CONSANGUINITY — Marriage b/w blood relatives

Sanguine = blood ; con - same

→ Consanguinity is problematic in recessive condition because chance for getting an affected child by consanguineous marriages are much more. This chance is more because the chances of having same type of recessive allele is much more higher in this case as compared to non-consanguineous marriage.

Consequences of consanguineous marriages -

This is higher in Japan, S. India, Middle East.
↓ 50 case studies; now stopped resulting in -
Deleterious alleles have been purged out now.

Generalized data of consequences of Consanguinity

1) Still birth.
2) Neonatal death.
3) Congenital abnormalities.
4) Abnormal offspring 3-5 % (in general population 2-3% whereas max. marriages are non-consanguineous

From 3rd degree relations consanguinity is not very good ~~much~~ but on later (4th, 5th, ...) generations it is more.

F = Co-efficient of breeding

1° of consanguinity relationships :

Monozygotic Twins.

Dizygotic Twin = Siblings

2° of consanguinity relationships.

uncle - niece = $\frac{1}{2}$

Aunt - nephew = $\left(\frac{1}{4}\right)$ $F = \frac{1}{8}$

$\frac{1}{4}$ raised by a factor of 2.

3° of consanguinity relations.

$\frac{1}{8}$ $F = \frac{1}{16}$

4° of consanguinity relationship

$\frac{1}{16}$ $F = \frac{1}{32}$

5° of consanguinity relationship.

Second

$\frac{1}{32}$ so $F = \frac{1}{64}$

$\frac{1}{32}$ raised by 2.

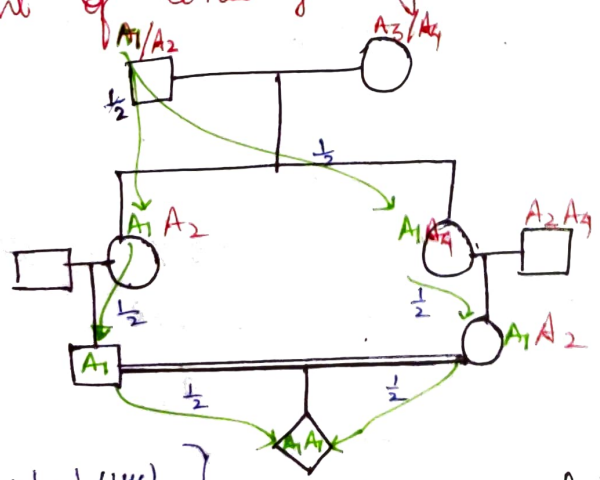
for autozom

In a general population, each individuals is recognized as 8-10 alleles, minimum. mutant 1. Mostly they are in heterozygous condition so not causing defect but on consanguinity their chances of being homozygous mutant too very much.

> Closer the relationship $\propto$ More population of alleles ~~thou it~~ shared
> $\propto$ Co-efficient of inbreeding.

Measurement of Consanguinity; Done on the basis of relationship type

A_1, A_2, A_3, A_4 → 4 different allele of same gene A



Here same A1 has descendant from a parent

p(any homozygous) = $4 \times \frac{1}{64} = \frac{1}{16}$

A_1A_1
A_2A_2
A_3A_3
A_4A_4

P $A_1A_1 = \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2}$ (LHS)

$\frac{1}{2} \times \frac{1}{2} \times \frac{1}{2}$ (RHS)

what is the chance for being A_1A_1 (homozygosity) for this foetus.

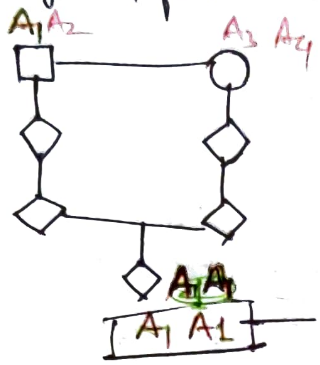
POSSIBILITY :

① They are not consanguineous, then parent should be ...

Here A_1A_1 state is from 2 diff ...

Identity by state (IBS) { IBS are ... among population.

② By consanguinity - After several generations of inbreeding



— same A_1A_1 (both A_1 are same) have been arrived in child.

NOTE: 4th onwards generation, we don't find inbreeding effect.

IBD : Coefficient of inbreeding.
↓ Identical
Identity by Descent

→ Genetic Isolates :

eg Ashkenazi Jews → Religious group (inhabited same place and bred among themselves. They had various inbreeding diseases. They harboured diseased alleles in themselves and consanguinity increased that frequency.

eg- ① Tay Sachs.

Frequency in general population $= \frac{1}{360,000}$

" " Askernazi Jews " $= \frac{1}{3600}$

② Familial breast cancers.

F value ~~is low~~ variation

Lowest in Canada .00004 to ~~.0007~~ .0000307

In Roman Catholics 0 — 0.00008

S. America 0 — 0.003

India 0 — 0.02

Japan (Very high) — 0.5% (f)