

# INHERITED GENETIC DISORDERS

SBI 3C

# CATEGORIES:

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- ▶ Genetic disorders can be caused by chromosomes or genes
- ▶ Inherited disorders is caused by a gene passed from parent to child
- ▶ Inherited disorders can be:
  - ▶ Dominant
  - ▶ Recessive
  - ▶ X-linked



# DOMINANT DISORDERS:

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- ▶ If one parent has the dominant gene disorder, there is a 50% chance that it will be passed to each child
- ▶ Examples:
  - ▶ Huntington's disease:
    - ▶ An inherited nerve disorder that causes loss of control of movements and mental function, usually starts around 35-50 years old
  - ▶ Polydactyly:
    - ▶ Extra fingers or toes



# RECESSIVE DISORDERS:

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- ▶ Both parents must carry the gene for the disorder
- ▶ If you have a recessive gene for a disorder, you are a **CARRIER**
  - ▶ You may show no symptoms but you can still pass it on to your children
- ▶ If both parents carry the recessive gene:
  - ▶ 25% chance that a child they have will have disorder
  - ▶ 50% chance that their child will be a carrier
  - ▶ 25% chance that a child will not get the gene at all



# EXAMPLES OF RECESSIVE DISORDERS:

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## ▶ Sickle Cell Disease

- ▶ Red blood cells have crescent shape
- ▶ Causes anemia and pain, most often in African Americans

## ▶ Tay-Sachs Disease:

- ▶ Causes mental retardation, blindness, seizures, and death usually by age 5
- ▶ Most often seen in people of eastern European Jewish descent, French Canadians, and Cajuns

## ▶ Cystic Fibrosis:

- ▶ Causes problems in digestion and breathing
- ▶ Occurs mostly in people of Northern European descent



# X-LINKED / SEX LINKED DISORDERS:

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- ▶ Disorders caused by genes on the X chromosome
- ▶ Usually abnormal gene is recessive
- ▶ Generally seen in males:
  - ▶ Women have 2 copies of X, therefore, one normal, dominant X chromosome
  - ▶ Men have one X chromosome, therefore, no dominant gene to cancel out recessive gene
- ▶ If a daughter has an X-linked disorder, her mother was a carrier, her father had the disorder
- ▶ If you are a carrier, 1 in 2 chance a son will have the disorder and a daughter will be a carrier



# X-LINKED / SEX LINKED DISORDERS:

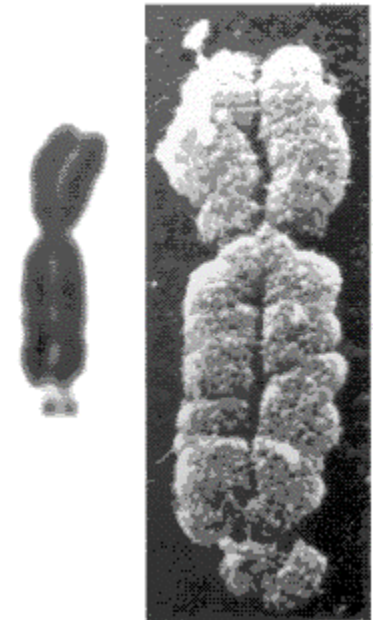
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- ▶ **Duchenne muscular dystrophy:**

- ▶ Most often affecting males
- ▶ Weakness, muscle wasting
- ▶ Death by around 30

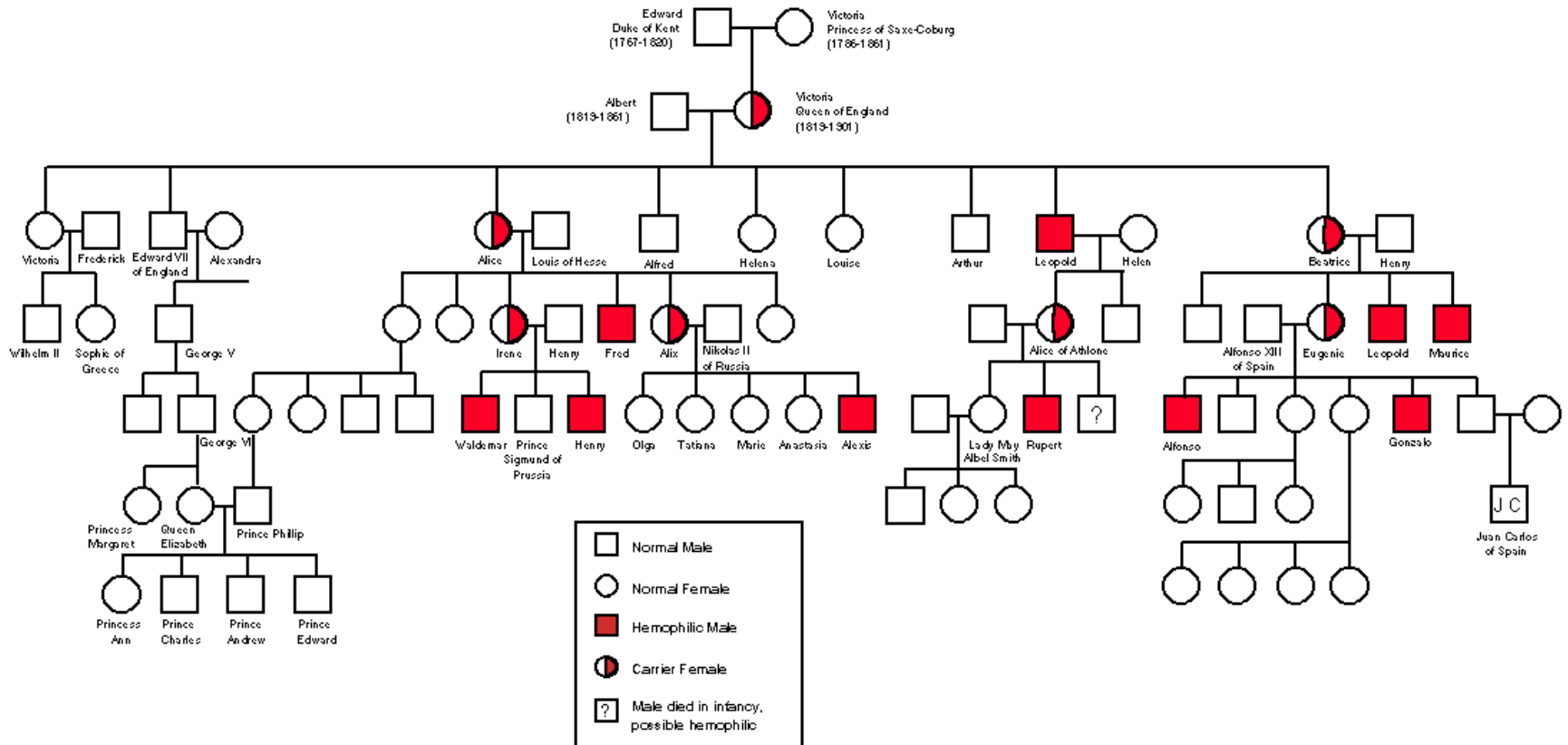
- ▶ **Fragile X Syndrome:**

- ▶ Most common cause of mental retardation
- ▶ Piece of X chromosome is not fully attached



# HEMOPHILIA:

- ▶ Blood lacks the substance needed to clot
- ▶ Known as the “Royal Disease” because it appeared in the descendants of Great Britain’s Queen Victoria





# PEDIGREES:

- ▶ A type of chart that shows patterns of transmission within a family
- ▶ Generally constructed after an undesirable genetic trait has appeared in a family, helps to determine if:
  - ▶ Gene is X-linked
  - ▶ Trait is dominant or recessive
  - ▶ Chance of transmission from parents to children

