

## Quantitative Traits: Quantifiable

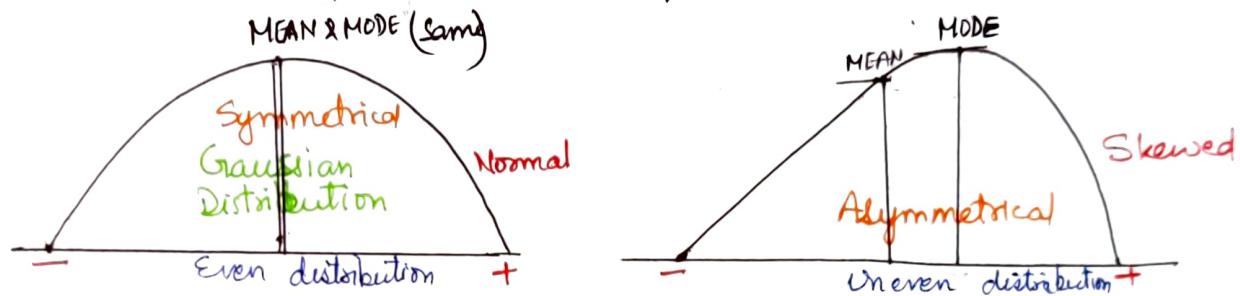
- Metric Trait (measurable)

Metric  
countable nos.  
no. of petals  
" "  
" "  
Continuous gradient (could be in fraction)  
e.g. height  
weight  
bristles  
tufts

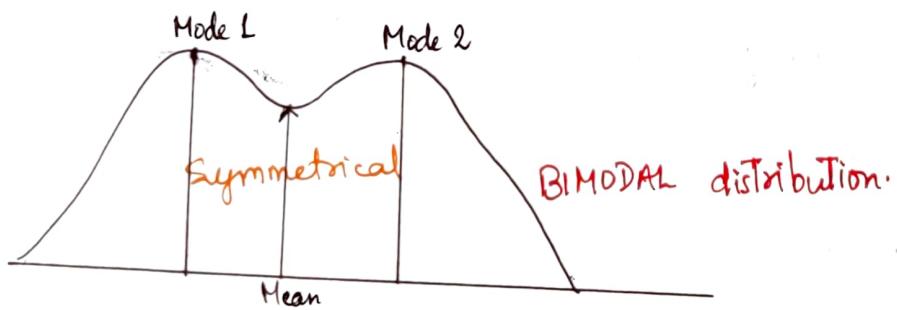
Mean = Sum of the phenotypes

Sum of no. of individuals whose values are summed:

MODE: Value at the peak of distribution and usually corresponding to the most frequent class of people in distribution.



Mode is peak value always, mean may be equal to mode or may not be.



MEDIAN: Class that divides the distribution in equal halves by number of individuals.

VARIANCE: A measurement of the degree to which individuals within the sample vary from one another.

$$S^2 = \text{Variance} (v) = \frac{\text{Summing the squared deviation from the mean}}{\text{no. of individuals} - 1} = \frac{\sum (x_i - \bar{x})^2}{n-1}$$

24/10/2011 Polygenic Trait examples -

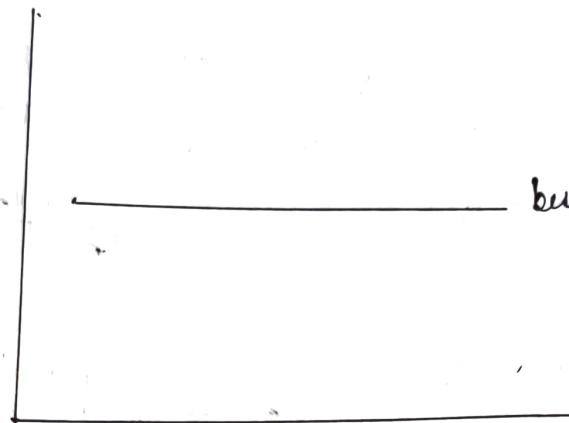
(i) Height -

Offspring  
value -  
Population  
mean



Mid parent value - Mean of population.

plots  
as relatives  
height of offspring  
of mid parent  
values means



Not corr.  
to

### TOOLS FOR ANALYSIS OF INHERITANCE OF POLYGENIC/ COMPLEX TRAITS

- First idea came that - only environment is describing inheritance of traits.
- Environment + Genetics.
- How much genetic (genes?)
- Which are the genes.
- What they do (Molecular pathogenesis)
  - ↓
  - clinical genetics.
  - Symptom → Genes.

① Family analysis

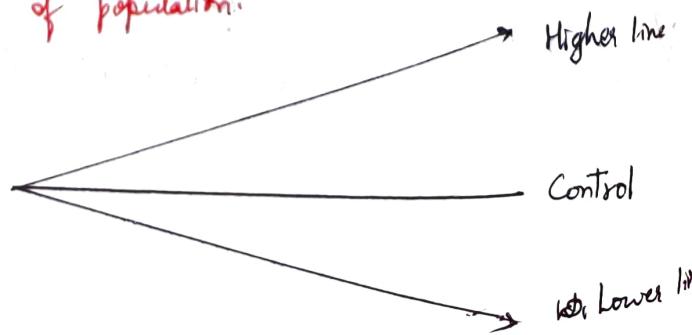
② Twin study.

③ Adoption study

Best strategy to study

is Twin - adoption study

*pure*  
Preparation of lines from mean character  
of population.



Lower line

Non-Mendelian characters are so called because it doesn't follow pedigree we can't make pedigree chart but it follows it there is inheritance of trait.

① **FAMILY STUDY.** Frequency of the Trait in individuals of different degrees of relationships.

Degree of genetic relation. Frequency of occurrence

1st Relatives (50%)	40%
2nd " (25%)	20%
3rd .. (12.5%)	10%

Sharing of genes

Directly proportional.

Frequency of occurrence as decreasing as the degree of relationship is increasing and closeness of relation is decreasing.

Familial aggregation also (Familial clustering)

② How much is the correlation b/w occurrence of trait and genetic relationships → is a/a **Familial Risk Ratio (FRR)** This measurement is calculated in terms of  $\lambda = \boxed{(\lambda)} = FRR$

$\lambda$  value has to be measured

$$\lambda_R = \frac{\text{Frequency of disorder in relatives of an affected individual}}{\text{Frequency of the disorder in the population}}$$

These are the polygenic i.e. Total gene pool.

Total alleles

Total no. of genes and alleles in a potentially reproductive gametes is

Gene pool.

In  $\lambda_R$   $R$  is general denotion of any substance relationship.

$$R = S, P, O, Mz, Dz, 1, 2, 3$$

S - Sibling, P - Parent, O - Offspring, Mz - Monozygotic,

Dz - Dizygotic

1, 2, 3 (is degree of relationship)

The degree of relationship remains effective till 3 then after it is not considered.

$$\lambda_R = 1 \text{ for maximum value of } \lambda_R$$

Chance of developing that trait is not different from general population but equal to any one of the individual

$\lambda_R > 1$  then the disorder is clustered <sup>within</sup> the family.

Familial Risk Ratio =  $\lambda$ .

$\lambda > 1$  then familial clustering.

$\geq MZ$   $\geq DZ$  for some disorders  
↓  
twins

Anorexia nervosa - Loss of appetite due to mental disturbance  
that he/she'll get obsessed on eating. But if it becomes severe then it is genetic.

$$\geq MZ = 710 \quad \geq DZ = 141$$

$$\text{General pop}^{1n} = 0.1$$

$$\text{Schizophrenia} = 4.8$$

$$G.P = 1$$

1/10/2011 Degree of family clustering =  $\geq R$   
(Familial Risk Ratio)

This value is calculated in population? This value is for a particular population.  
It will vary on changing the population.

	Risk for Relatives <small>observed data</small>	Population incidence <small>For Schizophrenia</small>	$\geq R$
Parents	14.12%	0.8	17.65 ( $\frac{14.12}{0.8}$ )
Sibs	8.51%	"	10.6
Childrens	12.31%	"	15.4
Uncle/Aunt <small>2<sup>nd</sup> relative</small>	2.01%	"	2.5

DTE: In complex trait, population study is essentially required. ~~the~~ a familial study may/may not be included. But in Mendelian trait only families are studied but not population at all.

Degree of relationship  $\propto$  ~~proportion~~ proportion of gene shared

- Whenever incidence level in family matches with population incidence level then it means the disease is not genetically related to particular family.
- A co-relation b/w ↑ in degree of relationship with degree of frequency so it is due to genetic component.

## ② TWIN STUDY:

Monzygotic (MZ)

Dizygotic (DZ)

One fertilized egg



from 2 foetuses

→ Either both ♀ %

→ Identical Twins

→ Gene sharing = 100%

→ Environmental sharing  
~100%

Two independently fertilized eggs

Two foetuses

These are just like siblings but growing together temporarily.

→ ♂ & ♀ both could be in same time-

→ Fraternal Twin

→ Gene sharing 50%

~100%

→ Monzygotic (MZ) twins

frequency = 3.5 / 1000 live birth (a globally constant value)

→ Frequency of DZ twin variable worldwide.

East Asia = 1 / 1000 live birth

Saharan Africa = 12.5 / 1000 live birth (much high)

Europe = 4.5 / 1000

→ If both twins are showing (whether they are MZ or DZ) the trait, then it is c/a **Concordance (C)**

If only one twin is showing the trait, then it is c/a **Discordance**

Level of Concordance: It is calculated