



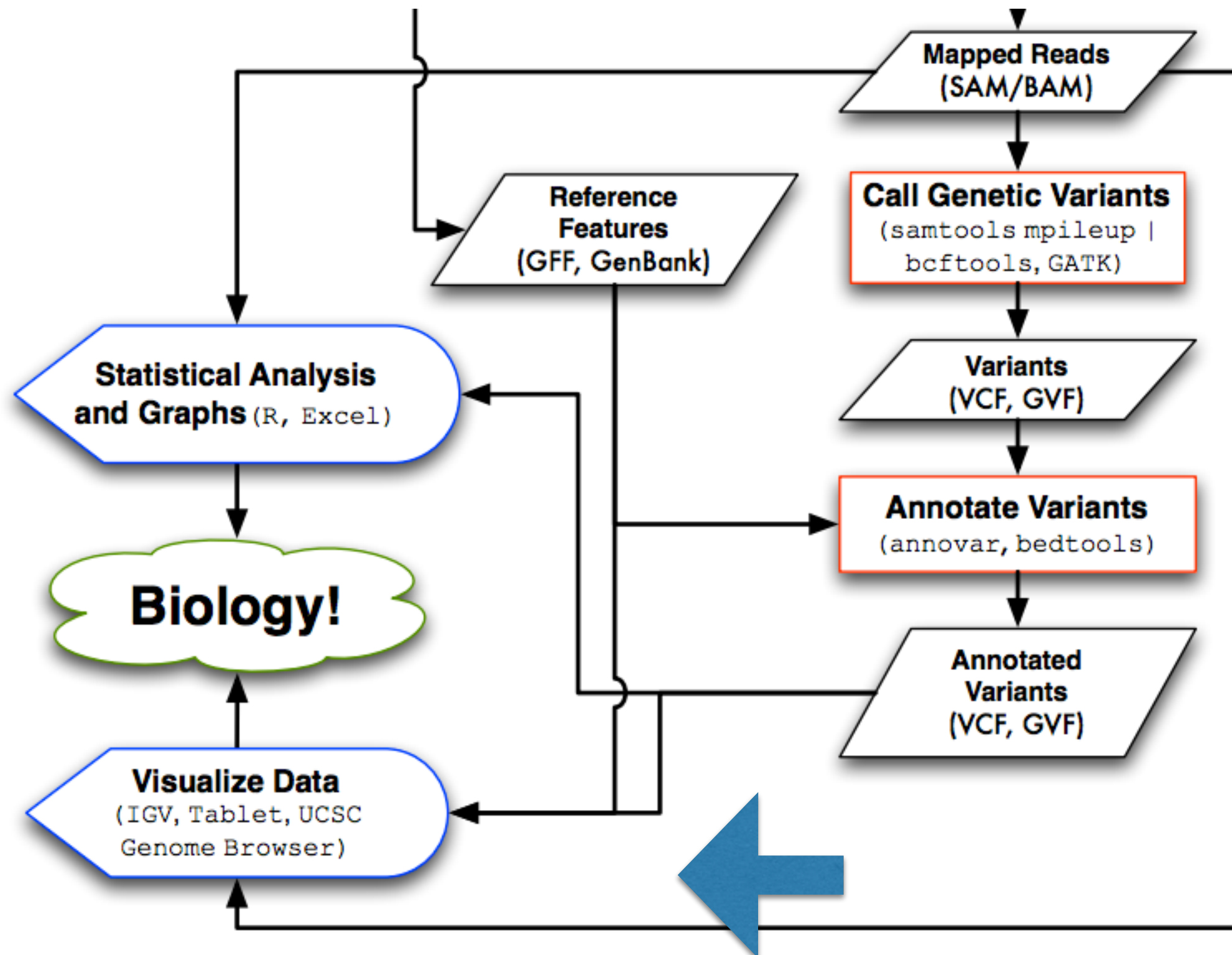
GENOMIKA

Priorização de variantes

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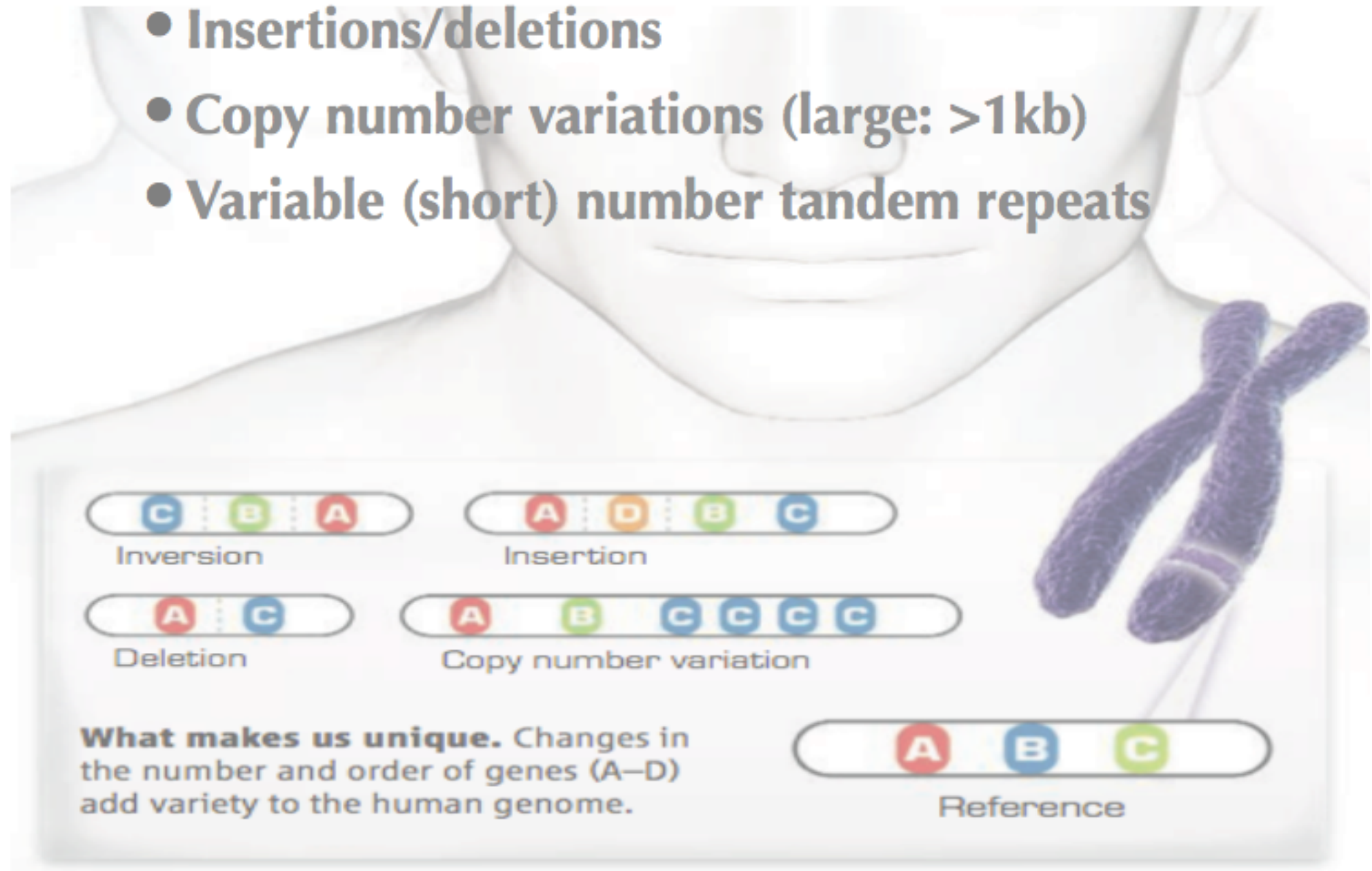
Pipeline



You are here!!

Genetic variation

- **Single nucleotide polymorphisms (SNP)**
- **Insertions/deletions**
- **Copy number variations (large: >1kb)**
- **Variable (short) number tandem repeats**



SNPs

Individual A:

ACGTGT**A**CGTGAAGGGGATTGGAATGGA
ACGTGT**A**CGTGAAGGGGATTGGAATGGA

Individual B:

ACGTGT**A**CGTGAAGGGGATTGGAATGGA
ACGTGT**G**CGTGAAGGGGATTGGAATGGA

Individual C:

ACGTGT**G**CGTGAAGGGGATTGGAATGGA
ACGTGT**G**CGTGAAGGGGATTGGAATGGA

SNP: One nucleotide difference occurring in at least 1% of a population

How many SNPs differ on average between you and you?

Human: 3 billion base-pairs

1. 0.01% (eg. 300k)
2. 0.1% (eg. 3 million)
3. 1% (eg. 30 million)
4. 10% (eg. 300 million)



How many SNPs differ on average between you and you?

Human: 3 billion base-pairs

~ one base pair out of every 1,000 will be different between any two individuals

1. 0.01% (eg. 300k)
2. 0.1% (eg. 3 million)
3. 1% (eg. 30 million)
4. 10% (eg. 300 million)



SNP and genotype

Human are diploid (we have 2 homologous copies of each chromosome)

At each SNP there are 3 possible genotypes:

- homozygous reference
- heterozygous
- homozygous non-reference

Homozygous = 2 identical alleles at given locus, eg. AA

Heterozygous = 2 different alleles at given locus, eg. AC

SNP information

- rs number
- location (chr:position)

Reference SNP(refSNP) Cluster Report: rs17822931 ** With probable-pathogenic...<http://www.ncbi.nlm.nih.gov/sites/varvu?gene=85320&rs=17822931> [detail] **

http://www.ncbi.nlm.nih.gov/ SNP/snp_ref.cgi?rs=17822931

NCBI dbSNP Short Genetic Variations

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books SNP

Search for SNP on NCBI Reference Assembly

Search Entrez: SNP t for Co

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Reference SNP(refSNP) Cluster Report: rs17822931 ** With probable-pathogenic allele [detail] **

RefSNP

Organism: human ([Homo sapiens](#))

Molecule Type: Genomic

Created/Updated in build: 123/135

Map to Genome Build: [37.3](#)

Validation Status: [View](#)

Citation: [PubMed](#)

Allele

Variation Class: SNV: single nucleotide variation

RefSNP Alleles: C/T

Allele Origin: G: Germline A: Germline

Ancestral Allele: C

Clinical Source: [View](#) [OMIM](#)

Clinical Significance: [With probable-pathogenic allele \[detail\]](#)

MAF/MinorAlleleCount: T=0.310/579

MAF Source: 1000 Genomes

HGVS Names

NC_000016.9:g.48258198C>T

NG_011522.1:g.15891G>A

NM_032583.3:c.538G>A

NM_033151.3:c.538G>A

NM_145186.2:c.538G>A

NP_115972.2:p.Gly180Arg

NP_149163.2:p.Gly180Arg

NP_660187.1:p.Gly180Arg

SNP Details are organized in the following sections:

[GeneView](#) [Map](#) [Submission](#) [Fasta](#) [Resource](#) [Diversity](#) [Validation](#)

Integrated Maps (Hint: click on 'Chr Pos' or 'Contig Pos' column value to see variation in NCBI sequence viewer)

Assembly	Genome Build	Chr	Chr Pos	Contig	Contig Pos	SNP to Chr	Contig allele	Contig to Chr	Neighbor SNP	Map Method
GRCh37.p5	37.3	16	48258198	NT_010498.15	1872397	+	C	+	view	blast
reference	36.3	16	46815699	NT_010498.15	1872397	+	C	+	view	blast
Celera	36.3	16	32785323	NW_926462.1	1830122	+	C	+	view	blast
HuRef	37.3	16	34148352	NW_001838268.2	3012385	+	G	-	view	blast
HuRef	36.3	16	34148352	NW_001838268.2	3012385	+	G	-	view	blast

GeneView

View more variation on this gene (click to hide).

☒ Clinical Source: ☐ in gene region ☒ cSNP ☐ has frequency ☐ double hit Co

Primary Assembly Mapping

Assembly	SNP to Chr	Chr	Chr position	Contig	Contig position	Allele
GRCh37.p5	+	16	48258198	NT_010498.15	1872397	C

RefSeqGene Mapping

RefSeqGene	Gene (ID)	SNP to RefSeqGene	Position	Allele
NG_011522.1	ABCC11 (85320)	-	15891	G

Gene Model(s)

SNP consequence



Coding SNPs:

- non-synonymous coding (missense)
 - change of an amino acid
- synonymous coding (silent mutation) – no change in protein sequence
- stop codon (nonsense) - premature stop codon
- frame-shift coding – insertion/deletion resulting in frame shift

Non-coding SNPs:

Mostly non-functional, but may affect:

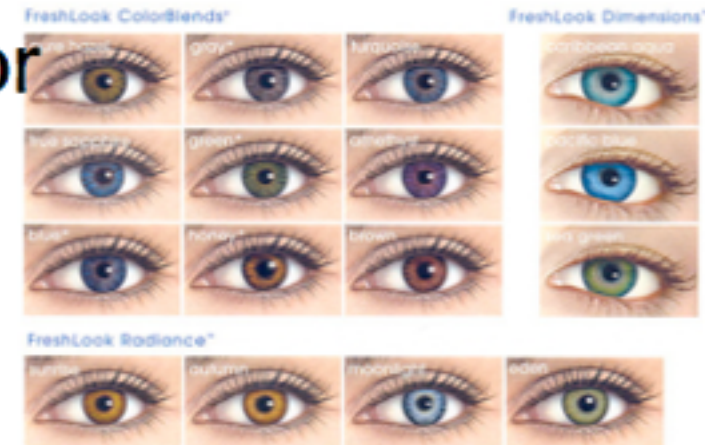
- regulatory region
- splice site
- transcription factor binding
- messenger RNA degradation
- sequence of non-coding RNA

Phenotype

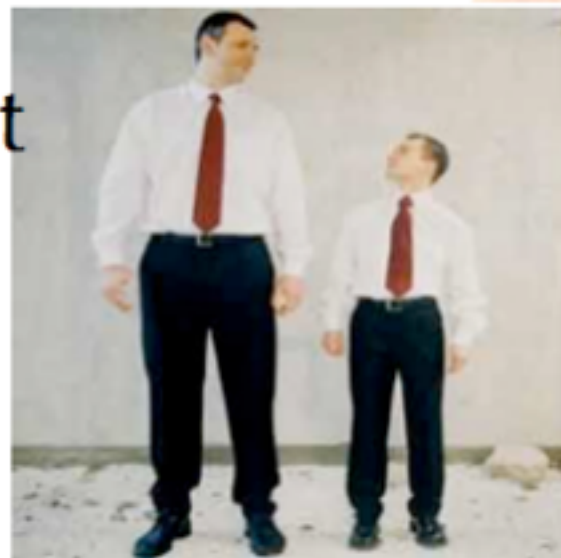
Phenotype = organism's observable characteristic or trait



eye color



height



An organism's genotype is a major influencing factor in the development of its phenotype



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