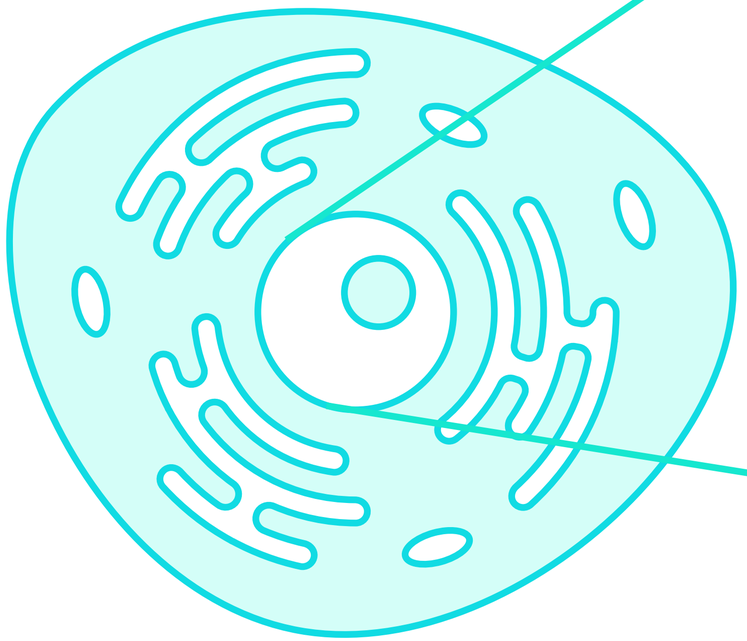
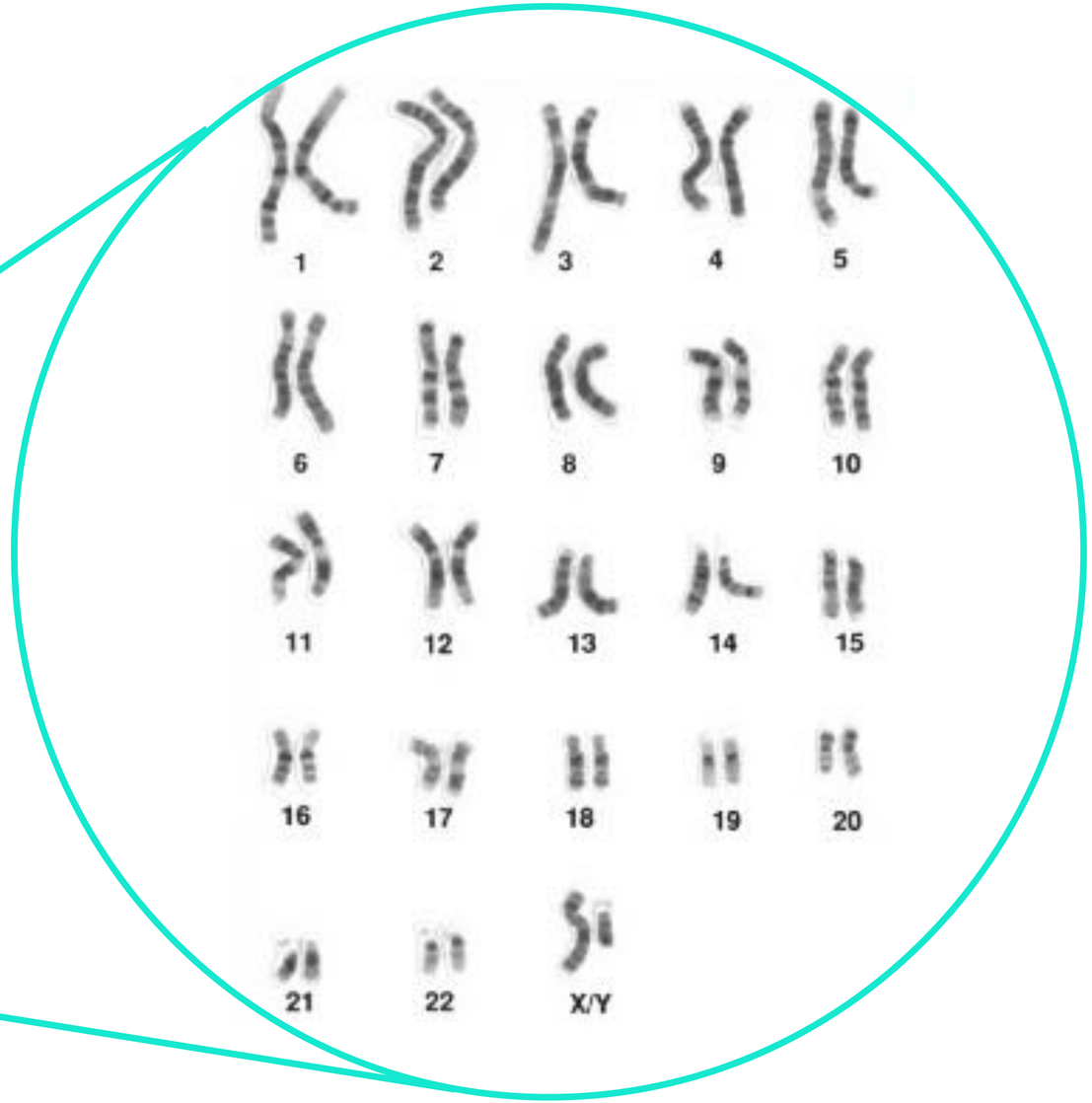
genetics

genetics

Chromosomes

Chromosomes are molecules of DNA. In humans they're found in the nucleus.





This is a **Karyogram** of a human's chromosomes. They are stained and arranged in decending order of size. Notice that the Y chromosome in the 23rd pair means these chromosome are from a male.

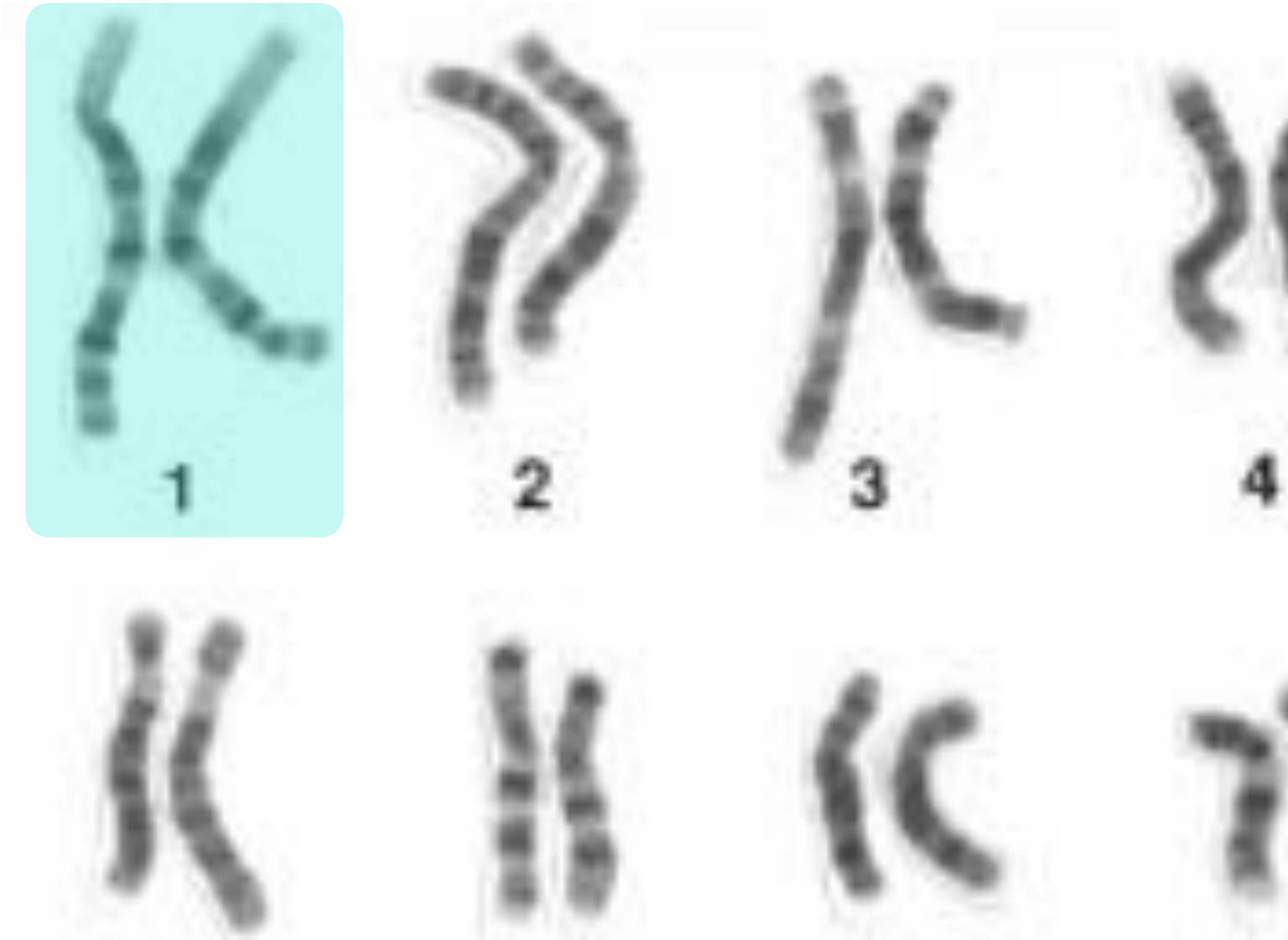
https://www.ncbi.nlm.nih.gov/books/NBK22266/

Chromosomes

In most human cells chromosomes come in pairs.

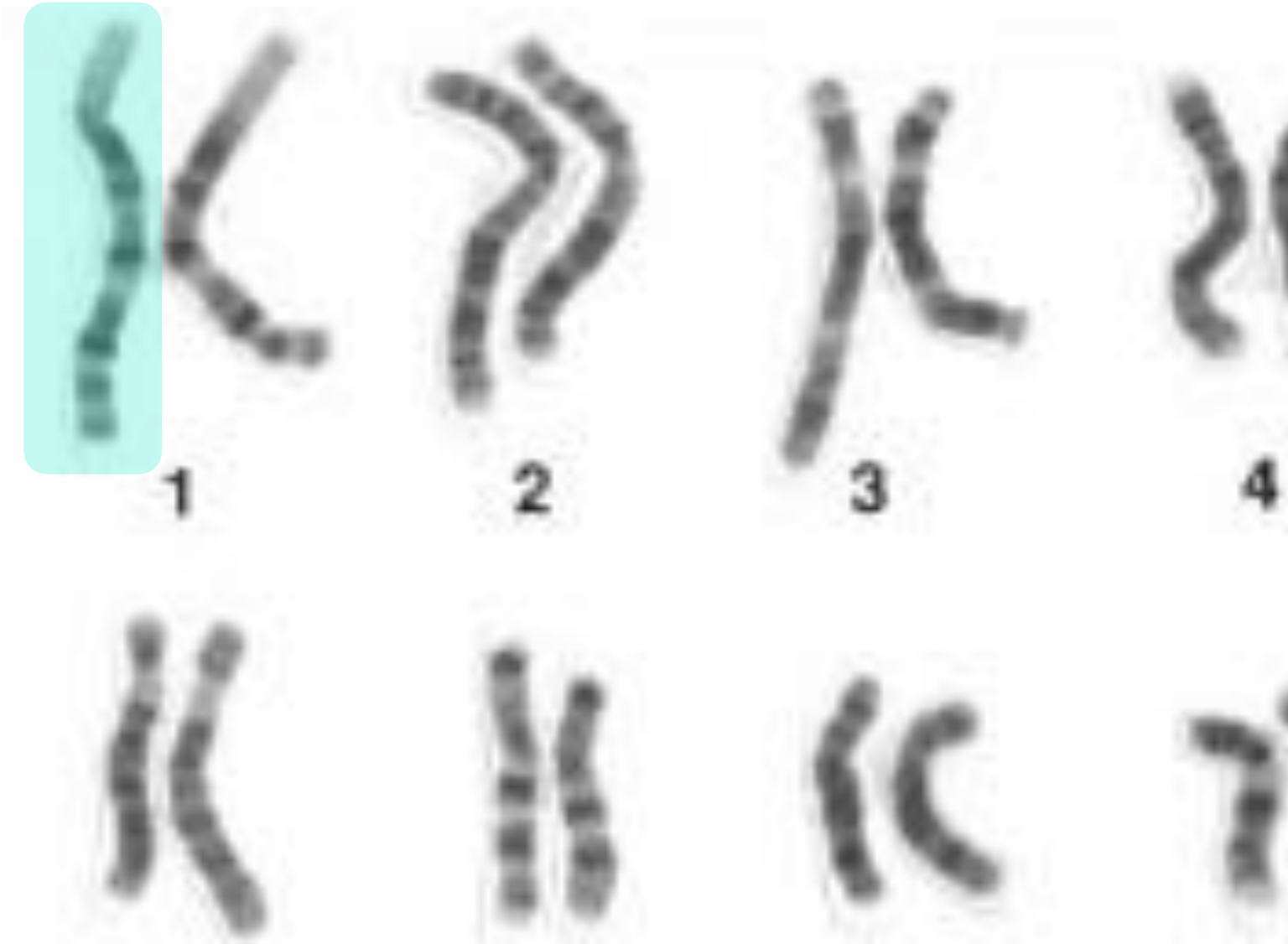
These cells are said to be **diploid**.

Cells with a single copy of each chromosome are called **haploid**.



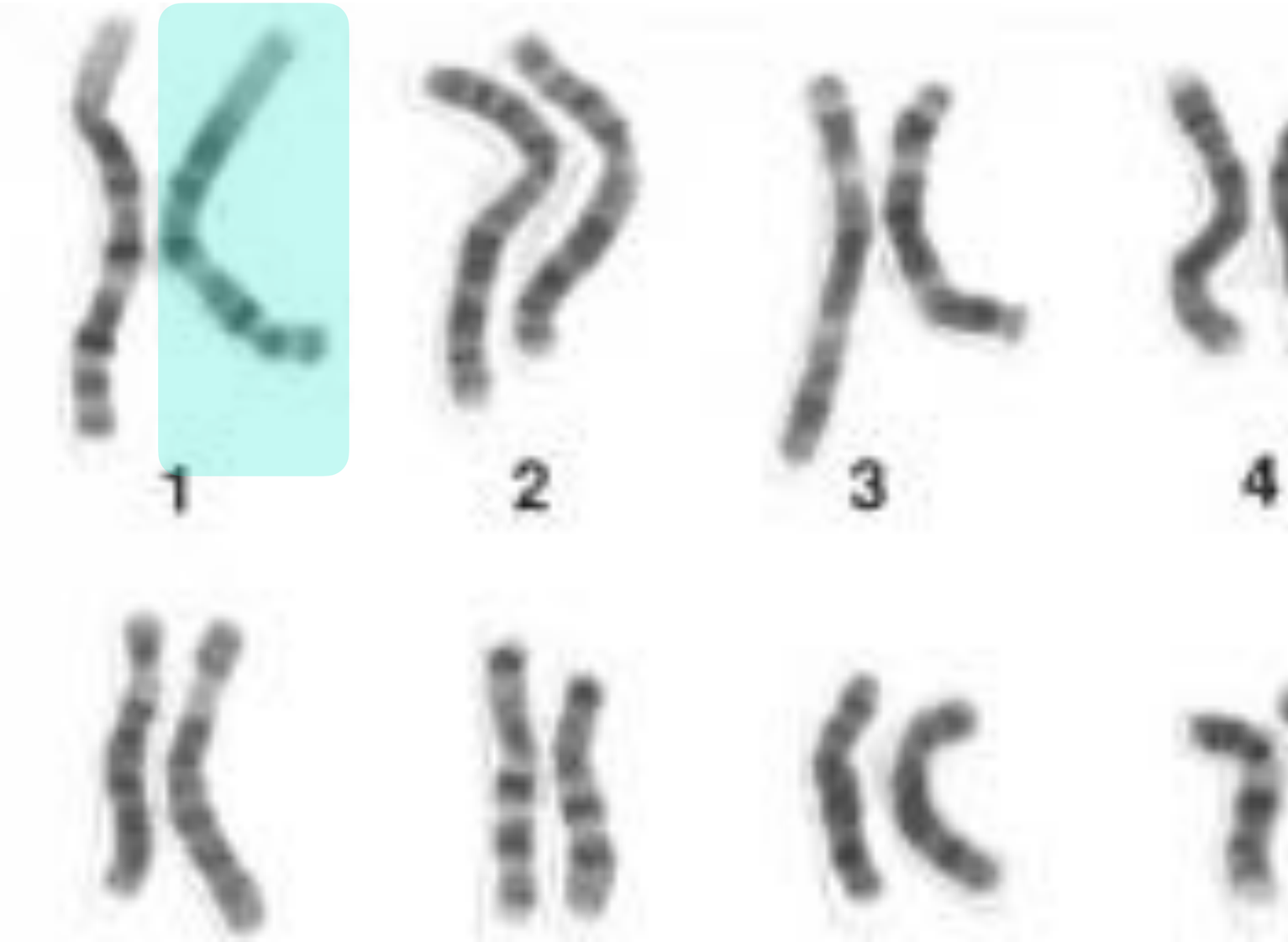
Chromosomes

In each pair one chromosome is inherited from the mother.



Chromosomes

And the other is inherited from the father.

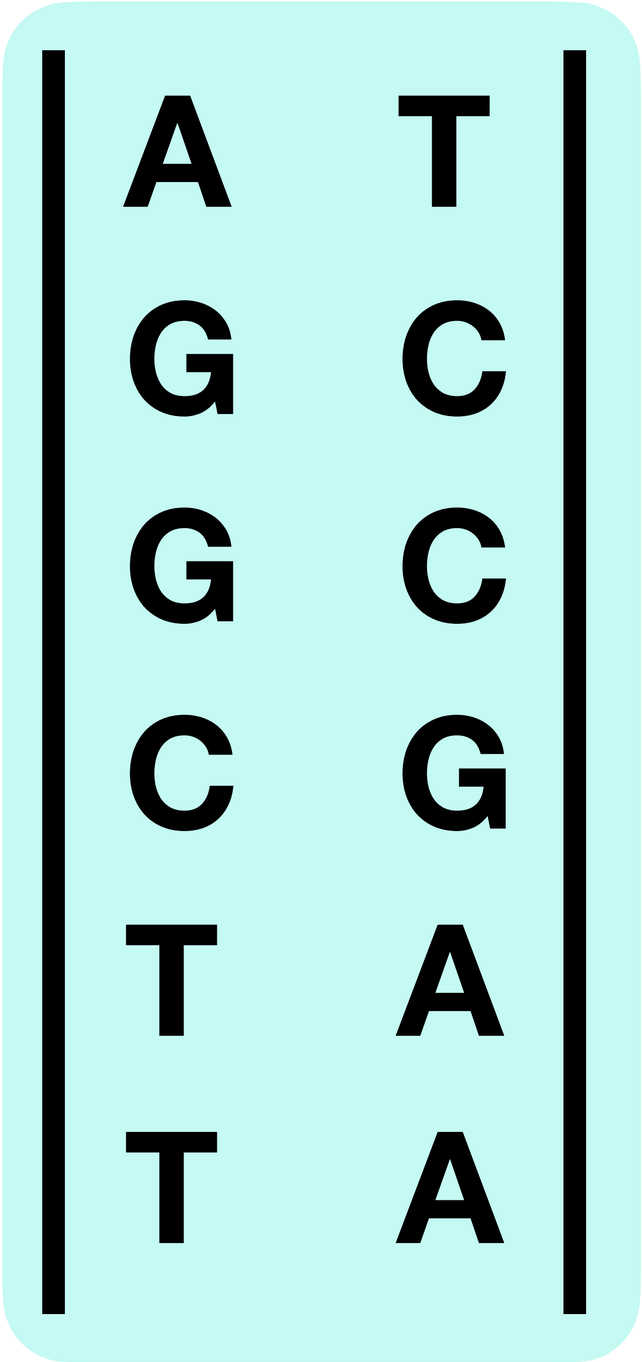


Chromosomes

A single chromosome is one very long molecule of DNA.

In humans chromosome 1 has 248,956,422 base pairs.

stretched to full length chromosome 1 would measure 85 millimeters.

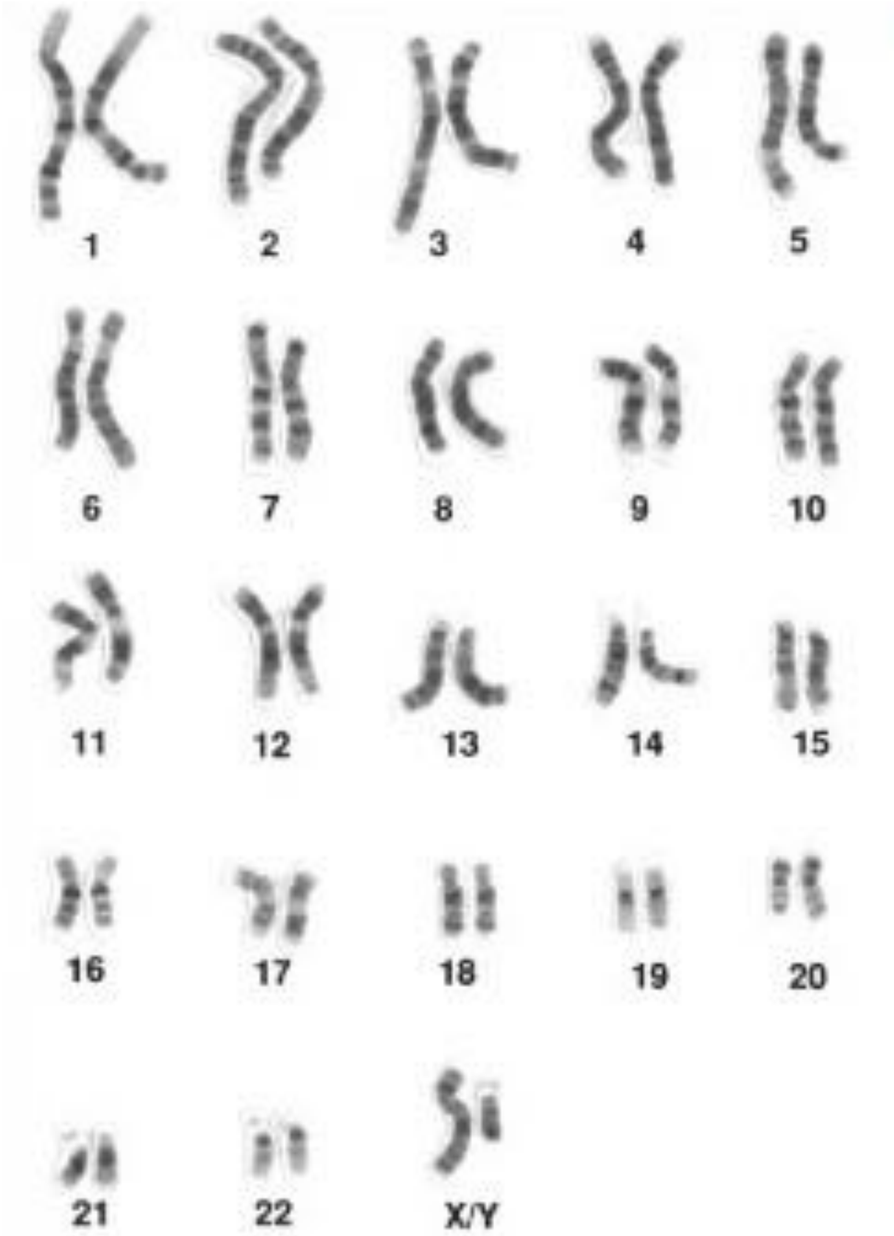


these letters are called nucleotides

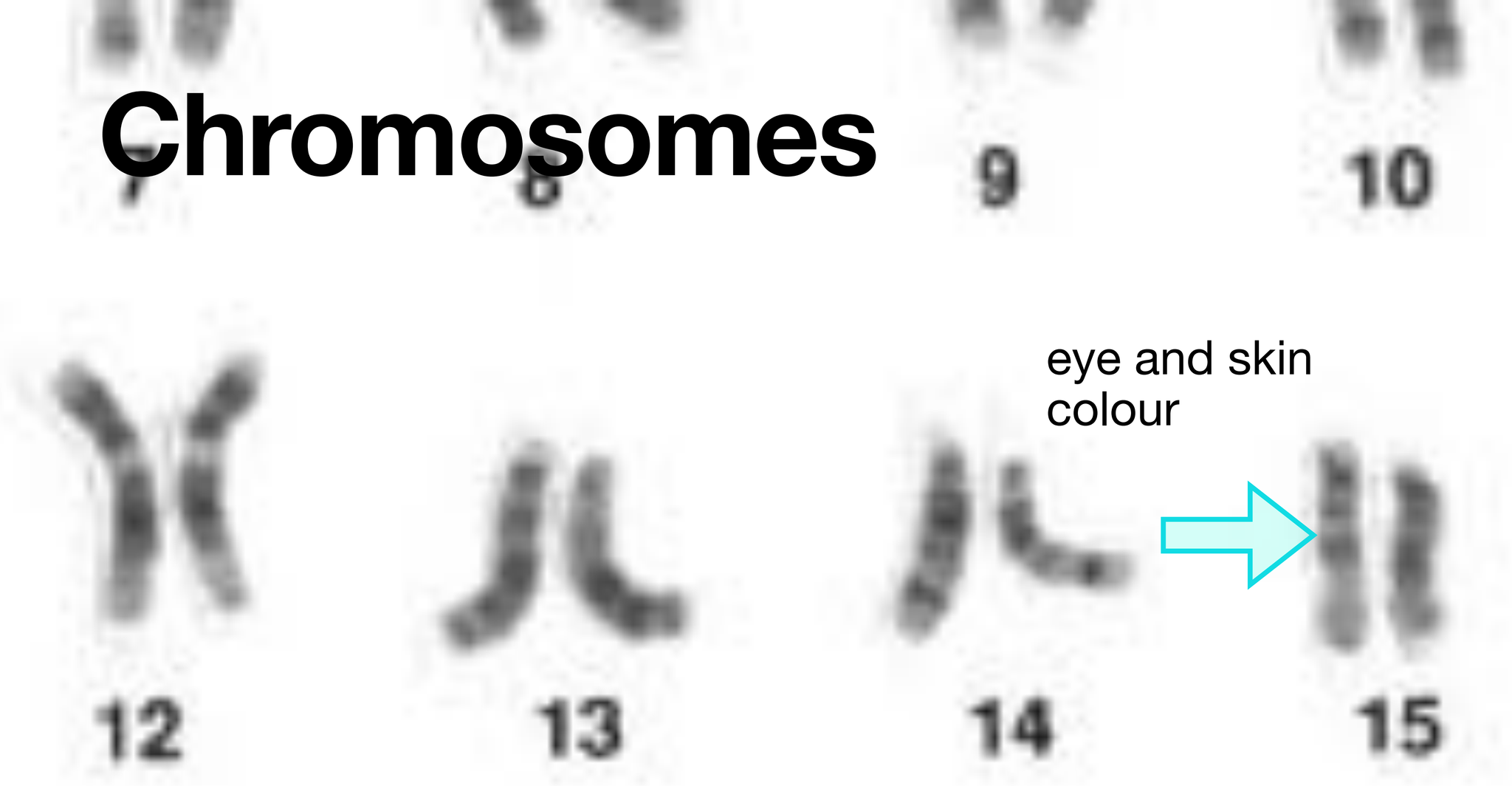
Chromosomes

Sections of DNA instructs a cell how to make proteins. These sections are called **genes**. Alternate versions of the same gene are called **alleles** of the gene. For instance the gene fro eye colour has alleles for blue, brown, green etc.

Different versions of a protien, function differently. Sometimes better, sometimes worse.



Chromosomes

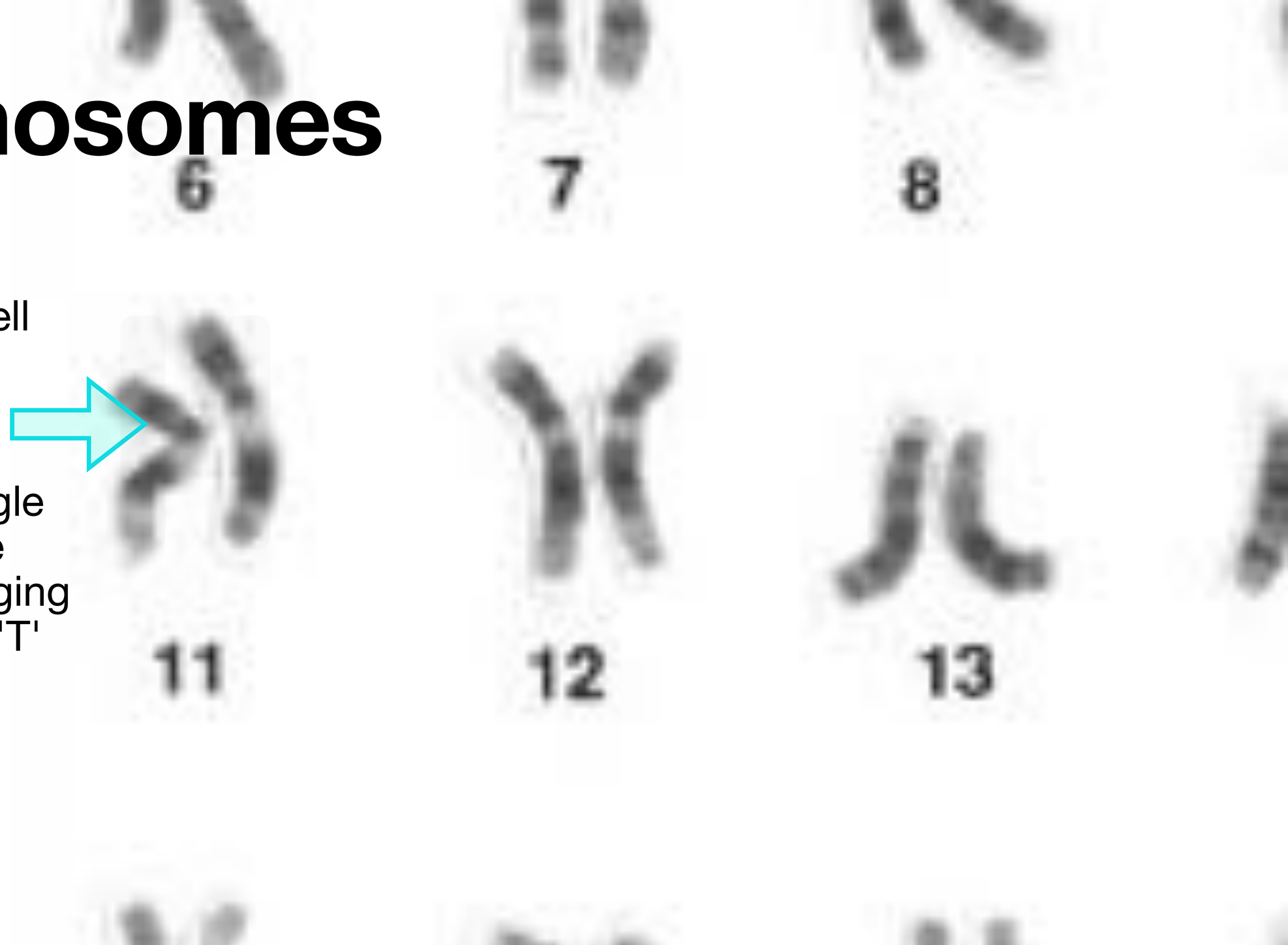


https://www.ncbi.nlm.nih.gov/genome/gdv/browser/genome/?id=GCF_000001405.39

Chromosomes

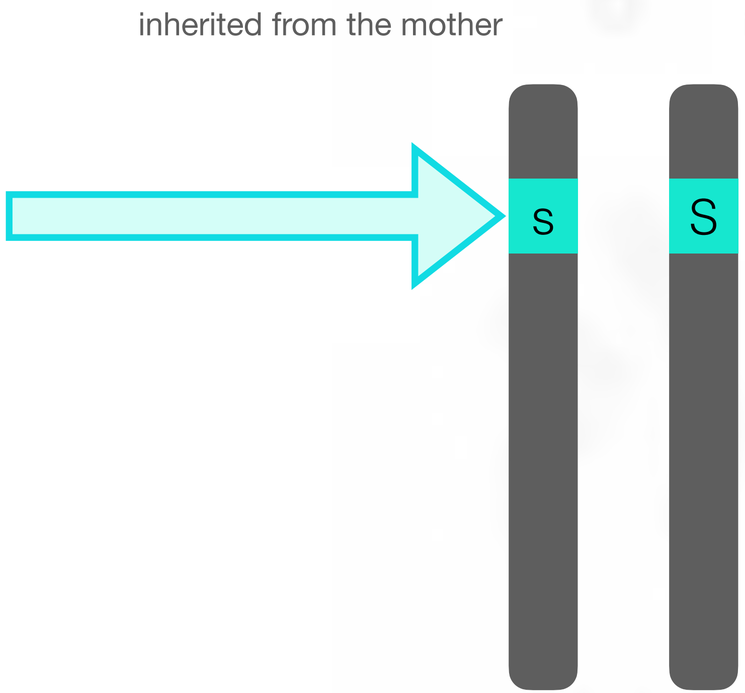
sickle cell anemia

caused by a single nucleotide in the HBB gene changing from an 'A' to a 'T'



Inheritance

Locus of the HBB gene. The damaged version 's' causes sickle cell anemia. A disease which causes blood cells to clump together if the blood oxygen falls too far, for instance during exercise.



s = sickle cell allele

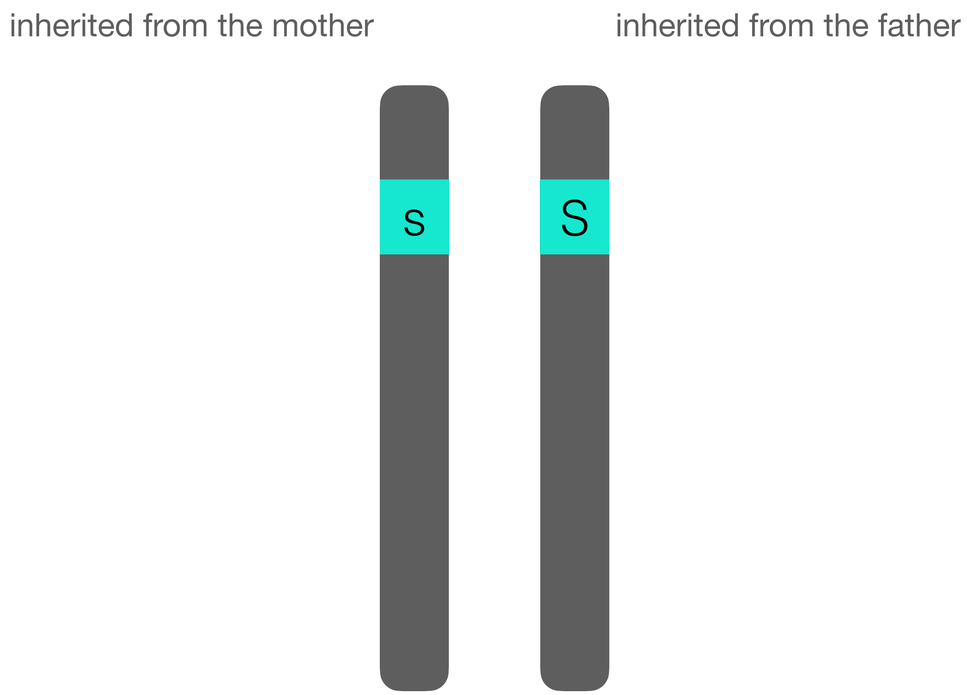
S = normal HBB allele

Inheritance

So does this individual have the disease?

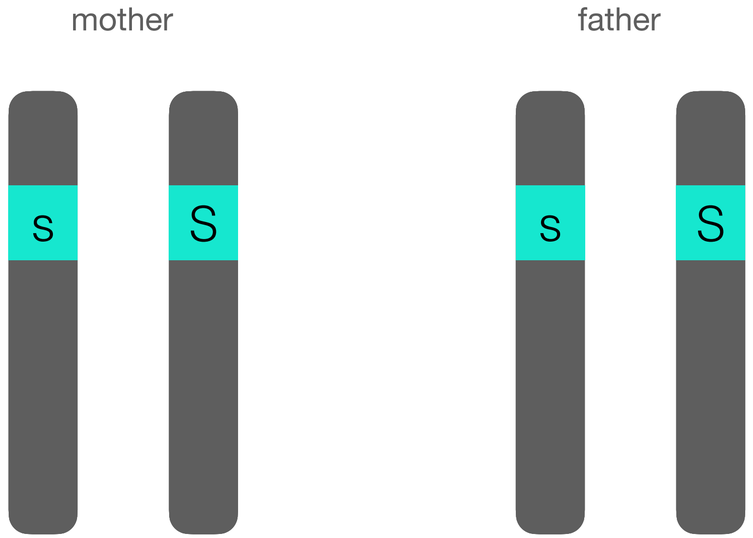
No.

Sickle cell anemia is **recessive**. You need to inherit two small 's' alleles, one from both parents, to get the disease.



Inheritance

Let's look at the parents.

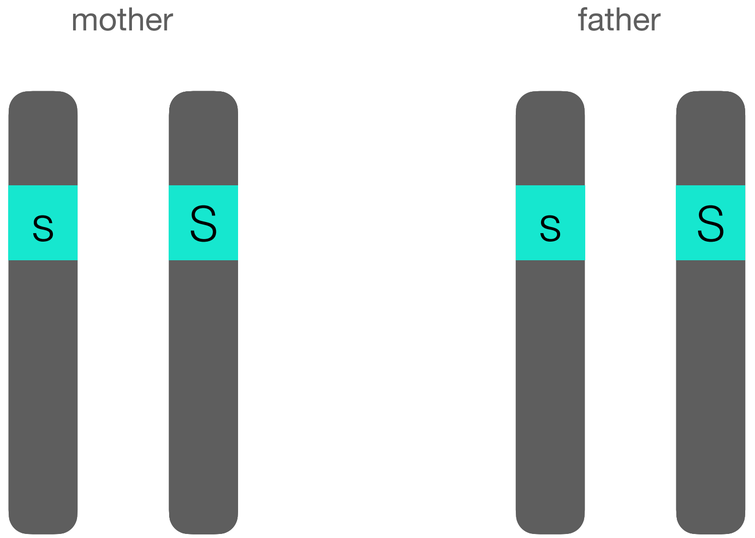


Suppose they are both unaffected carriers of sicke cell.

There are 4 possibilities for their children...

Inheritance

Let's look at the parents.



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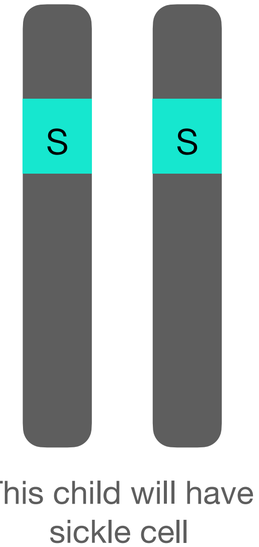
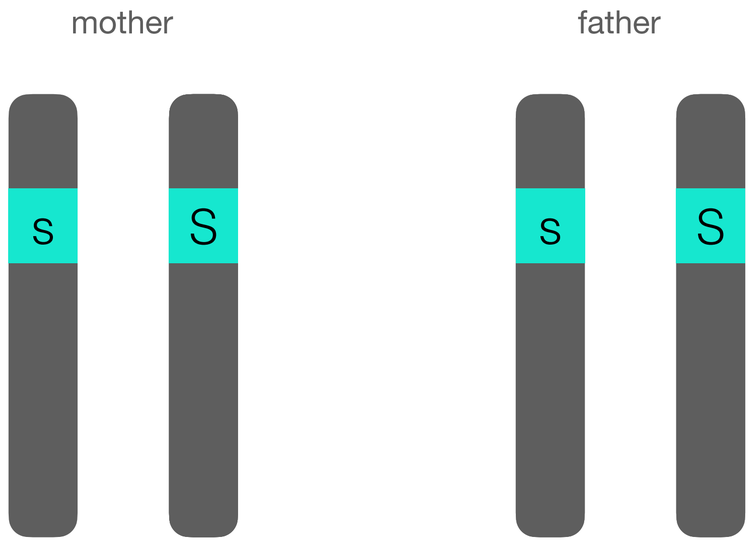


This child will have sickle cell

Inheritance

Let's look at the parents.



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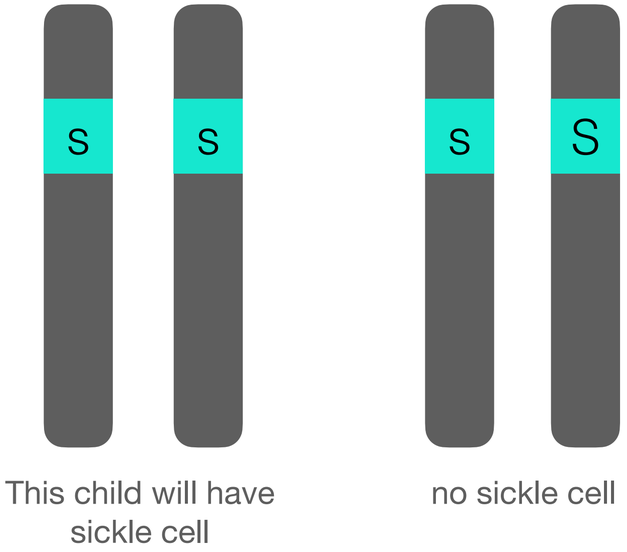

Inheritance

Let's look at the parents.

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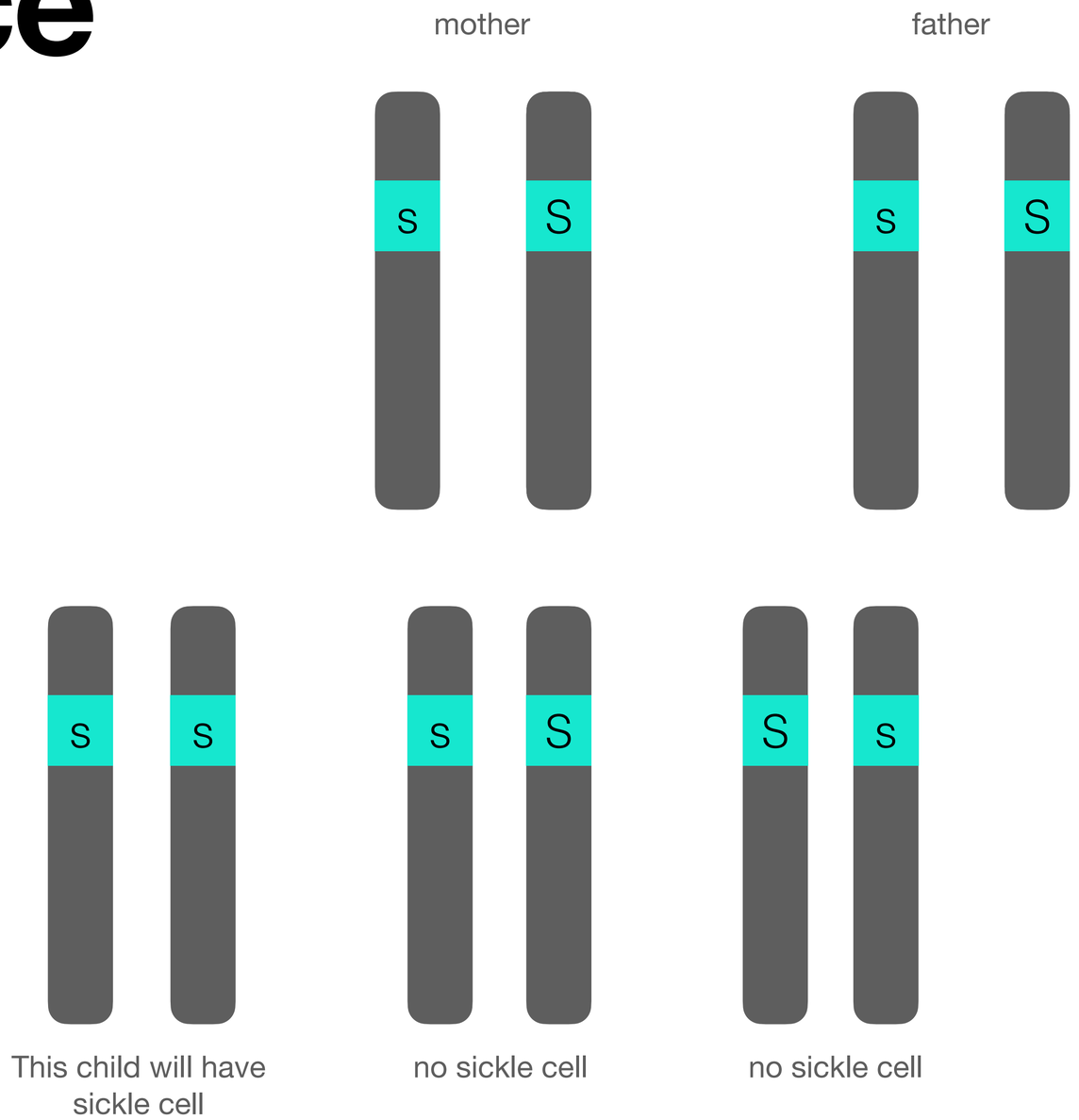

Inheritance

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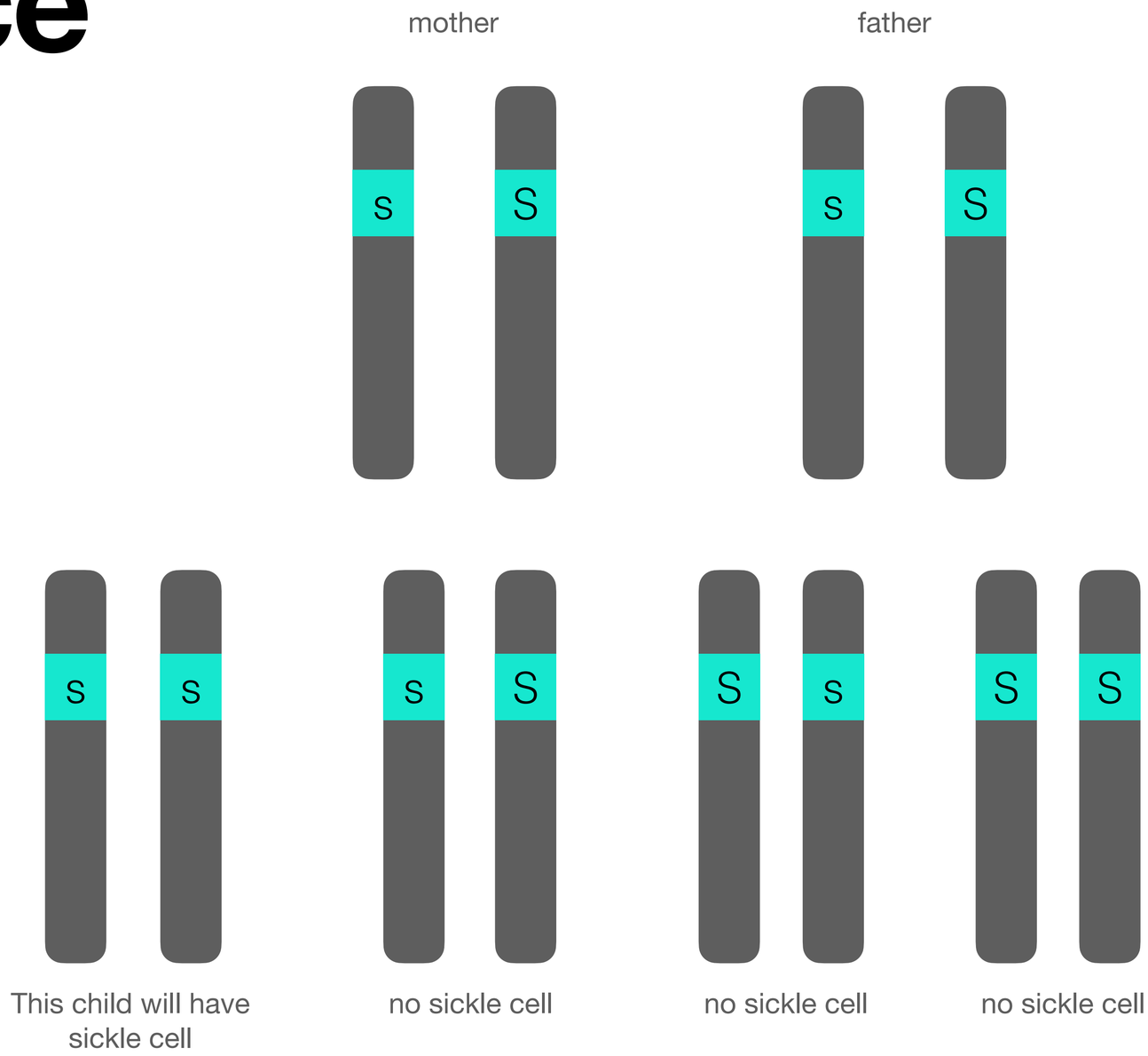
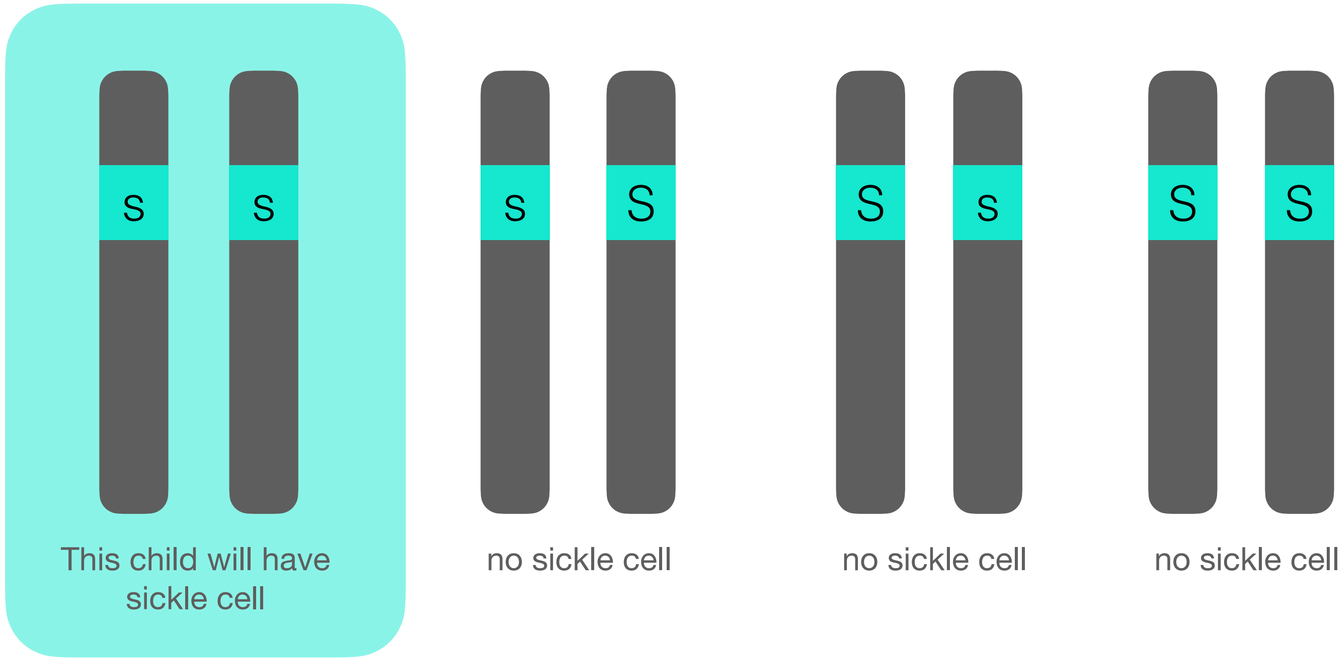


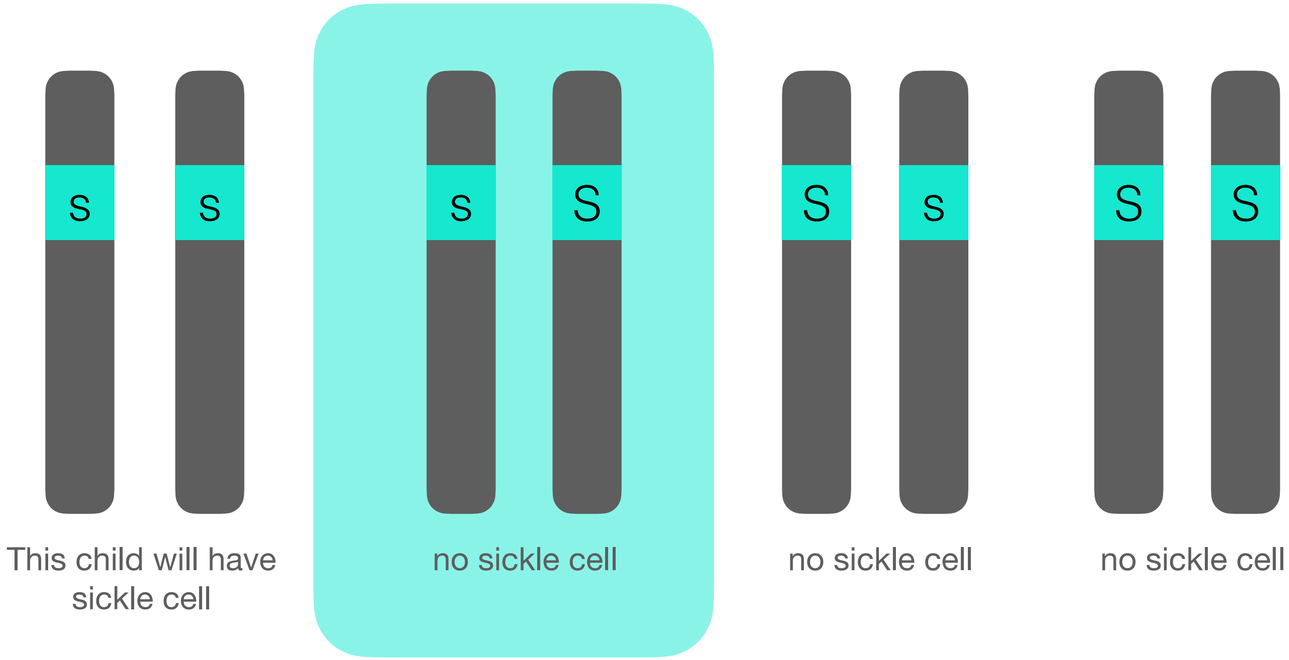
Inheritance



the same allele from both parents

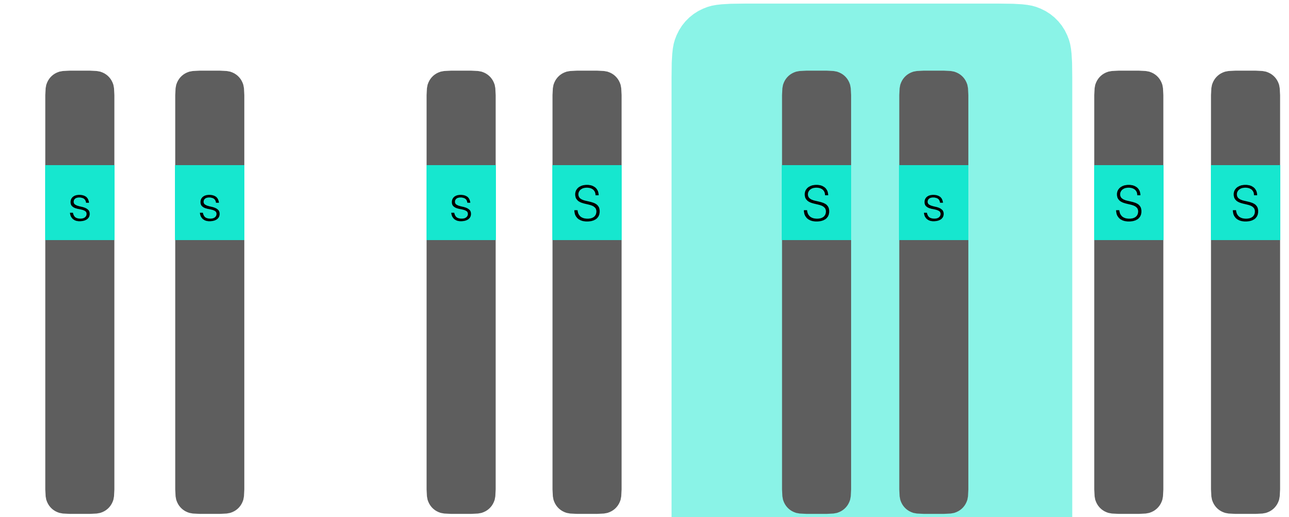
we call this **homozygous**.

Inheritance

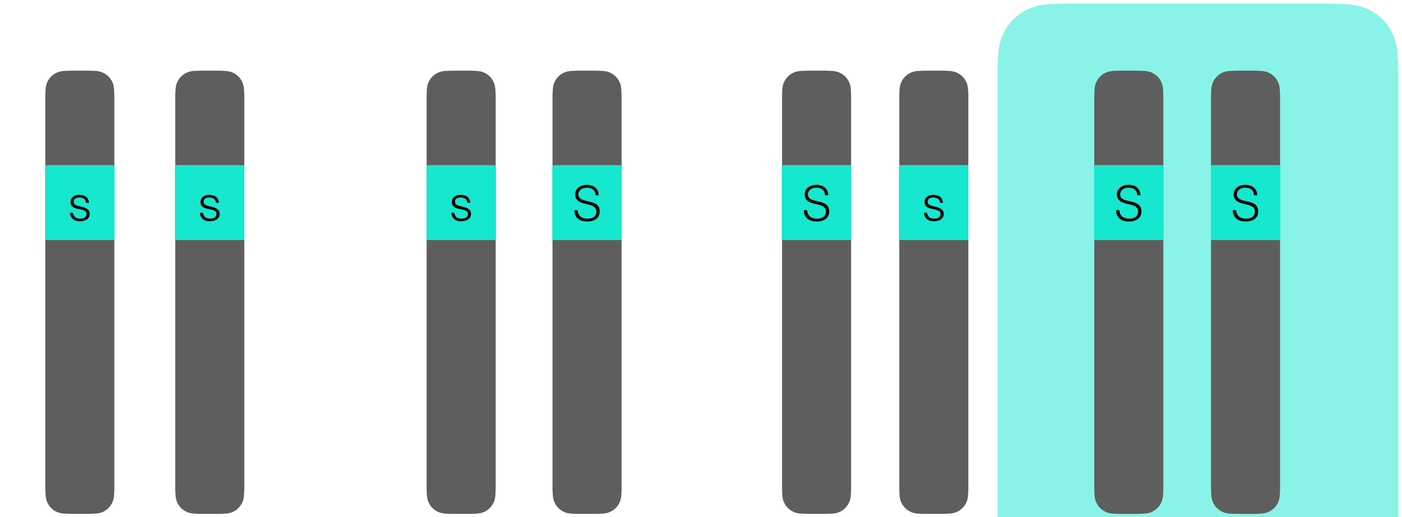


we call this **heterozygous**.

Inheritance

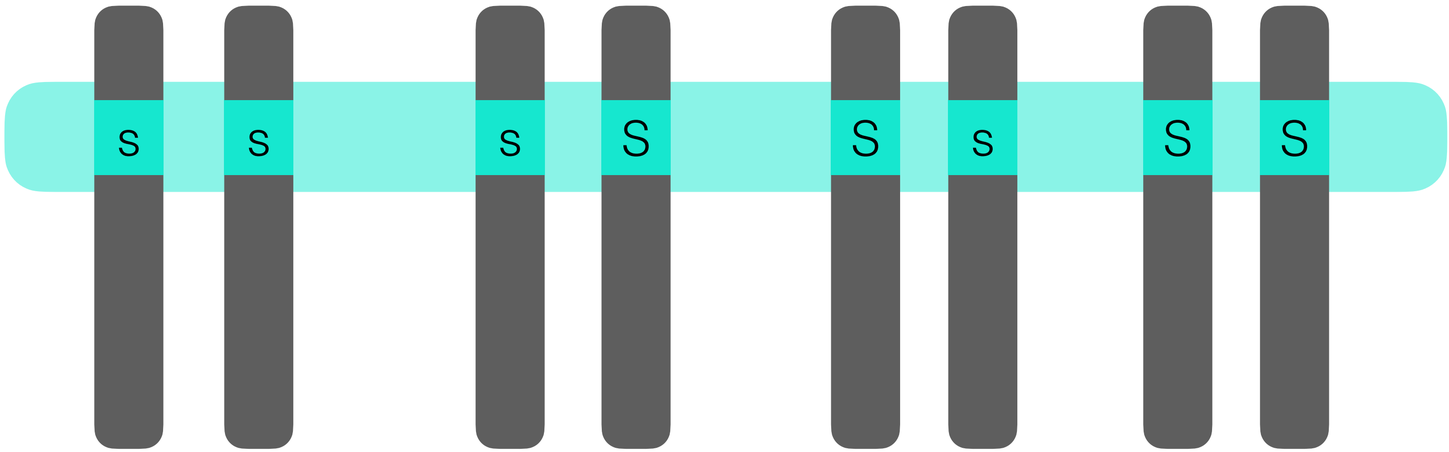


Inheritance



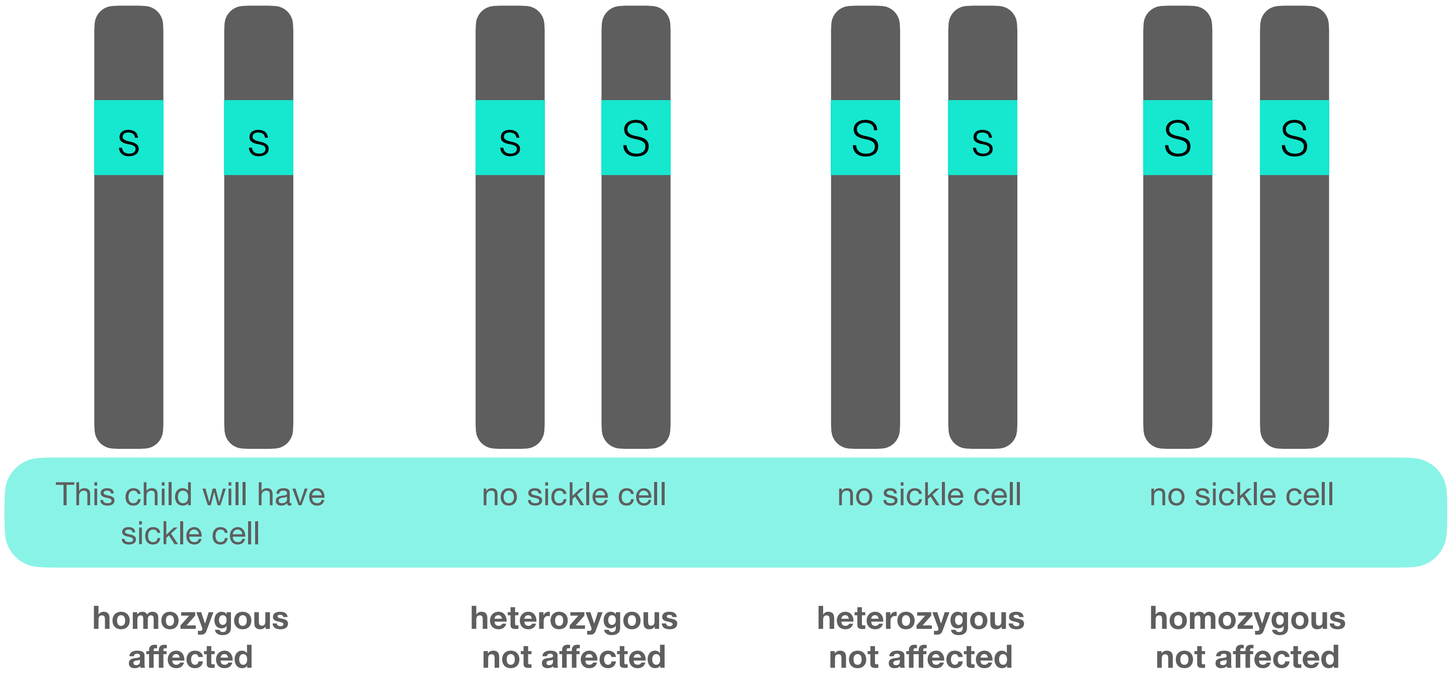
Inheritance

Genotype: the genetic make up of an individual.



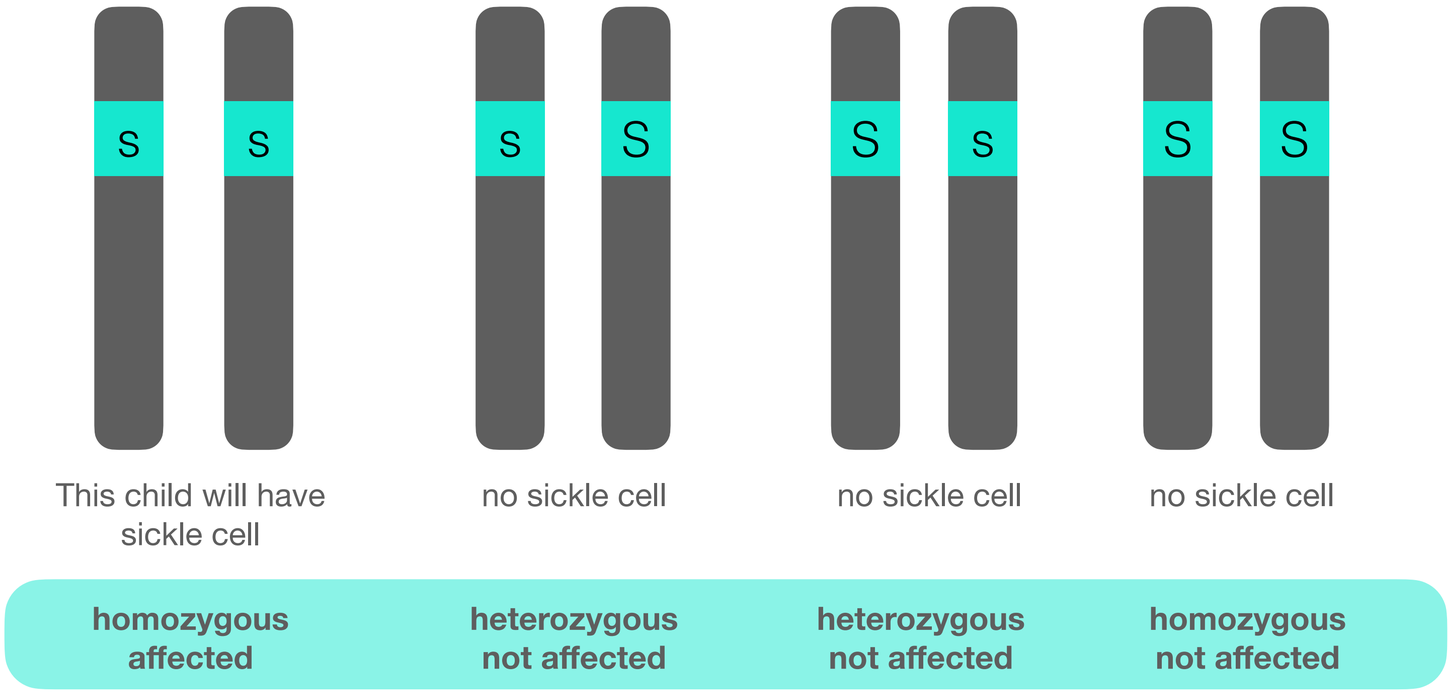
Inheritance

Phenotype: the physical characteristics of an individual.



Inheritance

Zygosity: whether both alleles are the same or not.



Inheritance

A quicker way is to use a Punnet square.

Inheritance

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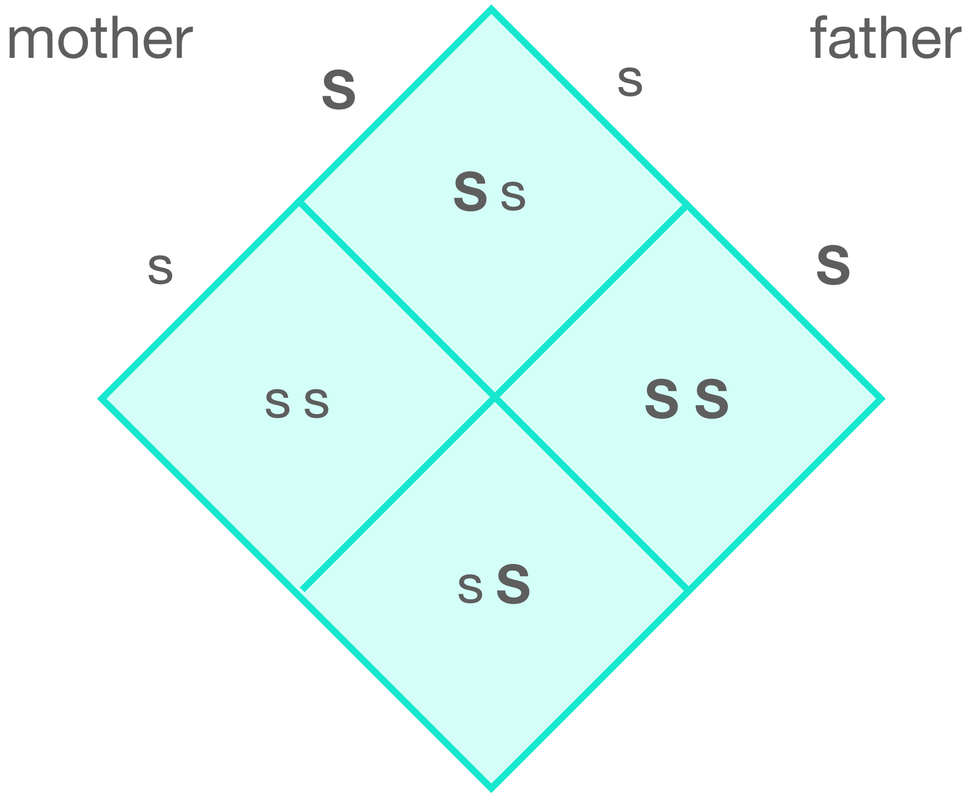
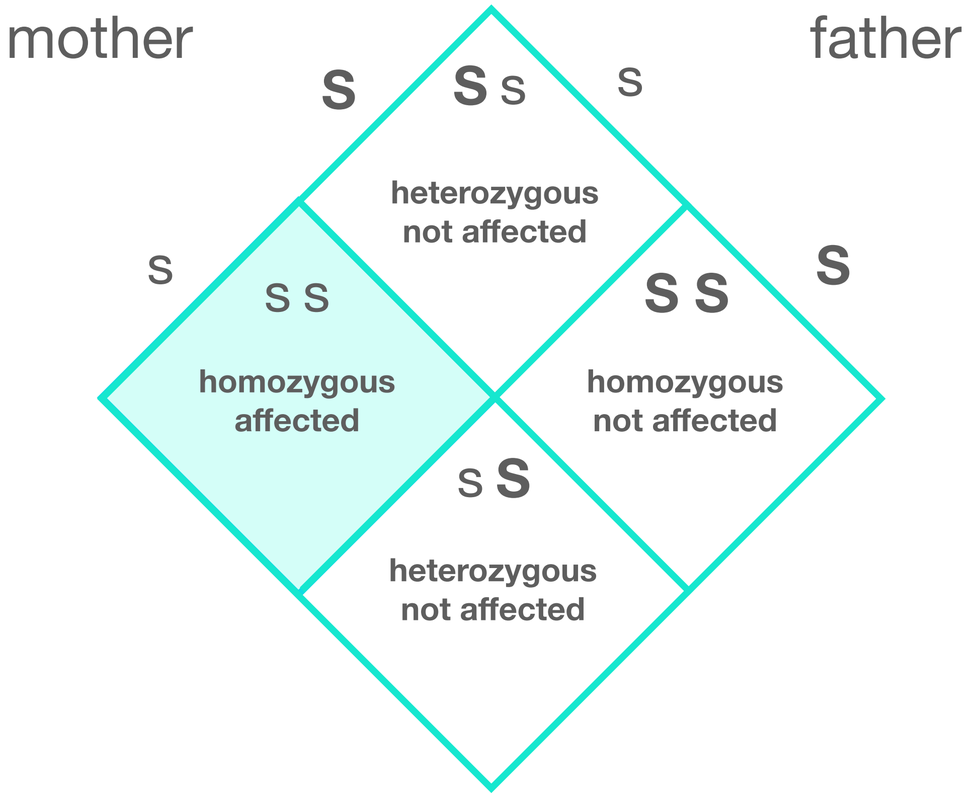


Inheritance

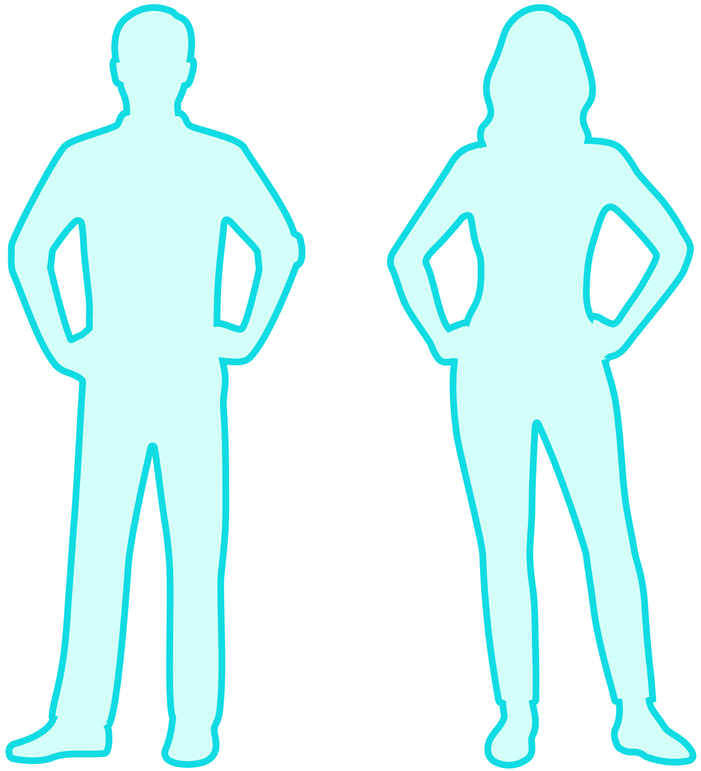
A quicker way is to use a Punnet square.



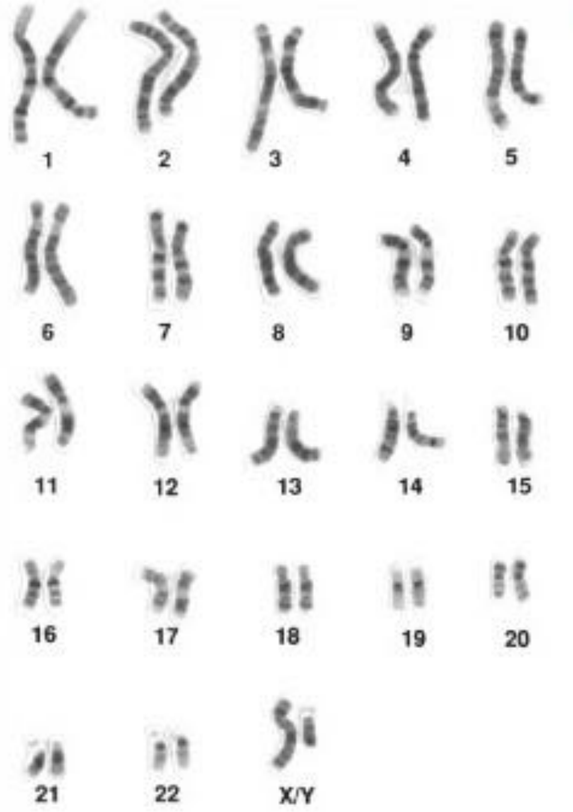
The probabability of being affected by sickle cell is 1/4 or 25%

Inheritance: gametes

in organisms that reproduce sexually most cells are **diploid**.

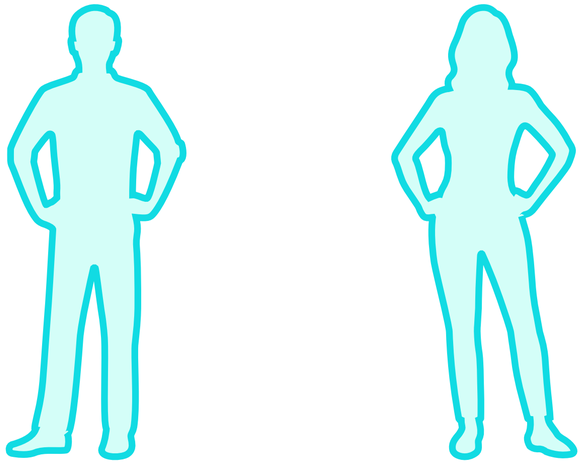


2 copies of each chromosome

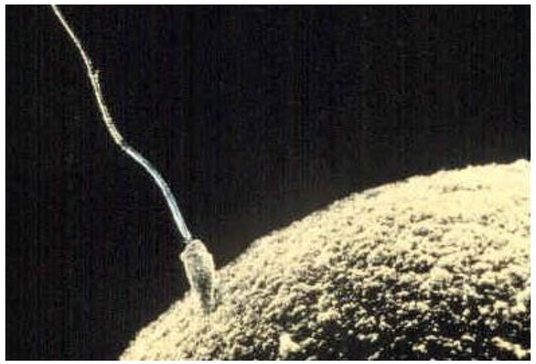


https://en.wikipedia.org/wiki/Sperm#/media/File:Sperm-egg.jpg

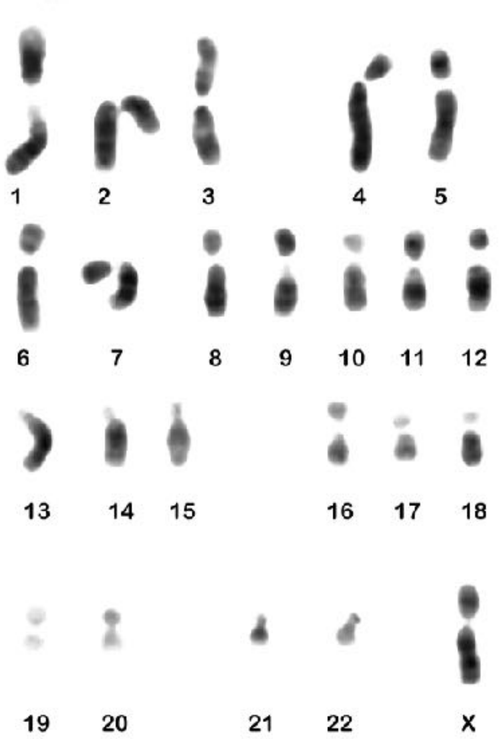
Inheritance: gametes



but the cells involved
in reproduction,
gametes, are **haploid**.



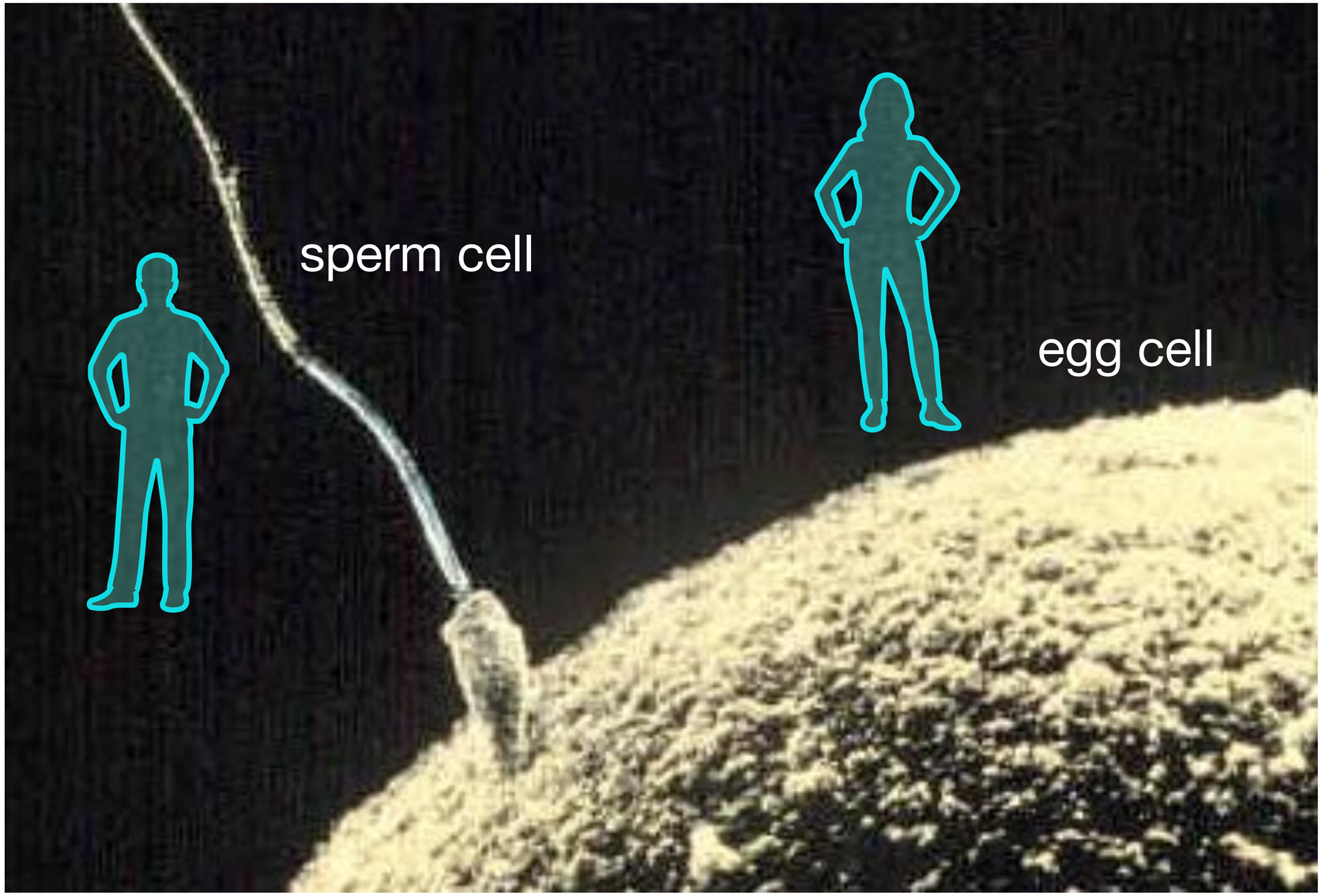
1 copy of each
chromosome



https://en.wikipedia.org/wiki/Sperm#/media/File:Sperm-egg.jpg

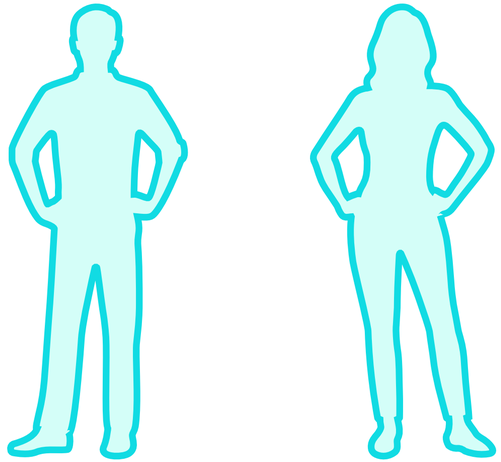
Inheritance: gametes

Gametes are produced by a type of cell division called **meiosis** which divides the pairs of chromosomes up amongst 2 daughter cells so they each get just 1 copy.



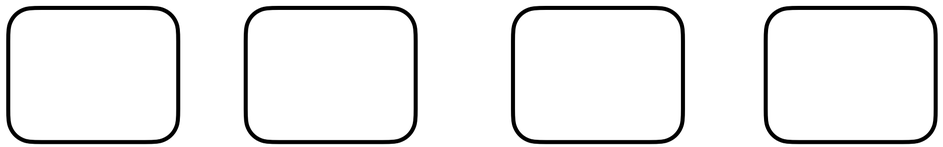
Inheritance: gametes

Looking at the example of sickle cell again



phenotype of gametes

phenotype of children



Inheritance: gametes

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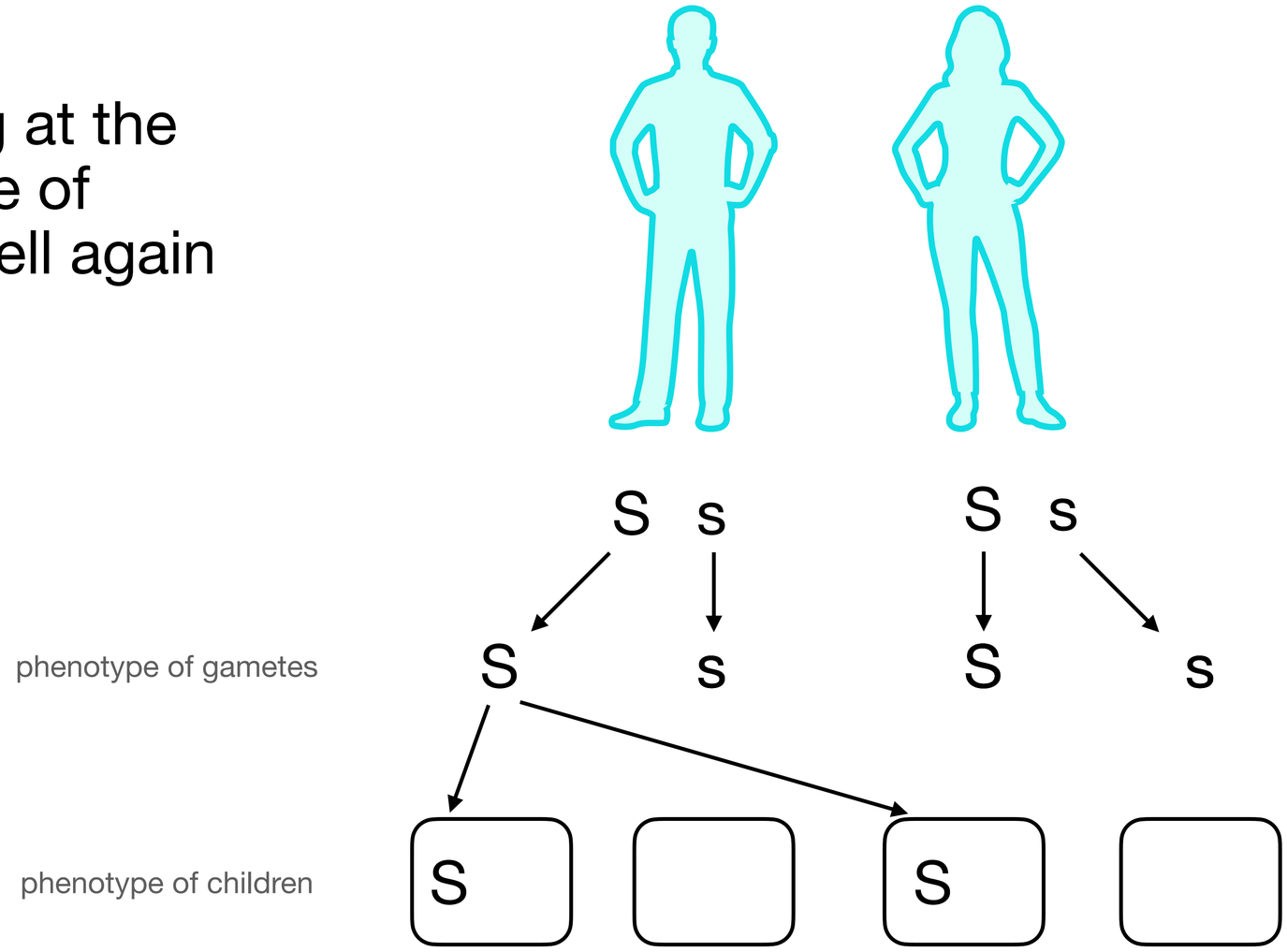

Inheritance: gametes

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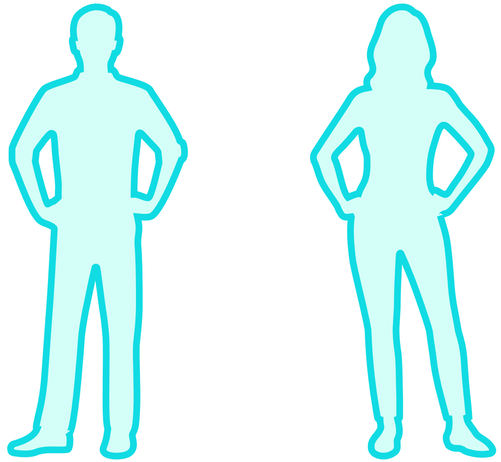

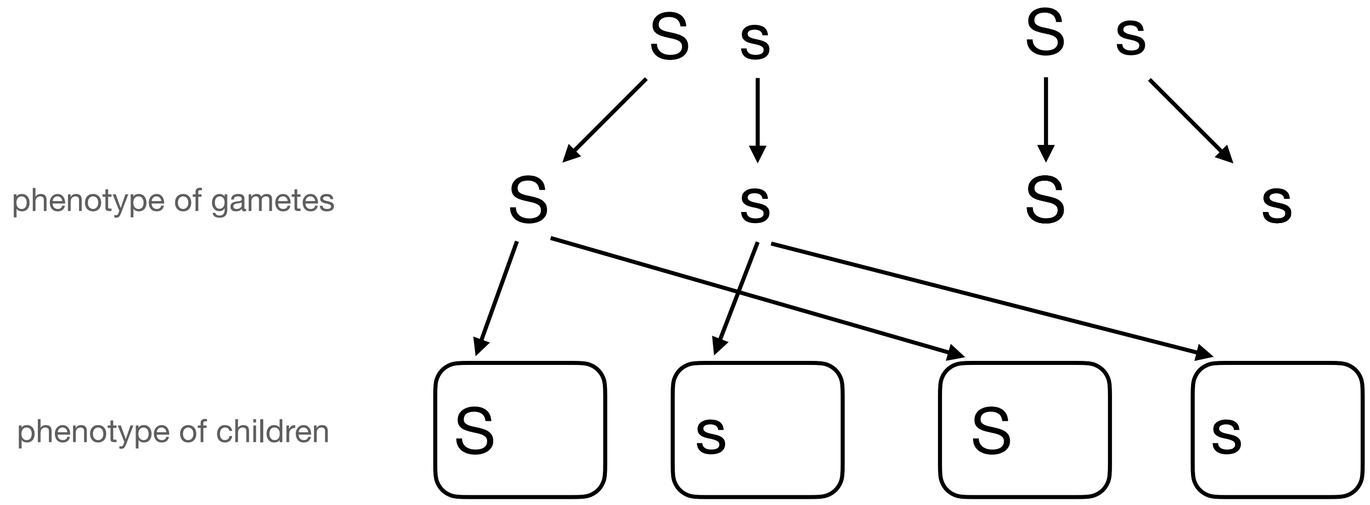

Inheritance: gametes

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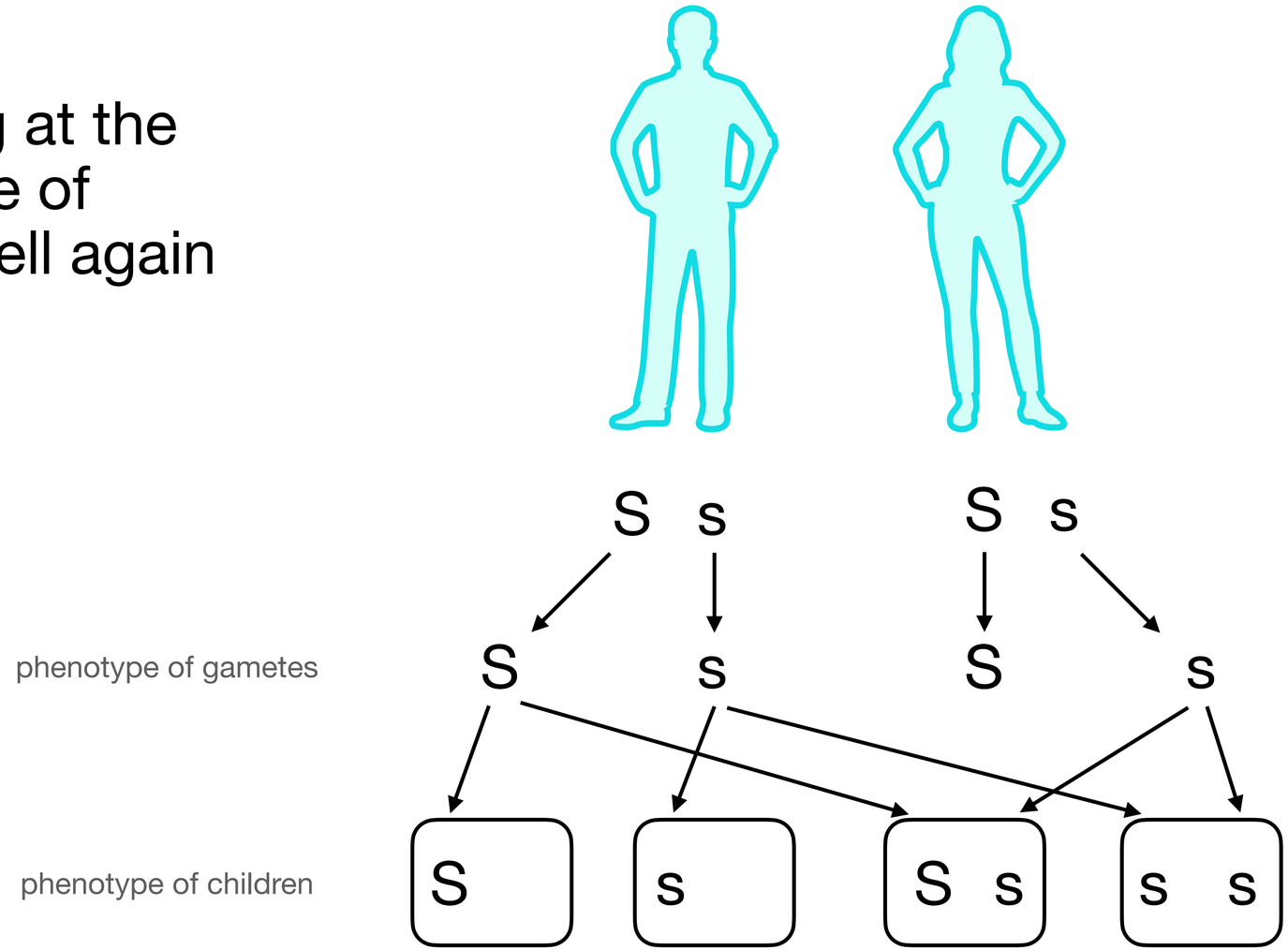

Inheritance: gametes

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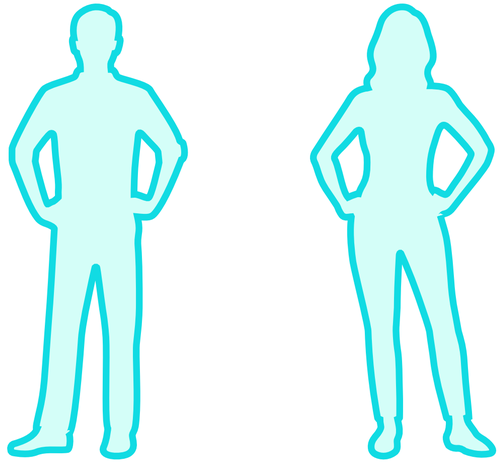

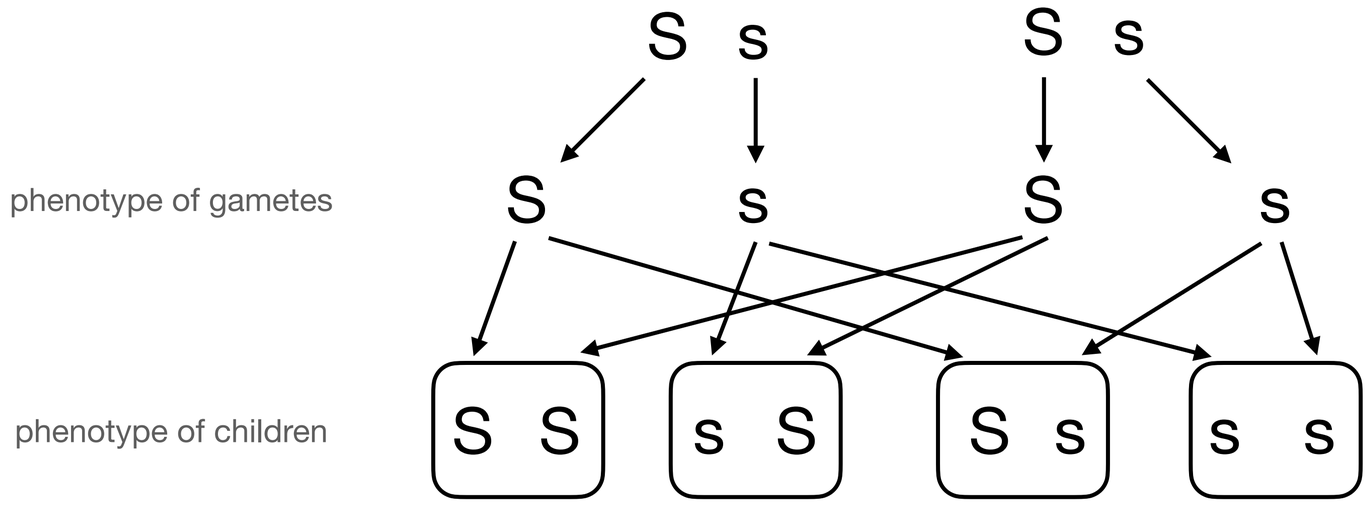

Inheritance: gametes

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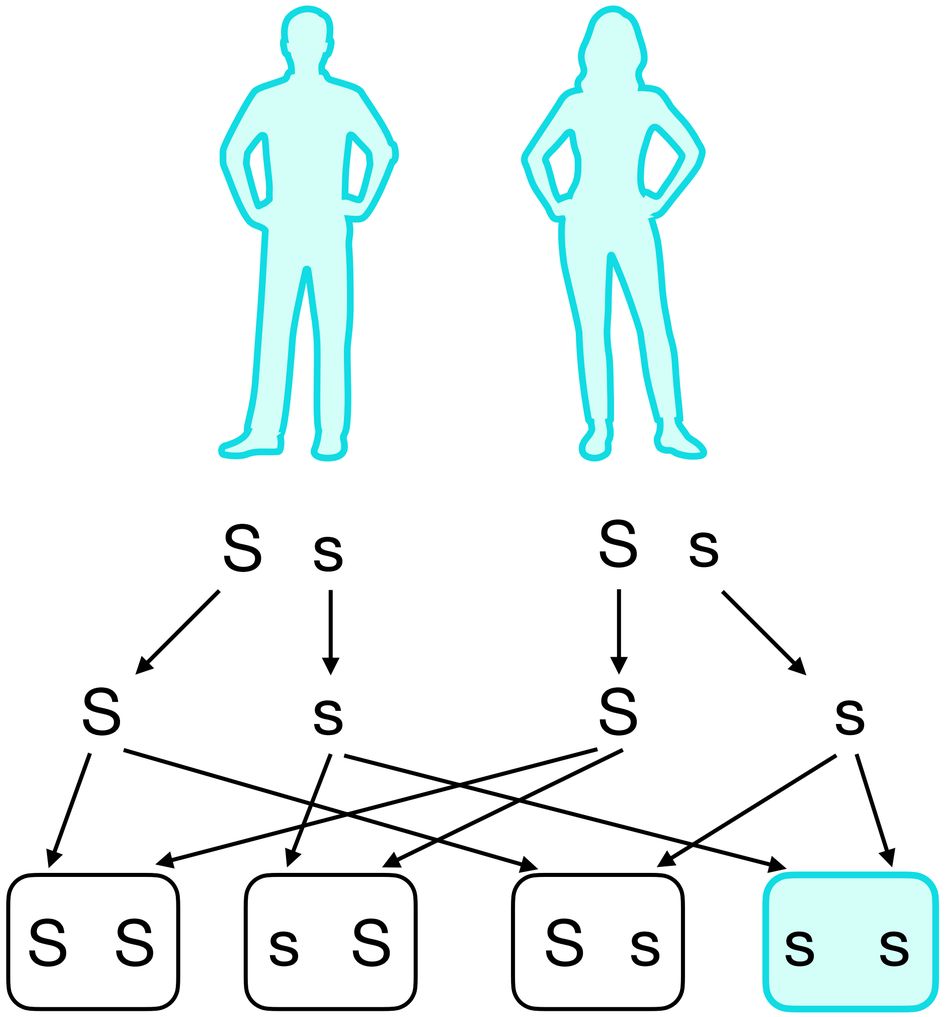

Like last time we see there's a 25% chance a child born to parents heterozygous for sickle cell will be affected by the disease.