University of Basrah
Al-Zahraa Medical College

Ministry of Higher education
and Scientific Research

Block: child health

Lecture: pattern of inheritance

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Nelson Textbook of Pediatrics, 20th edition.
Nelson Essentials of Pediatrics, 7th Edition 2015
Pediatric Decision-Making Strategies
Illustrated Textbook of Pediatrics
Short Atlas in Pediatrics

Patterns of Inheritance

Learning outcomes

LO1: To understand different modes of inheritance and genetic disorders
LO2: To interpret a genetic pedigree
LO3: Autosomal Dominant
LO4: Autosomal Recessive
LO5: X- linked Recessive
LO6: X- linked Dominant
LO7: Y- linked
LO8: Mitochondrial Inheritance
LO9: Multifactorial Inheritance

Hotspot: Inheritance patterns trace the transmission of genetically encoded traits, conditions or diseases to offspring.

THE PATTERNS OF GENETIC INHERITANCE	LO1
Mendelian	Non- Mendelian
Autosomal Dominant	**Mitochondrial**
Autosomal Recessive	**Multifactorial**
X-linked Recessive	
X-linked Dominant	
Y-linked	

How to evaluate a pedigree LO2

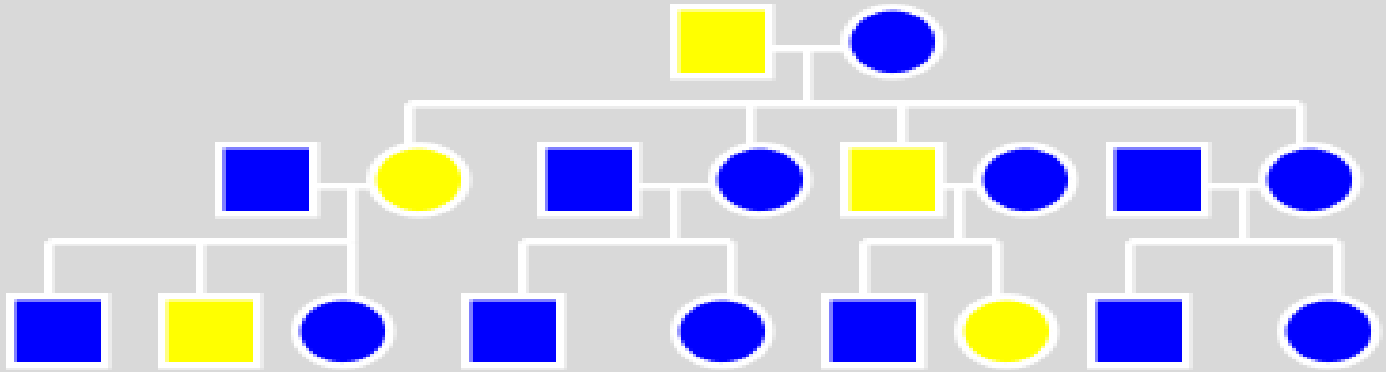
Transmission: Are there affected family members in every generation (vertical pattern) or in only a single generation (horizontal pattern)?

Sex Differences: What is the ratio of affected males to females?

Segregation: Is disease/gene being passed through unaffected females? Is there male to male transmission? What % of children are affected (e.g all of sons but none of daughters)?

Autosomal Dominant

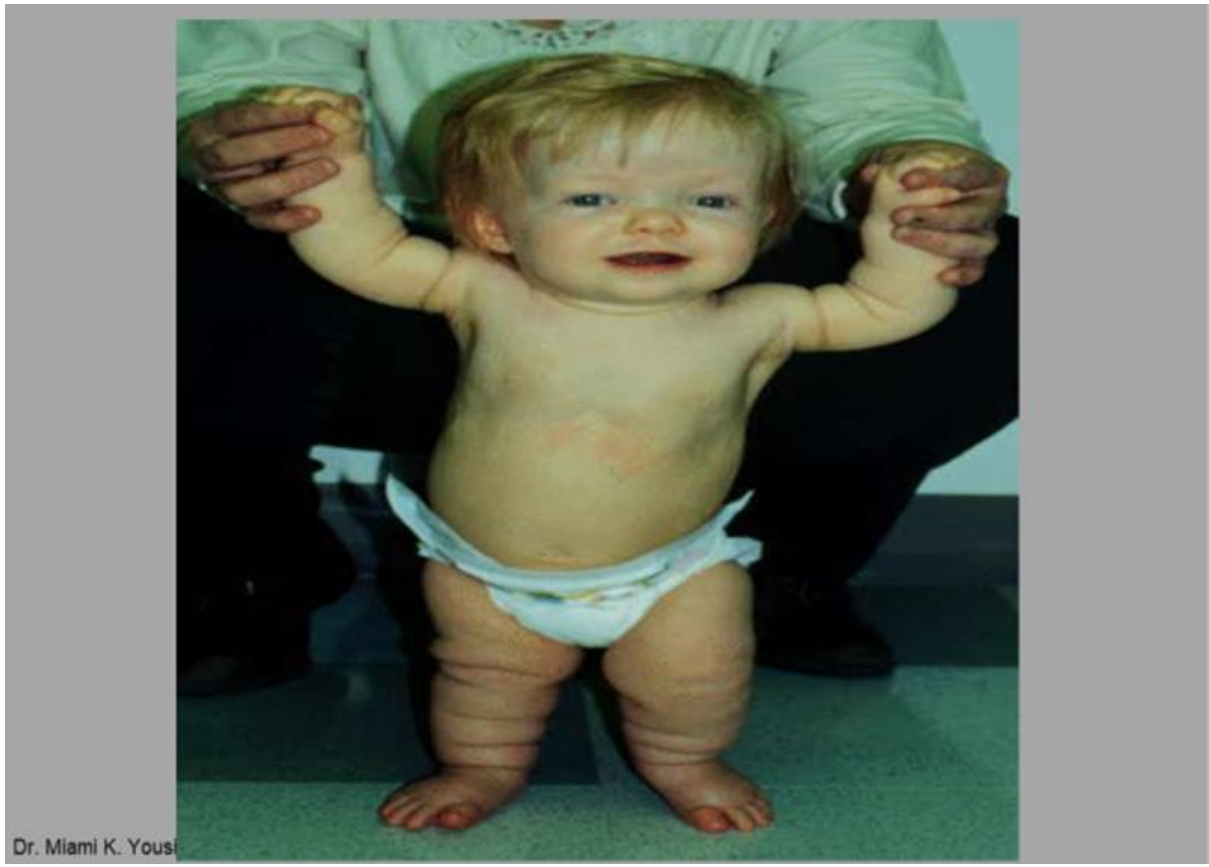
LO3



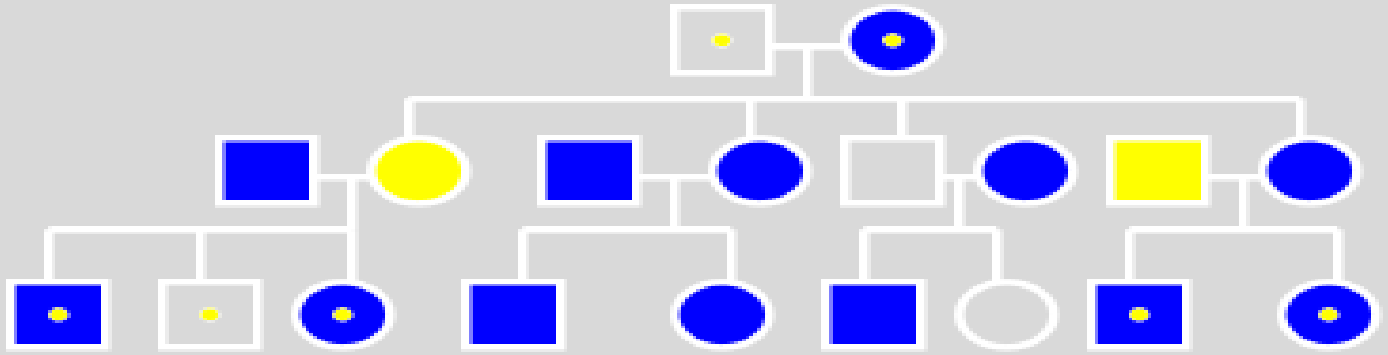
- Vertical pattern: multiple generations affected
- Males and females equally likely to be affected
- See male to male transmission
- Each child of an affected individual has a 50% chance to be affected
- Every affected child has an affected parent

Mother's Gametes \ Father's Gametes	D	d
d	Dd	dd
d	Dd	dd

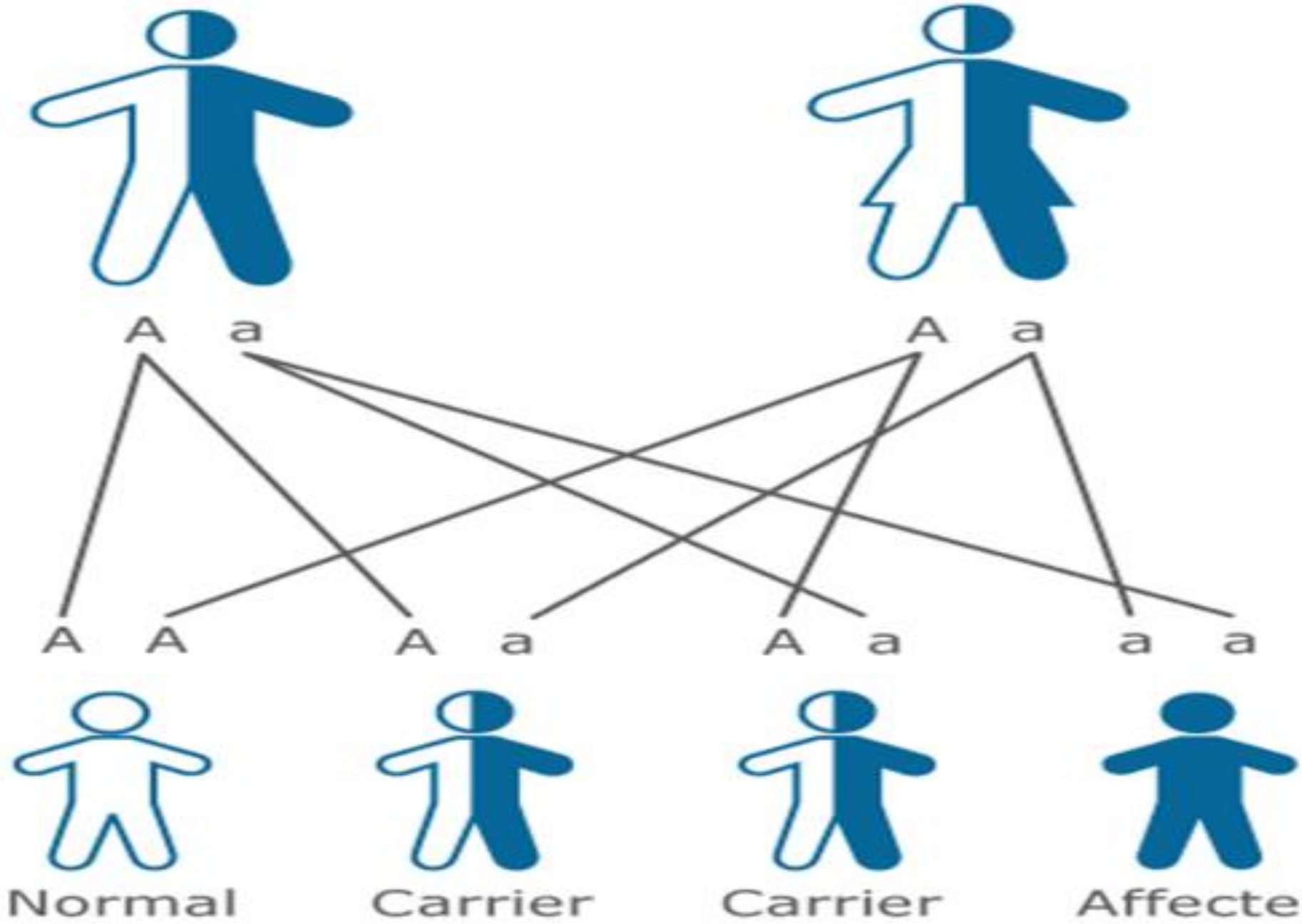
Achondroplasia



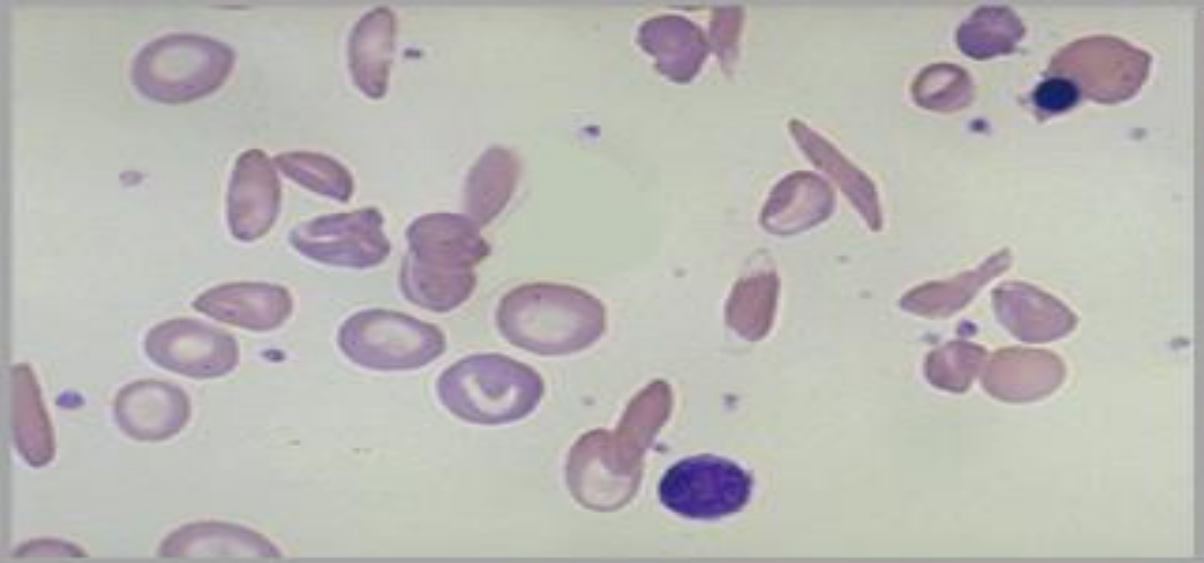
Autosomal Recessive LO4



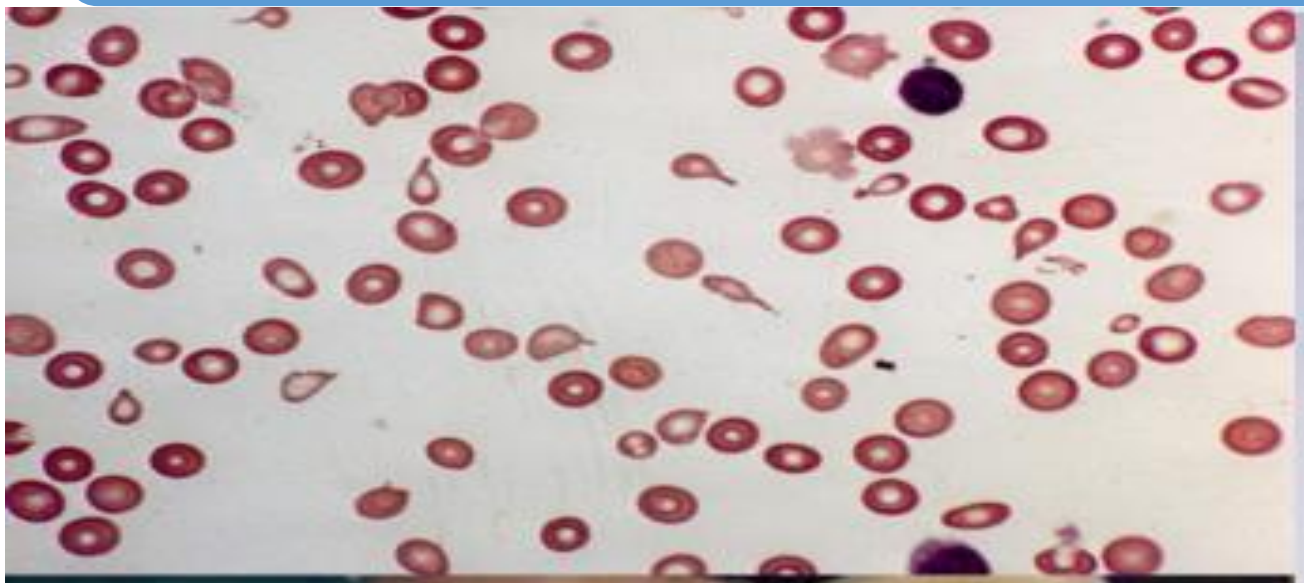
- Horizontal pattern: single generation affected.
- Males and females equally likely to be affected
- Parents of affected child are unaffected gene carriers and have a 1 in 4 or 25% recurrence risk
- 50% chance to be carrier, 25% normal
- Children of affected individuals are obligate carriers.

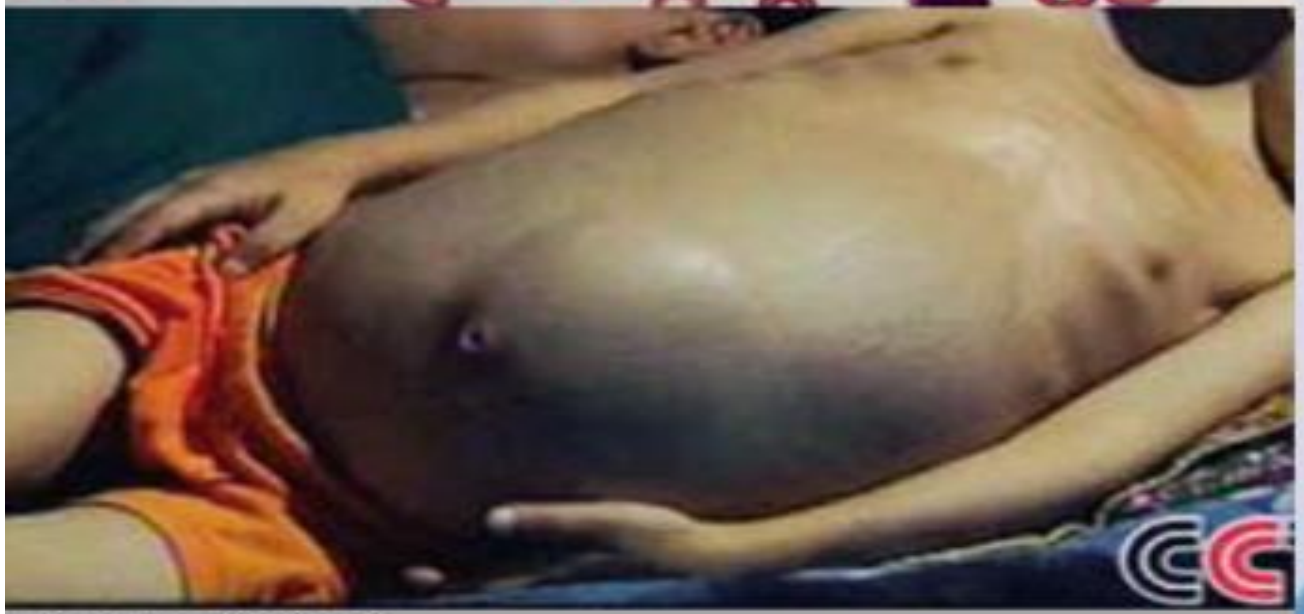

A a
A a
A A A a A a a a
Normal
Carrier
Carrier
Affected

Sickle Cell Anemia



Thalassemia





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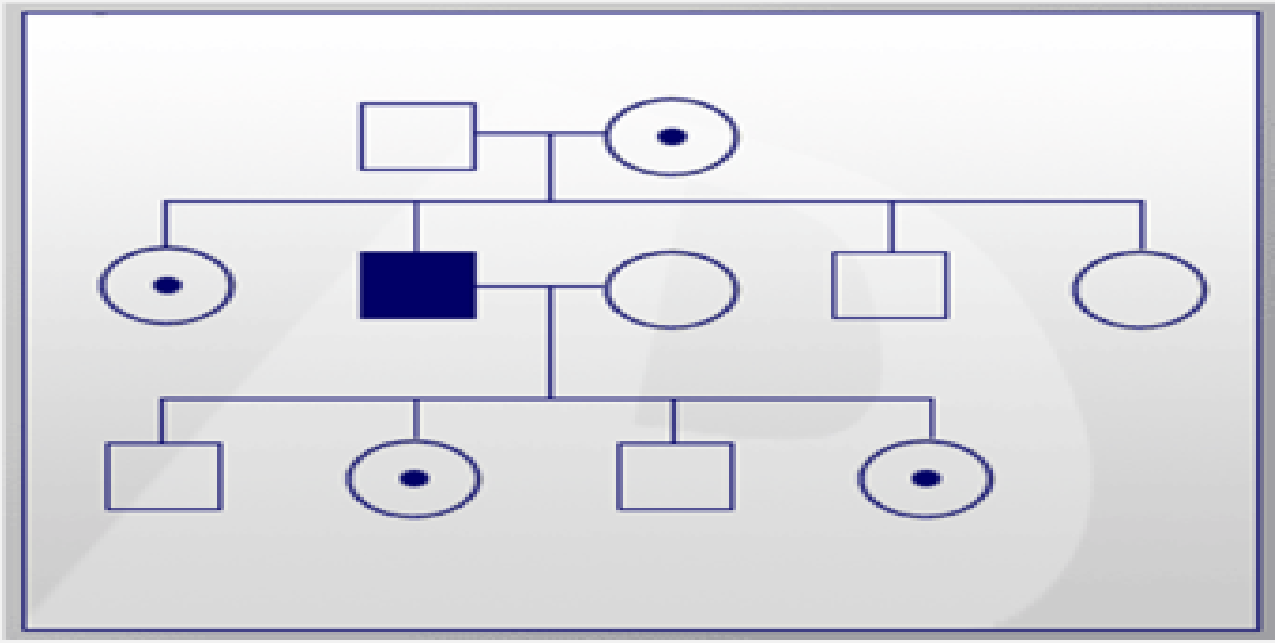
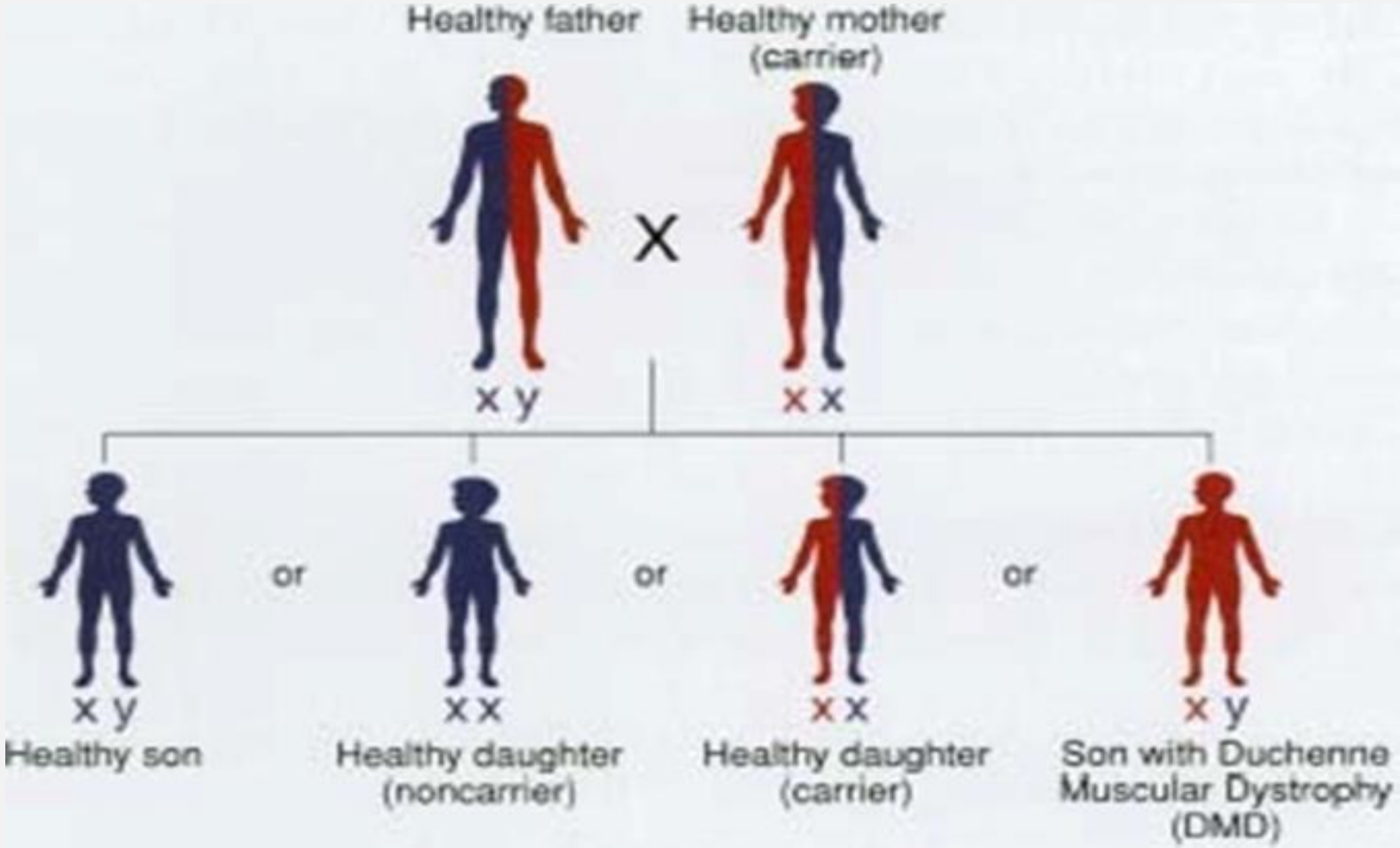
Phenylketonuria

Neonatal Screening test

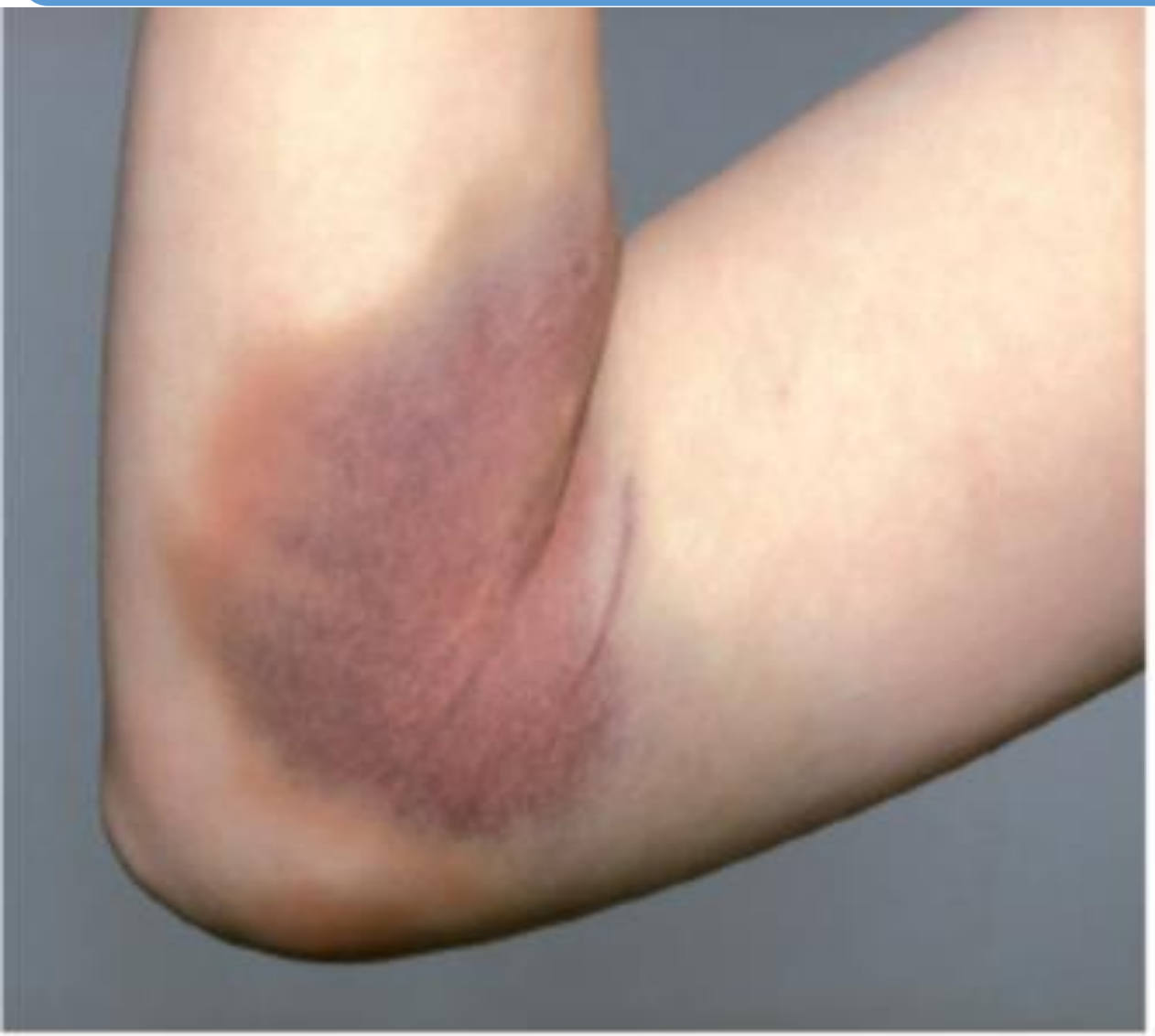
X- linked Recessive LO5

- Males are more often affected than females.
- Affected males pass the gene to all of their daughters and none of their sons (NO male-to-male transmission)
- Daughters of carrier females have a 50% chance to be unaffected carriers. Sons of carrier females have 50% chance to be affected.
- Affected males in the family are related to each other through carrier females

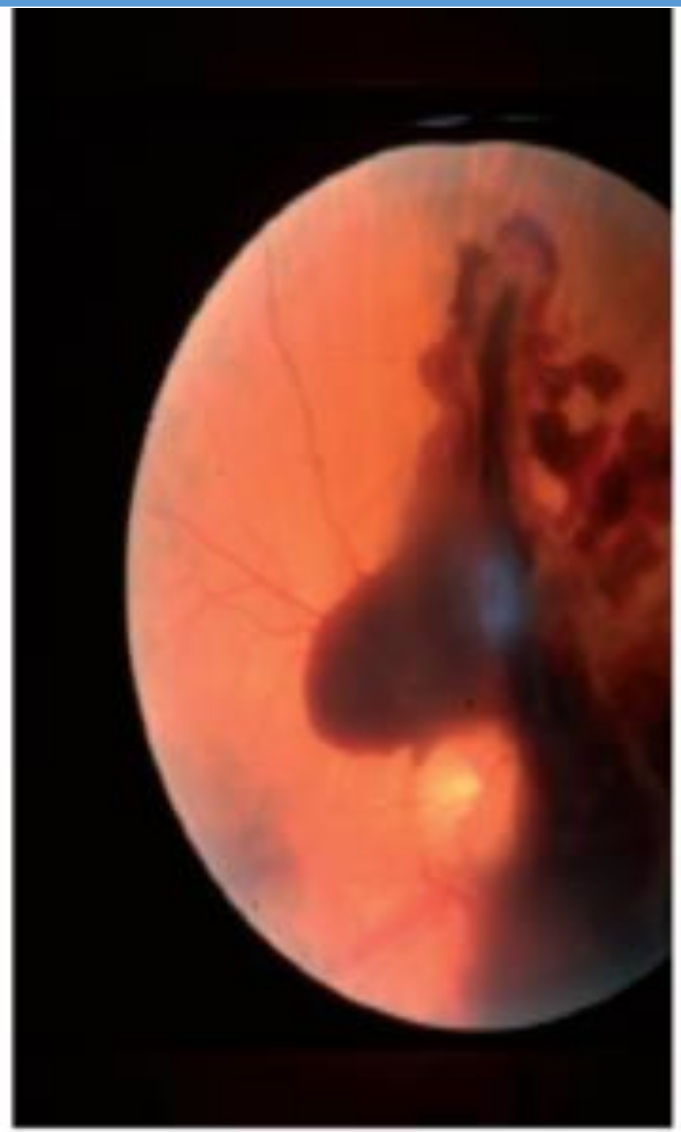



Healthy father
Healthy mother
(carrier)
X
x y
x x
or
or
or
x y
Healthy son
x x
Healthy daughter
(noncarrier)
x x
Healthy daughter
(carrier)
x y
Son with Duchenne
Muscular Dystrophy
(DMD)

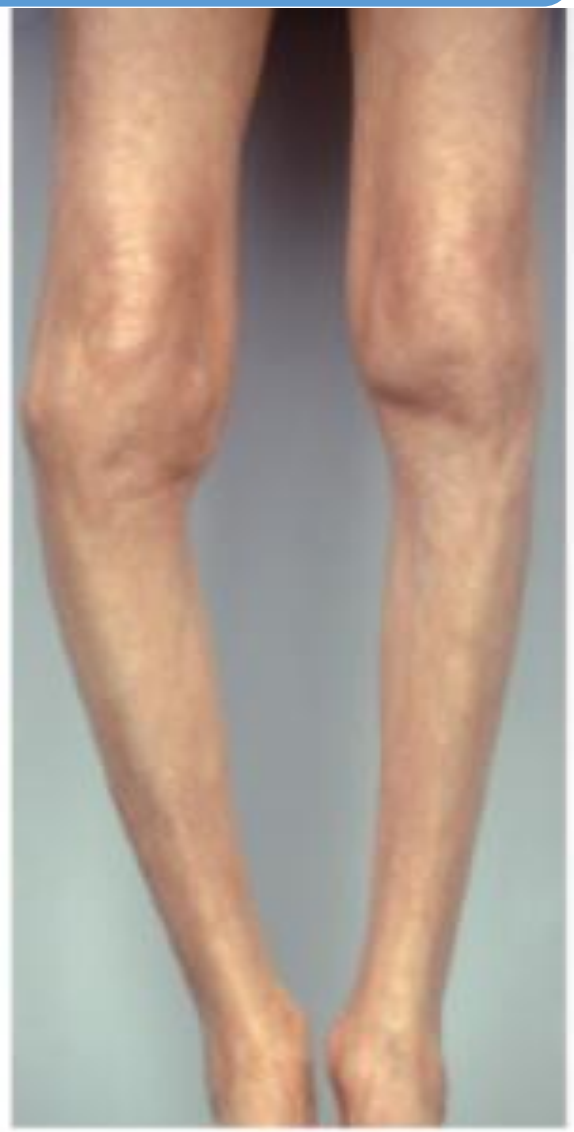
Hemophilia



(a)

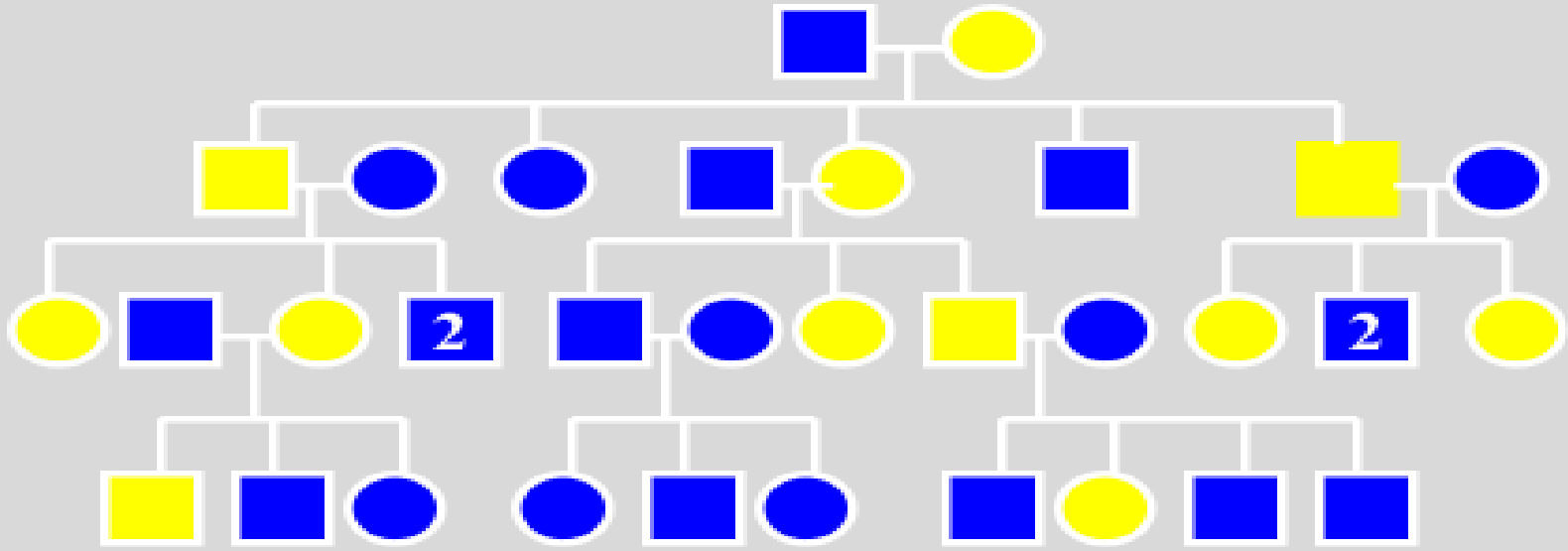


(b)



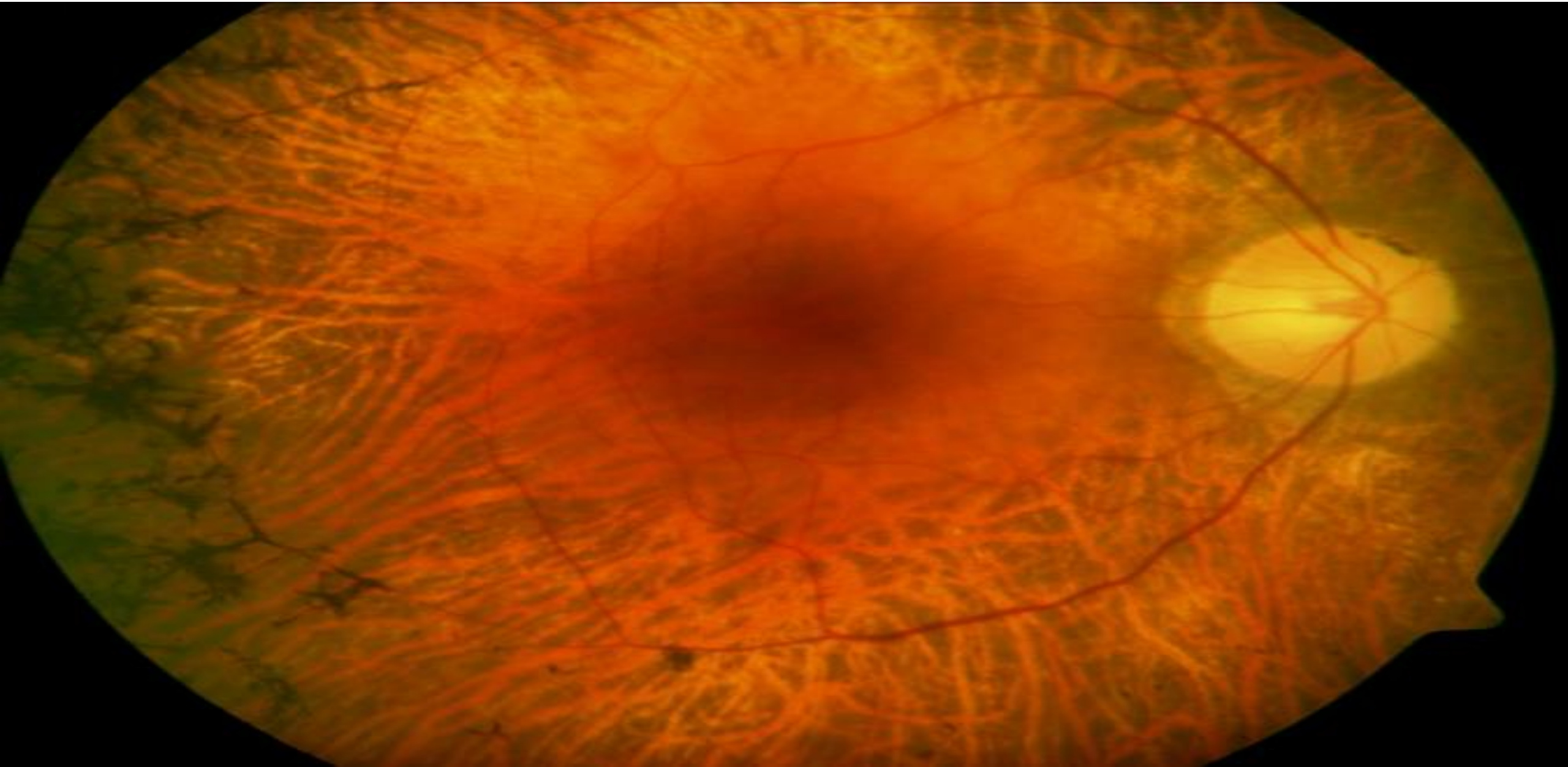
(c)

X- linked Dominant LO6

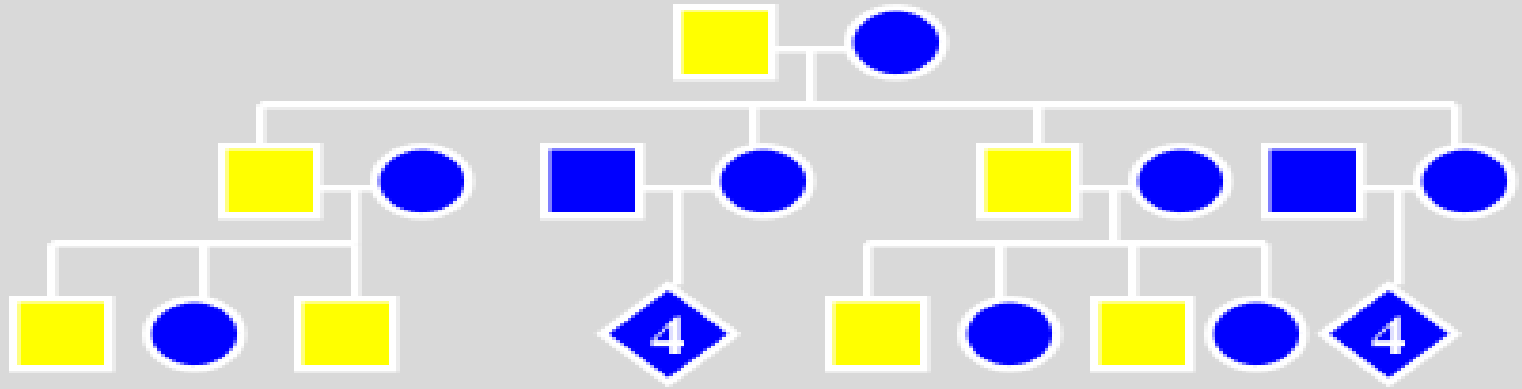


- For rare conditions, females are about 2x as likely to be affected than males. May be lethal in males and usually milder, but variable, in females.
- Affected males pass the gene to all of their daughters, who will be affected, and to none of their sons (NO male-to-male transmission)
- Sons and daughters of affected females have 50% chance of being affected (similar to autosomal dominant)

Retinitis Pigmentosa



Y- Linked LO7



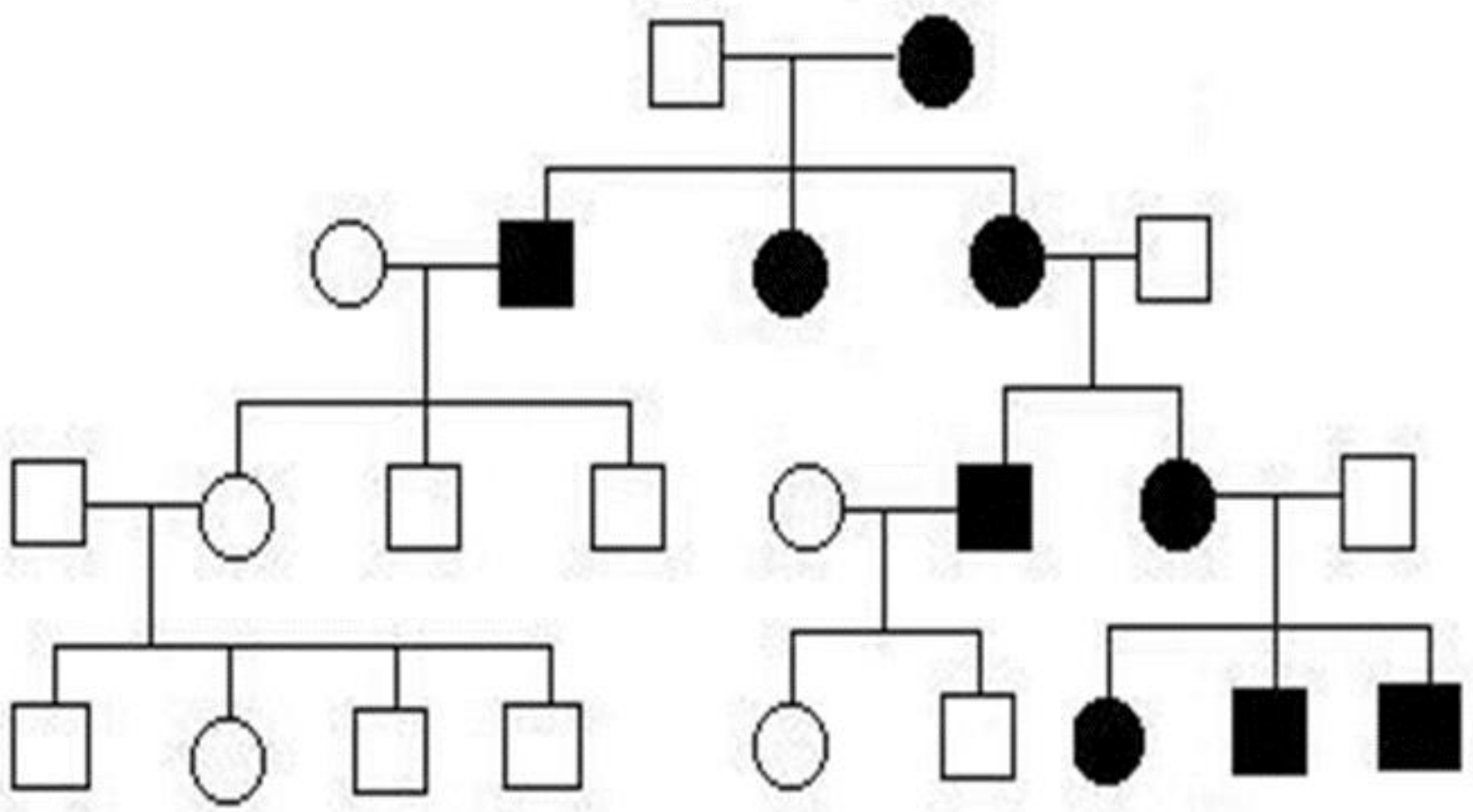
- Only males are affected
- Affected males pass the disease gene to all their sons and to none of their daughters

Mitochondrial Inheritance LO8

Mitochondria are only inherited from the mother's egg, thus only females can transmit the trait to offspring, however they pass it on to all of their offspring. The primary function of mitochondria is conversion of molecule into usable energy. Thus many diseases transmitted by mitochondrial inheritance affect organs with high-energy use such as the heart, skeletal muscle, liver, and kidneys.

- Mitochondrial Myopathy
- DAD (Diabetes Mellitus And Deafness)
- MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, Stroke like symptoms)

Mitochondrial Inheritance



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Multifactorial Inheritance LO9

Most diseases have multifactorial inheritance patterns. As the name implies, multifactorial conditions are not caused by a single gene, but rather are a result of interplay between genetic factors and environmental factors. Diseases with multifactorial inheritance are not genetically determined, but rather a genetic mutation may predispose an individual to a disease.

Conditions with multifactorial inheritance

- heart disease
- some cancers
- neural tube defects
- insulin-dependent diabetes mellitus
- intelligence
- Schizophrenia

Assignment

1- what is the difference between a genetic disease or condition and a congenital disease

2- write two other conditions as examples of AD inheritance

3- in what condition a female may exhibit signs of X-linked trait

Thank you