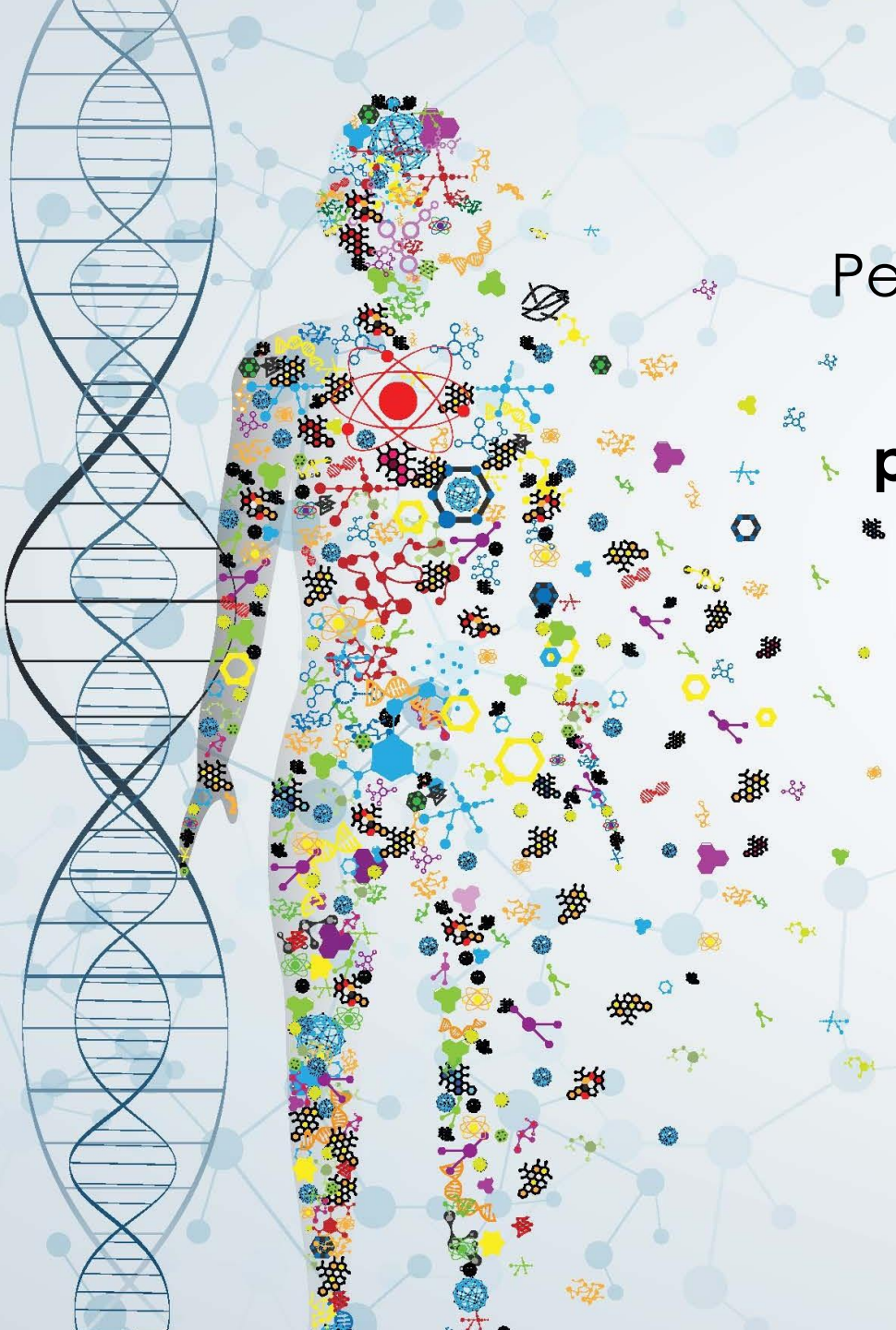
Introduction to Genetics

Sasha Mannion Maggs
MSc (Med) Human Genetics

DNALYSIS
PREDICTIVE GENE PROFILING



Personalized healthcare focuses on the **personalisation** of diet, exercise, lifestyle, supplement and prescription recommendations according to an individual's unique **genetic code**

Introduction to Genetics

What is DNA?

What is a gene?

Genetic mutation

Genetic variation

Nomenclature and Terminology

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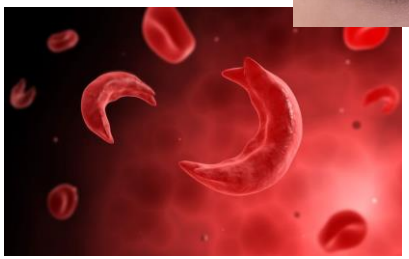
Traits

- Each individual is unique
- Visible traits

- Non-visible



- Inherited (genetic)
- Genes and environment

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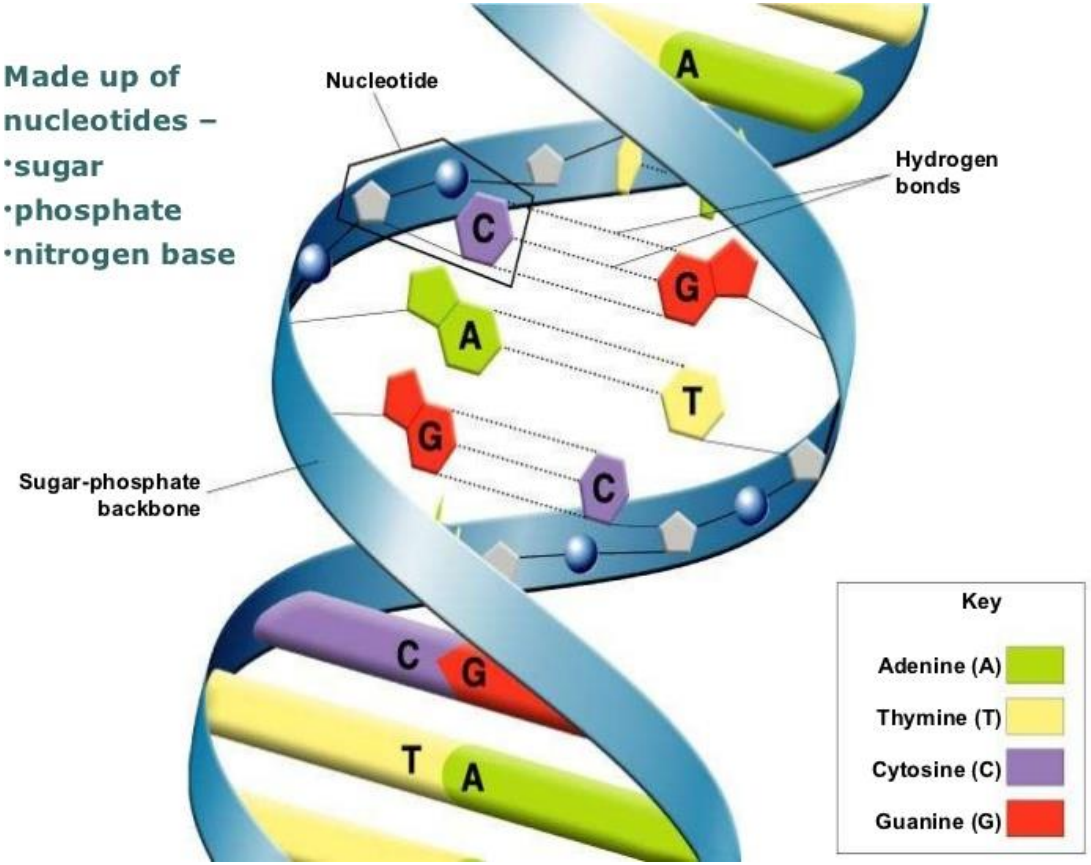
DNA

Made up of nucleotides –
- sugar
- phosphate
- nitrogen base



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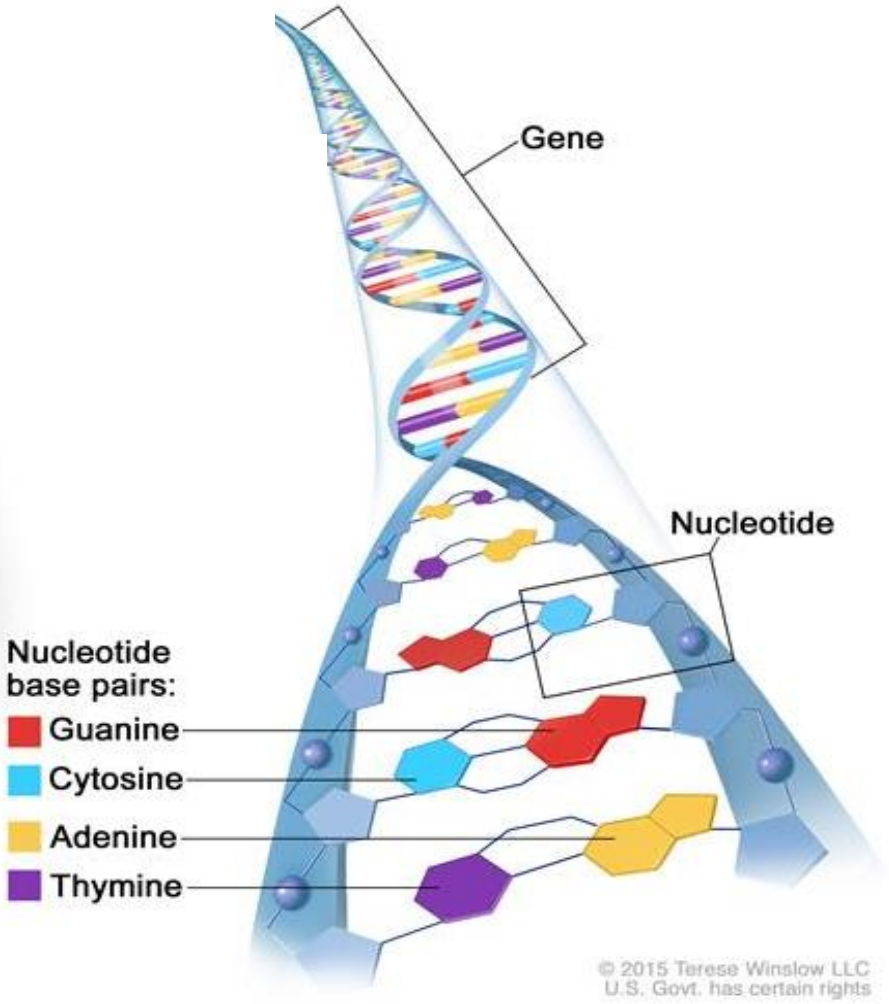
Gene



A gene is the basic physical and functional unit of heredity

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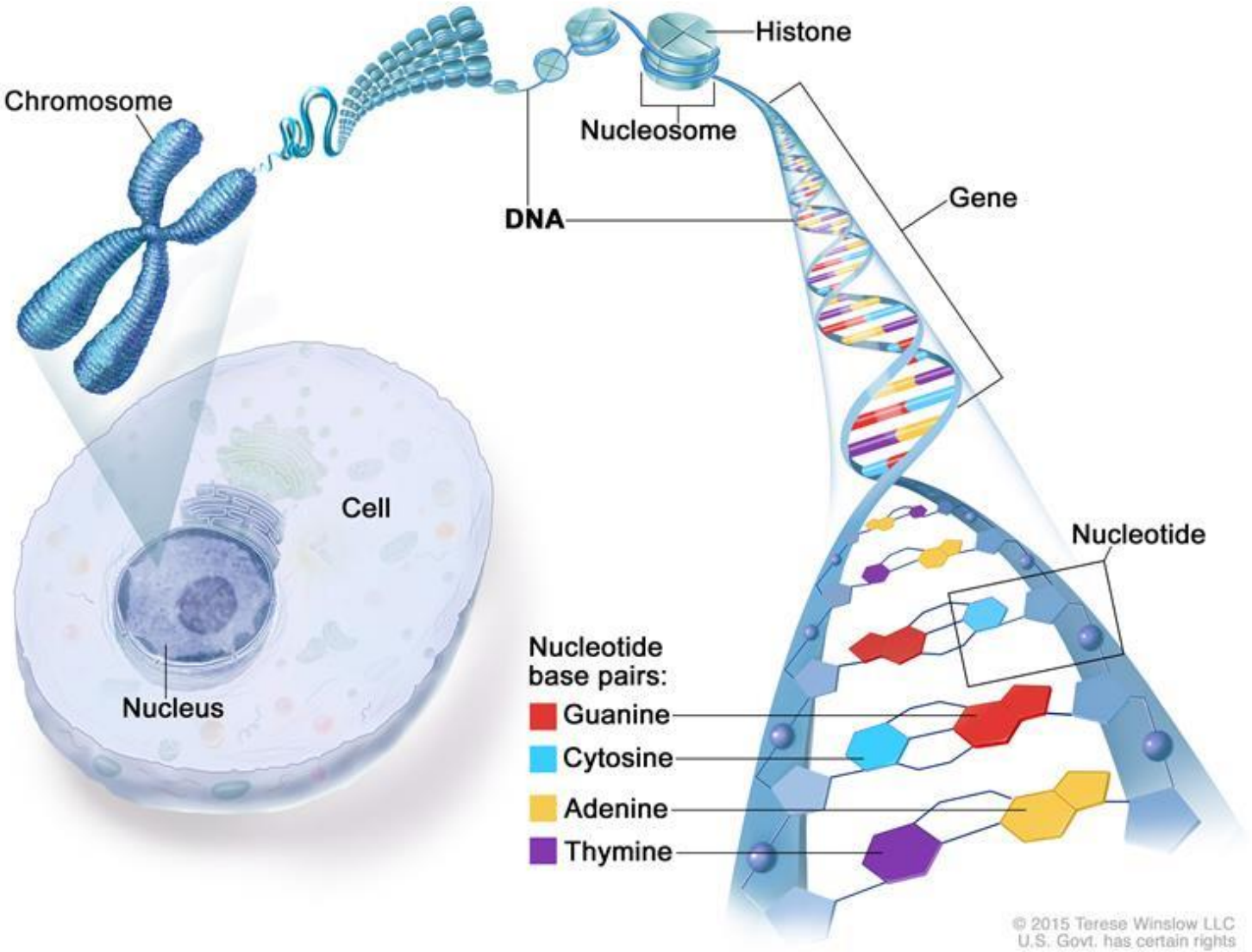
DNA Structure



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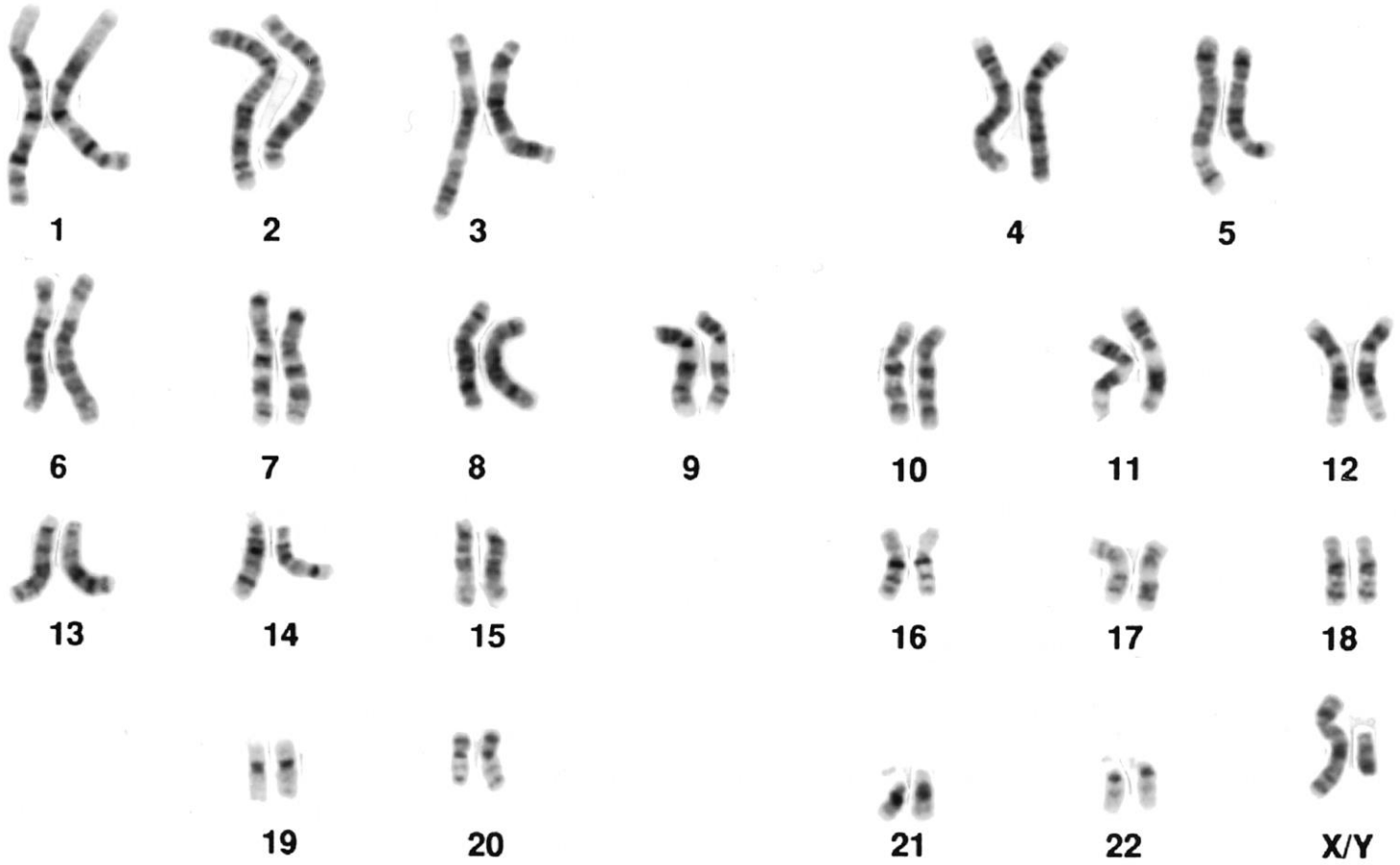
Chromosomes



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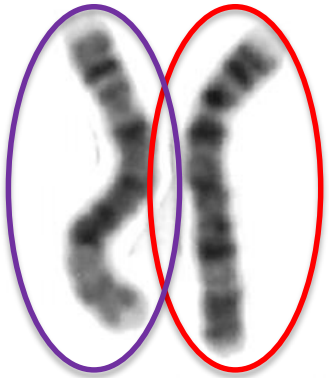

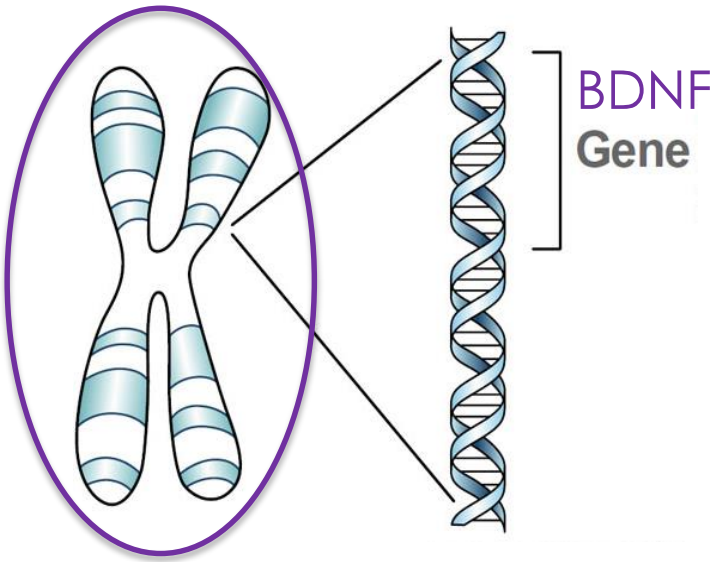

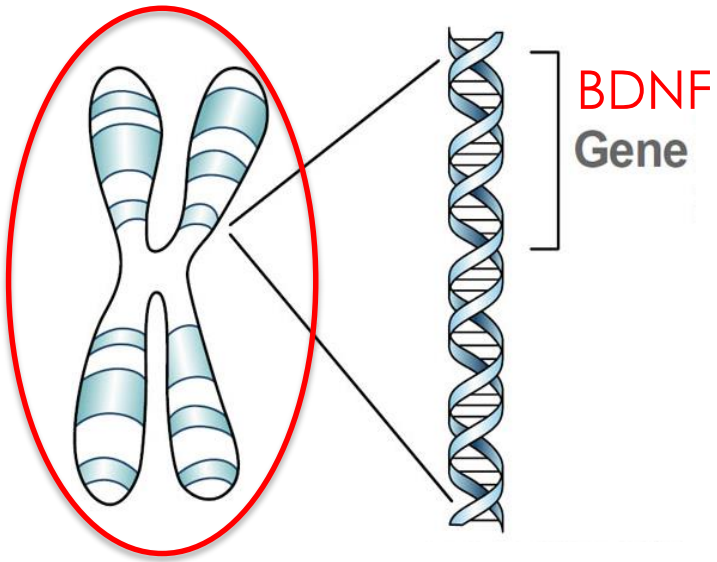

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BDNF gene

BDNF gene

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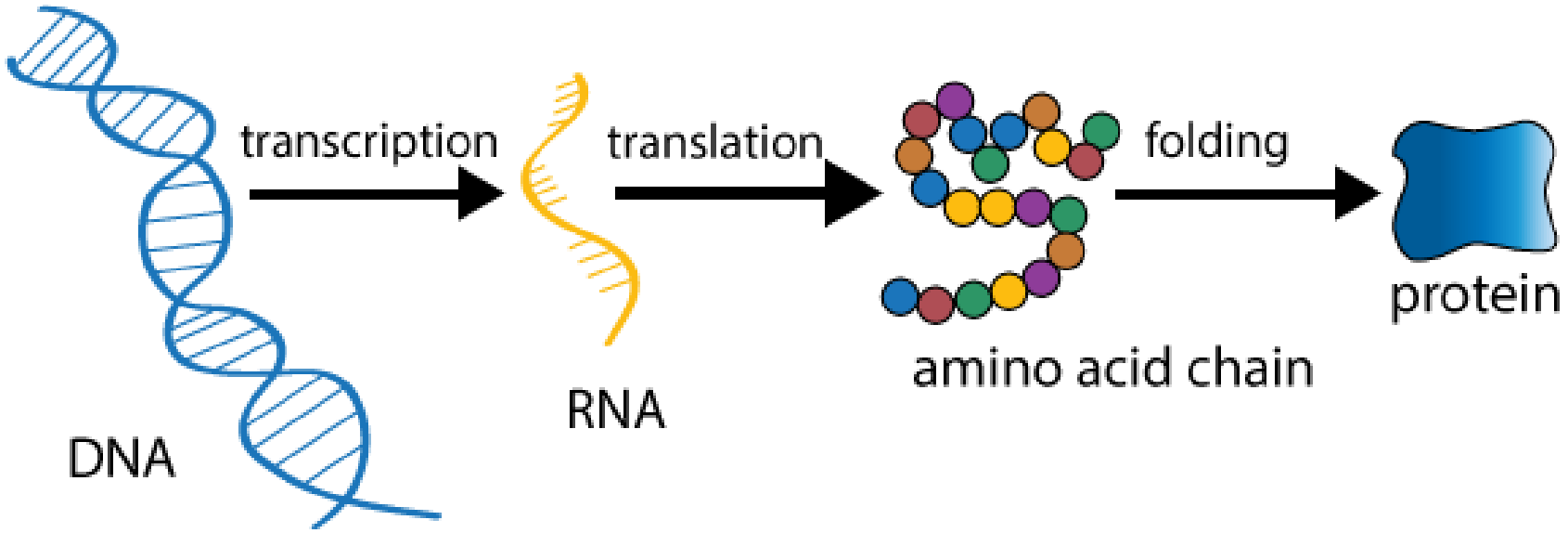
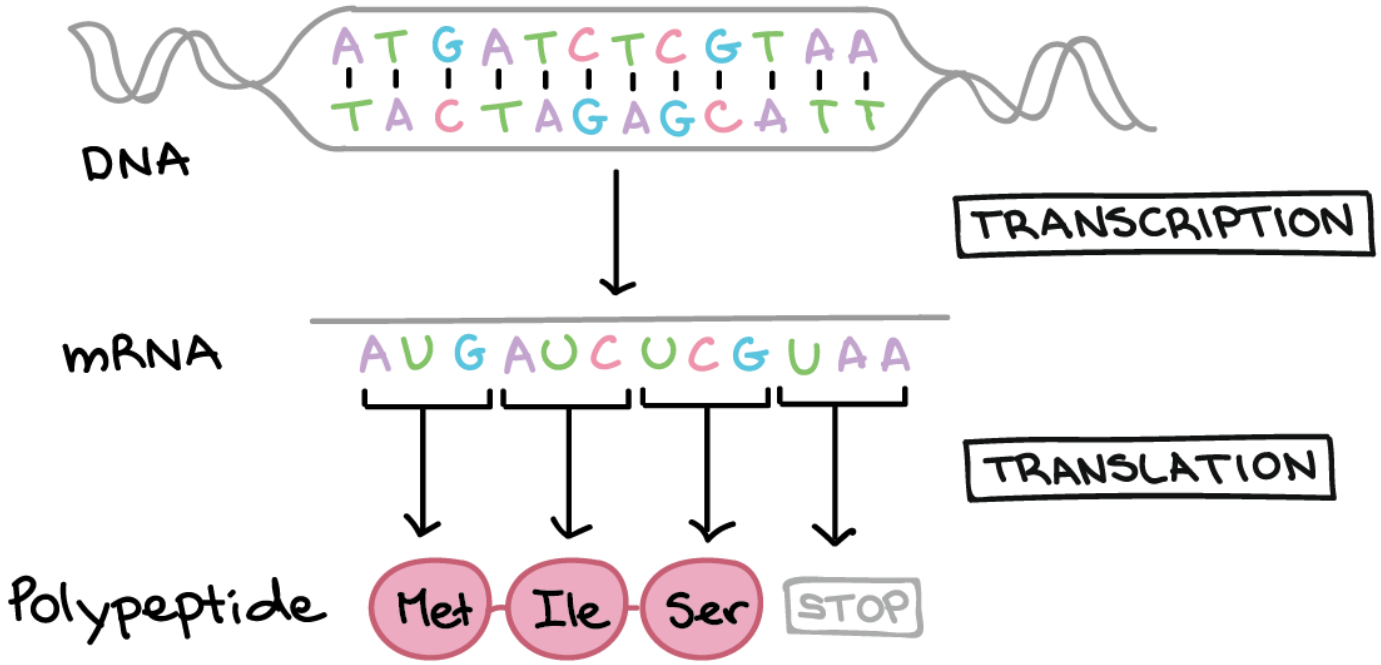
Central dogma of biology



BDNF gene → BDNF protein

Central dogma of biology



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DNA sequence variation

- The DNA of any two individuals is 99.9% identical
- The majority of the 0.1% variation has no functional significance
- Small genetic **variations** can influence the ability of genes to perform their required functions

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Genetic Mutation

- A mutation is a change in the genomic sequence of an organism that causes a discernible change in the characteristics of an organism
- High Penetrance (cause-effect relationship)
- Convey high probability that the effect will be expressed and "visible"
- Can be a single base-pair, entire gene, large segment of a chromosome or an entire chromosome

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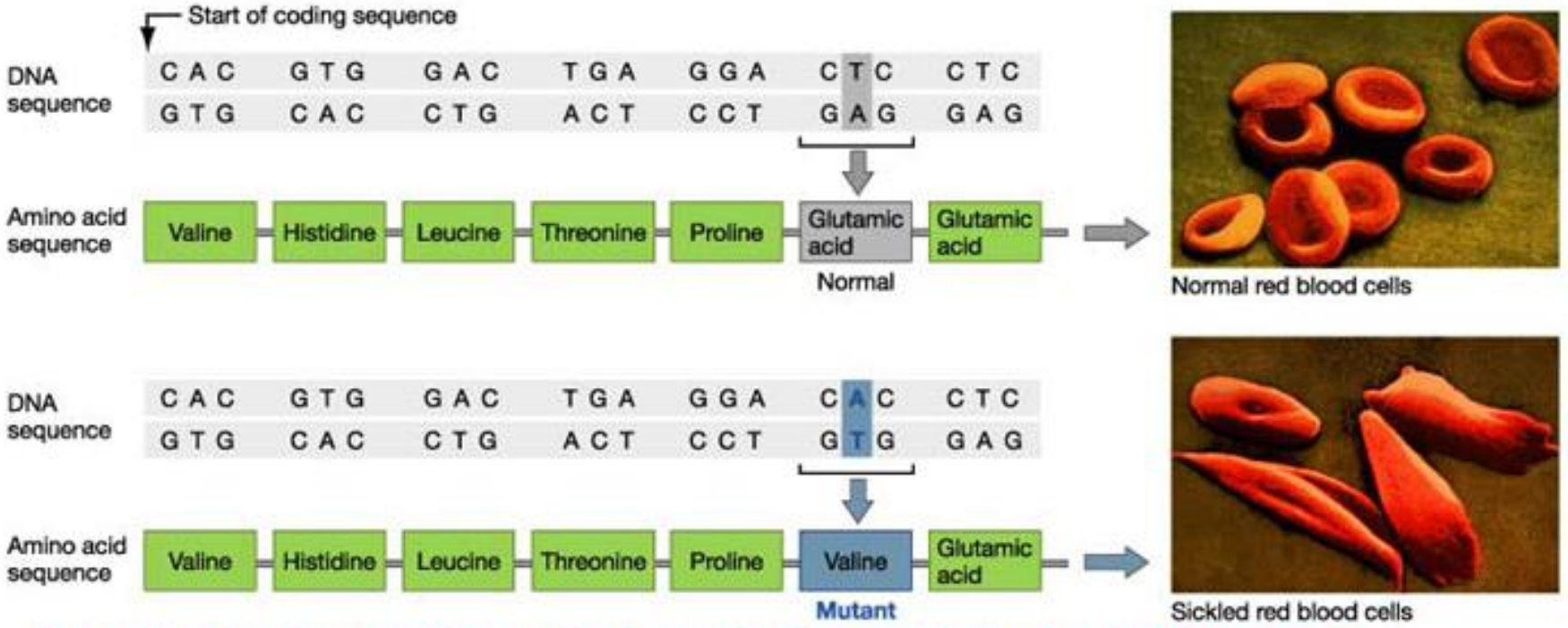
Genetic Mutation

Causes *disease*



The change in amino acid sequence causes hemoglobin molecules to crystallize when oxygen levels in the blood are low. As a result, red blood cells sickle and get stuck in small blood vessels.

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Genetic Variation

- Low Penetrance
- Minor impact, does not cause disease on its own
- Convey susceptibility to disease
- Usually single nucleotide polymorphisms or a small sequence of nucleotides

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Genetic Variation

CYP1A2
gene variant

CVD

Gene x Environment

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SNP

- **S**ingle **N**ucleotide **P**olymorphism
- A gene variant that is caused by a change in a single nucleotide/base
- These tiny variations in our genome make a significant difference, by affecting the ability of our genes to perform critical functions

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BDNF
rs6265
196 C>T
Val66Met

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Nomenclature

BDNF	gene name
rs6265	identification number
196 C>T	position of the nucleotide variation and possible alleles
Val66Met	position of the amino acid variation

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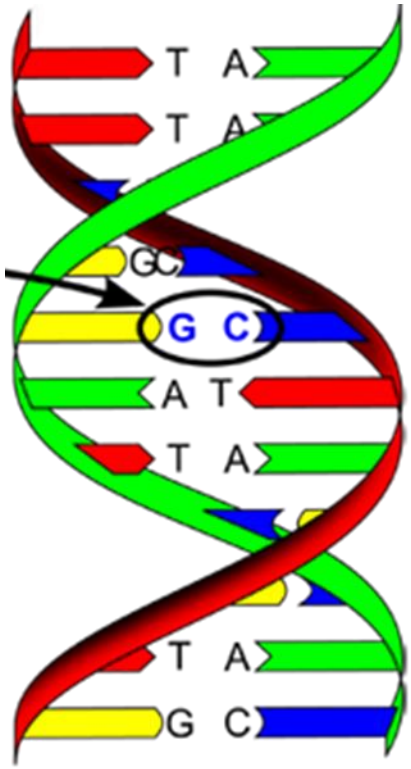
BDNF 196 C>T



C

BDNF 196 C>T

T

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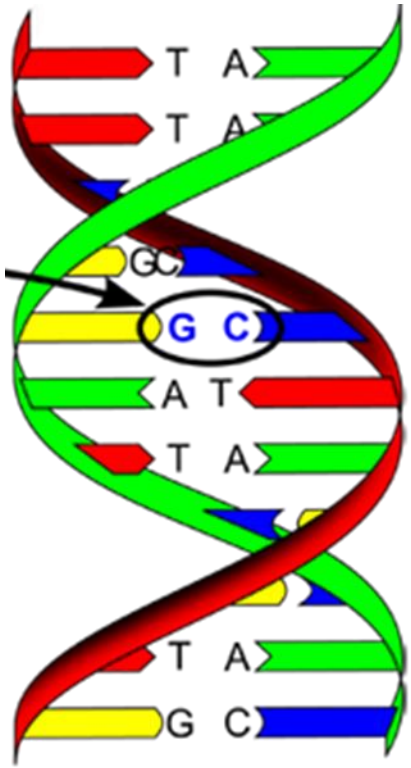
BDNF 196 C>T

(Allele 1)



C

BDNF 196 C>T

(Allele 2)

T

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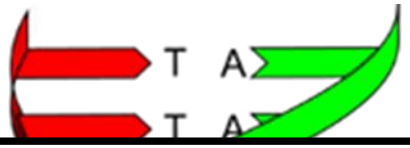
BDNF 196 C>T

(Allele 1)

BDNF 196 C>T

(Allele 2)

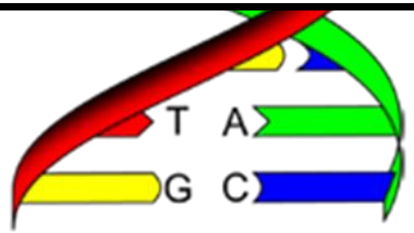


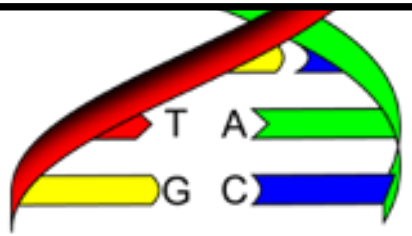
BDNF 196 C>T

Genotype

CT

Heterozygous





C

T

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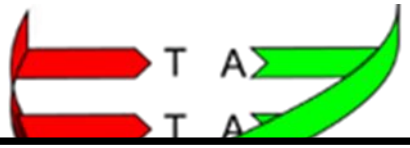
BDNF 196 C>T

(Allele 1)

BDNF 196 C>T

(Allele 2)

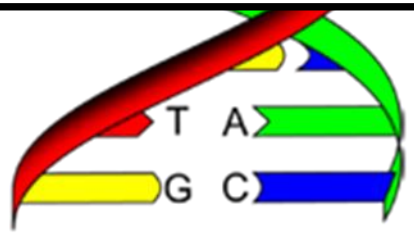


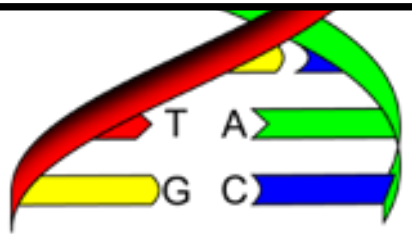
BDNF 196 C>T

Genotype

CC

Homozygous





C

C

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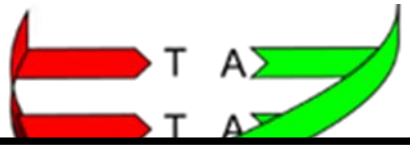
BDNF 196 C>T

(Allele 1)

BDNF 196 C>T

(Allele 2)

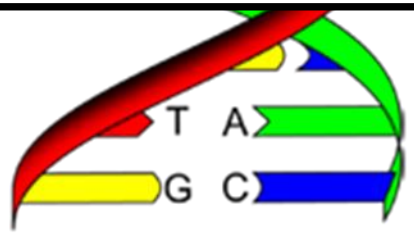


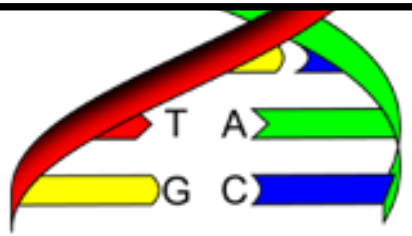
BDNF 196 C>T

Genotype

TT

Homozygous





T

T

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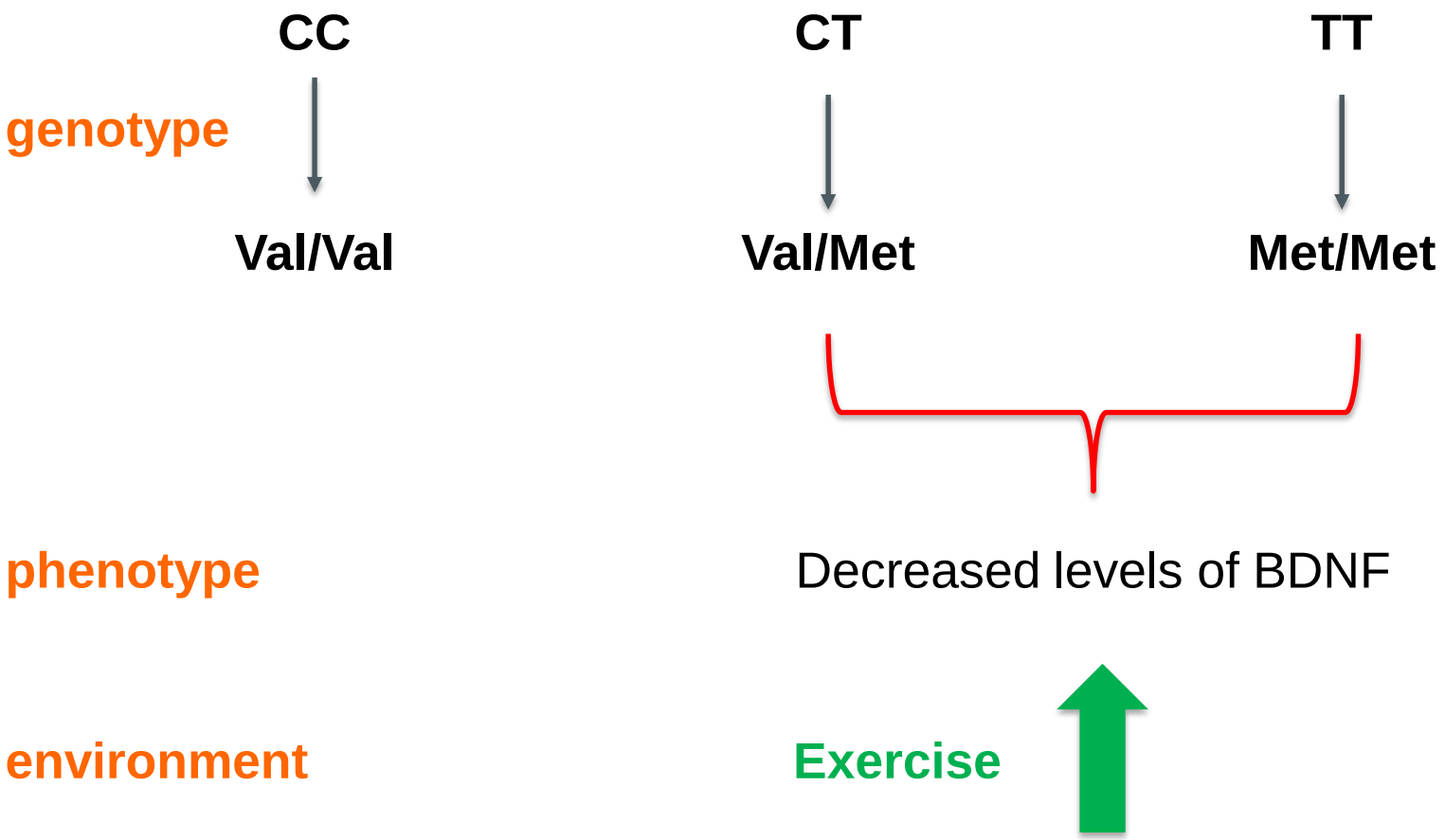
BDNF 196 C>T Val66Met



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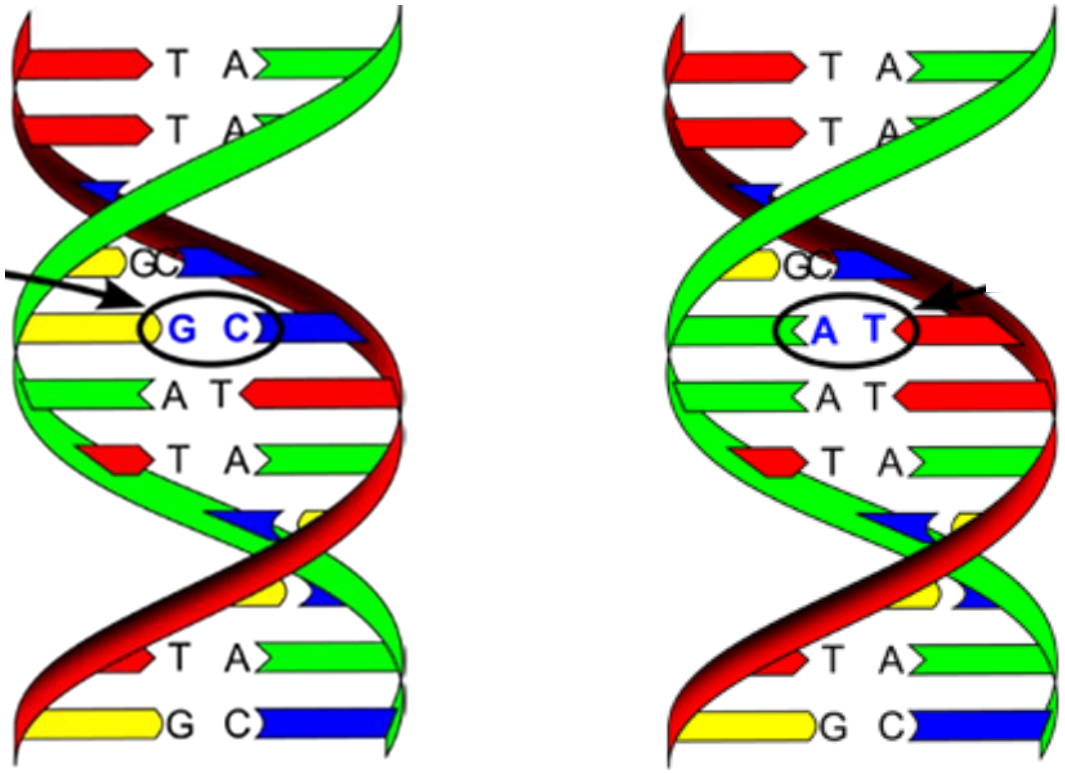
Complimentary alleles

BDNF 196 C>T



Recap

DNA is the carrier of genetic information passed from parent to offspring; made up of the 4 nucleotides A, T, C and G

Genes are base pairs in varying sequences and the basic physical and functional unit of heredity

Genes are located on **chromosomes**; humans have 23 pairs of chromosomes

A **SNP** is a common genetic variation between individuals involving 1 nucleotide

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Recap

An **allele** is one of a number of alternative forms of the same gene or same genetic locus

A **genotype** is the genetic makeup of a cell, an organism, or an individual usually with reference to a specific characteristic under consideration.

A **phenotype** is the composite of an organism's observable characteristics or traits, such as its morphology, development, biochemical or physiological properties, phenology, behaviour, and products of behaviour

SNP inclusion criteria

- ✓ Significant evidence linking the gene to a phenotypic outcome
- ✓ SNP within gene exerts direct influence over specific biochemical processes that create known symptoms or disease
- ✓ Effect of the SNP is modifiable by environmental factors such as diet, nutrients, exercise or lifestyle
  - ✓ Nutrients modifying gene activity are readily available
- ✓ SNP relatively common and relevant to the population
- ✓ Impact of SNP and clinical interventions measurable
- ✓ Potential negative effects of specific nutrient intake with certain genetic variants

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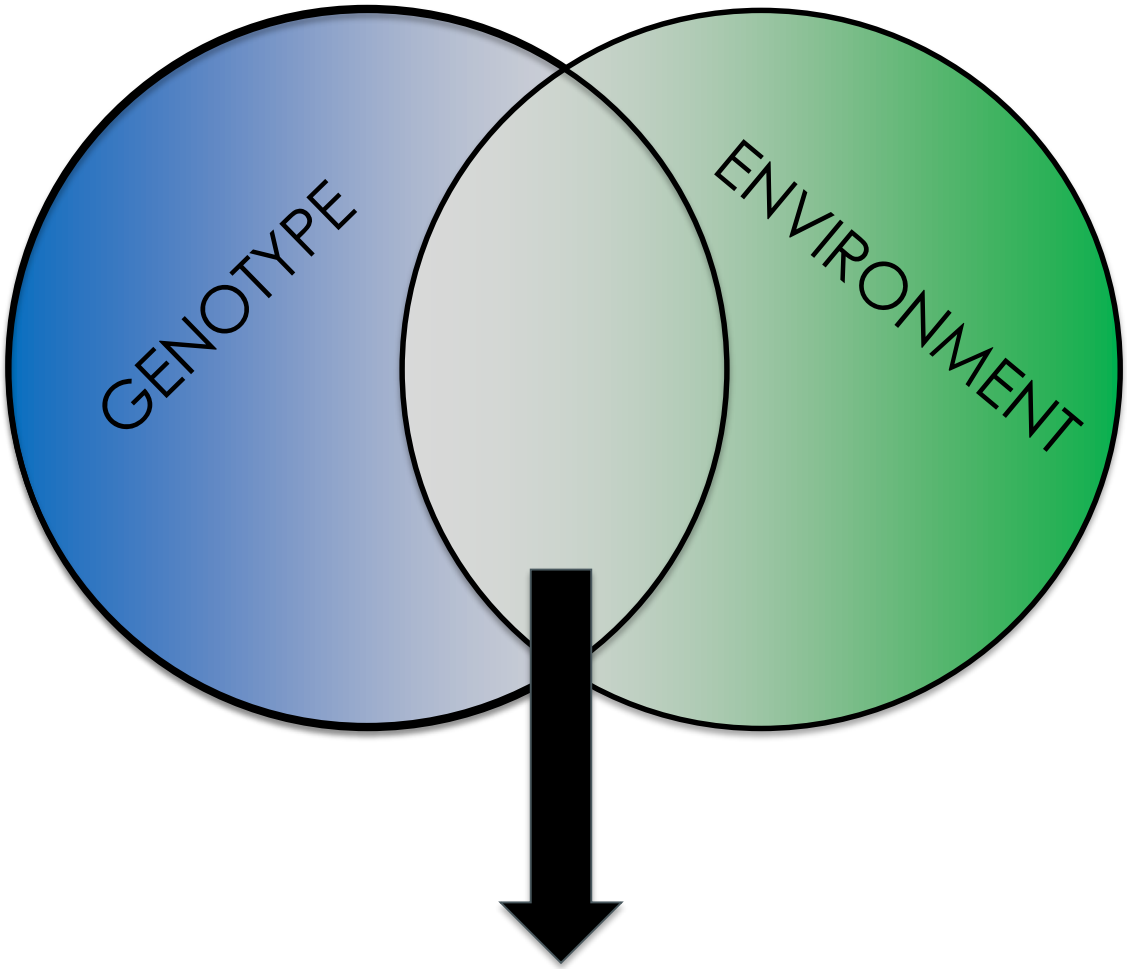

PHENOTYPE

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