Pedigree Analysis

Pedigree:

Pictorial representation of a family history, a family tree that outlines the inheritance of one or more characteristics.

Proband:

The first affected person from whom pedigree analysis starts.

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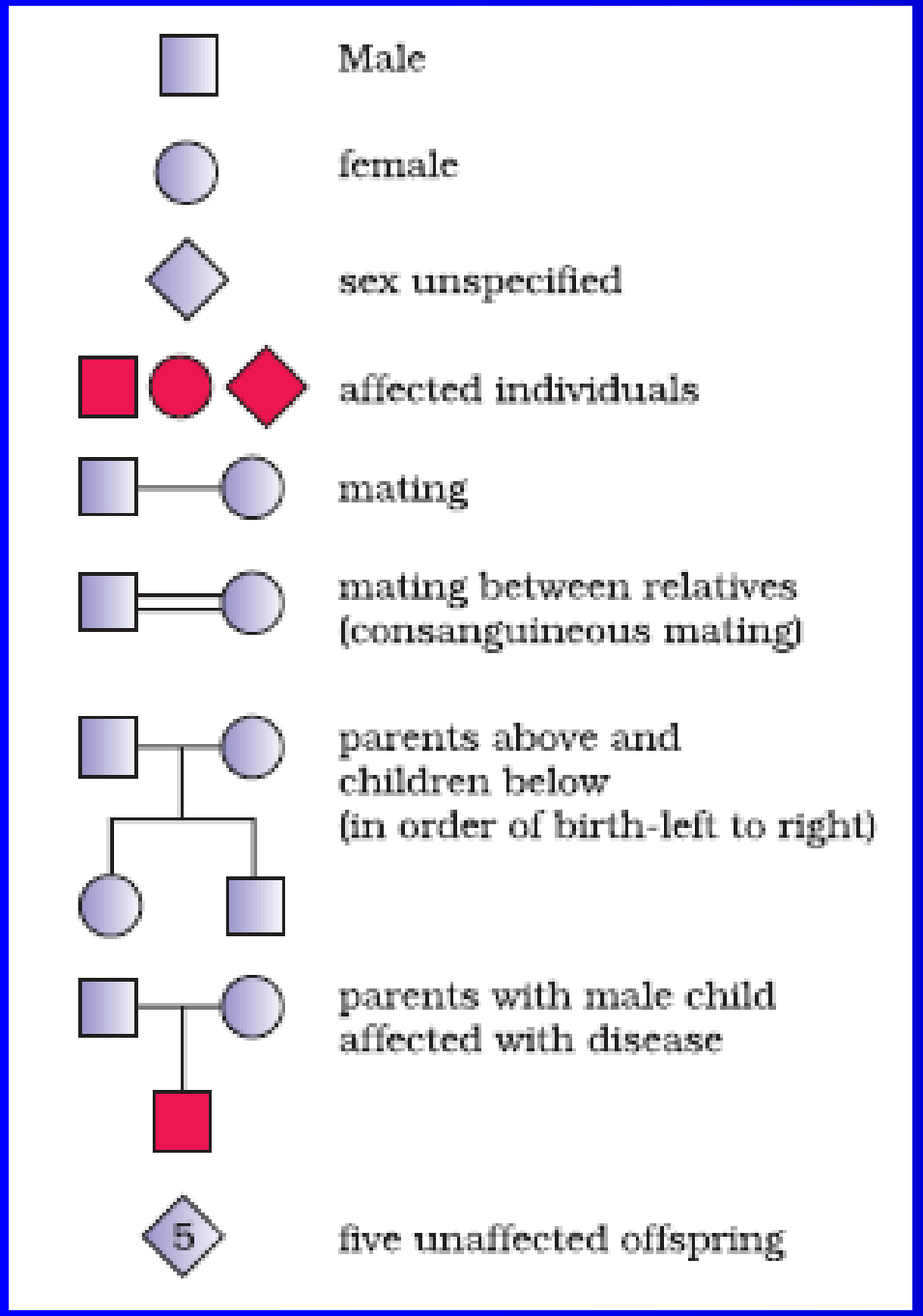

Symbols used in the human pedigree analysis

Rules of Pedigree

Rules of Pedigree

	Autosomal Dominant	Autosomal Recessive	X-Linked Dominant	X-Linked Recessive
1	Males and females are equally affected **(Autosomal)**	Males and females are equally affected **(Autosomal)**	Males and females are unequally affected **(X-Linked)** **Females** are more affected than males	Males and females are unequally affected **(X-Linked)** **Males** are more affected than females
2	Trait never skips generations **(Dominant)**	Trait skips generations **(Recessive)**	Trait never skips generations **(Dominant)**	Trait skips generations **(Recessive)**
3	Both the affected parents can have unaffected children	Unaffected parents (carriers) can have affected children	Affected father will have affected daughters	Unaffected mother (carrier) will have affected sons
4	It is caused by a dominant gene present in an autosome	It is caused by a recessive gene present in an autosome	It is caused by a dominant gene present in an allosome	It is caused by a recessive gene present in an allosome
	Both the parents affected ⬇ Children unaffected	Both the parents unaffected ⬇ Children affected	Affected Father ⬇ Affected Daughters	Unaffected Mother ⬇ Affected Sons
	No carriers	Carriers occur at heterozygous condition	No carriers	Females are carriers at heterozygous condition
	Huntington's disease Familial Hypercholesterolemia	Sickle cell anemia, Cystic fibrosis, Phenylketonuria (PKU)	Hypophatemic Rickets (Vitamin D resistant rickets)	Hemophilia, Colour blindness.

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Some Examples of Mendelian Disorders

Mendelian Disorders

Autosomal Dominant	Autosomal Recessive	X-Linked Dominant	X-Linked Recessive
Myotonic dystrophia Huntington's disease	Sickle Cell Anemia Thalassemia Cystic Fibrosis Phenylketonuria (PKU)	Hypophatemic Rickets (Vitamin D Resistant Rickets)	Haemophilia Colour Blindness

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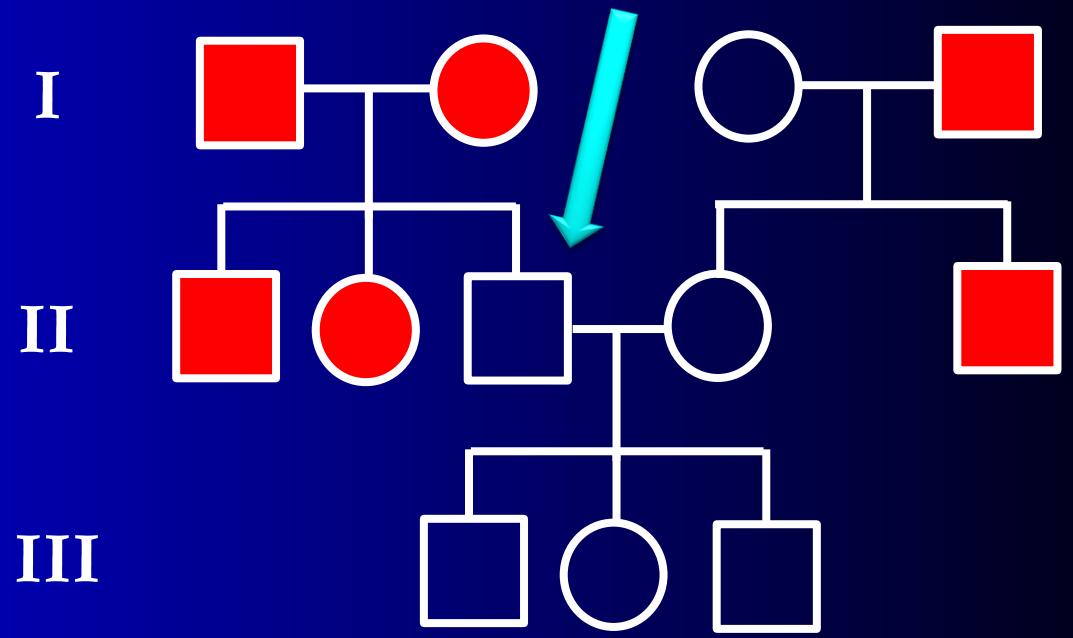
Autosomal Dominant Inheritance

Autosomal Dominant Trait

- Males and females are equally affected.
- It does not skip the generation.
- Affected parents can have unaffected children. (both affected father and mother)



Examples: Huntington's disease, Familial Hypercholesterolemia, Brachydactyly, polydactyly, dimple in the cheek

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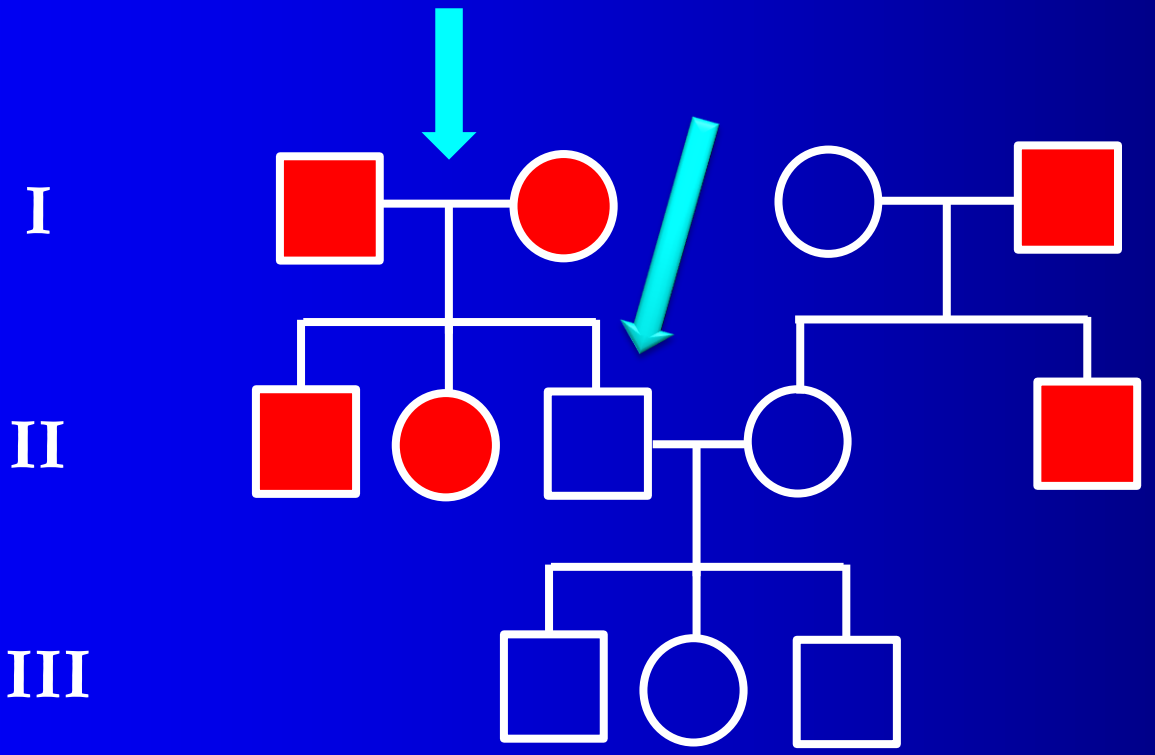
Autosomal Dominant Inheritance

Both the parents are affected but one of the sons is unaffected



Examples: Huntington's disease, Familial Hypercholesterolemia, Brachydactyly, polydactyly, dimple in the cheek

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Autosomal Dominant Inheritance

The gene is present in any one of the autosomes and the normal allele is recessive (a) to its mutant allele (A).

Examples: Huntington's disease, Familial Hypercholesterolemia, Brachydactyly, polydactyly, dimple in the cheek.

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Autosomal Dominant Trait

Autosomal dominant inheritance is due to mutation in Gene ‘a’

Genotype of affected individual: Aa or AA.

Genotype of unaffected or normal individual: aa

Affected offspring has at least one affected parent

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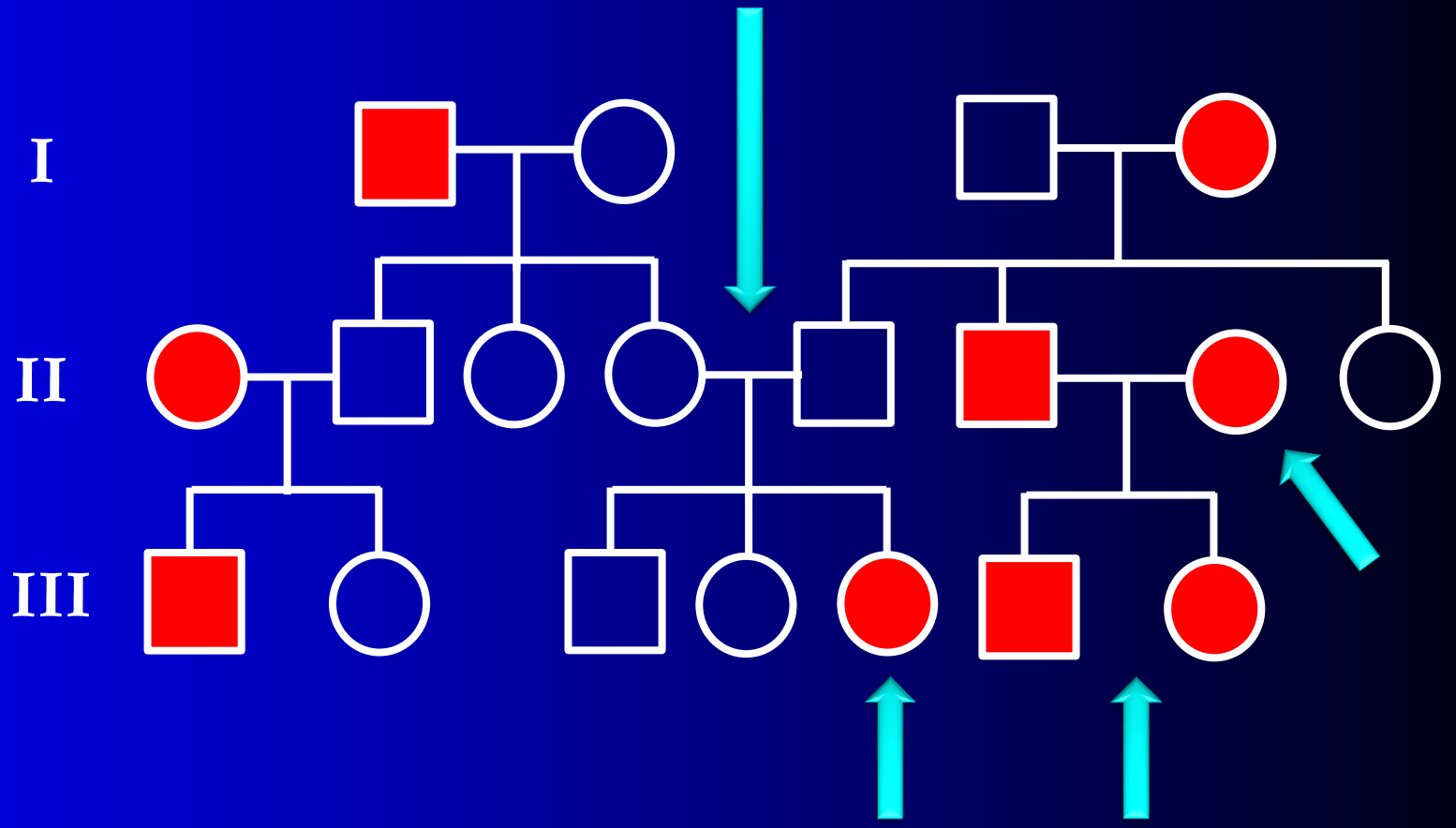
Autosomal Recessive Inheritance

Autosomal Recessive Inheritance

- Males and females are equally affected.
- Trait skips the generations.
- Both the parents are unaffected but the child is affected.
- If both the parents were affected, all the children would be affected.
- Traits are often found in pedigrees with consanguineous marriages.



Examples: Sickle Cell Anemia, Cystic Fibrosis, Phenylketonuria (PKU)

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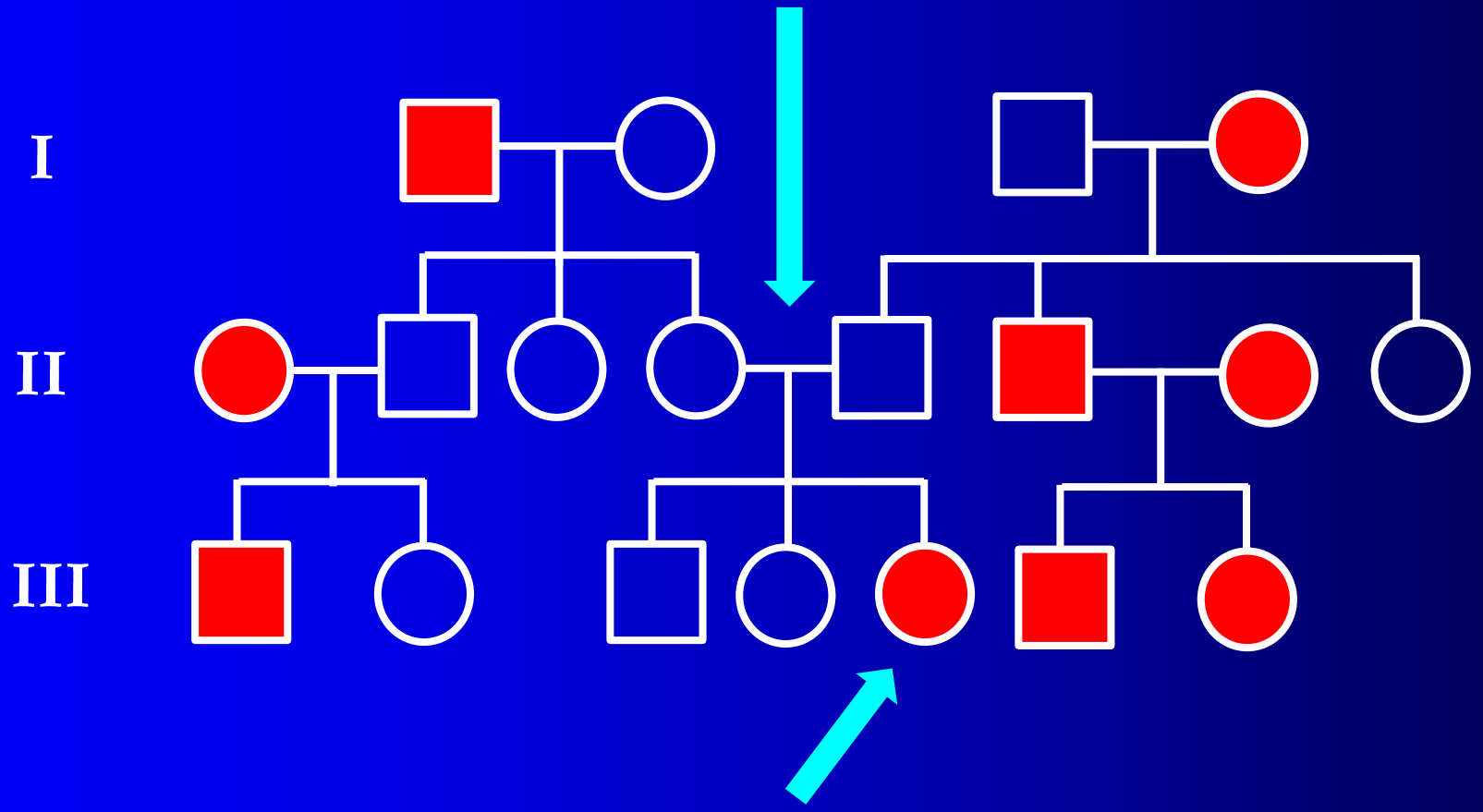
Autosomal Recessive Inheritance

Both the parents are unaffected but the child is affected



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Autosomal Recessive Trait

The mutant allele is recessive (a) to its wild type allele (A).

Genotype of affected individual: aa.

Genotype of unaffected or normal individual: AA or Aa

Affected offspring gets the disease causing allele from both parents

i.e. parents are at least heterozygous for the disease.

Examples: Sickle cell anemia, cystic fibrosis, phenylketonuria (PKU)

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Autosomal Recessive Inheritance

When a normal person marries an affected individual, if all the children were normal, it indicates that the **normal parent is homozygously dominant.**

When a normal person marries an affected individual, if one or more of the children were affected, it indicates that the **normal parent is heterozygously dominant.**

Approximately half of the children are affected.

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Sex Linked Dominant Inheritance

Sex Linked Dominant Inheritance

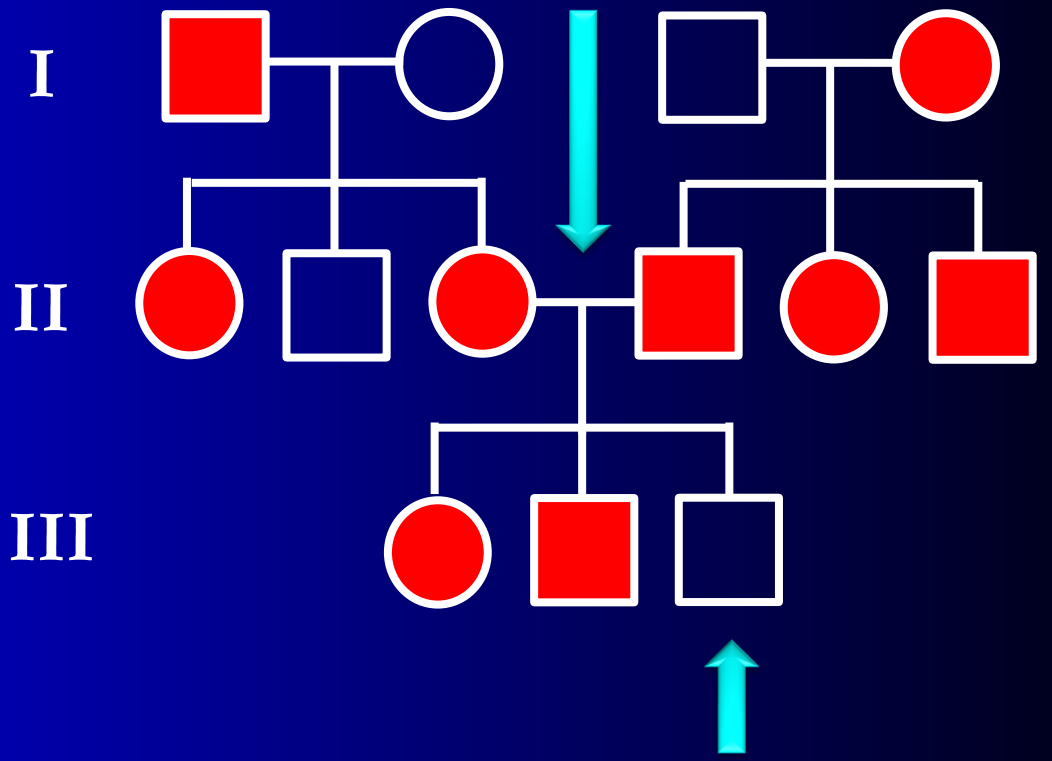
Females are more affected.

Trait never skips generations

Affected parents can have unaffected children.

All daughters of an affected father will be affected.

Disease transfers from father to daughter and from mother to both sons and daughters.



Example: Hypophatemic Rickets (Vitamin D Resistant Rickets)

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Sex Linked Dominant Inheritance

For a male child to be affected the mother must be affected.

For a female child to be affected, either father or mother must be affected. (Any one of the parents must be affected)

Approximately half of the sons of an affected mother are affected, showing mother is heterozygous.

Example: Hypophatemic Rickets (Vitamin D Resistant Rickets)

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Sex Linked Dominant Trait

Genotype of affected males: X^AY.

Genotype of affected females: X^AX^A or X^AX^a

Genotype of normal male: X^a Y.

Genotype of normal female: X^a X^a

Affected father transmits trait to all the daughters but not to sons.

Affected mother (if heterozygous) passes the trait to 50% of her daughters and 50% of her sons.

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Sex Linked Recessive Inheritance

Sex Linked Recessive Inheritance

Male are more affected than females.

Trait skips generations

Disease transfers from

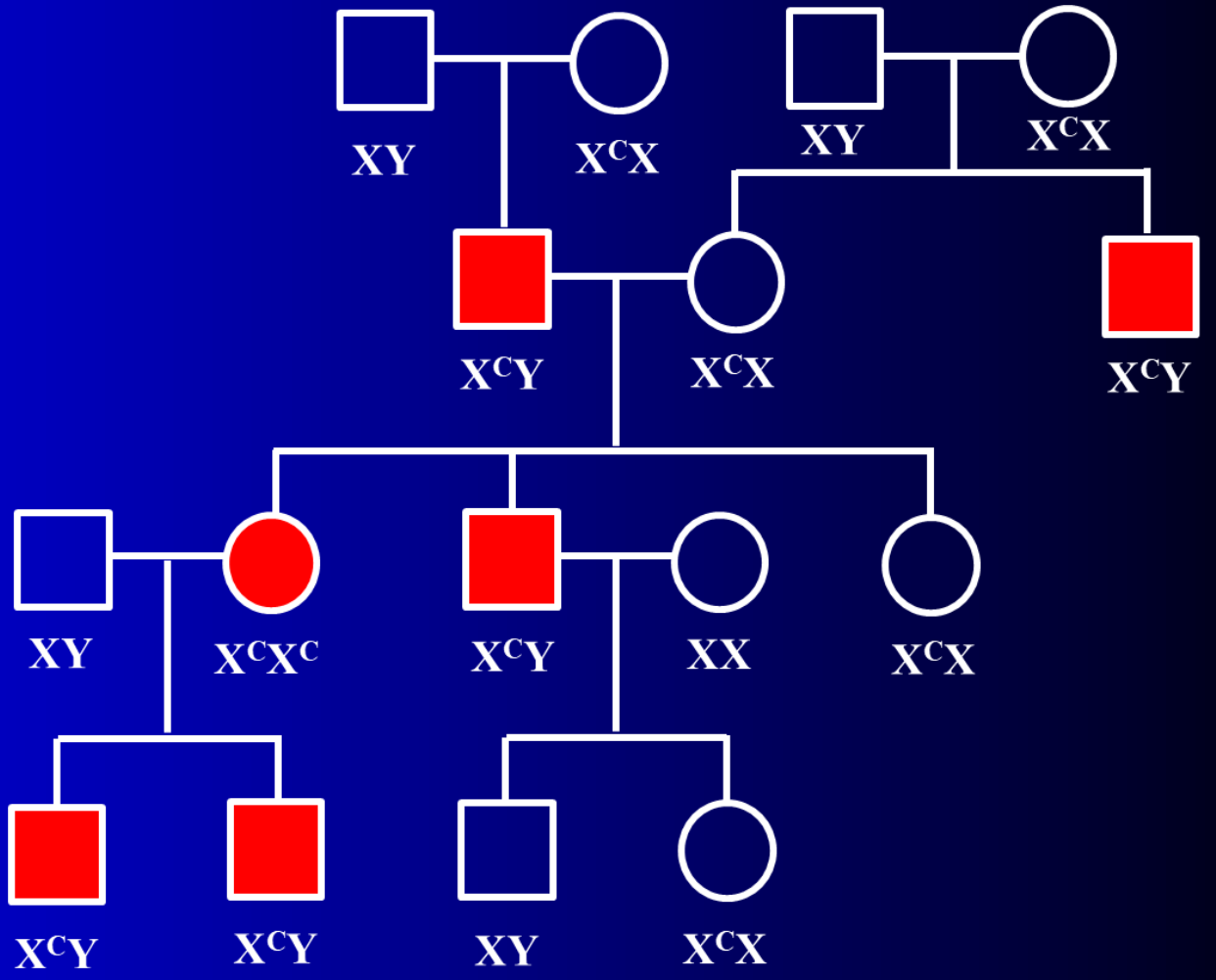
Mother to son and

Father to daughter. (crisscross inheritance)

All the sons of an affected mother will be affected. (Mother to sons)

Disease never transfers from father to son.

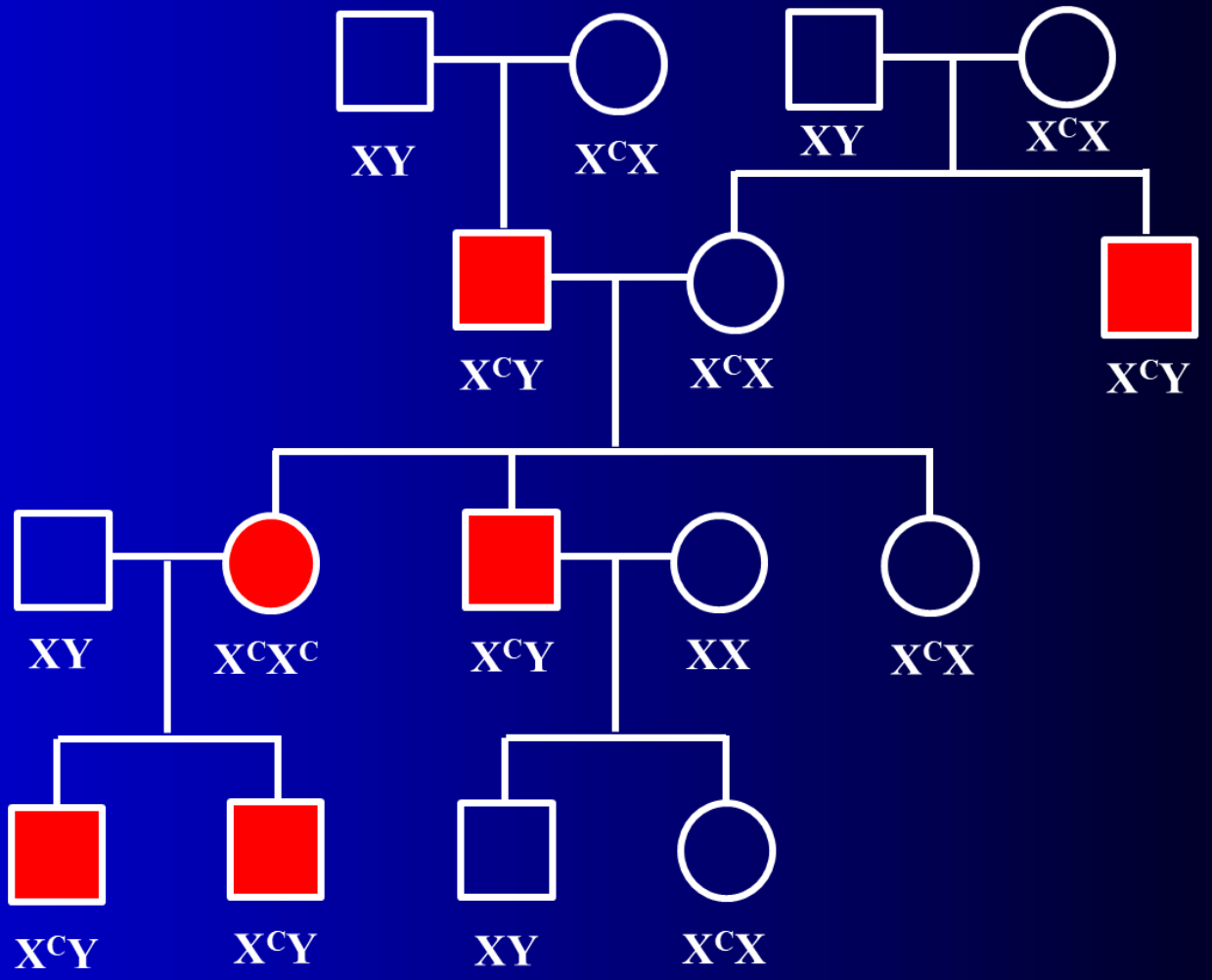
Examples: Hemophilia, Colour Blindness



Unaffected mother (carrier) will have affected sons

Sex Linked Dominant Trait

- Unaffected parents can have affected children.
- Affected father does not pass to his sons but passes to all daughters.
- Affected mother passes the trait to all her sons.



Examples: Hemophilia, Colour Blindness

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Sex Linked Recessive Trait

Genotype of affected males: $X^a Y$

Genotype of affected females: $X^a X^a$

Genotype of normal male: $X^A Y$

Genotype of normal female: $X^A X^a$ or $X^A X^A$

Affected mother transmits the trait to all the sons.

Carrier mother transmits the trait to 50% of her sons.

Examples: Hemophilia, Colour Blindness.

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Sex Linked Recessive Inheritance

For a **male child** to be affected the **mother must be affected** or a carrier.

Usually affected sons are born to unaffected mother. (Carrier)

All the sons of an affected mother must be affected.

For a female child to be affected, the **father must be affected** and the **mother may be either affected or a carrier.**

Approximately half of the sons of **carrier mother are affected.**

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God Bless You!