lakhs i.e.  $10^{-6}$ .

## Varieties developed through spontaneous mutations

Sr. no.	Crop	Variety
1	Rice	GEB-24, Dee-Geo-Woo-Gen
2	Wheat	Norin
3	Groundnut	TMV-10
4	Sorghum	Co-4 (coimbatore 4)



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# Induced mutations:

Mutations can be induced artificially through treatment with either **physical or chemical mutagens**. The exploitation of induced mutations for crop improvement is called **mutation breeding**.

## Examples of popular induced mutants in crop plants

Sr. no.	Crop	Mutant variety	Parent	Mutagen
1	Rice	Jagannath,	T-141	X-rays
		Mahsuri mutant	Mahsuri	$\gamma$ -rays
2	Wheat	Sharbati sonara	Sonara 64	UV rays
		NP-836	NP-799	x rays
3	French Beans	Pusa Parvati	Wax podded	x-rays
		Pusa Lal Meeruti	Meeruti	x-rays
4	Tomato	S-12	Sioux	$\gamma$ -rays
5	Castor	Aruna	HC-6	Thermal neutrons
6	. Cotton	MCU 7	1143 EE	x-rays
		MCU 10	MCU 4	$\gamma$ -rays



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