The Science of Genetics

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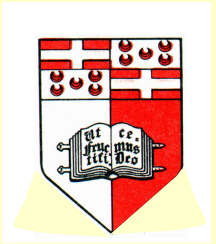
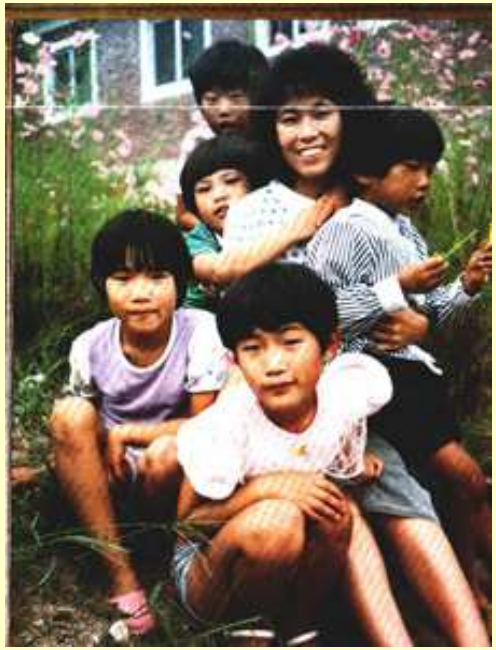


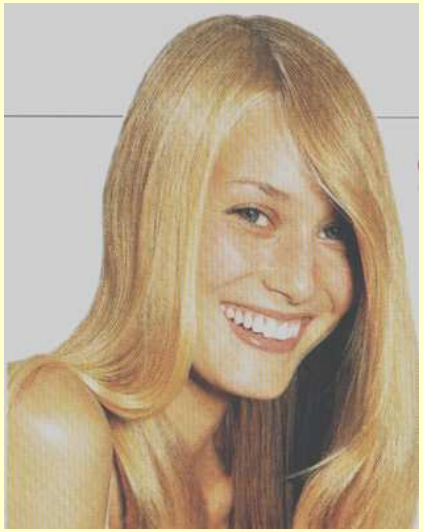




Genetics is diversity- Genetics is life





Human diversity is

What we look like

How we live

How we think

Language

Religion

Culture

… an interaction between genetics and environmental experiences

Beneath
the striking diversity
is
a an even more striking
similarity...

- same anatomy,
- same physiology
- same biochemistry

…. which extends
beyond
human beings

I’m not so different!

I have a very similar
- anatomy,
- physiology
- biochemistry

I even gave my heart for transplantation to humans.

Human diversity also includes features that are not easily visible

Specialized areas of genetics
These are some examples.

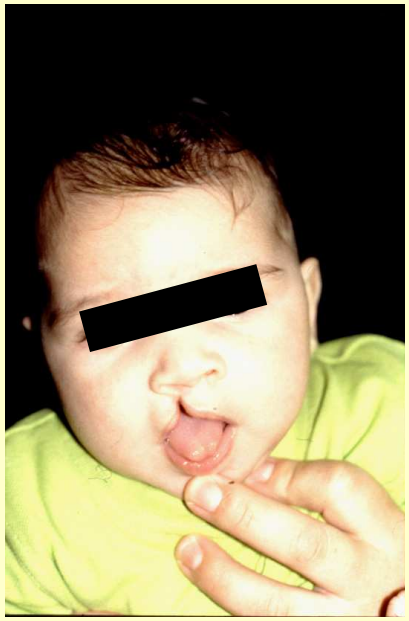
Behaviour	Neuropsychiatric genetics
Development	Developmental genetics
Blood & tissue types	Immunogenetics
Enzymes	Biochemical genetics
Metabolism of drugs	Pharmacogenetics

Among human diversity is that caused by disease conditions

Achondroplasia (dwarfism)

Vitamin D-Resistant Rickets

Collodion baby



Cleft lip

Diseases Are Very Important Aspects Of Genetics

Genetic diseases may involve:

External features (developmental disorders)

- dysmorphology
- congenital anomalies

Behaviour (neuropsychiatric disorders)

- mental retardation
- hyperactivity-attention deficit disorders
- schizophrenia, biphasic disorders
- addiction to alcohol and drugs

Biochemical defects (metabolic diseases)

- e.g. gangliosidosis, hypercholesterolaemia
- drug intolerance

Genetics has been crucial in understanding many basic aspects of life.

How cells function
Molecular biology
Developmental mechanisms
Ageing
Evolution

50 Years of Work on the Molecular Structure of Genes Has Culminated in the Human Genome Project.

A genome

is the total complement of genes present in an organism.

The Human Genome Project defined the complete genetic sequence in which are contained all the secrets of life.

The Human Genome Contains the Blueprint of Human Life.

The Human Genome

"the language in which God created life….
….. the complexity, the beauty, the wonder of God's most divine and sacred gift.“

(W.J. Clinton, 2000)

The Human Genome

A sequence that defines our identities.

“William Jefferson Clinton .....
was famously caught out by it.”

(Richard Aedy, 2000)

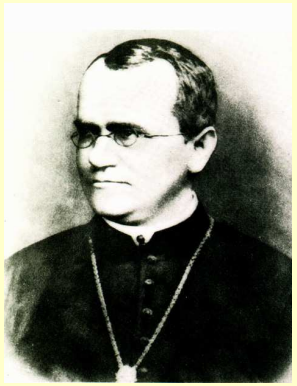
The Human Genome Project has opened up new avenues

- Exploring and understanding diseases
  - diagnosis, treatment and prevention
- Deeper understanding of human life
- Control of the transmission of human life
- Normal and abnormal human development
- Evolution

The human genome is expected to have a tremendous impact on health and disease and a far-reaching influence on the social aspects of life.

The Science of Genetics Developed Over the Last 150 Years

Three Eminent Persons in Genetics



Gregor Mendel (1822-1884) studied different characteristics: proposed the hypothesis of one gene - one characteristic.
A unifying concept of variation

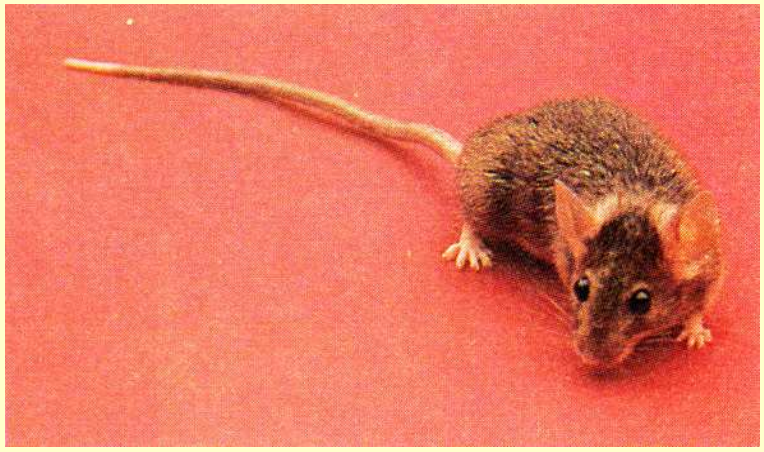
Charles Darwin (1809-1882) proposed the theory of evolution and common ancestry and noted similarities among organisms.
There is similarity amidst diversity

Thomas Hunt Morgan (1866-1945) analysed how genes are related to one another,
The beginnings of gene mapping.

Two Prominent Organisms Used in the Study of Genetics

Drosophila, the fruit fly
was the protagonist in genetics.

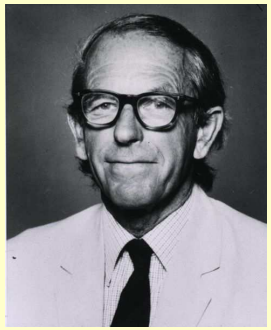
- It has a short generation time
- produces hundreds of offspring
- has giant chromosomes



The mouse, a quiet intruder, was an intermediate in the study of mammalian genetics.
Extensively used for generating transgenic organisms including human genes

Eminent Persons in Genomics

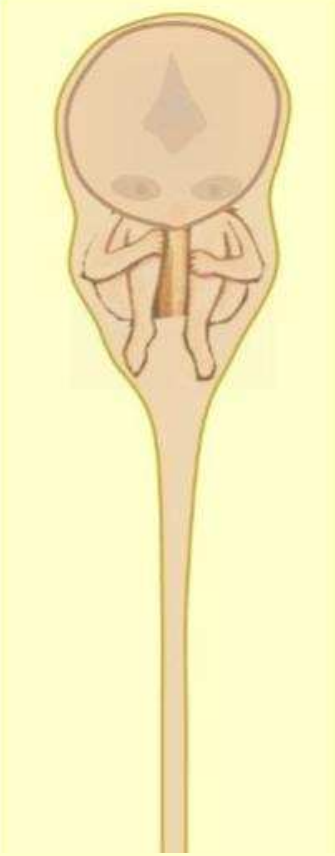

James Watson and Francis Crick (1953) proposed the molecular structure of DNA
The foundations of molecular genetics



Frederick Sanger (1977) first to determine a gene sequence (in a bacteriophage)
The foundations of gene sequencing

Francis Collins (1989) sequenced a human gene (of cystic fibrosis); directed the Human Genome Project
The foundations of Genomics

A Radical Change in the Concept of Life

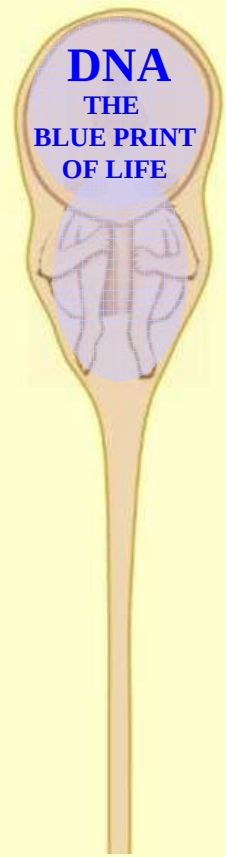


Early concepts of life

The sperm contains a tiny homunculus, a miniature human being, which is nurtured and grows in the mother's womb.

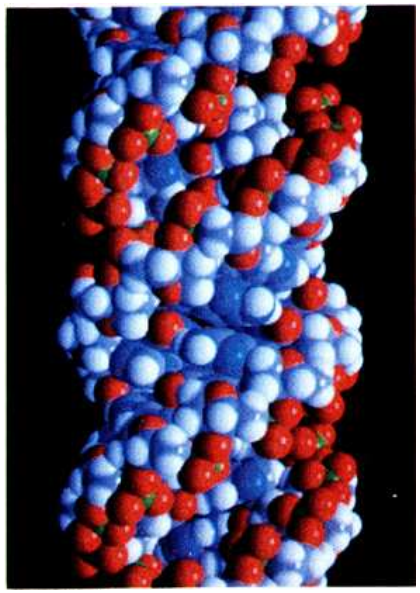
Modern concepts of life

DNA contains all the information to form a living organism with equal contributions from both parents - the blueprint of life

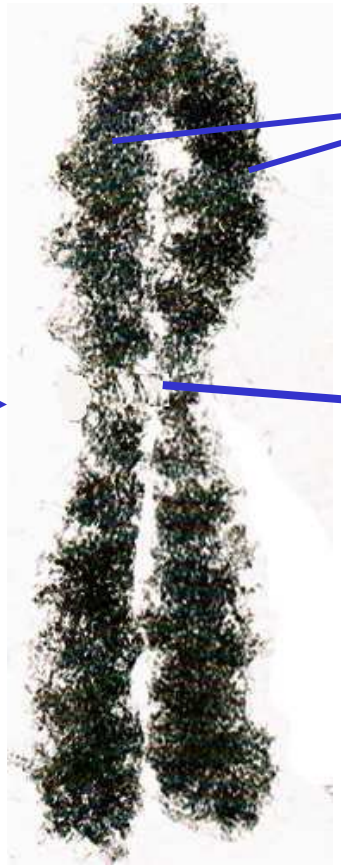


The Genetic Material

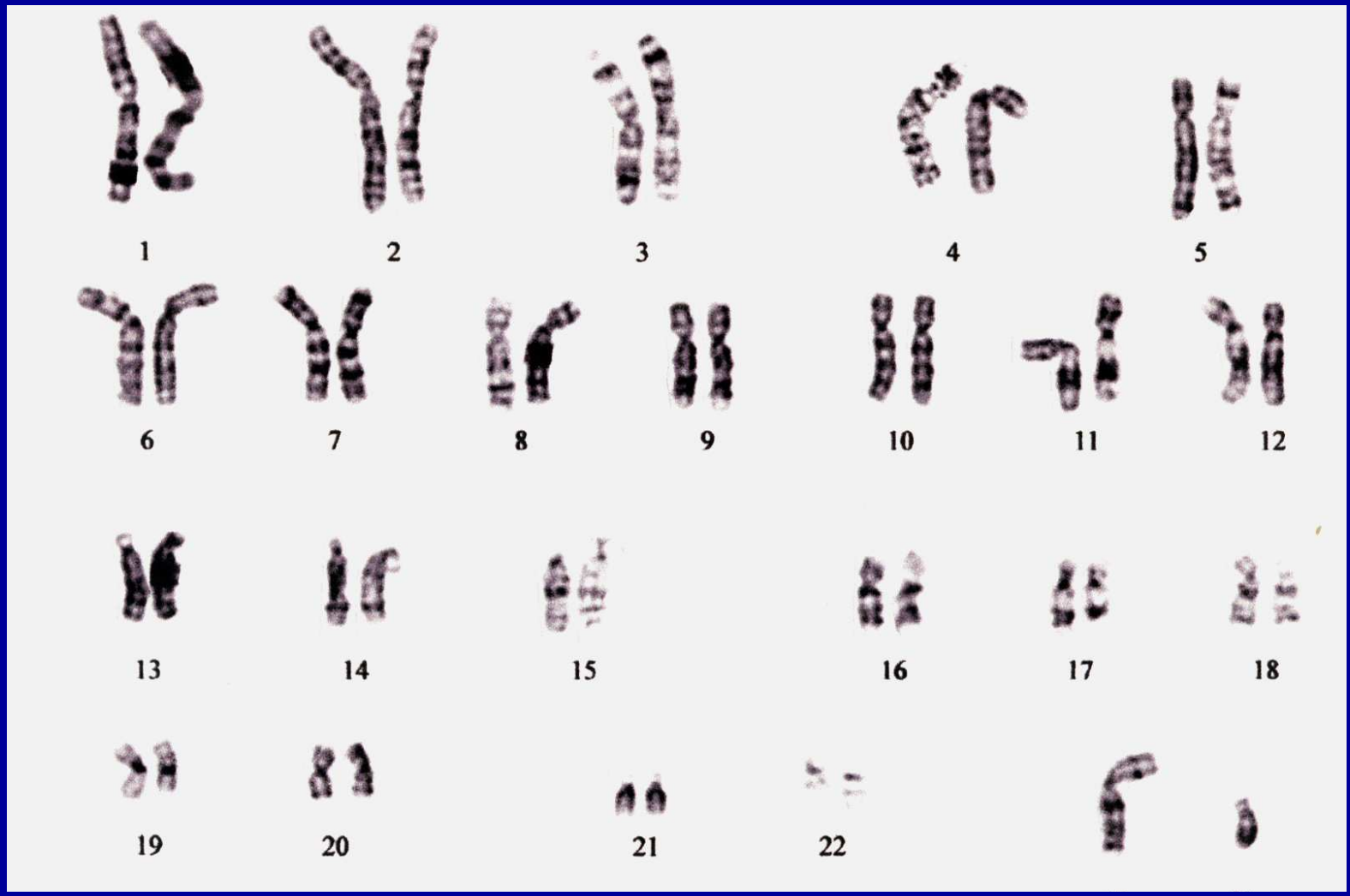
The DNA molecule constitutes the genetic material



It is packaged in the form of chromosomes, each consisting of two chromatids joined by a centromere

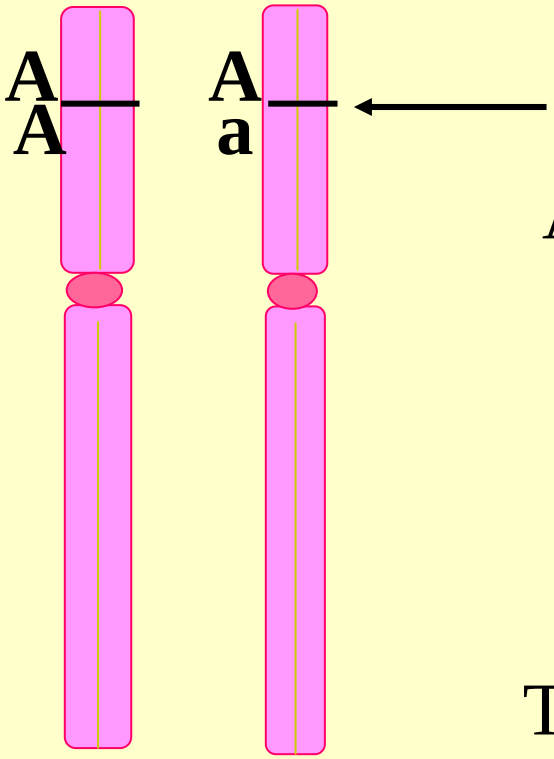


The Human Karyotype



Chromosomes occur in homologous pairs that are identical in size, shape and arrangement of genes.

A gene locus is the specific position of a particular gene on a chromosome.



Homologous pair

Alleles are the alternative forms of a gene that occupy a specific locus.

Alleles: A, a, A', a' …etc

There may be several alleles but only one pair is present in a particular individual

If there are 2 alleles: A a

There are 3 possible pair combinations :

AA	**Aa**	**aa**
Homozygous	**Heterozygous**	**Homozygous**

In AA and aa the alleles are identical or **homozygous**

In Aa the alleles are different or **heterozygous**

These are the **genotypes**.

The genotype is the allelic or genetic constitution of a particular individual.

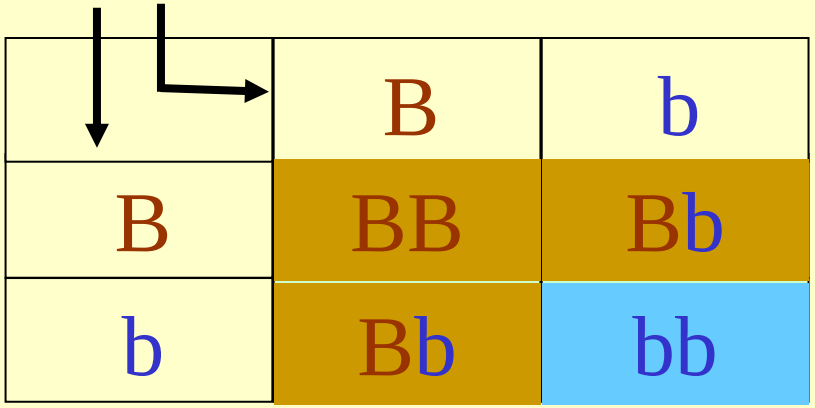
Example: Eye Colour

ALLELES	**B -gene for brown eyes** **b -gene for blue eyes**		
GENOTYPES	**BB**	**Bb**	**bb**
PHENOTYPES	**Brown eyes**	**Brown eyes**	**Blue eyes**

The phenotype refers to the characteristics manifest in an individual, including the morphological, behavioural,biochemical and other manifestations .

Construct a Punnett Square to work out how genotypes are inherited in the offspring

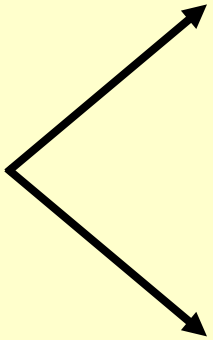
genotype of parents



	Phenotypes	Genotypes
Parents:	both brown eyes	both heterozygous
Offspring:	3 brown eyes	- 1 homozygous; 2 heterozygous
	1 blue eyes	- homozygous

Alternative Punnett Squares

Brown eyes
x
Brown eyes



	B	B
B	BB	BB
b	Bb	Bb

	B	b
B	BB	Bb
b	Bb	bb

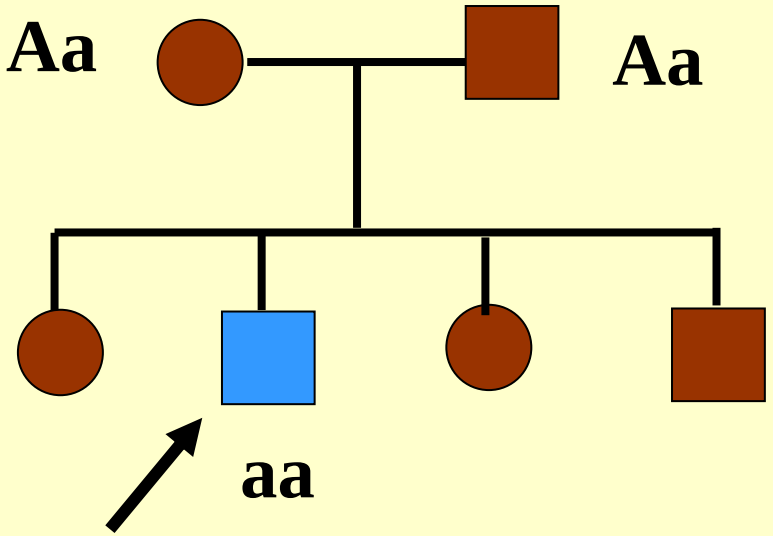
Brown eyes
x
Blue eyes

	b	b
B	B b	B b
b	b b	b b

Construct a pedigree

A pedigree shows the phenotypes

Genotypes can be added if known



Propositus: the individual through whom the family came to be investigated


The
path of man
The diversity of nature

Mendel's Laws

1. Law of **segregation**
   - Each characteristic is determined by two factors (or genes), which segregate in the offspring
2. Law of **independent assortment**
   - each gene is inherited independently of others