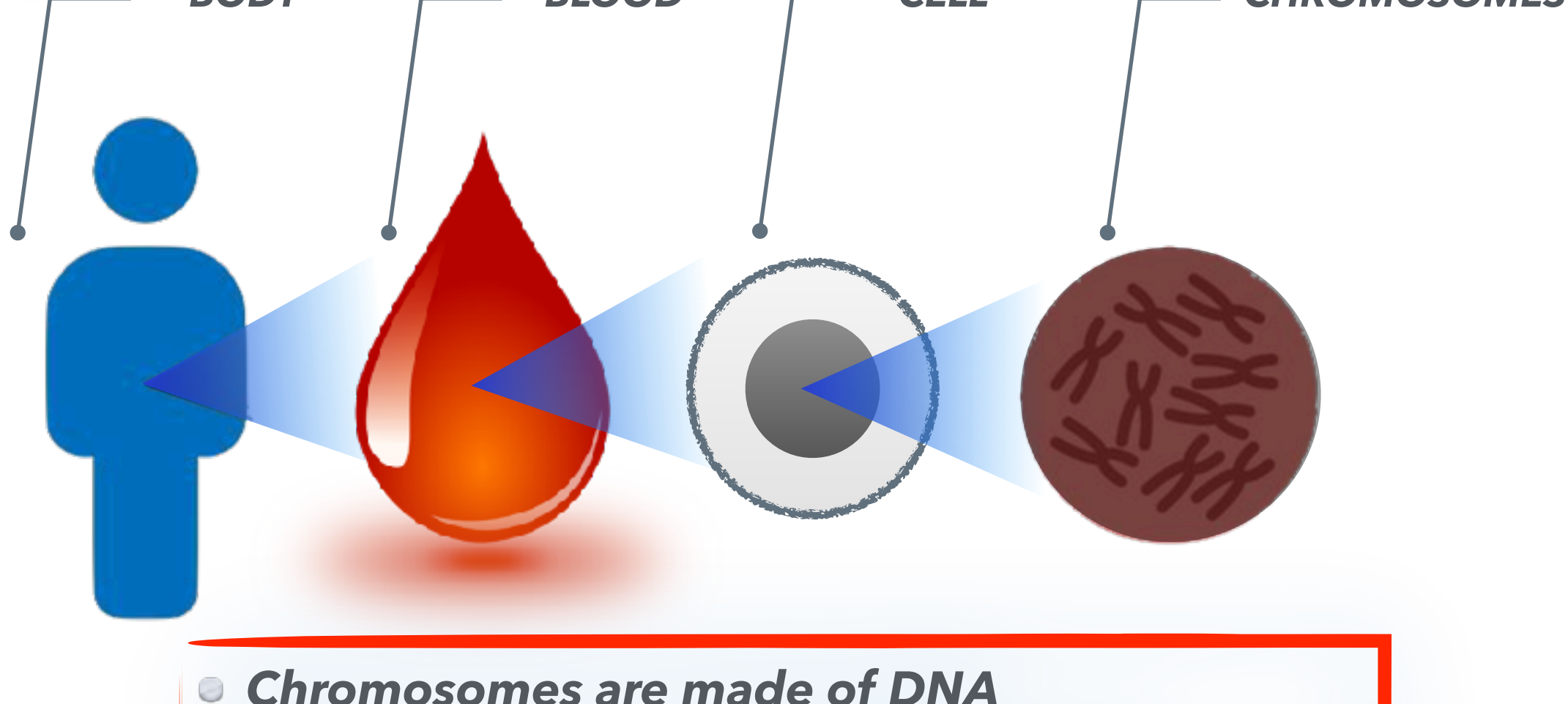


SYNGAP1 GENETIC INFOGRAPHIC

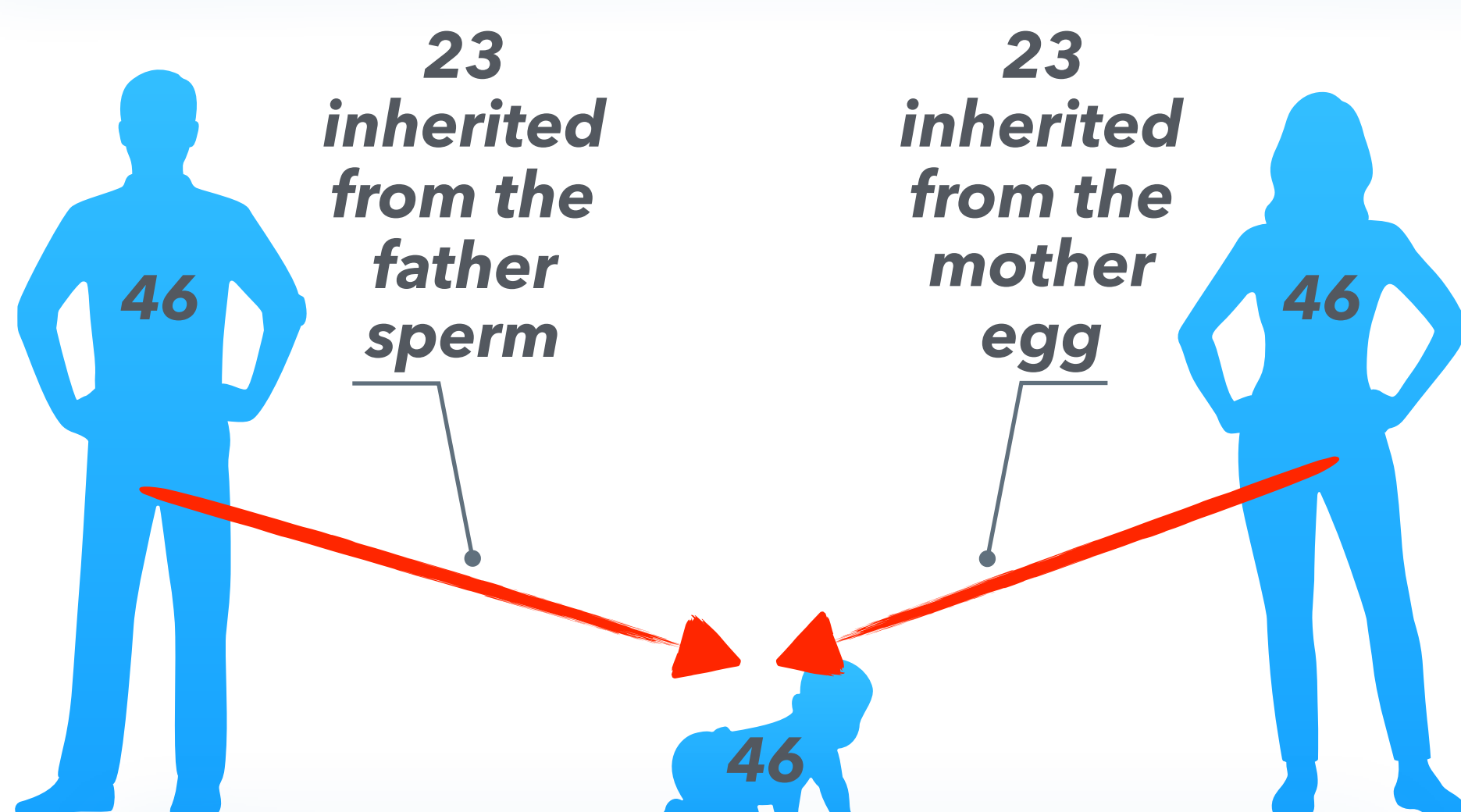


DNA is stored in all of our cells



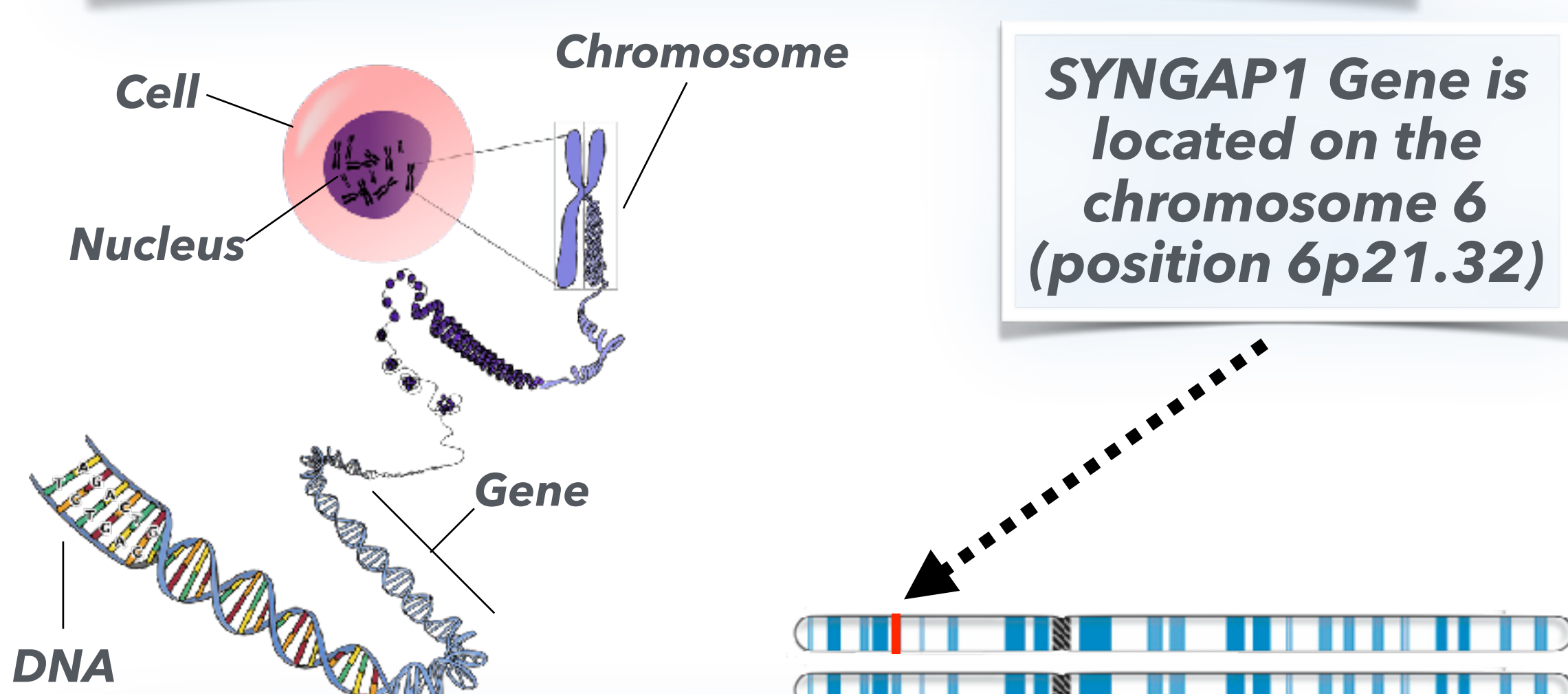
- Chromosomes are made of DNA
- DNA stands for DeoxyriboNucleic Acid

We inherit half of our chromosomes from each parent



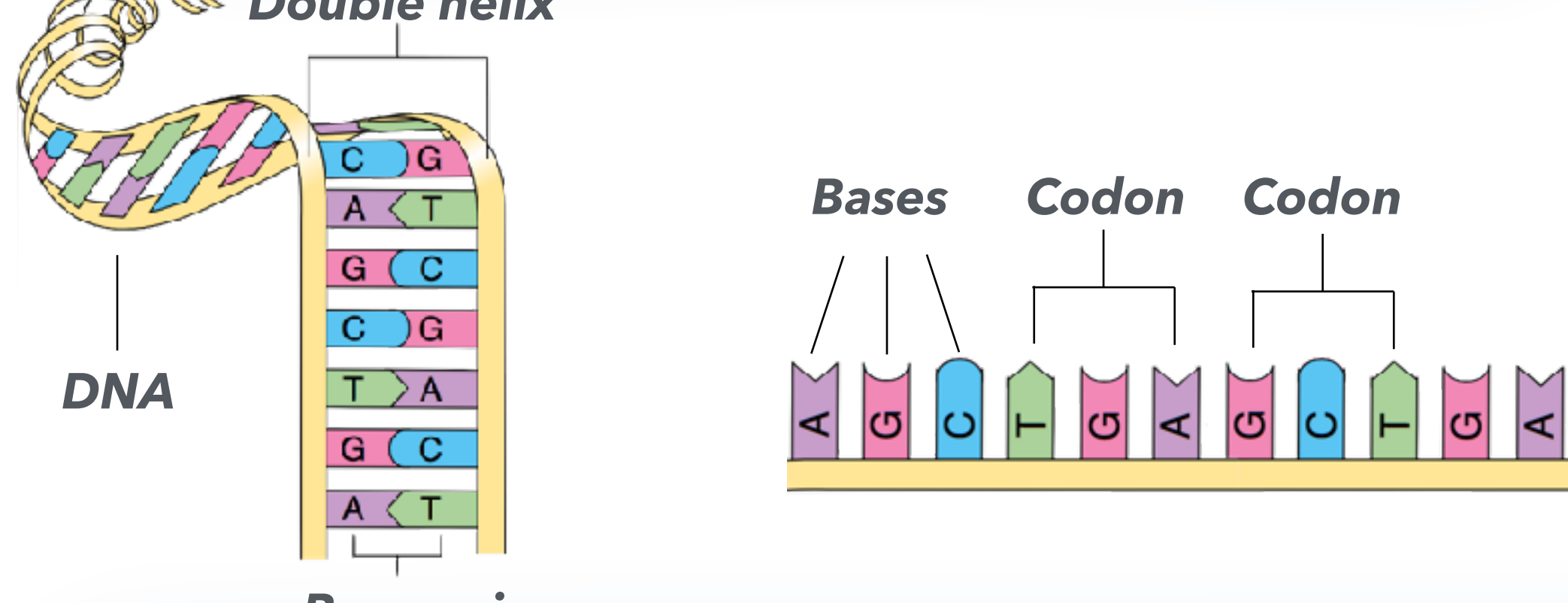
- Chromosomes are paired (46 chromosomes = 23 pairs)
- X/Y define the sex of the child (XX for a girl and XY for a boy)
- Picture of chromosomes lined up in pairs is called a karyotype

A chromosome contains genes A gene is a part of DNA



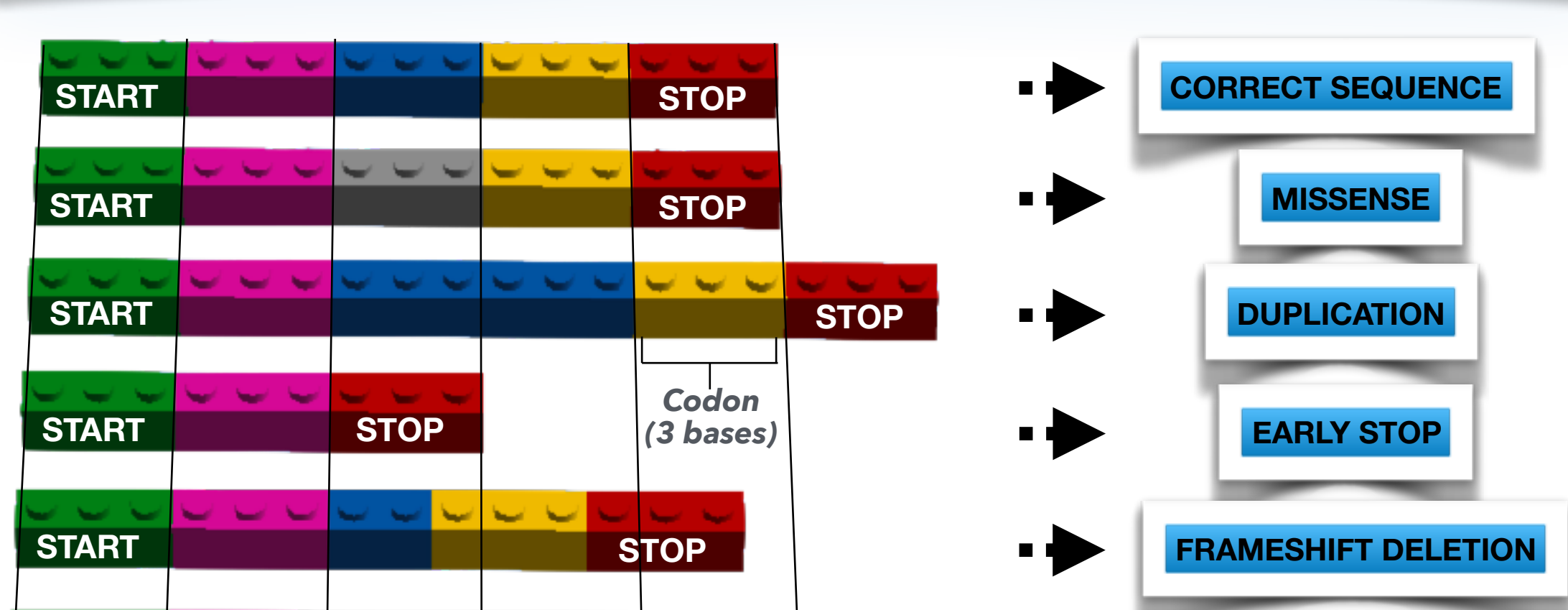
- A gene is segments of the DNA that function together
- Human DNA contains around 20000 different genes
- Genes produce proteins which are human body building blocks
- Most DNA is "non-coding" and does not produce proteins
- Only 2% of the DNA encodes proteins

DNA is a long string of bases



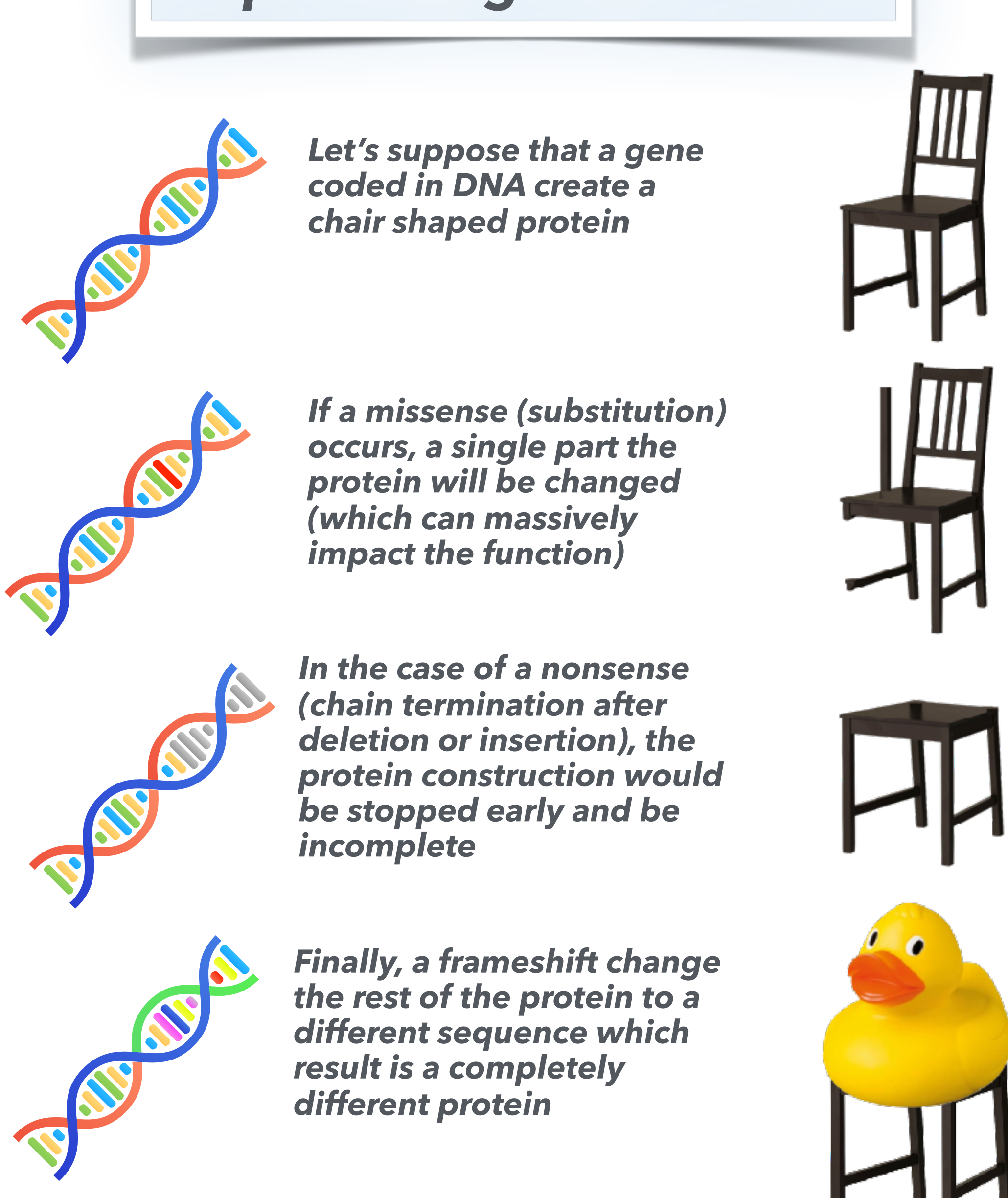
- DNA is like a long thread made of "base pairs" A-T and G-C only
- Genes contain the information to make proteins. Mechanism that make proteins read the message in three-base sets called codons
- One set of three means "start", others specify the 20 different amino acids (gene building blocks) or "stop"
- Changes in a gene sequence alter the protein it encodes
- The complete list of all bases is called genome

Main types of genetic mutation



- Insertion/deletion can cause a simple change or can cause a frameshift, which also causes a stop
- Most Syngap mutations are "de novo" (new/random, not inherited from the parents)
- Rarely, a mosaicism can occur. Reproductive cells with a syngap mutation from the otherwise unaffected parent can cause syngap syndrome in the child
- Speak to your geneticist about the possibility of being mosaic if you are having more children

Impact of a gene mutation



- Based on the known genetic results of syngap patients, any of the mutations will have an impact but there is no known correlation between the mutation and the severity of the condition

More information on:
www.syngapglobal.net

And
Facebook Syngap
Global Network

Note : information page was written by Syngap parents, not medical professionals.

