

Terminology

- **GENES**: Segments of DNA [That code for characteristics]
↳ Eg. AGCTA
CTTAA ↳ eg. Genes that code for eye color
- **PHENOTYPE**: Observable/Physically Expressed traits
↳ Eg. Blue eyes, yellow flowers
- **ALLELES**: Different variations of the same gene
↳ Eg. B, b

Biology Refreshers

- 1) DNA is DOUBLE-STRANDED → Two strands twist together to form the iconic helix shape
↳ hence why DNA sequences are written on two lines
- 2) DNA building blocks are one of A G C or T.
That's why these are the only four letters you'll see in DNA sequences

NOTES

- 1) **Mutations** are the source of new alleles. They aren't inherently bad.
 - Some are benign → Eg. Blue or Green eyes
 - Other 'mutations' or alleles are less benign or even harmful → Eg. Lactose-intolerance or Cystic FibrosisThe line between what's 'normal' and what isn't is fuzzy for benign variations.
Less so for non-benign mutations.
- 2) In humans, often a combination of two alleles determine the phenotype of a gene because we get one allele from each parent.
or
genotype
But genes can still have over two alleles (hence why there are more than 2 or 4 eye colors)

Chromatids are just really well packed DNA strands

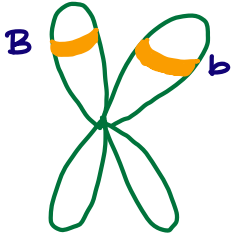


This is a **chromatid**.
It makes up $\frac{1}{2}$ of a chromosome.
You get one from each parent.
Humans have 23 chromosomes



← This is a gene on a chromatid

It contains info
about which **allele** of a gene is encoded
Eg. B

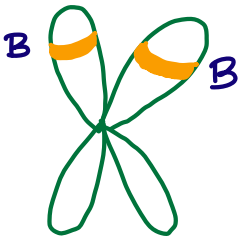


Chromosome

All together, this person has the
allele combination (called the **GENOTYPE**) Bb.

If the **gene** in question is responsible for hair color
and B is the **allele** for brown hair and
b is the **allele** for blond hair, then this individual (maybe)
has the **phenotype** dirty-blond hair.

Compared to :



This individual has the **genotype** BB and has the **phenotype** brown hair
for the **SAME gene**

Both individuals carry the **SAME gene**,
but with **different variations**.

Eg. If you buy a blue bag and a friend buys a green bag,

you're both buying bags (i.e have the same genes),

but they're in different colors (have different phenotypes/are
variations of the gene)

Note : The hair gene could have other alleles in addition to B & b

Mutations: Think of these as 'harmful' or 'abnormal' alleles.

Eg. Let's start from the top: *Lactase is an enzyme that lets you digest lactose

[DNA SEGMENTS] GENE

Allele 1

AGACTTA
TCTGAAT

Allele 2

AGACATA
TCTGTAT

* Note the slight variation in DNA sequences. Here, Allele 2 is a 'mutated' form of the 'normal' Allele 1 *

* The ID given to this gene is its rsID *

GENE PURPOSE

↓
Controlling Lactase
→ Production

↓
Codes for the sufficient
production of lactase

↓
Responsible for the
digestion of lactose

ALLELE

m

↓
Codes for the insufficient
production of lactase

↓
Responsible for the inability to
digest lactose (lactose intolerance)

M

PERSON A

m

PARENT 1 ALLELE

m

PARENT 2 ALLELE

mm

GENOTYPE

can consume lactose

None

PHENOTYPE

'DISEASE'

PERSON B

M

M

MM

indigestion after consuming lactose

Lactose intolerance

↑
I don't know if lactose
intolerance counts as a
'disease' but the same
logic applies to diseases

* didn't deal with Mm genotypes
since it requires explanation of
unnecessary terminology