

ABHIJIT RAY AIR 50 UPSC CSE 2021

ANTHROPOLOGY PAPER 1 - 9.1 to 9.6

If you have made your own notes, please do go through mine once and note down the value addition and extra things. Trust me, you will be benefited as these notes are made from extensive sources.

— A ~~genetic marker~~ is a chromosome. It helps identify individual or species.

Uses:

- ① Genetic disorders inheritance
- ② Trace genealogy
- ③ Genetic map

GENETIC MARKER

INTRO

Blood groups obey Mendelian rules of inheritance with gratifying precision. Blood is made up of Plasma, RBC, WBC, platelets.

Genetic markers in RBC

- ✓ Antigen: Blood group ABO, MNS, Rh etc
- ✓ Hemoglobins - HbA, HbS, HbC, HbF, Tn
- ✓ Enzymes - G6PD

+ HLA
+ Gm
+ plasma
HP Tf

ABO system 1900 by Landsteiner

① Mendelian inheritance
② Easy to detect
③ Set of simply inherited variations, easy to detect and unaltered by age, sex and environment.
④ Not affected by diet, sex and environment.

1904, Behrendtals established the mode of inheritance of blood groups (Behrendtals)

3 alleles	A, B, O	O → recessive, A & B co-dominant
Genotypes	I ^A , I ^B , I ^O	Phenotypes
OO		O
AB		AB
AA		A
AO		
BB		B
BO		

1900/1924

According to **(Byrd)** there are some advantages of using blood groups for racial classification:

Inherited acc. to Mendelian principles

② Unaltered by food, nutrition, climate, medical condition

③ Frequency in pop'ls stable.

④ They probably arose very early

⑤ Sharply distinguished blood group.

⑤ Thru
⑥ Cor
trib

Blood pr
A (I^A)
B (I^B)
AB (I^A, I^B)
O (I^O)

Distribu

A → Asia, Eu, Am
B → North India, S. Cen. Amer

Eu ← B → At

Rh

1940

If wa
monkey
Hence

Those

Also,
system

Initially
as a

RhRh, R

JTRO

ence with
ma, RBC,

+ HLA
+ Gm
+ plasma
HP Tf

co-dominant
and un-
inheriting of

co-dominant

00/1924

advantages
classification:

clinical condition

- ⑤ There was a correlation b/w gene frequency and some disease distribution of blood group.
- ⑥ Considerable correlation b/w geography and the distribution of blood group.

Blood group	Antigen	Antibody	Accept	Donated
A ($I^A I^A, I^A I^O$)	A	Anti B	A, O	A, AB
B ($I^B I^B, I^B I^O$)	B	Anti A	B, O	B, AB
AB ($I^A I^B$)	A & B	No antibody	All	AB
O ($I^O I^O$)	None	Both Anti A & Anti B	O	All

Distribution:

A → Asia, Eu, Australia

B → North India and central Asia

$$A \rightarrow 0.215 \quad B \rightarrow 0.162 \quad O \rightarrow 0.623$$

O → American Indians (90%), Australian (90%)

B → Sub-Himalayan (25-30%), Europe (15%)

African (40%)

$B \leftarrow A \downarrow A \uparrow$ A → Primarily in Europe

Aust East to West →

Blood grp → Gastric & duodenal cancer

A → Pancreatic & breast cancer, Diabetes

Rh system

$A \uparrow B \downarrow$

O → gastric & duodenal cancer

A → diabetes, Pancreatic cancer

or Ovarian cancer

1940 Landsteiner and Wiener

It was first observed when blood of a rhesus monkey was injected in a rabbit and serum was obtained. Hence it is named Rhesus factor.

Those who possess the factor are Rh⁺ rest Rh⁻

Also, Rh system is independent of ABO and MNS systems.

Initially it was assumed that Rh factor was inherited as a simple Mendelian pair.

Rh⁺, Rh⁻ (Rht), rh⁺, rh⁻ (Rhθ)

1940 | 1947

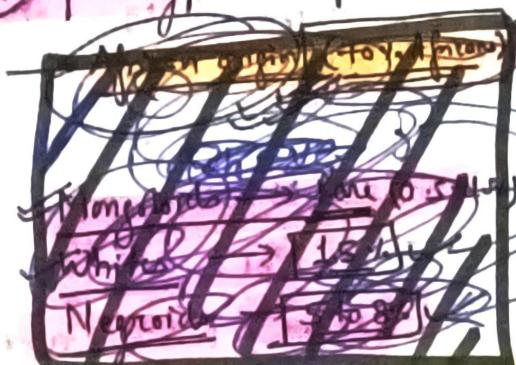
Rh^+ or Rh^- is actually
 $Rh(D)$ presence or absence

1947, Fisher claimed that Rh types are determined by a series of three pairs of alleles, $C/c, D/d, E/e$. These genes are inherited in groups of three located on a single chromosome.

Thus 27 genotypes and eight phenotypes are possible.

Genotype	Fisher
RR	cDe
Rr'	Cde
$r'r''$	cdE
RR_1	CDE
RR_2	cDE
Rr_1	CdE
Rr_2	CDE
$r'r_2$	cde

(21) 8



Native American/Aust blood group: 99%
 East Asia - 98% European - 80%

Rh^+

Erythroblastosis / Rh-incompatibility

Rh \ominus mother

Rh \oplus Baby

(has Rh antigen)

Rh antibodies are spontaneously generated in response to Rh antigen in Rh \ominus person.

Monoploid 99%

Baloch Pyrenees 60%

No natural antibodies

Response
 Rh antibodies are formed

could not cross placenta

Post birth / abortion

the blood laden with Rh antibody covers the mother's uterus.

In case of 2nd birth this blood antibody may cause blushing of fetus.

Within 72 hr of birth mother is given Anti Rh bovine injection.

Diego system

most are Diego negative

only Native Americans (40%) and East Asians (+ve) → shows East Asian origin of native

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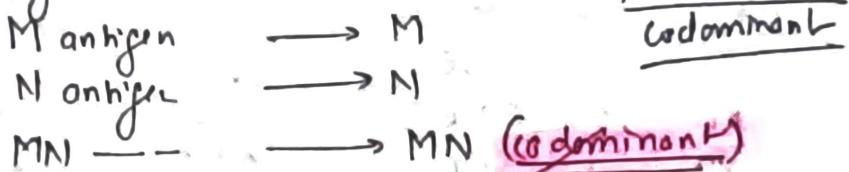
absence

#

MNS system

No natural anti bodies
No effect on transfusion of blood
All have M or N or both

1927, Landsteiner and Levine found \Leftrightarrow human antigen
M and N with no natural antibodies and hence they
have no effect on transfusion of blood.
Either of M or N or both is present in all human
beings.



1947, Sanger and Race found two more antigens S and
and they occur along M and N antigens.
Thus, 4 genotypes and 3 phenotypes have been
identified. $(10) \rightarrow (3)$

M, N, S, s

Genotypes	Phenotypes
MSMS	MS
MSMs	
M _s M _s	

MSNS
MSN_s
M_sNS
M_sN_s

NSNS
NSN_s
N_sNS

(MSN)

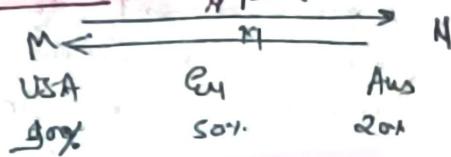
(NS)

1927 / 1947

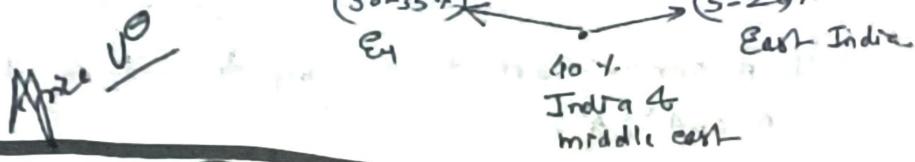
native American exclusive

M → ~~American Indians exclusive to North America~~

N → Most common in Australia and Pacific



(S) → Higher in India and middle east



#

Gm groups Steinberg

Gm factor is serologically detectable variations of certain antibody proteins (immunoglobulins) of the serum

Gm specificities (Gm no.) are due to variations in H chain of IgG only. S Antigens → Ig GAMED (IgG4) ↴ 2 Y chains

Similar to Rh factor, Gm groups are also transmitted in groups.

✓ Gm (3, 5, 13, 14) is common in Europeans but infrequent elsewhere

✓ Gm (1, 13, 17) found in eastern Asian people

✓ Gm (1, 15) found all over the world

✓ Steinberg did extensive anthropological work on Gm factors.

Inherited in families

it is a complex of genes or chromosomes which encode for cell surface proteins that regulate immune system

HLA - Histo compatibility system

When skin and other tissues are grafted from an individual to another, immunological rejection leads to destruction of the graft.

This graft rejection is less severe if both are related.

This occurs due to inherited difference in HLA antigens

HLA antigens are decided by two closely linked loci LA and B.

At least 14 LA antigens and 17 B Antigens have been found in Europeans.

Similar to Rh factor and Gm factor, HLA are also inherited in groups.

The frequencies of some HLA alleles show considerable geographical variation.

Frequencies in Europe is uniform but those in Australia and New Guinea show considerable variation.

Hemoglobins

Variation → age
→ easily mutated
→ environmental formation

It is a variable, it varies with age and easily mutated.

Variations in type of Hb can be correlated to the environmental conditions and also linked to epidemiological problems.

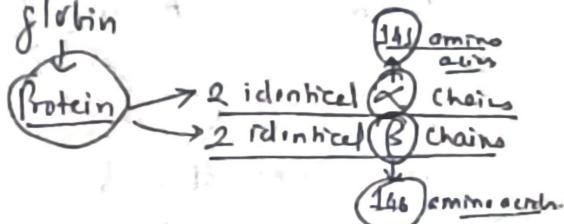
Four types: Embryonic, fetal, adult, A₂

disappears
in first month
of embryonic life
by erythropoiesis
(1+ remains)

Major
Minor
(A₂)

Structure: Heme + globin

Organic molecule with Fe



Mutations: Changes in protein chains leads to

HbS, HbE, HbC and Thalassemia

HbS: Sickle cell → ↓ in O₂ capacity

Heredity

B chain
must also be balanced

← Hb A/HbS
Polymorphism HbS/HbS

in Malaria environment

Normal Malaria

1901



Not found in New world, Europe and Most of Asia.

HbE: 1954, Indonesian archipelago

Thailand, Cambodia, Viet. Laos, Malya, Burma, Assam, Meghalaya S.E. Asia

It has been correlated to sprout of agriculture

Homozygotic HbE → reduced fertility

Heterozygotic HbE → protection against malaria.

20000
deaths
y~

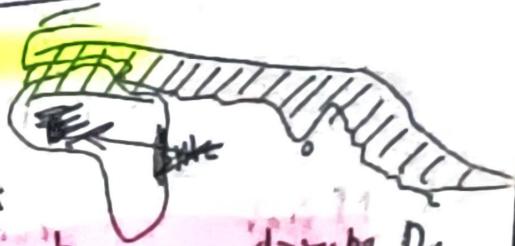
Two variants G

HbC: It is an anomaly in β chains.
Usually found in West Africa.

Thalasssemia: Two Greek words (thalassos = sea and hamia = blood)
Distribution: Around Mediterranean and Tropical India, Indonesian Archipelago.

30000
deaths/
year

Although it is associated with haemoglobin but it is not an abnormal Hb. It is a hereditary disorder. It has presence disrupts the synthesis of HbA and promotes abnormalities in haemoglobin.



Thalasssemia
↓
P thalasssemia
(2 genes)
↓
major (homozygous)
minor (heterozygous)

α Hb genes
(4 genes)

disrupts
synthesis of HbA

and promotes abnormalities
in HbA and HbB

Auto-somal
recessive

It causes severe anaemia (red blood cells are destroyed)

G6PD

Glucose - 6 - phosphate dehydrogenase

is a red blood cell enzyme which metabolizes

glutathione in RBCs.
Inherited X-linked

The deficiency is called G6PD deficiency

Distribution coincides with falciparum malaria.

30000
deaths/
yr

Two variants

G6PD-A — 10% in African, Indian, Chinese

G6PD-Mediterranean — Middle east, Mediterranean

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both present against me

X linked recessive

Genetic markers in Plasma:

Both Plasma and Serum contain several soluble proteins. These proteins can be separated by electrophoresis.

Haptoglobin: (Hp)

two genes \rightarrow Hp^A and Hp^B

Hp^{AA} , Hp^{BB} , Hp^{AB} or Hp^0
Name Nitro → co-dominant

They follow mendelian principle of inheritance

~~Alleles~~ In many populations they are absent (Hp^0) - African
 $Hp^A \rightarrow$ ~~Alleles~~ Central and South America

Transferrin (Tf): β -globins, They have chemical

property of binding iron. 20 molecular varieties of Tf have been identified and each is associated with an autosome allele located on some locus.

TfC is most common and found in all population of a world.

Serum =
Plasma -
clotting
factors

Autosom
Autosomal
Sex-link
Sex-Link

Parent

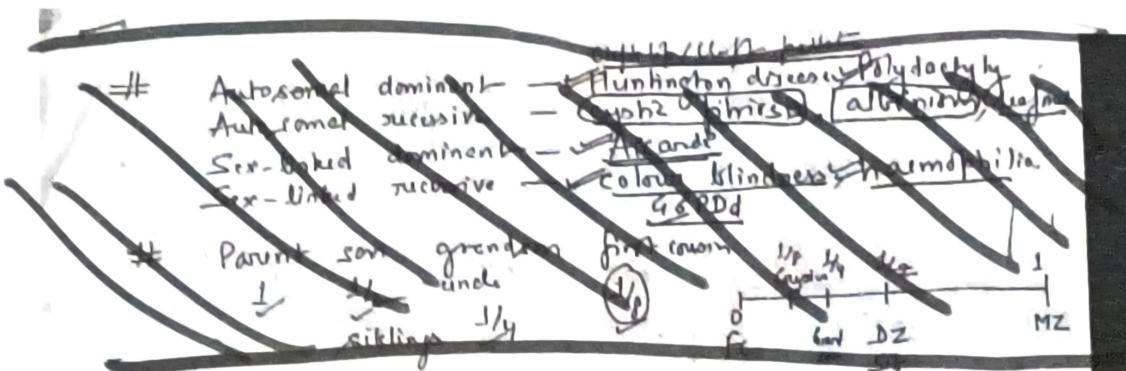
IMMUN

- These are antigen-antibodies
- These are

- Each is
- Light
- Heavy protein

- Either
- not b-
- Antibod

All 5
their
and th
allotypes



IMMUNOLOGICAL STUDIES

- These methods to study man-family are based on antigen-antibody reaction. (IgA, IgG, IgD, IgE, IgM)
- There are five types of antibodies — IgA, IgG, IgD, IgE, IgM
- Each is made up of heavy protein chains and light protein chains.
- Light → Kappa (κ) ✓, Lambda (λ) ✓
 Heavy → Gamma (γ) ✓ IgG
 proteins alpha (α) ✓ IgA (GAMMA)
 Delta (δ) ✓ IgD
 Epsilon (ϵ) ✓ IgE
 Mu (μ) ✓ IgM
- Either of κ and λ are present. They both are not present simultaneously.
- Antibody chain → constant region ✓
 → variable region ✓
- All 5 antibodies are present in all humans but their constant part varies due to genetic differences and this variation is called immunoglobulin allotypes. ← allotype

Polygenic traits: Those traits that show variation between two extremes and are influenced by multiple genes.

Lethal genes → those that kill the possessor

time
domestic
sub-lethal
semi LD
Dominant
Recessive
① Rec. lethal
② Dom. lethal
③ Sub lethal
④ Semi lethal
⑤ Epilepsy
⑥ Huntington disease
HLS

Dominant lethal → heterozygous and homozygous

Sub lethal → epilepsy

Semi lethal → Huntington disease

Recessive lethal → homozygous

Sub lethal → Thalassemia

Semi lethal → Haemophilia

T
H

Foster child:

To find out effect of environment.

Random selection
(genetic component)
out

→ sent to {good
bad} homes

Time → IQ tested

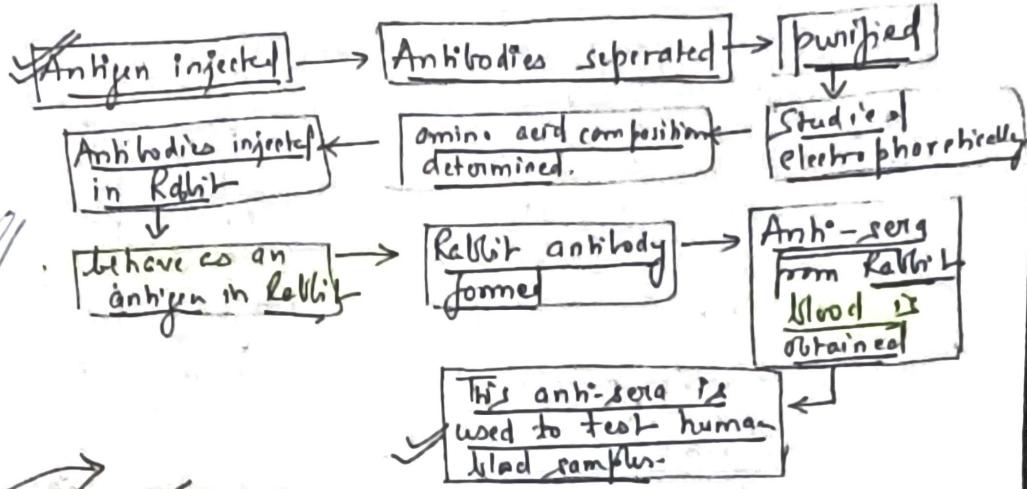
In practice, it is never free from biases

I. Chicago studies: Adopted children

Grothman → 45 → 112
Avg → 39 → 105
Poor → 27 → 96

shows effect of environment on IQ

II. Minnesota ground home study:
Effect of both environment & genetics



e.g. Inverse factors. There are 3 inv. factors - 1, 2, 3
 Population survey with anti-serum factors.
 ① and ② are common in Venezuelan Indians and rare in Europeans

Variations
 ④ Gm system - IgG
 ⑤ Am-system - IgA
 ⑥ Inv-system - R
 ⑦ Z-system

These variations are due to multiple allelism

BIOCHEMICAL STUDIES

Methods employed to separate proteins, DNA and RNA are called biochemical methods

① Separation and identification of proteins:

→ Gel-filtration: Separation on the basis of molecular weight

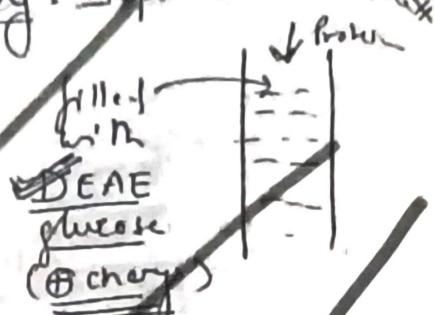
Polyacrylamide (separates)

Protein
 ↓
 Beads of polyacrylamide (tiny holes)
 (Large protein molecules)

~~Q. Non-exchange Chromatography : Separation on basis of charge.~~

Proteins with $\text{+} \Theta$ charge are strongly bound.

Mix is washed with buffer solution of diff. pH.

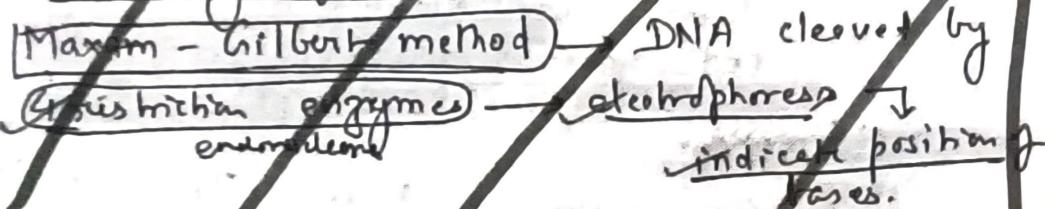


~~3. Electrophoresis : Charge and molecular weight~~

~~Electric field of acrylamide gel. (Agarose gel)~~

~~Weight = Charge \propto speed of migration~~

~~(B) Sequencing of DNA :~~



~~(C) Southern, Northern and Western Blotting~~

~~DNA~~

~~RNA~~

~~Restriction~~

~~Electrophoresis~~

~~Blotted on nitrocellulose filter paper~~

~~Hybridized with labelled probe~~

~~Western Blotting~~

~~Protein~~

~~Electrophoresis~~

~~Blotted on nitrocellulose filter paper~~

~~Treated with antibody~~

~~Antigen-Antibody complex~~

~~Reacted and labelled~~

PHYSIOLOGICAL VARIATIONS

Haemoglobin (Hb)

Present in RBC



Organic molecule with Fe

↓ Protein \rightarrow 2 α chains
2 β chains

Basic function is to bind O_2 and transport it to cells

Variations in Hb level are due to —

- ① Age
- ② Sex
- ③ Ethnographical diversity
- ④ Nutritional status.

It is measured in gm/dL

① Variations based on Sex :-

female have lower Hb level compared to males because of monthly blood loss to menstruation and lower RBC count.

② Ethnographic variation :-

- Verma (1976) (Andamanese) found both male and female anaemic.
- Chatterjee (1952) found 50% female, 57% child male, 62.5% child female anaemic.

③ Variations based on age :- In 10-14 years, both M and F have similar Hb level (13.9 & 13.8) but post puberty Male \rightarrow 15 female \rightarrow 13.3

④ Environment

According to most studies, various ethnic groups show gradual rise in the Hb-level with Age upto 30 yrs.

Pulse Rate :

70 - 72 - men

infantile - 110 - 140

78 - 82 - women

elderly 50 - 70

Sex

age

Sensory Variation

→ PTC - Phenylthiocarbamide
(TT, Tt, tt) → non taster.
Red and Green colour blindness
(X-linked recessive)

Variations in R&G Colour blindness → genes

Variation in PTC → genes based on TT, Tt, tt
and based on Sex: Females are found to be more sensitive to taste
and the differences are statistically significant. Muslims of India do not show sex-based differences in the PTC tasting.

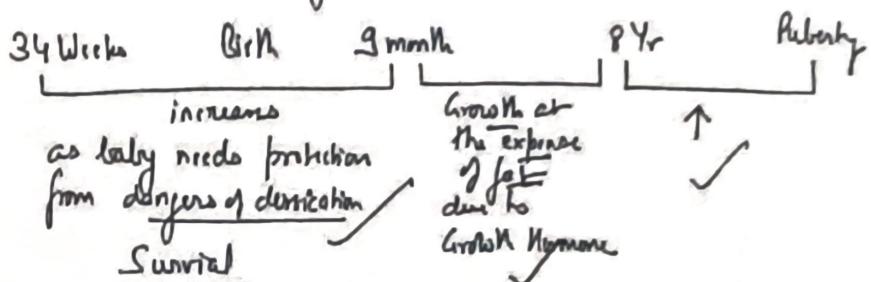
statistical correlation
b/w PTC tasting
and thyroid issue chance

fat level

Subcutaneous fat is unevenly distributed over the body.
Fat level varies based on -

- ① Age
- ② Gender
- ③ Genes
- ④ Environment

(a) Variations based on age:



(b) Variations based on sex:

Post puberty, male fat deposition reduced while it continued in trunk area of females.

Survival: To save heat lost due to menstruation and child birth.

(c) Variations due to Genetics and Environment:

No. and distribution is decided by Genetics

Filling of these cells decided by Environment and nutritional factors

By Gangatic plains → fertile land → no need to store fat ↓ less No. of fat cells.

Deserts of Rajasthan → extreme → more no. of fat cells

(d) Body fat is closely related to cardio vascular problems.

Respiratory rate:

11

American Society of Human Genetics (1975) - genetic counselling is a communication process which deals with the human problems associated with the risk of occurrence of a genetic disorder in a family.

GENETIC COUNSELLING

Explain counselling by a geneticist regarding genetic problems is called genetic counselling.

Two types of people visit a geneticist - already married or about to get married. Both have history of genetic disease in the family. The one with genetic disease is called probands. But their infected, they become inbreeds. They do refrain from consulting a geneticist.

There are five stages -

1. Pedigree construction
2. Examination
3. Diagnosis
4. Counselling
5. follow up

Step 1: Pedigree analysis:

Geneticist has a list of more than 5000 genetic diseases along with their mode of inheritance, symptoms, socio-economic - psychological effects.

But he/she is pedigree analyst to ascertain inheritance pattern.

Eg. Heredity could be autosomal dominant, recessive or via mutation also.

Make a pedigree diagram)

Step 2: Confirmation is done via medical techniques like skin punch test etc.

Step 3: Diagnosis: Pedigree and medical investigation may lead to a confident diagnosis or further investigation/interview may be required.

After a detailed and confirmed diagnosis only further steps are taken.

Step 4: Counselling interview - It should be non-judgmental and non-directive. It involves informing the family about socio-economic - psychological disabilities.

(1) Counsultation without mode of inheritance probabilities of occurrence of disease.

(2) Treatment and therapy at pre-natal stage.

(3) Cleft lip - cleft palate, alternative → genetic testing for birth defects.

(4) Edentophilia → only childhood

(5) Duchenne muscular dystrophy → abnormalities.

- Huntington disease → 60-70 years of age

Autosomal dominant - retinitis pigmentosa, H. Don't do

Autosomal recessive - albinism, deaf mutes, etc.

X-linked recessive - Hemophilia, color blindness, etc.

• X-linked dominant - Huntington's disease

* Cleft lip → plastic surgery

* ADH deficiency → gene mapping (all human genes, etc.)

* Sickle cell anaemia → gene mapping

If autosomal recessive → advise them to marry those relatives.

Step 5: Follow up

Frequently the scope of genetic counselling is limited but its significance will be amplified many folds once gene therapy is fully developed.

- Limitations of Genetic counselling:

 - 1) There can be cases of no diagnosis (conclusive)
 - 2) Possibility of incomplete or incorrect diagnosis due to complex system of genes
 - 3) Limited or inadequate information.
 - 4) Strong heredity being supported
 - 5) Limited knowledge of genetic behaviour.

PEDIGREE ANALYSIS

PEDIGREE ANALYSIS [familial method]

Introduction: General information collected from the family and summarized via pedigree (mapping family tree) formulation and its analysis helps to find inheritance pattern of a genetic disorder.

This genetic information is stored in genes which are present on chromosomes. Humans have 22 pairs of autosomal (homologous) chromosomes and 1 pair of (X & Y) sex chromosomes.

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These genes can be surviving (expressing only if homozygous) or dominant (always expressing). Any genetic trait can be traced via a pedigree pedigree beginning with an infected person called

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1. AUTOSOMAL DOMINANT

Aq	Aq	Aq
q	Aq	q
		q

A	AA	AA
a	aA	aq



~~Each affected individual has an affected parent~~
~~Unaffected relatives will not have an affected~~

In Grm II it is not necessary to elaborate all. Those only are formal in II for making Grm III.

based on photopiles

2. AUTOSOMAL RECESSIVE

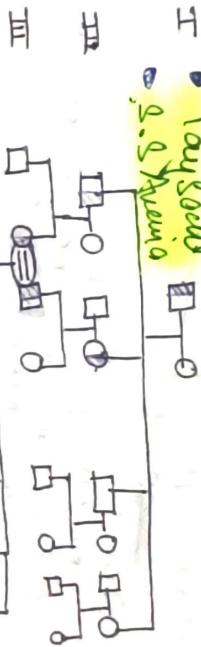
Albinism, deaf mutism

- albinism
- Thalassemia

A	A	a
A	AA	aa
a	Aa	aa

Q.S.

- Tay Sachs
- S.S. Phenyl



III

1. Expressed only if homozygous.
2. Almost never is the gene present in both parents.

3. Sibling affected. Unaffected have 1 in 4 chance
of being affected.

4. Parental consanguinity.

5. Linked inheritance: Colantidium hamophilus

Experiments on F6-PD_d

X ^h	X
X ^h X	XX
Y	X ^h Y
X ^h Y	XY

X ^h	X ^h
X ^h X	X ^h X
Y	X ^h Y
X ^h Y	XY

X^hY → affected male → ♂
X^hX → carrier female → ♀
X^hX^h → affected female → ♀

X ^h	X
X ^h X	X ^h X
Y	X ^h Y
X ^h Y	XY

X ^h	X
X ^h X	X ^h X
Y	X ^h Y
X ^h Y	XY

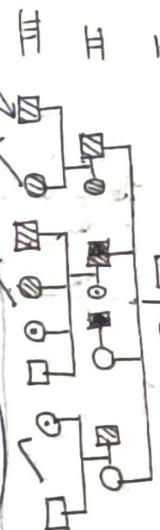
X ^h	X
X ^h X	X ^h X
Y	X ^h Y
X ^h Y	XY

- Haemophilia

- R.G. colour blindness
- G6PD defn

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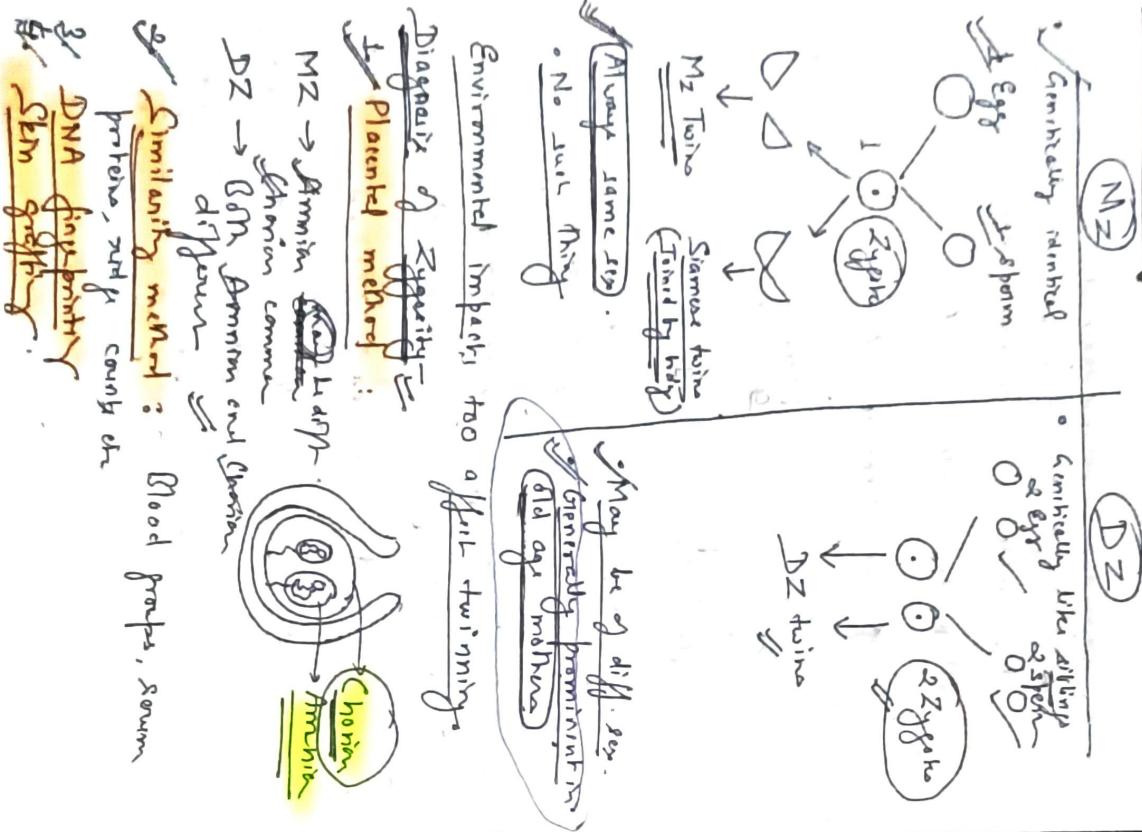
X linked recessive = Affected Sons + H. in mothers.



II



TWIN METHOD



Study of heredity in twins

Aimed to find heredity of ~~combined traits~~ (ht, intelligence)

Aimed to find heredity of ~~different characters~~

- High concordance in MZ and low in DZ \Rightarrow Environmentally determined
- Both MZ and DZ twins are studied and their extent of concordance known.

MZ (concordant)	DZ (concordant)
89.6%	28.1%
Setting up	Setting up
White colour	White colour
22.1%	22.1%

Reining studies: This is more of a statistical study by finding out mean and variance for various traits

$V_{DZ\text{RA}} \rightarrow \text{Total variance (envt.)}$

$V_{MZ\text{RA}} \rightarrow \text{environmental variance (similar and } |V_{DZ\text{RA}} - V_{MZ\text{RA}}| = \text{genetic variance}$

$$\text{Heredity} = \frac{V_g}{V_e + V_g} = \frac{V_{DZ\text{RA}} - V_{MZ\text{RA}}}{V_{DZ\text{RA}}}$$

\therefore ratio \rightarrow closer to 1 \rightarrow heredity \downarrow closer to 0 \rightarrow half environmental

care but

Leonard Heston - 1966

Schizophrenia - genes determine

not env.

Are twin studies authority?

Seuness

Validity of twin studies have been questioned on following aspects -

1. It neither specifies the genetic component nor the environmental component.

2. Assumption that MZ twins are identical can be questionable. Identical supply may vary.

3. Heritability estimates may not be applied to all aspects as some of them may have been significantly influenced by intrauterine development.

4. Environment of the two twins may not be representative of the environment of the population.

5. Such studies don't consider intra-population variations.

6. Heritability estimates of complex phenotypes like IQ and behavior is difficult because of complex interaction of heredity and environment.

Concordance rate

$$C' = \frac{C + 2C''}{C + 2C'' + d}$$

C \Rightarrow No. of concordant pairs

C'' \Rightarrow No. of concordant pairs in which both members are discordant independently.

d \Rightarrow No. of discordant pairs.

Intro: Kline (1921) provided threefold way of twins in genetic research.
 1. Inheritance of normal variability,
 2. Mental determination of offspring,
 3. Degree of genetic determinism of environment is determined.

Co-Twin Control Method:
 A new method of study of twins has evolved recently where MZ twins and DZ twins are used in controlled environments.

A scholar studied effect on IQ by psychological methods.

Twins adopted from mothers here with low IQ given training and tested from training they show improvement in IQ due to their co-hab.

Twin methods

Define Twins MZ

1. I.Q. < 12

2. DZ > 22

3. DZ < 22

4. DZ > 12

5. DZ < 12

6. DZ < 12

7. DZ < 12

8. DZ < 12

9. DZ < 12

10. DZ < 12

11. DZ < 12

12. DZ < 12

13. DZ < 12

14. DZ < 12

15. DZ < 12

16. DZ < 12

17. DZ < 12

18. DZ < 12

Twin methods

Define Twins MZ

1. I.Q. < 12

2. DZ > 22

3. DZ < 22

4. DZ > 12

5. DZ < 12

6. DZ < 12

7. DZ < 12

8. DZ < 12

9. DZ < 12

10. DZ < 12

11. DZ < 12

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17. DZ < 12

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Twin methods

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It is an epigenetic mechanism of which gene cause gene to be expressed differently based on which parent it come

discovered by

David Solter, Azim Surani in mid 1980s

GENETIC IMPRINTING

Genetic imprinting is defined as differential inheritance of genes maternal from mother and paternal

Mendel gave the theory of equivalence which says there is no difference in expression of same gene inherited from male or female.

But in humans, some characters are inherited markedly different from male and female parents.

Eg: Huntington's Chorea + autosomal dominant
Source can be both male or female (paternal or maternal). Symtoms same, but severity and time of expression different

② Genetic imprinting suggests that in certain cases a gene defect will only produce a phenotype if inherited from a particular parent

Eg: deletion of chromosome pair

inherited with symptoms in male same
Paternally derived claim naturally derived with no symptoms

chromosome pair

developmental delay

- stereotyped behavior

- mental retardation

- reduced tonus

- mother's body is largely unaffected

- improved growth

- go neither growth nor retardation

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Characteristics of genomic imprinting:

1. Imprinted genomes show variability - type and magnitude
2. Genomic imprints are erasable - can抹去 highlight methylated genes from mother and erase them back to its original form in his genes before passing them on to its offspring
3. Genome imprint are species specific
4. Genomic imprinting is imitated in many diseases
5. **Genome imprinting is not a rule**

Nature and mechanism of genome imprinting

It is done via differential methylation of DNA in nucleus of somatic cells. Enzyme methylase adds CH_3 group to cytosine position of cytosine and thus guanine is also methylated



Applications -

1) It has helped to understand various genetic disease and their relative inheritance from a particular parentage.

2) Methylation is further used in genetic engineering through recombinant DNA technology

such as - insulin production, growth hormone, somatotropin, etc.

3) Gene cloning

4) Gene therapy

Conclusion IVP tech → uniparentalism - as normal imprinting is affected

auto or diff

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also called Malthusian fitness

GENETIC LOAD

$L = \frac{D}{I}$ - formula
 $D = \text{Fitness} - \text{Average fitness}$
 $I = \text{Population size}$

Genetic load is defined to be the relative decrease in the average fitness of a population with respect to the fitness of those individuals of the population who exhibit maximum fitness.

$$L = I - W$$

$L \rightarrow$ Genetic load
 $I \rightarrow$ Maximum fitness } of the same population.
 $W \rightarrow$ Average fitness

Generally, it has been observed that more \rightarrow the number of mutations are primitive and harmful. The more harmful the number of mutants, less will be generalized. i.e. if increased by mutations and natural selection can play role less in its formation, it only \rightarrow an \downarrow their number.

Effector of Genetic Load

Earlier view: Haldane said \uparrow in GL will be harmful for population \Rightarrow eliminate mutant viruses.

Optimum conductor \rightarrow is decided by environment due to rapid environmental changes, optimum genome changes and in order to better adjust and adapt to new environment, the greater the genetic load. The latter chance of survival.

Modern view:
② Population fitness is decided by carrying capacity and over crowding

Relevance:

too many seedlings in a small area \rightarrow all will die.
If some are stronger, others weak \rightarrow stronger will survive.
N.S. \rightarrow thinning out

Genetic load causes to the thinning out of the population and thus ensures survival.

GENE MAPPING

Gene mapping is the procedure employed to study where gene is located on what place on which chromosome.

Linkage map

Cytoplasmic inheritance

Physical map

To find relative distance between genes on a chromosome.

Distance between genes in centimorgans

Linkage

distance between genes on a chromosome.

GENETIC EFFECT OF CONSANGUINEOUS AND COWIN MARRIES

BY N.

Consanguineous marriages are marriages between closely related individuals having common ancestors a few generations back.

However, consanguinity may vary. Marriages between father and daughter, mother and son or brother and sister (i.e. primary kind) is highly consanguineous. They are mostly prohibited by social norms.

- First cousin marriage is relatively less consanguineous and is allowed in many Middle Eastern and Indian countries.
- Consanguineous marriage and inheritance of defective gene

Genetic defects caused by dominant genes. (e.g. dominant genes - recessive genes)

Consanguinity has no effect on inheritance of dominant and codominant genes as, if they are present in heterozygous or homozygous condition they will always express and have a normal phenotype visible.



Both Aq → 45%

However, consanguinity has a well marked effect on successive inheritance. → autosome → autosomal recessive trait → few linked → haem, leukaemia etc. According to Mendel, successive inheritance in homozygous condition. Consanguinity enhances probability of defective homozygosity.

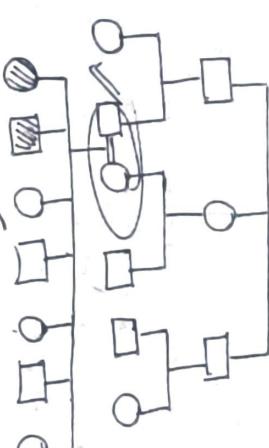
④ INHERITANCE OF ALBINISM: Lack of Melanin

(AUTOSOMAL RECESSIVE TRAIT)

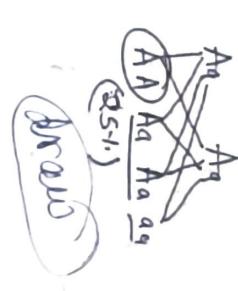
③ Inheritance of Six-fingered Dwarfishism: Amputee people marry amongst themselves. Six-fingered dwarfishism is rare among other people but common among Amish people due to consanguinity. But consequences of close consanguineous marriage are usually marked for inheritance of a sex-linked trait. e.g. colorblindness.



II



III



AA Aa Aa aa
(45%)

Draw

Pedigree of autosomal recessive trait

gentle coming → gives
surprise

Should first cousin marriages be banned?

Cannabis, hemp → many uses. Many uses are allowed and at home. Cannabis is mainly used in Middle Eastern, India and Pakistan societies.

white considerin' ginnin' time that we
we most consider length of time that we
tradition have existed. Moral
leading to conorwin

new
moderation
popn will over
a care of
consequential
popn

Mayer forte → \leftarrow stability

marriages

Studies

- Muslim + Hindu and Sikhs (1984)
- (Bhagatpur, Bihar)
 - Mortality in Hindus → 36.2
 - outbred → 28.7
- Ibrahim + Tomil Nady - Rao and Tamhane (1997, 1998)
 - (2000 years of inbreeding)
- The study did not find any significant rise in mortality or fall in fertility

GENETIC SCREENING

Genetic screening refers to the application of techniques for the purpose of detecting carriers of deleterious genes or chromosomal abnormalities.

Objectives:
- Identification of individuals with genetic disease →
- To identify potential high risk individual of having
- offspring with defects.

Also, following of **Experiments**...

Some diseases are diagnosed at birth, others during childhood. Some in adolescence and even in old age. Diabetes mellitus is detected by a simple sugar test in the new born babies.

Treatment is unavailable, skin retention is advised.
Fay Sachs disease - diagnosed at foetal stage

Limitations:
possible risk of procedure

2. Facilities for treating unavailable or costly
3. May lead to inhumanity of Down syndrome people
U.S. is limited in its scope and applicability and
abortion are not morally accepted.
Also, many diseases could not be diagnosed in utero.

卷之三

Organization of Rare Diseases in France

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so many new to be treated
of by a physician.

confidentiality
informed consent
a void proxy election
will be honored before

Informed consent
→ avoid psychologig
problems to be solved before

Scanned

1

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124

Karyotype is produced by nuclei picture of chromosomes are taken in order to determine chromosome complements, including no and any abnormalities.

abnormalities

CHROMOSOME METHOD AND KARYOTYPING

Steps involved are:

Stagnating cells in metaphase:

① Phyto-Haemagglutinin (PHA) used to separate WBC and RBC and induces mitosis

Fixation — prevents formation of spindle fiber and bursts cells in metaphase. It also promotes contraction and Widening of chromosomes

Accumulation of sufficient cells

Preparation of cellular suspension and fixation:

Hypotonic solution causes cytoplasmic swelling and prevents disjunction of metaphase chromosomes (Na or K Citrate, KCl)

Tetrahydrofuran (THF) and glacial acetic acid mixture (3:1) is used for fixation (hardening of chromosomes)

Slide preparation: Rubbing and drying cells in a suspension.

Q-Banding:

Quinacrine mustard, fluorescent binds selectively with guanine residues in DNA following polymorphic regions are distinguished by it—

XX YY XX YY XX XX
13-15 (D)

XX XX XX XX XX
16-18 (E)

6-12 and X (C)

XX XX XX
(E) A

XX XX
45(B)

XX XX
19-20 (F)

XX XX
21-22 and Y (G)

Quinacrine mustard, fluorescent

binds, selectively with guanine residues in DNA following polymorphic regions are distinguished by it—

3-4 → metacentric region

13-15, 21, 22 → short arms

Long arm of Y chromosome

Limitations

Need of a fluorescent microscope

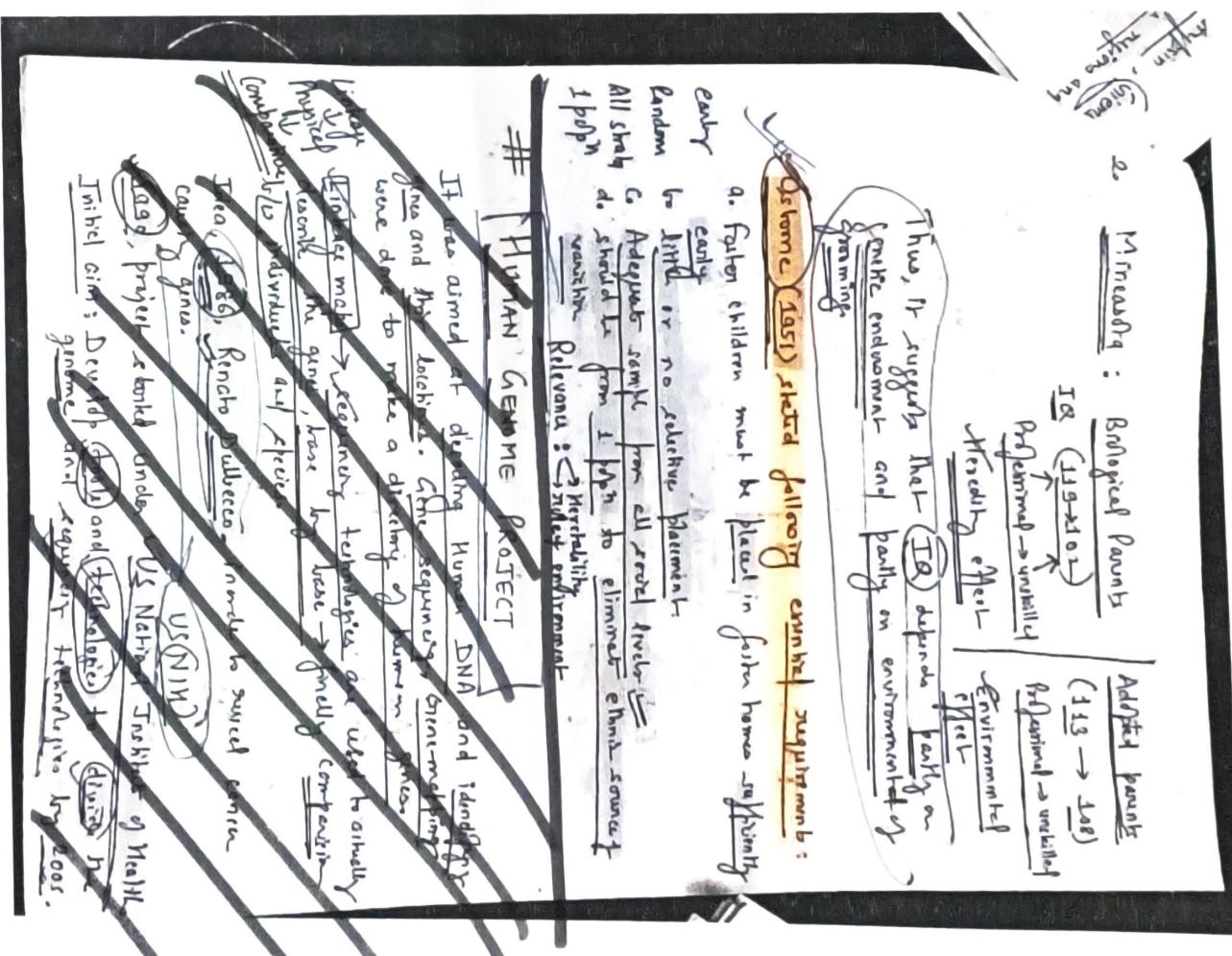
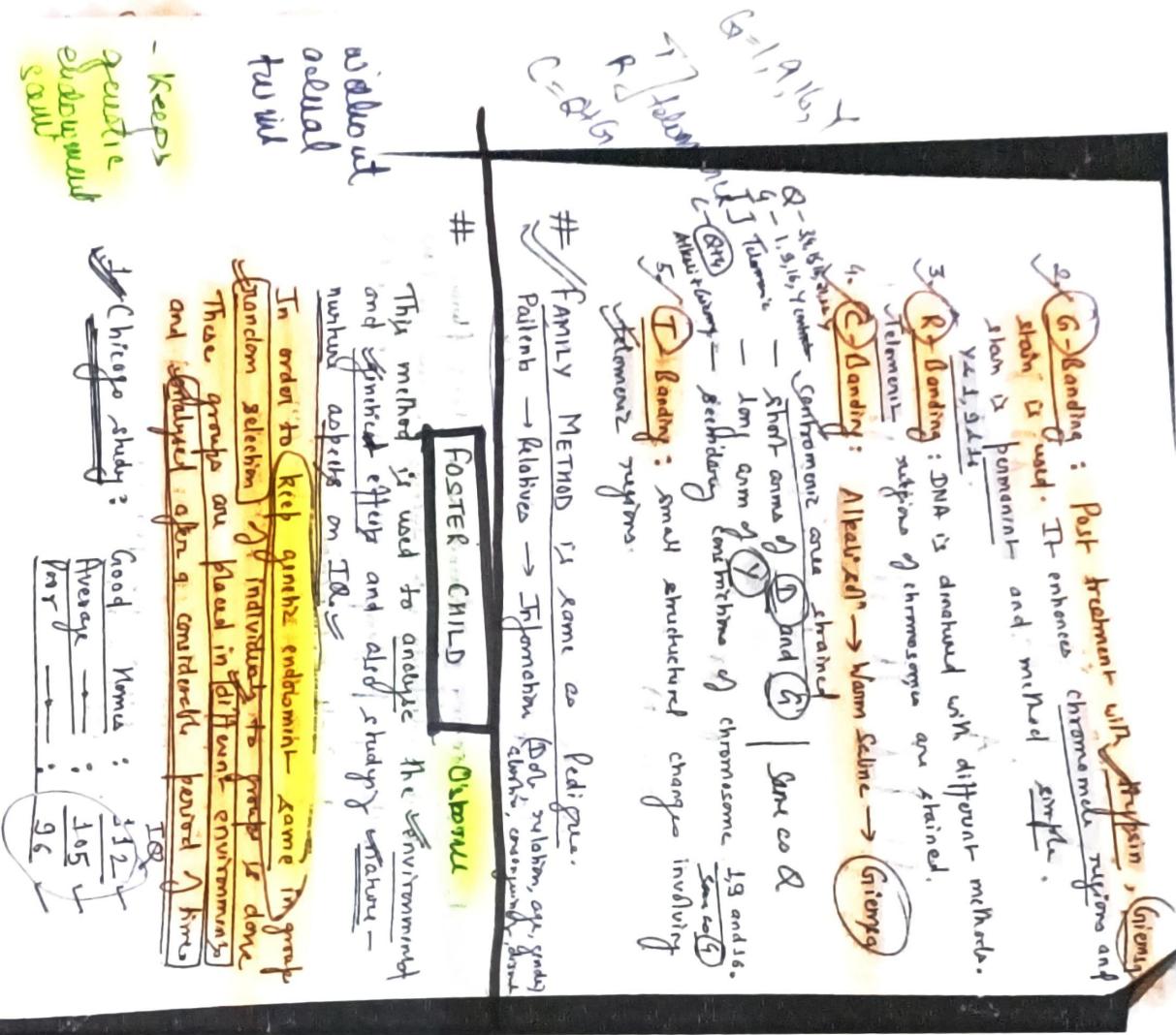
Stain is not permanent

Rapid quenching of fluorescence

$$A = \boxed{3, 9, 13, 14, 15, 21, 22} \quad \boxed{Y}$$

centromere short arm long arm

abnormalities in some monosomy that can be detected detection - dup deletion - genes rearranged insertion - translocation



(Some basic concepts of Genetics and cell division)

Chromosome (coloured + bodies)

Least no. of genes are in Y chromosome (49 genes)

Genome

Sum total of all genes variability of a species is called genome.

Genome

Sum total of all gene variability of a population is called genome.

Genome & Chromosome

Each species have a particular no. of chromosomes and is represented by ~~gen~~

Human 23 pairs

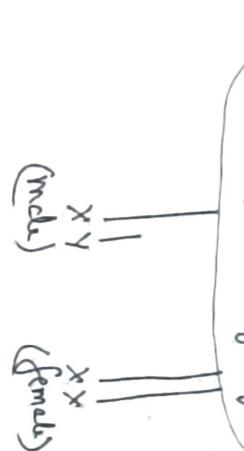
Sex pair

Sex-chromosome

In autosomal chromosomes, in each pair the two members are alike (same no. and same sequence of gene is present). In a pair of members look alike but call them homologous pairs. Thus all 22 autosomal pairs are homologous.

In sex-chromosome we have X and Y. Y is short Y₃rd of the length of X.

Two in female all 23 pairs are homologous.



Cell division → mitosis → meiosis

① Mitosis

No. of chromosomes double and then they separate



daughter cells have same no. of chromosomes as mother cells.

Pro → regulate
Mito → division spindle fibers
Mit → Separates (nucleus)
Tel → Cell separates.

Chromosome

Function: To maintain the no. of chromosomes in every cell of an individual

② Meiosis → Meiosis I → Meiosis II

All pairs arrange themselves at the centre of the cell.

function: To maintain the no. of chromosomes constant in every generation

n from male / n from female

Length wise arrangement of chromosomes



members of each pair move to opposite ends
This is also called Zygote formation.

Miosis is followed by series of mitosis to give 3×10^9 mm

E.g. 21+X, 50+22+Y

21-22-X are the smallest
and they lead to trisomy or malfunctions due to deficiency

A particular character, such as colour, length etc are controlled by a particular gene

Different forms or variations in genes are called Allotenia.
e.g. Blue \rightarrow dark hair \rightarrow fair

Blood \rightarrow 3 Allotenia A, B, O

RBC, T, T₁, T₂, T₃

	A	B	O
A	AA	AB	AO
B	BA	BB	BO
O	BO	BO	OO

3 Allotenia

Genotypes \Rightarrow Phenotypes

When two alleles are present in 1 chromosome one dominates other and other becomes recessive.

Genotype

AA	A	AB	AB	BB	B
AO		AO	AB	BO	BO
BA		BA	AB	BB	B
BB		BO	BO	BB	B

a genetic disease resulting from a chromosome abnormality

Small head — (2) 22 and Y = most vulnerability

34

Chromosomal abnormalities:

2

Sex-chromosomal aberrations

At times during [Anaphase], the 2 members of a chromosome fail to separate and lead to

Recombinant non disjunction

Now,

$$\begin{array}{l} \text{22+XX} + 22+X \rightarrow 22P + \underline{\text{XX}}X \quad [\text{Super female}] \\ \text{22+XX} + 22+Y \rightarrow 22P + \underline{\text{XY}}Y \quad [\text{Klinefelter}] \\ 22+XY + 22+X \rightarrow 22P + XYX \end{array}$$

$$\begin{array}{l} 22+XY + 22+Y \rightarrow 22P + XYY \quad [\text{Toots}] \\ 22+0 + 22+X \rightarrow 22P + \underline{\text{XO}} \quad [\text{Turner}] \end{array}$$

In all these questions explain disjunction and meiosis

and make diagram of disjunction.

Widened pelvis
tall upper limb
poor coordination

(47)

22 + XXX

male hypogonadism hypogonadism

(47)

22 + XY

male Klinefelter

(47)

poor coordination

(47)

22 + XXY

male Klinefelter

(47)

poor coordination

(47)

22 + YY

male Turner

(47)

poor coordination

(47)

poor coordination

(47)

poor coordination

poor coordination

(47)

</div

Very important word and you

unbalanced translocation can be induced

Kidney malfunctions
Heart defects
Infection but

Intellectual disability

Developmental delay
Walking ability
Small head
Widely spaced eyes
Underdeveloped testes.

Normal heart
malformations.
Most die before birth.
Still born.

Life span 5-15 days
87 survive more than 14 yr
due to age too

Treatment → No cure

Cri-du-chat syndrome

piece of chromosome 5P deleted / 5P-5P syndrome

Jerome Lejeune
Spurred by
mentioning for described by Jerome

cause translocation / delusion

on SP
CTNUOZ gene
loss - linked

Symptoms: Normal after the characteristic cry of
none. because of ~~afford~~ ~~longer & never~~ cry.
Cat-like cry.

to loose
mental
ability

② feeding problem due to difficulty in sucking &
swallowing

③ low birth weight and poor growth

④ some deformities, speech and motor disabilities

⑤ behavioral problems —

Hypothalamic
Hypothalamus

extra oral drooling
small head and ~~small~~ pink eyes

Thyroidal hormone

46 XY - m

graduate on long arm of X

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46 XX - F

Felix de Loos

1/4000 - M

but true explained

1/1000 - F

most cases

most cases

① Maternal imprinting
Genes cannot be switched on and off
as genes switch off or on growth is done
switching on genes → temporary
transmission

No infinity

② Genes → proteins
genes → proteins

Genes cannot be switched on and off
as genes switch off or on growth is done
switching on genes → temporary
transmission

We induce ② copy of every gene
(clone)

Maternally imprinted genes MEG
Paternally imprinted genes PE

Genomic imprinting is a result of DNA methylation

that arises from methyl groups
It does not change the sequence of DNA
Makes an allele unusable

Some imprinting causes problems
Imprinting disorders:

Imprinting disorders:

But since in human

we inherit one from
mother other

both paternal

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1/4000 - 1/1000

most cases

#

Limitations of Mendelian:

① Gene interaction
Phenomenon called epistasis
2 genes decide 1 phenotype

gene
will affect other gene present at another locus
not added

Suppression: Stabilic
Gene
Suppressor: Hypostatic

Example: Bombay blood group.
Bombay blood group + O despite 1 parent AB.
Sex linked inheritance

* This is due to the fact that XY are heterozygous
and one of different lengths. Thus only 1 allele
is present so even if it is recessive it will

show phenotype.
X dominant
X recessive
linked e.g. (dom. allele X recessive allele)
Hemophilia

② Sex influenced characters:
Sex influenced characters: Expression depends on hormones -
How the expression of gene is dependent on
hormonal stimulation and release with size
e.g. Testes
Gonadal development



Sex Limited characters:

How the expression is completely dependent on sex e.g. Milk production

③ Extra nuclear inheritance: Mitochondrial DNA is passed on by female and thus does not follow mendelian inheritance patterns.

④ Incomplete dominance: Heterozygous individuals will be dominant and recessive individuals show intermediate phenotypes.
Tay Sachs disease - half normal

⑤ Co-dominance - AB system, MN etc
both alleles fully expressed w/ heterozygous condition

⑥ Crossing over
Interphase

Polygenic inheritance and quantitative inheritance
Skin colour not constant. (Dartmouth 1913)

⑦ Dominance
Co-dominance
Incomplete dominance
Recessive
Sex linked inheritance
Epistasis - control of trait by two or more genes
get linked inheritance from what ever trait is overriden
e.g. DNA polymerase
Int. DNA polymerase
Int. DNA polymerase

Lepusot - bluish-grey in middle coat colour. A yellowish brown in tail

Lethal / Sub-lethal / Semi + Lethal inheritance

Lethal, Sub-lethal, semi-lethal genes are just

on the road of "vulnerability" and "susceptibility".

Classification

① Based on time of expression

Sometimes Lethals: vulnerability and susceptibility (=)

either genetic damage before or after birth

Sub-lethals: How the gene expresses before

the reproduction etc and the damage does

Epilethal

3. Semi-lethals: How the gene expresses after

the damage has attained reproductive maturity.

e.g. Huntington disease, Maunophilia

② Based on effectivity and dominance:

③ Incomplete dominance lethal

e.g. HbS HbS HbA HbS HbA HbA

(X) Incomplete dominance

If severe HbS HbS or HbA HbA can occur at any time \Rightarrow

④ Dominant lethal: $\frac{1}{100}$ Kinanthropometry \rightarrow death

e.g. Huntington \rightarrow dominant autosomal lethality. Huntington expression at 40 years of age.

Semi-lethal disease

Ebola: sub-lethal

mental deficit, tumors, abnormal skin growth

Huntington: express only in Huntington conditions

Huntington — sex linked recessive

• all males die \hookrightarrow Males $\times \text{Y}$

• only Huntington females die. $\times \text{X}^{\text{H}}$

⑤ Conditional lethals: Expression is pronounced

only under certain environmental conditions \Rightarrow

Rh — erythrodactylus foetidus

Mother $\xrightarrow{\ominus}$ Child

\ominus Child \oplus

Antidiarrhoeal \rightarrow BGM \rightarrow Placenta cut

\oplus No Antidiarrhoeal

Allantois: Placenta \rightarrow dead, Humans \rightarrow not killed under high temperature

Case study - Mar 2021

Australian reindeer call for release

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Australian reindeer - occurred of producing 4 own children - but had actually - CALM mutation - heart condition

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Elimination of lethal genes can be done by identifying carriers via pedigree analysis and preventing them from breeding

- genetic screening

- pedigree counseling

but fear of eugenics

Conclusion

Polygenic inheritance

Nikom - Ehrlich (1909) developed multiple gene hypothesis
When several genes present at different loci in chromosomes or at different chromosomes determine a single character. It is called polygenic inheritance.

Since quantity of these genes produce more particular allele form decide the magnitude of expression directly polygenic inheritance is also called as quantitative inheritance.

Q. Skin colour (studied by Davenport (1913))

Eye colour

Height

Hair colour

TQ

Skin colour → melanin

Melanin, colour → melanin. directly proportional to

No. of contributing genes

P

Normal

Agouti

White

Black

Pleiotropism

It is a condition where a single gene is responsible for multiple characters.

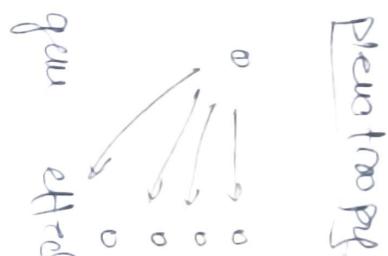
Drosophila melanogaster

→ produce accumulation of phosphatase in blood

It also controls hair pigmentation,翅膀 pigmentation & mental retardation

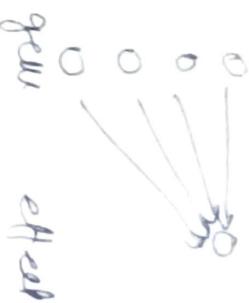
Penetrance:

It is a % of individuals who express trait they are supposed to express based on their genotype or phenotype.



Penetrance

Monogenetic trait



Polygenic trait

gs

RACE AND RACISM

Race is a Biological concept and Racism is a Cultural concept

In brief: Race represent an important part of history & development of Phys. A & Soc. A, which started randomly in Africa

but also from a major push by Colonial powers who wanted to spread Hinduism as a superior race.

Past 2 yrs, UNO commissioned a scientific study on race by a team headed by John Monteith. He came up with conclusions mentioned in UNESCO Statement on Race which besides other things included — "Race has no scientific basis".

Thus pure race in humans does not exist. There are no harmful effects of inter racial marriages. Race do not differ significantly with respect to other capacities and capabilities.

Heredity is the genetic composition of an individual. Environment refers to everything except genes.

In fact heredity as such does not play any role in race formation. Its only function is to pass the genetic variation to next generation. W/o variation, there won't be any change in allele frequencies.

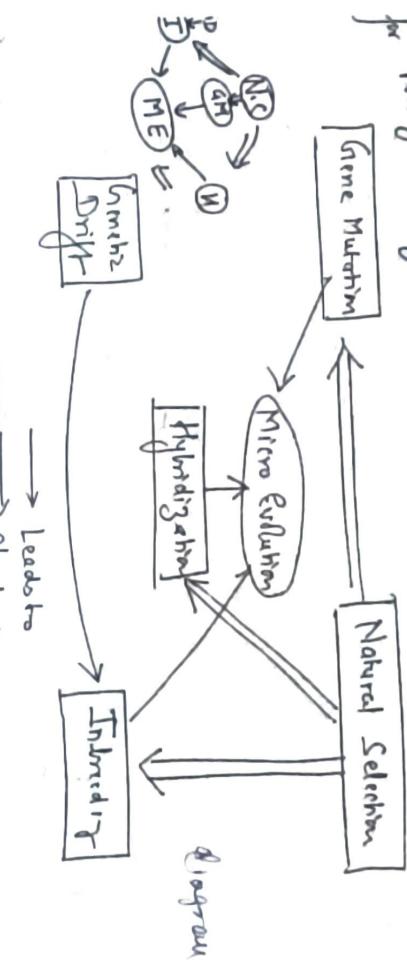
But environment is dynamic.

Genetic mutation - caused by environment

Natural Selection — is Natural

My. Influ., Soc. Env. — Cultural (man made part of environment)

unique
gene pool
process

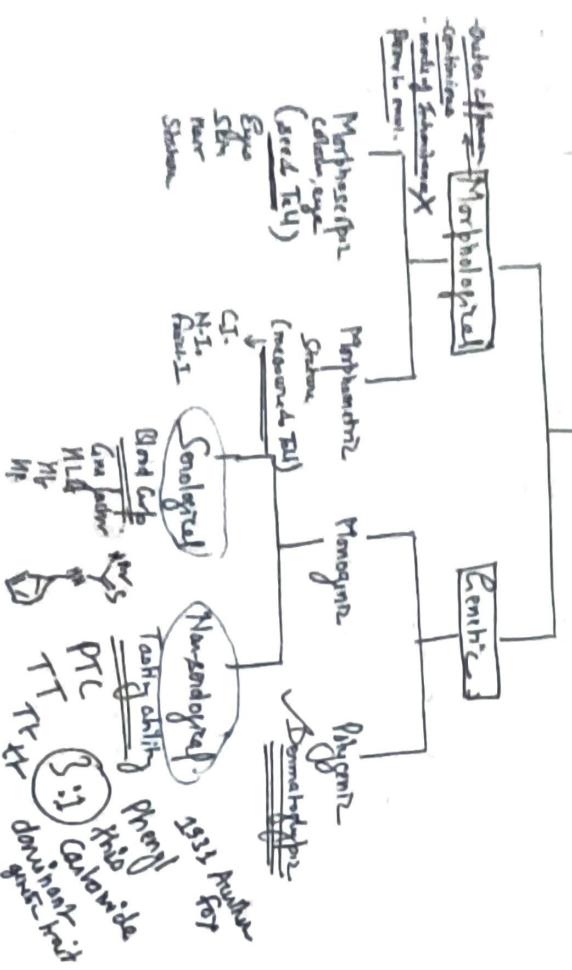


Thus, it's the interaction between environment & heredity that leads to new formation (synthesis)

Conclusion: According to Lorenzen (1921), human diff. can be classified into 7 basic geographically. He concluded that only 15% variation can be attributed to racial diff. 85% are due to differences b/w man & man, family & family.

Finally, on fully developed Human Genome Project it is proposed concluded that all human pop are 99.9% similar. Some variation of 0.1% are not enough to warrant classification.

Racial Criteria:



Morphometrics:

Eyes - Vepicanthus fold → mongoloid, out dr. skin - Keratodermia (white) → European
Yellow retinoid → Mongoloids
Xanthochrism

Hair - Smooth → Mongoloid ✓
Wavy & curly → Caucasian ✓
Wavy → Negroide ✓

Stature - Environment & Nutrition.

Morphometric:

$$\textcircled{1} \quad \text{Cephalic Index} = \frac{B}{L} \times 100 \quad \begin{array}{l} \text{Dolichcephalic} < 74.0 \\ \text{Mesoccephalic} \\ \text{Brachycephalic} > 80.0 \end{array}$$

$$\textcircled{2} \quad \text{frontal Index} = \frac{L}{B} \times 100 \quad \begin{array}{l} \text{Endocranial} < 84.0 \\ \text{Exocranial} (84-87.5) \\ \text{Opistocranial} > 87.5 \end{array}$$

$$\textcircled{3} \quad \text{Nasal Index} = \frac{B}{L} \times 100 \quad \begin{array}{l} \text{Broad & short} \rightarrow \text{Negroid} \\ \text{Narrow} \rightarrow \text{European} \end{array}$$

General criteria

Mnemonic → Blood group

(1)

It is thus, very difficult to make any generalization.

(2)

divisions of people into 'races' based on blood group.

① Blood group

Blood groups were known since 1900; but didn't classify who done 3 decades later by Beyd and he classified the world in (6) — Early Europeans (Europe), Central Europe, Africa, Asia, America, Australia, full of Europeans.

Summary of principle Blood Group Distribution

ABO

O (62.3%), A (25.4%), B (10.2%) — Abundant in America, India, Africa.

MNS-U

Rh (N) → Americans, (N) → Australians, M & N elements Ue → only in Africa.

Rh

Rh (N) absent or rare in most of the world
Ls+ Ecu., R+ Ecu. → Africa (40%)

Duffy

Absent in Polynesians (not common)
S. Amer., S. Amer., S. Amer.

Diego

Diego (N) → Americas, Indians — (absent in Africa)

Kidd

Kidd (N) → West Africa and American Indians (not common)

Mnemonic → Polygon → Dermatoglyphics.

2. Dermatoglyphics:

Under it we study ridges on the skin of finger, palm, toe & sole. Ridges: mostly studies have been done on fingers & palm. Thus, we will restrict to the discussion of dermatoglyphics in palms & fingers only.

① Finger Dermatoglyphics:

Ridges form definite patterns called Angerull pattern:
4 types:

② Arches



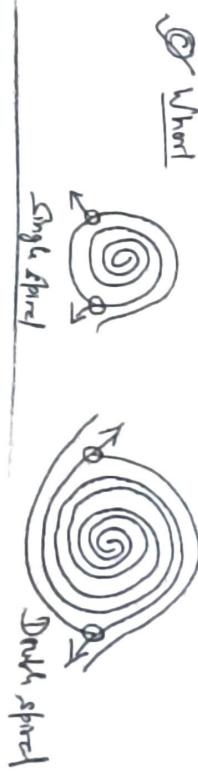
No double arch.

③ Loops



Triradius →

Arches	Whorls
0	1
1	2
2 or more	3 or more



④ Complex



On the basis of above human popn can be classified

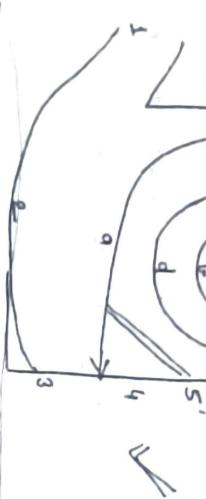
- Total ridge count, differential ridge count,
- pattern intensity index etc.

⑤ Palm dermatoglyphics:

Our ridges on each finger base A, B, C, D

Rule: Sturdy palm main lines called a, b, c, d

Palm is divided into 13 regions.



Popn can be divided into races using —

Main line formula ✓

Black line index ✓

Thinner ridge count etc. ✓

But, UNESCO said, variations within the popn of a race
much more than the differences b/w races. Thus, redundant

Now, if quinque is adopted of pure race exist.

- Introducing — Harmy + UNESCO
- Definition of Race → Similar
- Criteria → Proper

- Conditions for racial classification (blend)
- Morpho our
- Emrie fit → MO etc.

— Conclusion → Human Genome Project got substituted fossils → gggg.
Less known and no longer used, instead Hyper somatic One protein follow human distribution continents divide

Ace. due

J.C. Boyd

for selecting a good criteria for racial classification

⑥ They should follow Mendelian

⑦ They should be discrete & not continuous.

⑧ They should not be easily influenced by environment

— Stone

⑨ They arose quick early in human evolution
of these first three are usually important

Caucasoid

Mongoloid

Negroid

ABO	large modulus of A	high incidence of B	red ridge modulus of A and B
Rh	Rh - more frequent	Rh - less rare	Rh - it modulus

Dermatoglyphic

line

Pattern

density

formula

Main line	marked transversally	longitudinal alignment	<u>Opposite hands alignment</u>
formula	11-9-7	9-7-5	7-5-5

3 (9.3)

Genetic polymorphism

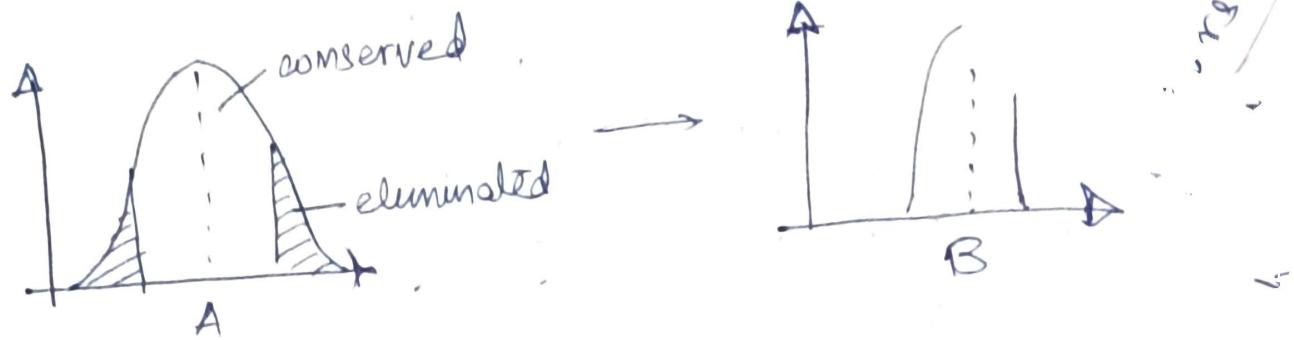
Defn by Cavalli-Sforza & Bodmer (1971)

Genetic polymorphism is the presence of several distinct ^{alleles at same locus} forms ~~of a gene~~ within a population with frequency greater than 1% i.e. several different forms of a gene that cannot be maintained by recurrent mutation.

- ~~- Phenotype polymorphism is the presence of several distinct forms of a phenotype in a population with frequency greater than 1% (only if $> 1\%$, we can be sure that natural selection has acted on it)~~

Monomorphism - also called stabilizing selection

- it is the presence of only one form of a gene or phenotype within a population
- it operates in constant or unchanging environment for long periods of time
- it introduces homogeneity in population
- favours average / normal individuals and eliminates over and under specialised individuals
- checks mutations that may lower the fitness in an unchanging environment
- rarely operate as environment ^{is rarely} constant



stabilizing selection

examples

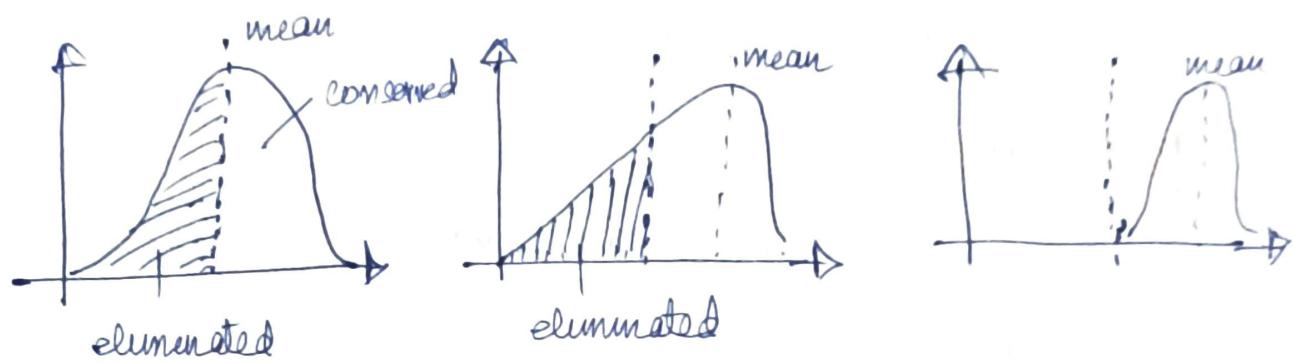
- Individuals not perfectly male or female are removed from population
- so that population shows only 2 discrete individuals with respect to sex. i.e. male or female.

Optimum birth weight in humans is 7.3 pounds and newborns less than 5.5 pounds or more than 10 pounds have higher mortality rate

Transient polymorphism - called directional selection

- when there are 2 alleles in gene pool if one is replacing another gradually under changing environmental conditions
- operates when environment is changing in a particular direction
- favours accumulation of those mutations

- increase fitness in new environment
- favour non average - i.e specialised phenotypes and eliminated normal or average
- brings progressive evolution



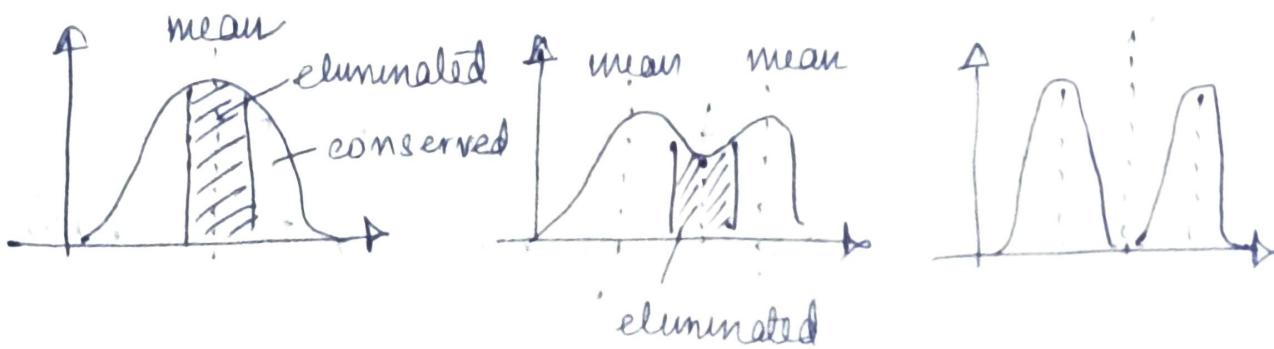
examples - mosquito resistant to DDT

↳ resistance of bacteria to certain drug

biston betularia → studied by Kettlewell in industrial ~~water~~ melanism in moth. When environment got polluted; mutation for black coloration of white moth spread as it protected from predators. So at a time when one form has not replaced other, polymorphic population exists. But over time polymorphism is replaced by monomorphism.

Balanced polymorphism or balanced s.

- maintenance of 2 or more allele frequencies with frequency greater than 1% in a population.
- sometimes, the result is heterozygote superiority.
- maintained through disruptive selection



- favour the extreme and eliminated members of mean expression - so two peaks seen
- very rare in nature but very important in bringing about evolutionary change

example

C HbA/Hbs has both HbA and Hbs

C Hbs is eliminated in US

C but in Africa - Hbs is preferred as it gives resistance to malaria

C fitness is higher than HbA/HbA and

HbS

heterozygote is best fit

- Thalassemia (β major or minor both types)
 β chain are defective), minor (α other chain defective)

Example

CD14 gene polymorphism in humans - one polymorphism lead to more CD14 protein as well as reduced levels of IgE protein - also varied based on allergen exposure - so help in treatment of asthma
CYPD gene polymorphism - lead to reduced DNA repair efficiency - increased risk of lung cancer

(GWAS)
Genome wide association studies - used to associate specific genetic variations with particular disease - use Manhattan plot to display single nucleotide polymorphisms.

Polymorphic vs Monomorphic Tr.

LO
very

~~Monom.~~



Polymorphic traits

- discrete phenotypes
- discontinuous
- controlled by single gene
- unifactorial
- Mendelian inheritance
- less modified by environment
- eg: ability to roll tongue longitudinally

polygenic
~~Monomorphic traits~~

- graded
- continuous
- controlled by multiple genes
- does not follow Mendelian inheritance
- strongly modified by environment
- eg: height, weight.

Advantages of polymorphism

- provides popn with alternate set of traits of character.
- very significant in changing environment.
- leads to natural selection.

Mendelian population

Dobzansky

Every individual ideally has an equal chance of mating with all other individuals of the population. But due to barriers such as caste, class, religion, they resort to inbreeding.

- So for population genetics, mendelian population is unit it need.

Mendelian populations

- aggregate of interbreeding individuals mating randomly.

~~Dobzansky~~ - said Mendelian popⁿ is a reproductive community of sexual and cross fertilizing individuals which share the same common gene pool.

gene pool

called Panmixia by Sewell Right.

↳ mendelian popⁿ by Dobzansky.

Nature of mendelian population

- mating at random.
- own gene pool.
- any gamete in this popⁿ has equal chance of mating with gamete of opposite sex in same group.

Factors that divide mendelian population

There
are

- geographic barriers
- linguistic barrier
- social
- economic
- educational.

m

- mendelian population is not restricted by absolute size if individual mate at random.
So mendelian popⁿ is an important tool to study law of inheritance and process of evolution

e.g. endogenous group in mendelian pop

Population genetics

- study of frequency of gene and genotypes in a mendelian popⁿ, i.e. popⁿ genetics
- it has been recognised that although inheritance of individual gene may be governed by Mendelian principle, the freqⁿ of individuals carrying true gene may depend on several factors like gene freqⁿ, popⁿ size

1908

Hardy Weinberg equilibrium

mathematician

physician

There are certain traits in human popⁿ

which remain in genetic equilibrium and do not evolve.

e.g. PTC test.

mathematical rule to explain when a popⁿ is undergoing evolution and when not.

Hardy Weinberg law gave an equation to explain the situation.

proposed and demonstrated

G. H. Hardy

Wilhelm Weinberg

before this, view was that dominant allele would increase in frequency.

assumption

there is genetic equilibrium

no evolution.

what is genetic equilibrium:

when evolution not happening - so gene frequency \rightarrow genotype freqⁿ is not changing over time.

no natural selⁿ

no mutation

infinitely large population

random mating

no differential reproduction

no immigration or emigration

Hardy Weinberg's Law

The relative frequencies of various kinds of genes in a large, randomly breeding, ~~panmictic~~ population, tend to remain constant from generation to generation in the situation of ~~no mutation, evolution, gene flow etc~~.

$$p^2 + 2pq + q^2 = 1$$

So if allele freqⁿ are known, then freqⁿ of 3 genotypes can be calculated

where p and q are allelic frequencies of dominant and recessive alleles

Example $p^2 \rightarrow$ genotype freqⁿ of AA
 $q^2 \rightarrow$ aa $2pq \rightarrow$ genotypes of Aa

parents

YY

Yy

yy

genotype frequency

0.49

0.42

0.09

total individuals

245

210

45

= 500.

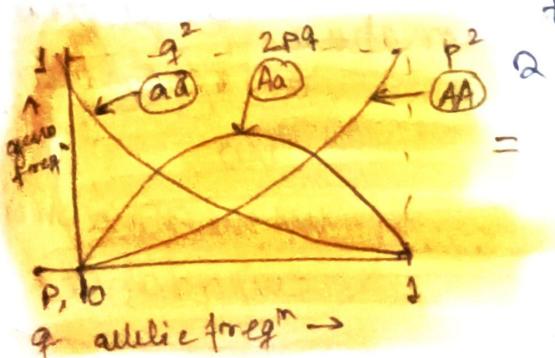


$$\text{so } Y =$$

$$\begin{array}{r} 245 \\ + \\ 245 \\ + \\ 45 \\ \hline 530 \end{array}$$

$$\begin{array}{r} 45 + 45 \\ + 210 \\ \hline 300 \end{array}$$

$$= 700$$



frequency $\frac{700Y}{1000} = [0.7 = P]$

$\frac{300y}{1000} = [0.3 = q]$

next generation.

Since random mating

♀	P	q
P	YY	Yy
q	Yy	yy

sperm can be Y or y.
egg ————— Y or y.

so

$$\begin{aligned} \text{so } p^2 + pq + pq + q^2 \\ = (0.7)^2 + (2 \times 0.7 \times 0.3) + (0.3)^2 \\ = 1. \end{aligned}$$

If not 1, then means the evolution is ongoing
deviation tested on Pearson Chi squared test.

Applications: and Fisher's Exact test \rightarrow computer needed

- this law helps us analyse if population is evolving or not. - measure evolution.
- calculate frequencies of specific allele.

and genotype such as deleterious recessive allele - so can help prevent disease like

Thalassemia like in UK

Epigenetics

- no sequence change of DNA

phenotype

- Epigenetics is the study of heritable changes in gene expression that do not involve change to the underlying DNA sequence.

- a change in phenotype without change in genotype

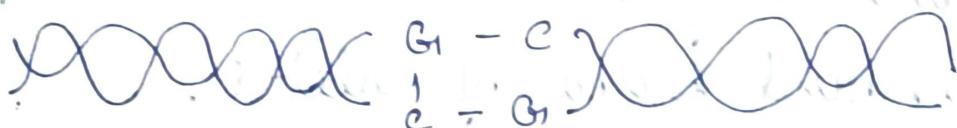


- epigenetic changes can switch genes on or off and determine which proteins are transcribed

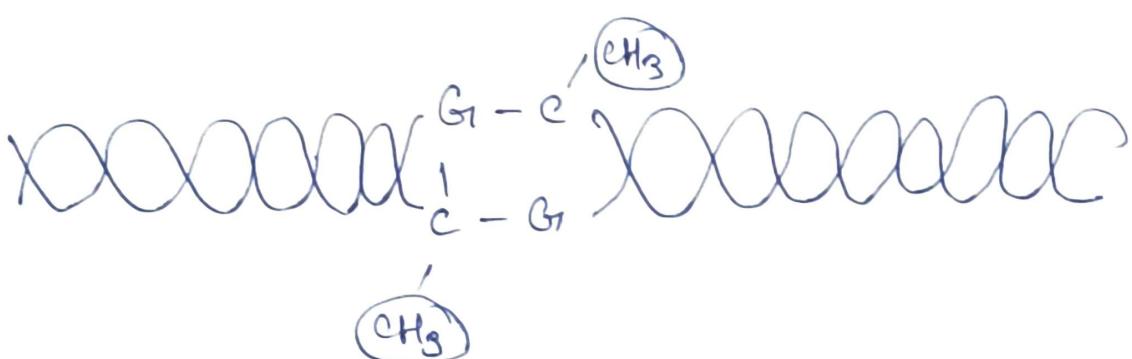
(Methods by which genes are affected)

3 ways

→ DNA methylation



↓ after methylation.



Such chemical changes make the gene more inactive - it modifies the gene's mechanism for transcription

~~modification~~ - by ~~acetylation~~ and
of proteins that is around it
~~ethylating~~, to make the chromatin condensed
acetylation hence inactive to transcribe.

- RNA associated silencing - can affect gene expression by causing heterochromatin to form.

Importance of epigenetics

- can switch genes on or off and determine which proteins are translated so can explain effect of environment on genes
can help better understand nature vs nurture debate - twin study different explained by M
- metab crestandin - due to fragile X chromosome caused by epigenetic changes
- help make better life style choices e.g. Vit B can protect against pollution.
- genome wide protein study

Euthenics

- seeks to improve already existing human beings which can be achieved by improving the environmental conditions eg. better nutrient intake, lesser pollution, better education

Eugenics

coined by Galton

- seeks to interfere at the inherent level of gene level.

- it is the practice or advocacy of improving the human species by selectively mating people with specific desirable hereditary traits.

- aims to reduce human suffering by "breeding out" disease, disabilities and so called undesirable characteristics from the human population

Origins

- Sir Francis Galton - if selection tried to protect weak, then at odds with forces of natural selection - will lead to "reversion towards mediocrity"

- Nazi war criminals tried to say US eugenics studies inspired them to do what genocidal

suggest us

Eugenics today: ethical concerns

- do modern day attempts to eradicate hereditary disorders equate to eugenics?
- genetic testing for purpose of disease eradication will likely involve a particular ethnic group due to the shared ancestry.
eg: Tay Sachs disease common in certain Jewish communities - kids die by age 4.
- can lead to racist ideologies like those of Nazi Germany
- overemphasis on eliminating disabilities in unborn children can mean bad for those already with the disability

→ particular social groups → discrimination
Tay Sachs - Jewish
already with disease

↳ positive eugenics:
increase frequency of beneficial
genes
eg: selective mating

↳ negative eugenics:
involve elimination of deleterious
genes from gene pool.
eg: haemophiliacs advised not
to reproduce.

Means of eugenics

(M)

- selective mating.
- whole animal cloning - SCNT.
- gene therapy - missing gene is produced or defective gene is killed.
- recombinant DNA - combining DNA from two different source.

To not fall into the negatives of eugenics, we must not be ethnocentric and focus on the anthropological definition of culture.

RRR

of culture.

(M)

DNA Tech. Bill - good step

specie



DNA fingerprinting

- Sir Alec Jeffreys 1985 invented it.

It is a modern application of DNA technology used in personnel identification. It is based on the principle that chemical structure of everyone's DNA is same, just the order of sequence of base pairs is different.

Principle and process

- There are so many millions of base pairs in each person's DNA that every person has a different sequence.
- but, so many millions of base pairs - can be very time consuming.
- instead shorter method - because repeating patterns in DNA called VNTRs. (variable number tandem repeats) which are known to vary among individuals a great deal.
- VNTRs are able to determine whether two DNA samples are from same person,

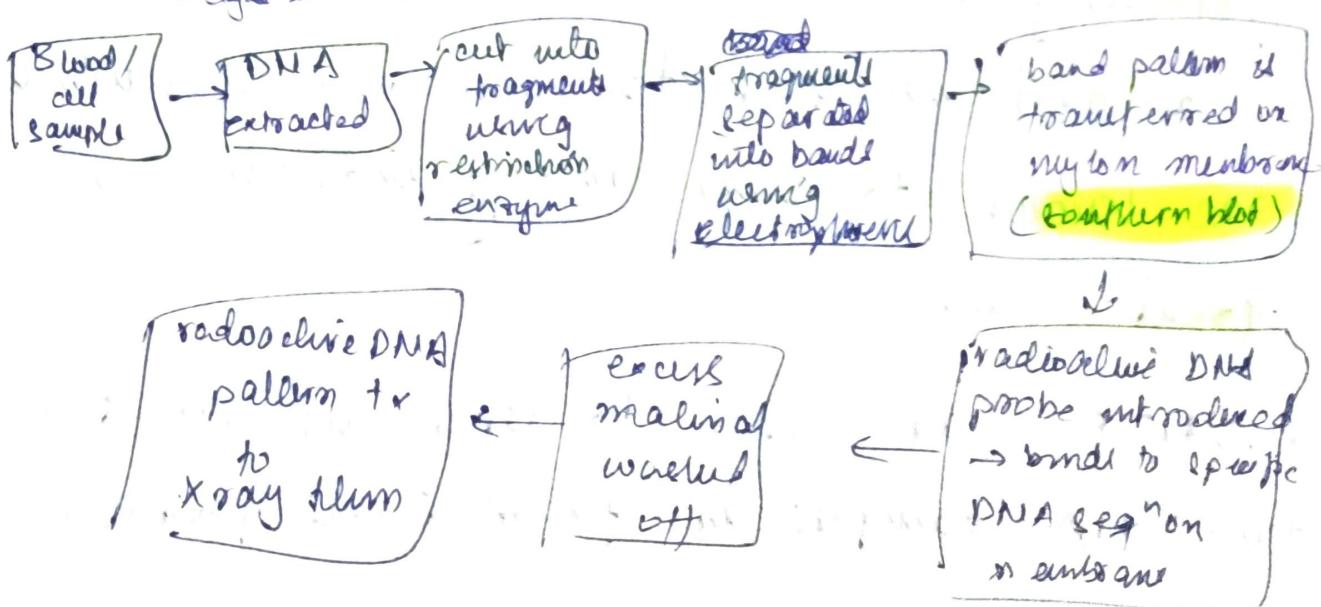
- ~~DNA~~ DNA finger printing - every human has unique sequence of DNA. i.e. very short nucleotide repeat. (VNTR).

Succesful

- Clyde Snow, identified a skeleton found in Boozed as that of Nazi war criminal Josef Mengle.
- Karen Burns - 9/11 victim identification and investigation of Rwandan genocide
- African Burial Ground Project - incontrovertible evidence of horrors of slavery in North America. ~~in~~ in New York
- N.D. Tuwan paternity diagnosis case

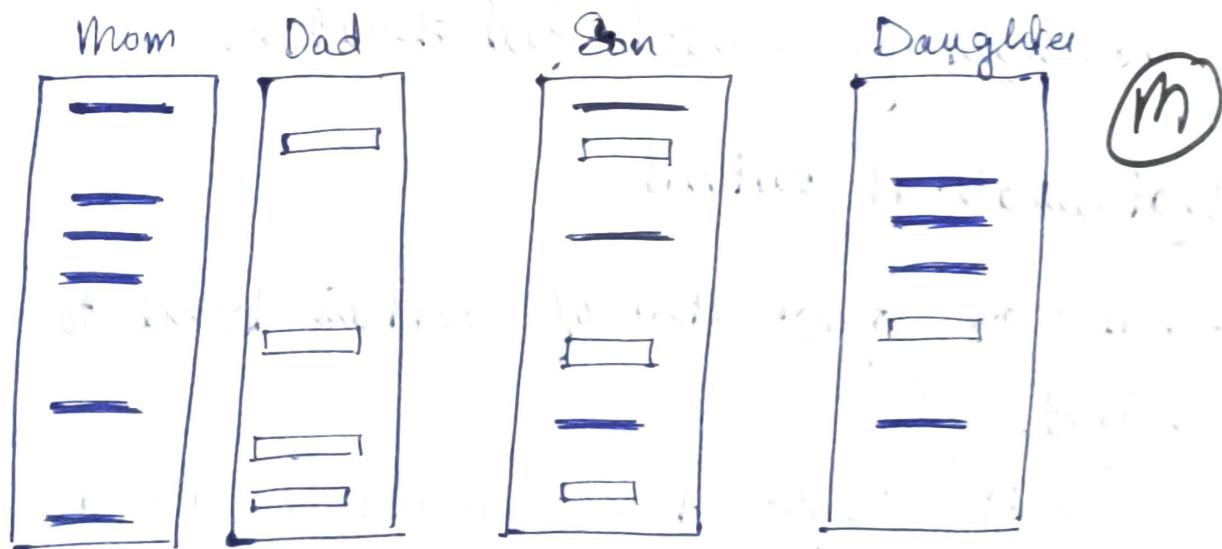
Ethical side of forensic anthropology is also important - need to bring responsible parties to justice - no conflict of interest

DNA finger printing process



~~and~~ persons or non related people.
VNTRs can contain anywhere from
20 to 100 base pairs.

- VNTRs are hereditary hence unique.
- So individual will have VNTRs inherited from mother or father, or a combination of both, but never a VNTR either of parents do not have.
- To determine if a person has particular VNTR, Southern blot is performed. And then it is probed along with a reference radioactive version of the VNTR in question.



* DNA tech belt

~~walshin~~

- Hb S is well distributed in Africa because it provides resistance against malaria.
- Hb E is well distributed in East Africa
- Hb F in East Asia

caucasoid

- * white (terracotta)
- * light blond hair
- * fine hair
- * wavy hair
- * light blue / light brown eyes

dolico to broady

11 subgroups

- mediterranean
- Indo African
- nordic
- alpine
- denasic
- caucasoid
- polynesian
- east Baltic

mongoloid

- yellow - yellow brown (xanthoderm)
- brown hair
- medium hair
- smooth hair
- light to dark brown eyes

* epicanthic fold
broady epiphalic

- classical or
central

Mongoloids

- arctic or
Northern -
mongoloid or
eskimosid

- southern or
Indo Malayan
mongoloid

American
Indian

negroid

- brown-black (melanoderm)
- black hair
- coarse hair
- woolly hair
- dark black
- eyes

prognathism

dolichcephalic *
protruding
occipital region

- African
Negroid

Oceanic
Negroid

northern
central
southern
Africa

Race and racism

Race

- * biological concept
- * scientific criteria
- * Morphometric, morphoscopic
endemic used
eg: cephalic index
- * difference in races
due to different
environmental
adaptations
- * no note in race

| intelligence
| cultural
| differences
| purity of
| blood.

Debunking racism

- blood groups vary independently of each other and of characteristic such as skin colour, intelligence etc.

eg: Eskimos, Portuguese, Australian aborigines

Racism

- * culturally defined concept
- * discriminatory effect
- * factors used
 - | purity of blood
 - | cultural superiority
 - | mental ability
- * wrong and unscientific thinking
- * creates xenophobic ideas

Group Blood

~~Ramberg clearly said that
no link between race and psychology~~

5. variable in blood group.

skin colour - adaptation to various climatic conditions.

- mental ability - brain size variation varies both inter and intra race - combination of genes and environment.

eg. frontal lobe - seat of ability - constant 44% of weight of brain

- cultural superiority - memory goes twice reared apart differ in mental ability.

UNESCO declaration about race

@ 1952

- all human beings belong to one species called homo sapiens
- all morphological and anatomical differences are the result of gene and environment and not because of only one of them.
- you cannot be grouped on the basis of nationality, religion, culture, language.
- intelligence or cultural differences are not the cause of racial differences.
- pure race has never existed - intermixture

Race crossing in man

- UNESCO has said there are no pure race
- interbreeding b/w two different races.
 - has been always going on.
 - interbreeding would result in admixture of these traits.

Example.

English Indian
Moplah in Kerala

- europeans have penetrated south west Asia and all regions of the world.
- mulatto - cross b/w negro and white

- African Americans

- mestizo - American Indians x white cross

Cause of race crossing mainly in S. America

- war
- colonialism
- expeditions
- slave + trade
- marriage alliances
- globalisation

- define race
- pure race - not
- race crossing

- UNESCO

Disability and impairment

Disability is the physical condition of a person in which he is unable to perform day to day functions.

Impairment is a phenomenon at the tissue level that impedes normal working of part of body due to disease or injury.

Anthropology is a holistic study so it is natural that anthropologists would be interested in the concept of disability and impairment to study it from various angles.

Anthropological understanding

the other → anthropologists understand how different cultures perceive others.

- the word "disabled" has been used to label people as different from people who are able bodied in one way or another.

- unlike social categories like gender, race etc, disability is a category one can enter anytime.

- although all disadvantaged groups

have a chance higher of becoming disabled

~~It~~ parallel to this can be drawn to the function of poverty by Amartya Sen - the inability (or disability) of the person to lead the life he or she cherishes.

→ what is considered to be a disability is different in different sociocultural settings.

~~Jayne Ginsburg~~ ^M said anthropology's key contribution to disability study is to understand social and material conditions that disable the full participation of all.

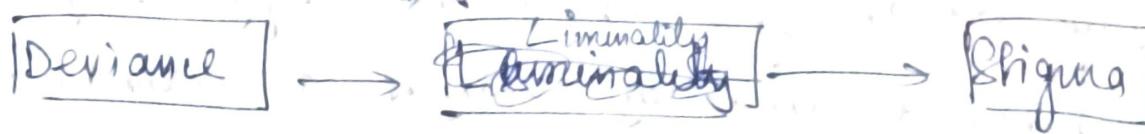
New anthropology says that "disability" is a category that is socially constructed.

~~Ruth Benedict~~ studied cross cultural comparisons - found that epilepsy (for eg) may be considered abnormal and undesirable in a sociocultural setting, but a highly valued psychiatric manifestation elsewhere. Her student ^{Mead} said disabled ppl are to be included as "normal". American for national ch study

★ Anthropologist Robert Edgerton was the first to explore mental retardation from an anthropological perspective.
"The Cloak of Competence"

- anthropology also contributed to understanding deviance, luminosity and stigma - and how it works in social context to make disability a category that is socially constructed.

e.g. work of Erving Goffman and disabled anthropologist Robert Murphy



person is evaluated as different/ loss of role.
socially valued as adverse
as disabled reaction by others.

Hence anthropologists have determined that disability has little to do with degree of functional loss or impairment - rather it is defined by societal standards and expectations for normal behaviour.

Applications

- in Indian context - can be used to improve policy making - e.g. improving social disability of manual scavengers so that they can be rehabilitated better.
- ↓ stigma of wt. disab.
- care & protection