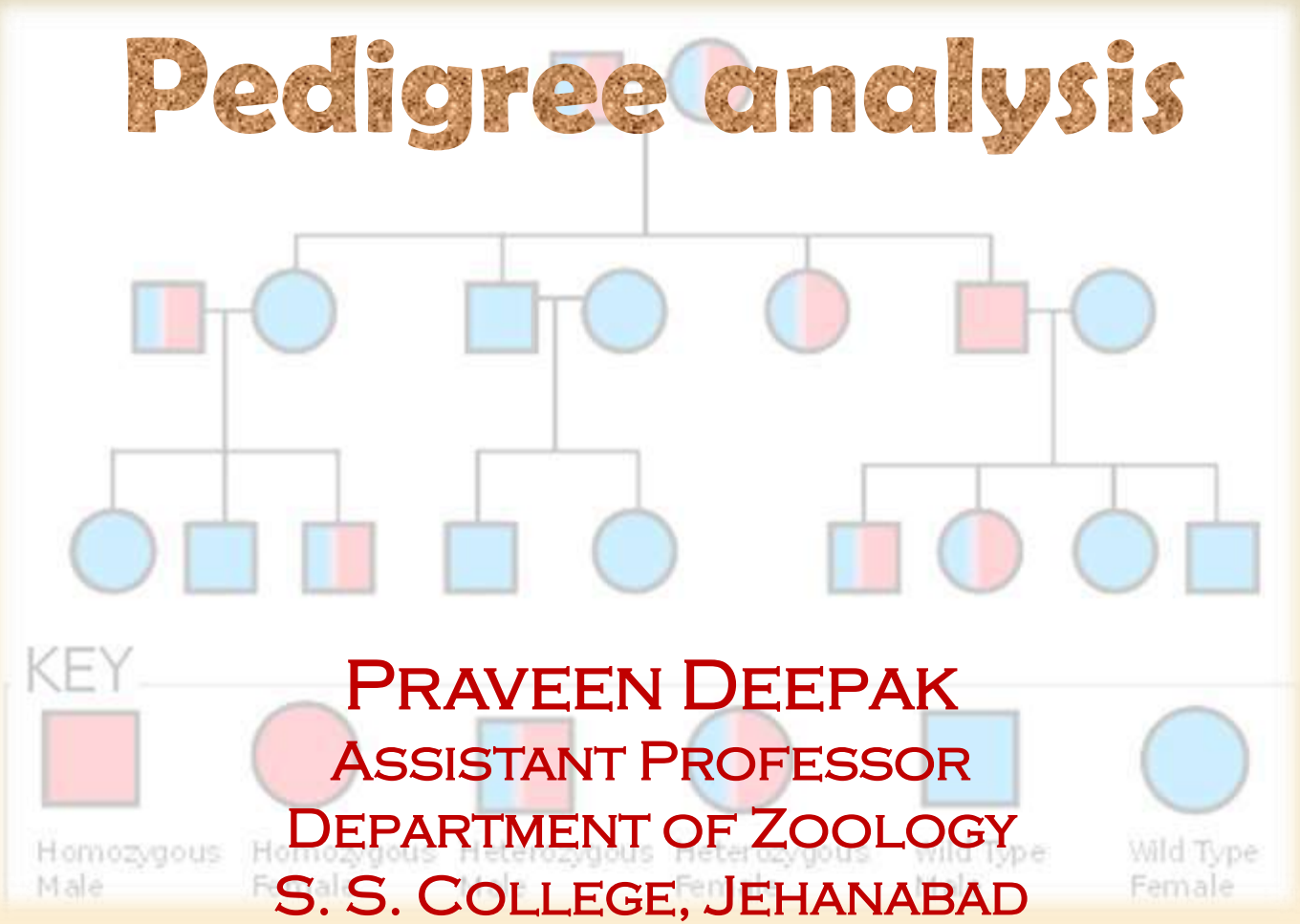
Pedigree analysis



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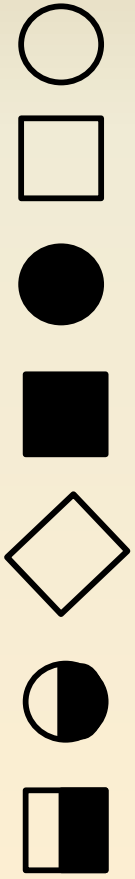
What is Pedigree Analysis?

- Literal meaning of “Pedigree” is “the recorded ancestry or lineage of a person or family or genealogy” or “वंशावली” in Hindi.
- It is an orderly presentation of family information.
- Basically, it is a graphical representation of the appearance of a particular trait or disease in related individuals along with the nature of the relationship using standard sets of symbols.
- It involves the construction of family trees that can be used to trace inheritance of a trait over several generations.
- “Crosses” and “mating” in human(s) are shown and analyzed in pedigree.
- Usually, it is performed in humans, sometimes also in dogs, and race horses.

Why Pedigree Analysis?

- Punnett square cannot be utilized to predict the variations and probabilities that can come from cross breeding in the case of human since its suitability is dependent on the population size..
- Humans have small families that means pedigree analysis is carried out in very small population.
- Human society have uncontrolled mating, often with heterozygotes.
- Need to determine the mode of inheritance: dominant, recessive, partial dominance, sex-linked, autosomal, mitochondrial, maternal effect.
- Need to determine the probability of an affected offspring for a given cross and to their subsequent generation.
- Helpful in following and diagnosing heritable traits.
- Useful in mapping (locating and isolating) genes responsible for certain traits.

Standard Symbols



Normal female

Normal male

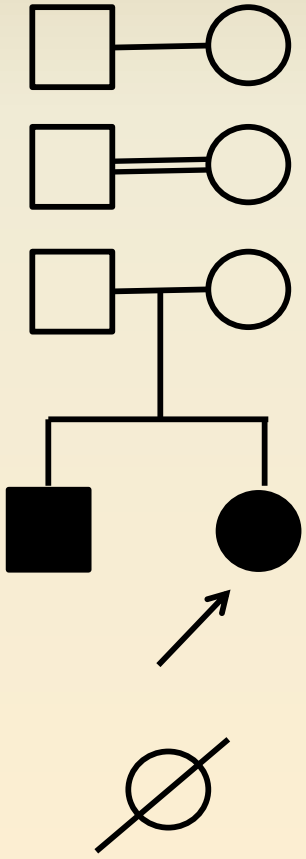
Female with trait

Male with trait

Sex unspecified

Carrier female

Carrier male



Mating

Consanguineous mating

Offspring

Arrow indicates that individual is proband)

Dead

Rules of Making Pedigree

- Use only standard set of symbols.
- Use only one generation per row; oldest at the top.
- Show siblings in order of their birth from left to right.
- Assign generation with a roman numerals, e.g. I, II, III, IV, etc.
- Assign individuals within a generation with an Arabic numerals, e.g. 1, 2, 3, 4, etc.
- Use arrow to indicate first affected individual as proband.

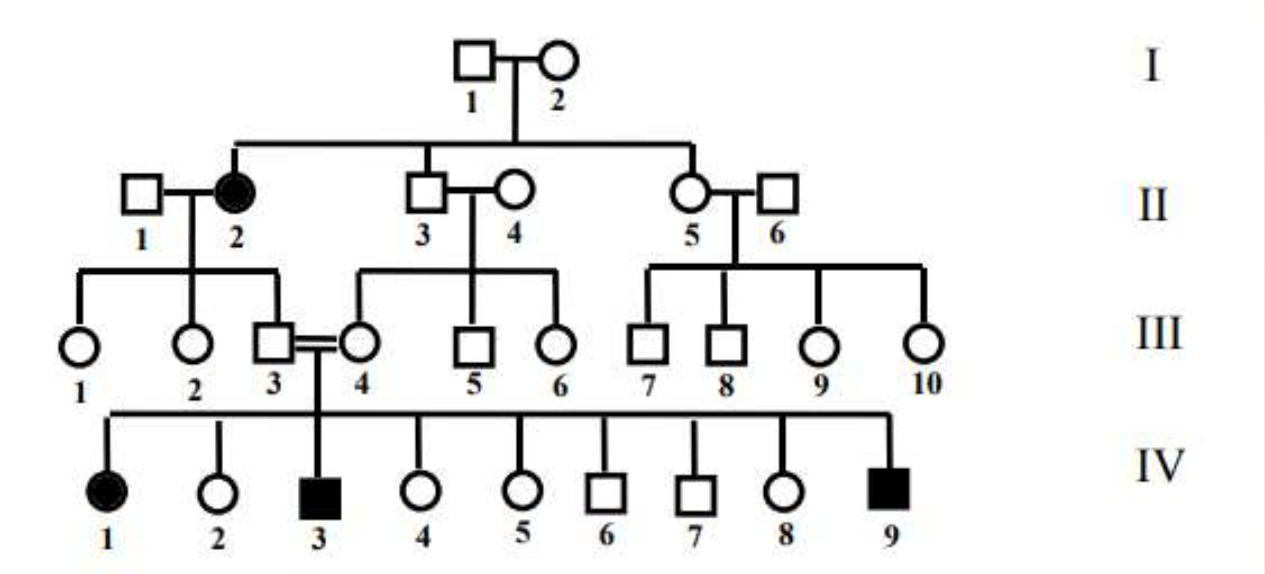
Outsider Rules

- The people whose parents are unknown, are known as “Outsiders”. For the outsider, some assumptions about their genotypes need to be made.
- Sometimes, the assumptions are proved wrong when the outsiders have children. Sometimes a given problem define the genotype of an outsider.

- *Outsider rule for dominant pedigree:* ***affected outsiders are assumed to be heterozygotes****.*
- *Outsider rule for recessive pedigree:* ***normal outsiders (unaffected) are assumed to be homozygotes.***

- Both of these rules are derived from the observation that mutant alleles are rare.

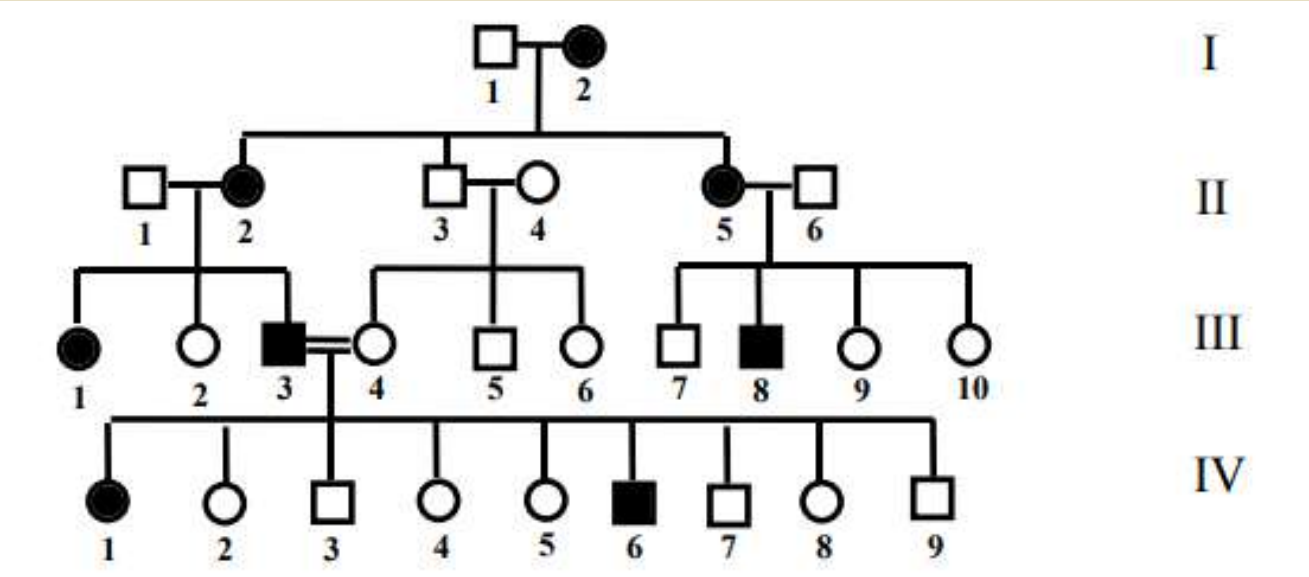
Autosomal Recessive



- All affected individuals are homozygotes.
- Unaffected outsiders are assumed to be homozygous normal.
- Both sexes are affected.
- Appearance of trait can skip generations.
- Example includes cystic fibrosis, albinism, etc.

Adapted from slideshare.com

Autosomal Dominant



- All unaffected individuals are homozygous for the normal recessive allele.
- Affected outsiders are assumed to be heterozygotes.
- Affected individuals might also be homozygotes.
- Both sexes are affected.
- Appearance of traits does not skip generations.
- Example includes myotonic dystrophy.

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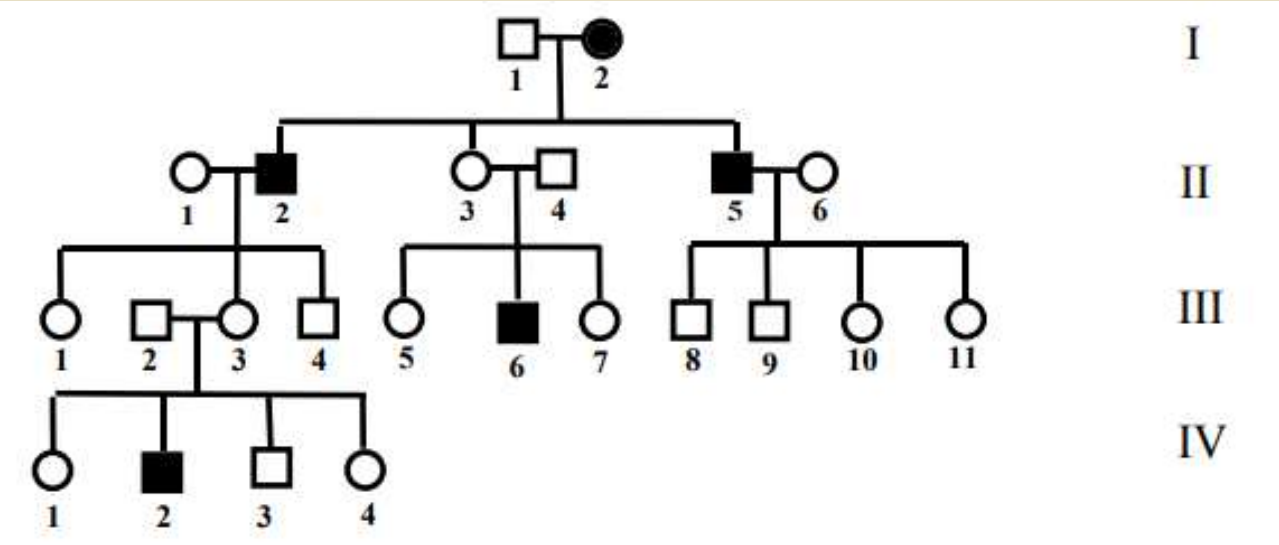
X-linked Recessive



- Males are more affected than females.
- Males never transmit to sons.
- Daughters of affected males always inherit recessive allele, thus daughters act as “carrier”.
- Traits can skip generations.
- Example include Color blindness, hemophilia, etc.

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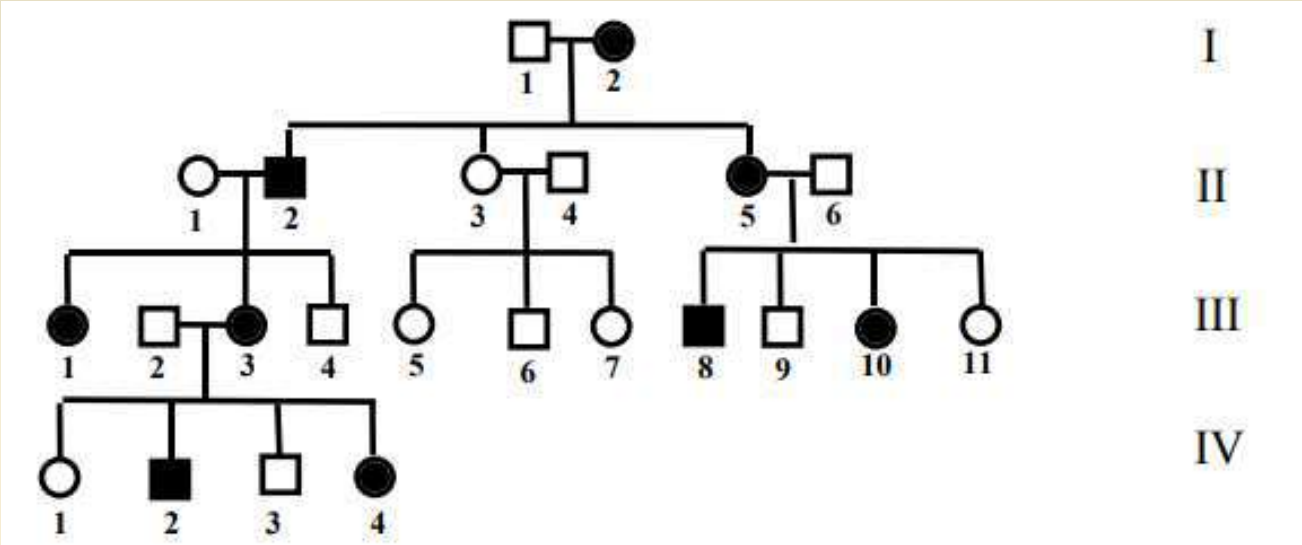
X-linked Dominant



- Both sexes are affected.
- Females transmit to daughters and sons.
- Males always transmit to daughters, but never to sons.
- Traits do not skin generations.
- Example includes Rett syndrome (mental retardation, neural degeneration).

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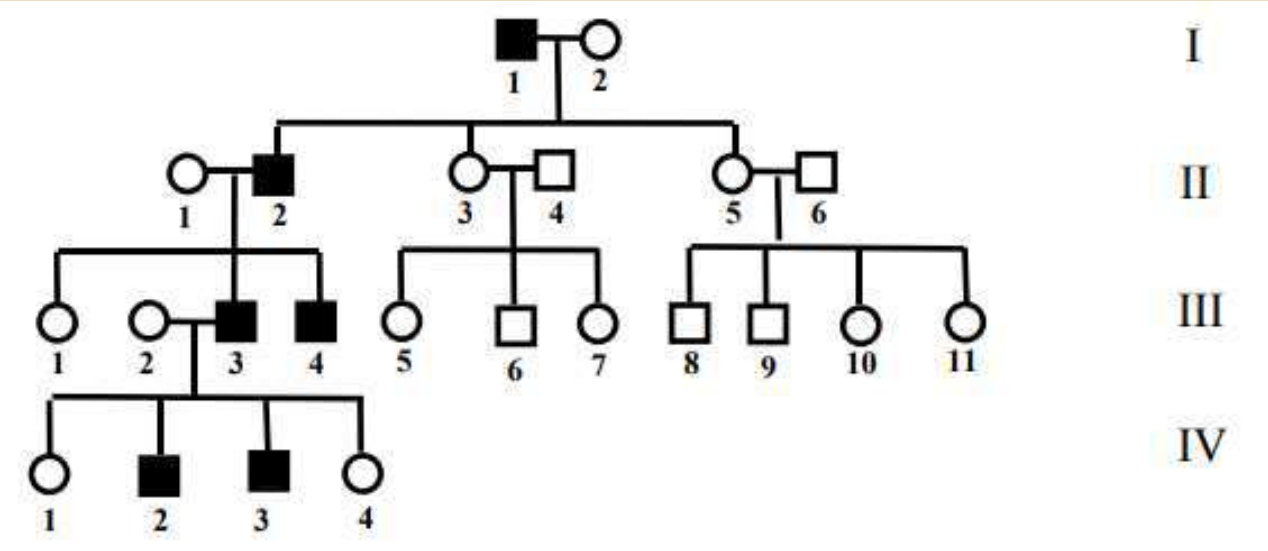
Y-linked



- Only males are affected.
- Males always transmit to sons.
- Example includes hypertrichosis

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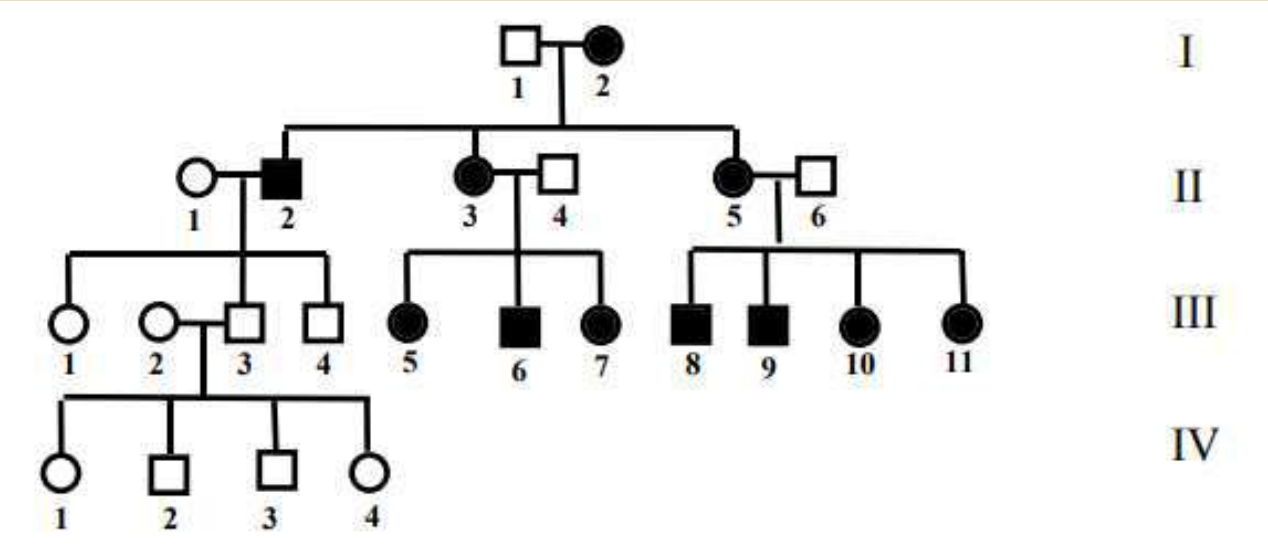
Mitochondrial Inheritance



- Both sexes are affected.
- Females transmit to all of their offspring.
- Males never transmit to any of their offspring.

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Identifying Dominant or Recessive Pedigree

- If two affected individuals have an unaffected child, it must be a dominant pedigree. In this condition, if **‘D’** is the dominant mutant allele and **‘d’** is the recessive normal allele, both parents are **‘Dd’** and the normal child is **‘dd’**.
- If two unaffected individuals have an affected child, it is a recessive pedigree. In this condition, if **‘R’** is the dominant normal allele and **‘r’** is the recessive mutant allele, both parents are **‘Rr’** and the affected child is **‘rr’**.
- If every affected person has an affected parent it is a dominant pedigree.

Gender Effects on Phenotype

- **Sex-limited inheritance**
  - Only one gender is capable of showing trait regardless of the genotype.
  - The genes involved are typically autosomal, but the expression of these genes is dependent on the gender of the individual.
  - Examples are beard growth, breast size, etc.
- **Sex-influenced inheritance**
  - The gender of the individual determines whether a particular phenotype assumes dominant or recessive state. A phenotype that is dominant in one gender is recessive in other gender.
  - Example is pattern baldness.

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Problems in Constructing a Pedigree

- poor medical records
- scattering of family members
- inaccurate and anecdotal information
- miscarriages and stillbirths
- infidelity / concealed adoptions
- variable expressivity of genotype (as phenotype)
- incomplete penetrance of genotype (showing phenotype)

Adapted from slideshare.com

There is no substitute for hard work.