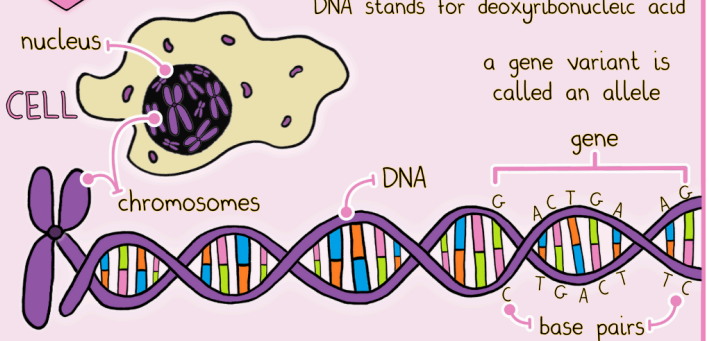
DNA and GENES

Humans have around 23,000 genes encoded in 23 chromosome pairs made of DNA in the nucleus of each cell in the body

DNA stands for deoxyribonucleic acid

a gene variant is called an allele



gametes (egg and sperm) each contain 23 chromosomes which pair up when the egg is fertilised

these gene pairs determine the characteristics that the child inherits from its parents, such as eye colour

VARIATION

Variation in gene expression makes each individual unique and creates biodiversity

DISCRETE

values in groups such as blood type or eye colour

A- B+ AB O

CONTINUOUS

values in a range of numbers such as height or weight

162 163 164 165 166

GENETIC

variation entirely encoded in genes and passed on from a parent such as eye colour or an inherited disorder

ENVIRONMENTAL & GENETIC

variation affected by a combination of both genetics and the environment such as height or weight

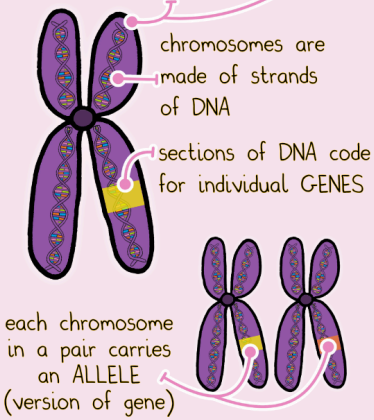
ENVIRONMENTAL

variation not encoded in genes and caused entirely by the environment such as scars or tattoos

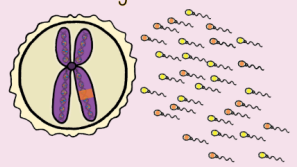
ALLELES

An allele is a variant of a gene that codes for different characteristics such as eye colour

each human is coded by 23 PAIRS of chromosomes



GAMETES (sperm or eggs) don't have pairs of chromosomes, so half carry one allele for each gene and half carry the other



one sperm fertilises one egg, so the child inherits one allele from each parent which gives the child a GENOTYPE

GENOTYPE and PHENOTYPE

A genotype is the combination of alleles that codes for a specific characteristic expressed as a phenotype

ALLELE ~ a variant of a gene that can be dominant or recessive

B capital letters always show the DOMINANT allele, i.e. for eye colour, capital B codes for brown eyes

b lower-case letters always show the RECESSIVE allele, i.e. for eye colour, lower-case b codes for blue eyes

GENOTYPE ~ the set of paired alleles that code for a trait

BB two dominant alleles are HOMOGYNOUS (homo = same type)

Bb one of each allele is HETEROGYNOUS (hetero = different type)

bb two recessive alleles are also HOMOGYNOUS

PHENOTYPE ~ the outward expression of genotype

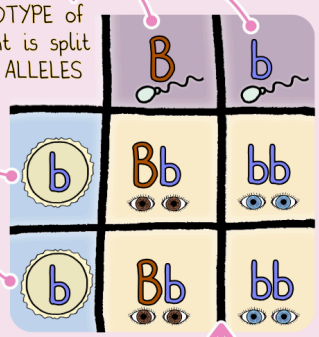
BB = BROWN **Bb** = BROWN **bb** = BLUE

the DOMINANT allele overrides the RECESSIVE allele, so the child will inherit brown eyes unless they receive a HOMOGYNOUS RECESSIVE code

PUNNETT SQUARES

The method used to calculate the probability of a child inheriting a genotype or phenotype from a parent

the GENOTYPE of each parent is split into two ALLELES



GENOTYPE = Bb
PHENOTYPE = brown eyes

eggs and sperm each carry ONE allele

a child gets one allele from each parent, one from the egg and the other from the sperm

GENOTYPE = bb
PHENOTYPE = blue eyes

child has brown eyes: Bb = 2 in 4 = 50%
child has blue eyes: bb = 2 in 4 = 50%

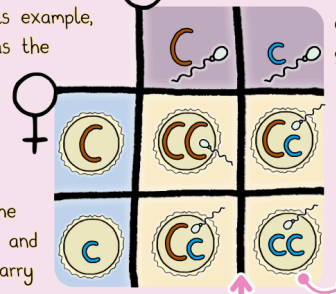
it is random chance which sperm fertilises which egg!

INHERITED DISORDERS

Genetic disorders are inherited from parents, e.g. cystic fibrosis which causes lung and digestive problems

a **Cc** genotype is heterogenous, which means both dominant and recessive versions of the gene exist: in this example, neither parent has the disease, but both are carriers

half of the sperm produced carry the dominant **C** gene and the other half carry the recessive **c** gene so it's a 50% chance of passing on the cystic fibrosis gene via the SPERM



half of the eggs produced carry the dominant **C** gene and the other half carry the recessive **c** gene so it's a 50% chance of passing on the cystic fibrosis gene via the EGG

cystic fibrosis is a homogenous recessive disorder, meaning two recessive genes, one from each parent, are required to develop the disorder

child has no disorder: CC = 1 in 4 = 25%
child carries the disorder: Cc = 2 in 4 = 50%
child develops the disorder: cc = 1 in 4 = 25%

www.steampink.education/inheritance