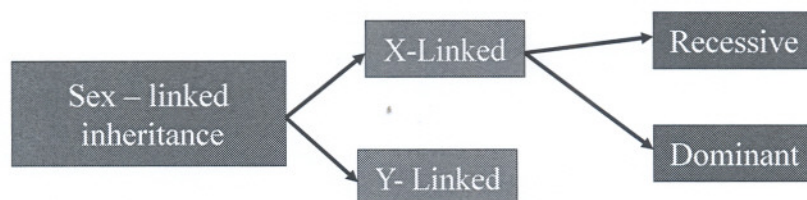


## Sex-Linked Inheritance



## Sex-Linked Inheritance



## Sex – Linked Inheritance

- This is the inheritance of a gene present on the sex chromosomes.
- The Inheritance Pattern is different from the autosomal inheritance.
- Inheritance is different in the males and females.



## Y – Linked Inheritance

- The gene is on the Y chromosomes
- The gene is passed from fathers to sons only
- Daughters are not affected
- Hairy ears in India
- Male are Hemizygous, the condition exhibits itself whether dominant or recessive

|        |   | Father |     |
|--------|---|--------|-----|
|        |   | X      | Y*  |
| Mother | X | XX     | XY* |
|        | X | XX     | XY* |



## X – Linked Inheritance

>1400 genes are located on X chromosome  
(~40% of them are thought to be  
associated with disease phenotypes)



### X-linked inheritance in male & female

|         | Genotype  | Phenotype             |
|---------|-----------|-----------------------|
| Males   | $X_H$     | Unaffected            |
|         | $X_h$     | Affected              |
| Females | $X_H/X_H$ | Homozygous unaffected |
|         | $X_H/X_h$ | Heterozygous          |
|         | $X_h/X_h$ | Homozygous affected   |

$X_H$  is the normal allele,  $X_h$  is the mutant allele





## **X – Linked Inheritance**

- The gene is present on the X chromosome
- The inheritance follows specific pattern
- Males have one X chromosome, and are hemizygous
- Females have 2 X chromosomes, they may be homozygous or heterozygous
- These disorders may be : recessive or dominant



## **X – Linked Recessive Inheritance**

- The incidence of the X-linked disease is higher in male than in female
- The trait is passed from an affected man through all his daughters to half their sons
- The trait is never transmitted directly from father to sons
- An affected women has affected sons and carrier daughters



# X – Linked Recessive Inheritance

## (1) Normal female, affected male

|       |    | Ova |     |
|-------|----|-----|-----|
|       |    | X   | X   |
| Sperm | X* | X*X | X*X |
|       | Y  | XY  | XY  |

All sons are normal  
All daughters carriers "not affected"



## (2) Carrier female, normal male:

|       |   | Ova |    |
|-------|---|-----|----|
|       |   | X*  | X  |
| Sperm | X | XX* | XX |
|       | Y | X*Y | XY |

50% sons affected  
50% daughters carriers

## (3) Homozygous female, normal male:

- All daughters carriers.
- All sons affected.



## **X - Linked Recessive Disorders**

- Albinism (Ocular)
- Fragile X syndrome
- Hemophilia A and B
- Lesch–Nyhan syndrome
- Mucopolysaccharidosis 11 (Hunter's syndrome)
- Muscular dystrophy (Duchenne and Becker's)
- G-6-PD deficiency
- Retinitis pigmentosa



## **G6PD Deficiency Prevalence & Heterozygote advantage:**

- **Prevalence:**
  - 5-25% in areas endemic to malaria
  - <0.5% in areas nonendemic to malaria
- **Heterozygotes for G6PD deficiency have some resistance to malaria → survival advantage**





## **G6PD Deficiency Inheritance Risk:**

- Each son of a mother carrying a G6PD mutation has a 50% chance of being affected
- Each daughter of a mother carrying a G6PD mutation has a 50% chance of being a carrier
- Each daughter of an affected father will be a carrier
- Each son of an affected father will be unaffected



## **X-Linked Dominant Disorders**

- The gene is on X Chromosome and is dominant
- The trait occurs at the same frequency in both males and females
- Hemizygous male and heterozygous females express the disease.



## \*\* Punnett square showing X - linked dominant type of Inheritance

(1) Affected male and normal female:

|       |    | OVA |     |   |
|-------|----|-----|-----|---|
|       |    | X   | X   |   |
| Sperm | X* | X*X | X*X |   |
|       | Y  | XY  | XY  | All daughters affected, all sons normal |

(2) Affected female (heterozygous) and normal male:

|       |   | OVA |    |                                       |
|-------|---|-----|----|---------------------------------------|
|       |   | X*  | X  |                                       |
| Sperm | X | XX* | XX |                                       |
|       | Y | X*Y | XY | 50% sons & 50% daughters are affected |



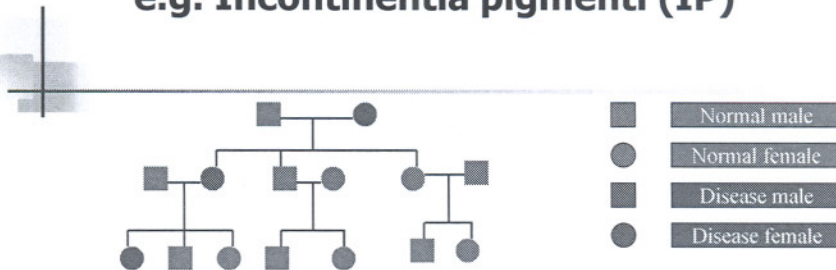
(3) Affected female (homozygous) and normal male:

|       |   | OVA |     |                       |
|-------|---|-----|-----|-----------------------|
|       |   | X*  | X*  |                       |
| Sperm | X | X*X | XX* |                       |
|       | Y | X*Y | X*Y | All children affected |





## X-linked dominant disorder e.g. Incontinentia pigmenti (IP)



### Lethal in males during the prenatal period



- Lethal in hemizygous males before birth:
  - Exclusive in females
  - Affected female produces
    - affected daughters
    - normal daughters
    - normal sons
- in equal proportions (1:1:1)

National Institute of Neurological Disorders and Stroke:  
[http://www.ninds.nih.gov/disorders/incontinentia\\_pigmenti/incontinentia\\_pigmenti.htm](http://www.ninds.nih.gov/disorders/incontinentia_pigmenti/incontinentia_pigmenti.htm)

