SAWA UNIVERSITY

اسم المادة و الكورس

LAB. 11 FAMILY PEDIGREE

Sawa University	جامعـــــــة ســـــــــاوة الاهلية
College of health and medical techniques	كليـــــة التقنيـــات الصحيـة والطبيـة
Department of Medical Laboratories	قسم تقنيات المختبرات الطبية
Third Stage	المرحلـــــــــــــة الثالثة

تدريسي المادة : م.د.عباس علي منشد
م.م. مهند ذجر اليونسي
م.م. ايات سلطان

الجانب العملي:الوراثة الطبية

Medical genetics-Practical

محاضرة رقم 11

Lecture No. 11

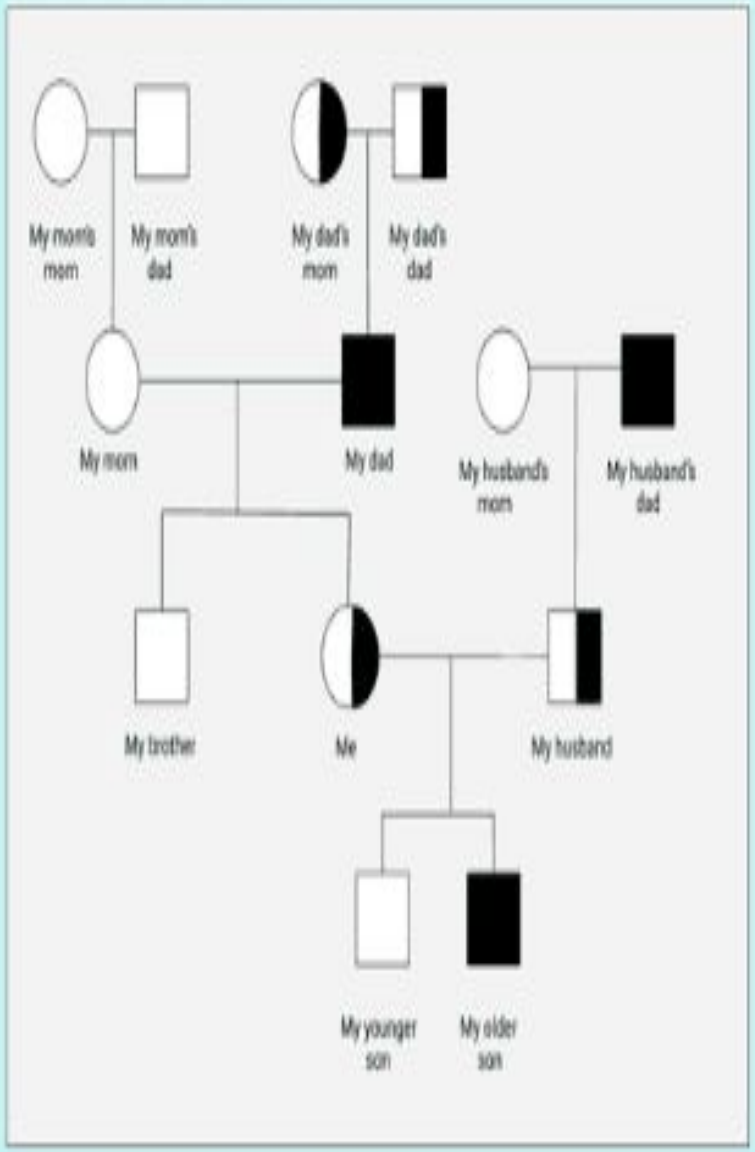
Objectives of karyotyping

MEDICAL GENETICS LAB.

By the end of this lab, students will be able to:

1. **Define Pedigree symbols.**
2. **Construct a family pedigree from a case.**
3. **Identify patterns of inheritance (Autosomal & Sex-linked).**
4. **Interpret pedigrees to determine genotype/phenotype.**
5. **Differentiate dominant, recessive, and X-linked traits.**

Exploring Genetics by Creating a Family Pedigree



Introduction

- **A family pedigree is a graphical representation of the inheritance of a trait, disorder, or genetic condition across multiple generations. It is one of the most essential tools in clinical genetics, medical diagnostics, and genetic counseling. Pedigrees allow clinicians to identify the mode of inheritance, assess recurrence risk, and detect carriers of genetic diseases.**

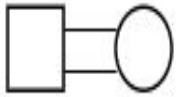
Standard Pedigree Symbols

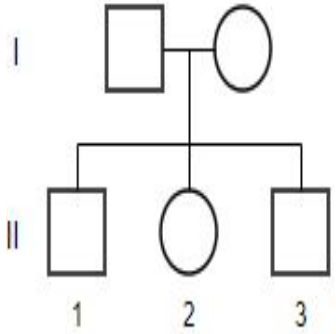
- Square = Male
- Circle = Female
- Diamond = Unknown sex
- Filled = Affected =
- Unfilled = Unaffected
- Slash / Deceased
- Double line = Consanguinity
- Proband = Marked by arrow
- Siblings connected by a horizontal line
- Generations = Roman numerals (I, II, III...)


Male


Female


Marriage

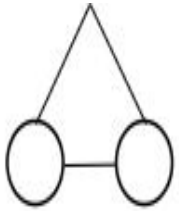

Consanguineous marriage

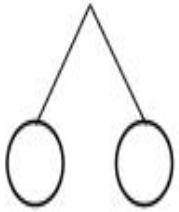


Roman numerals symbolize generations.

Arabic numbers symbolize individuals within a given generation.

Birth order, within each group of offspring, is drawn left to right, first-born to last-born.


Identical twins


Fraternal twins females


Affected male and female


Affected by history female and male


Carrier


Deceased female and male

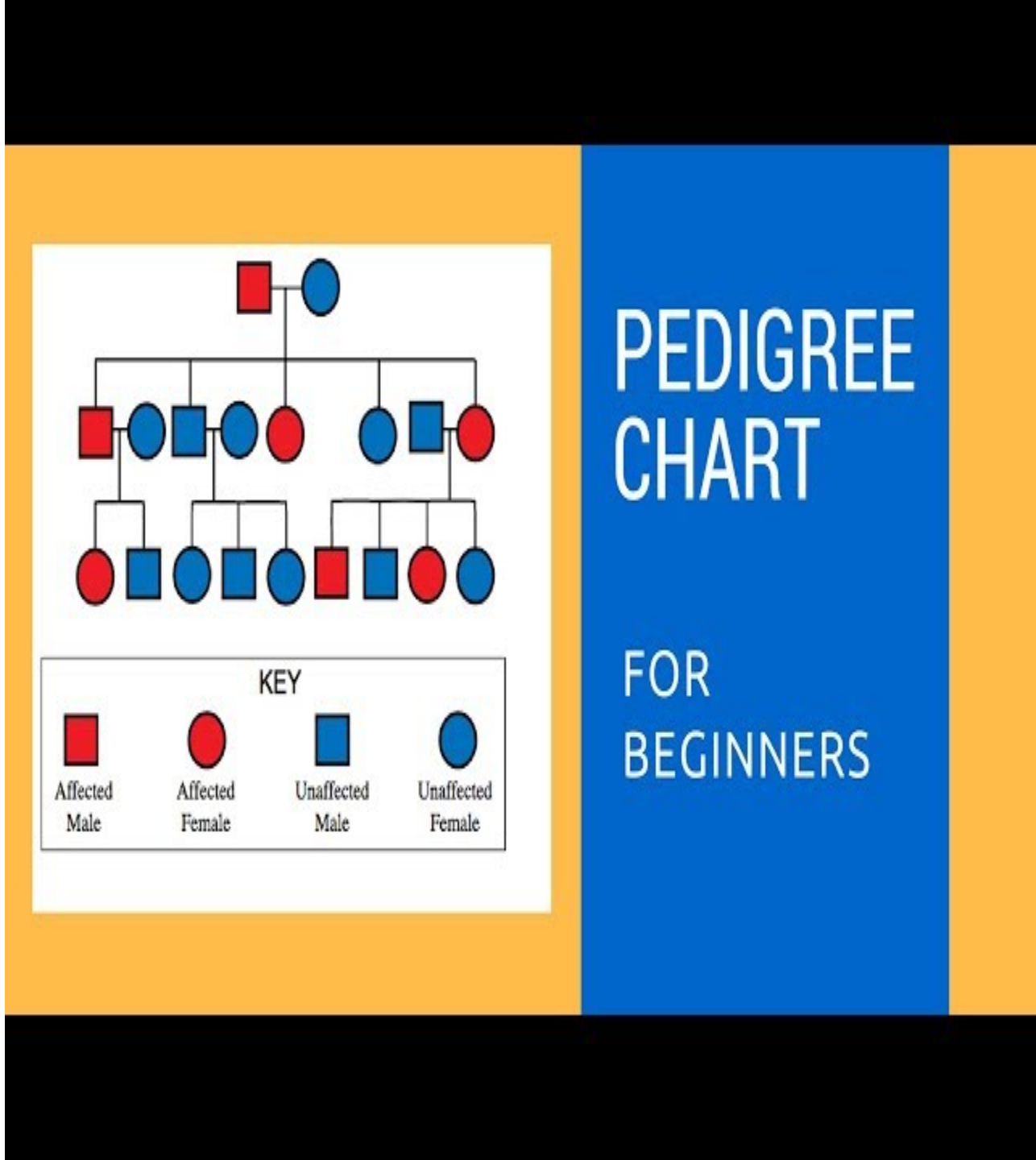

Unknown sex

How to Read a Pedigree

for understanding pedigrees

MEDICAL GENETICS LAB

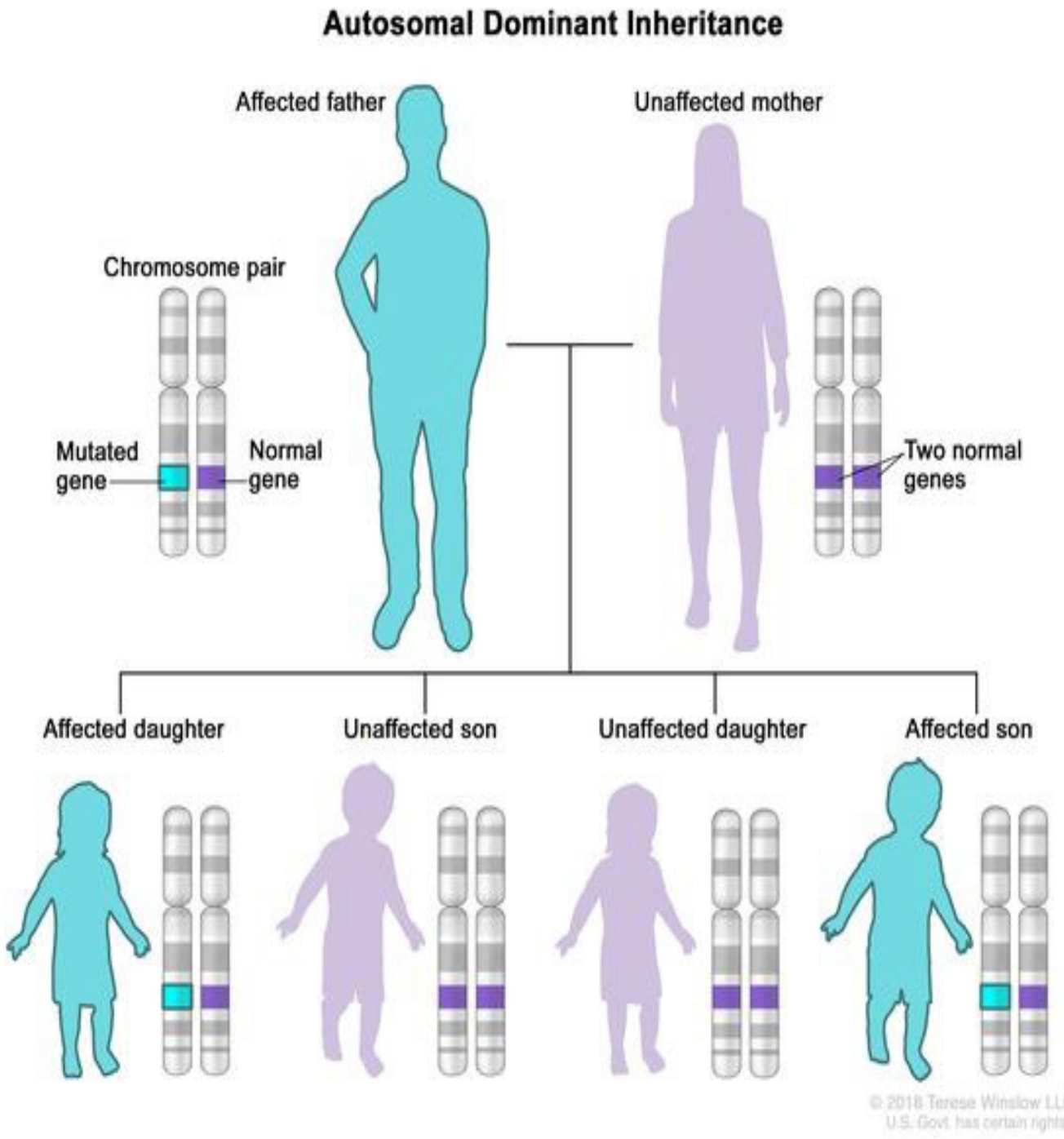
1. Trace the inheritance pattern through generations
2. Compare affected vs. unaffected
3. Check male vs female distribution
4. Ask: Is there vertical transmission?
5. Ask: Is consanguinity present?



Mendelian Inheritance Patterns

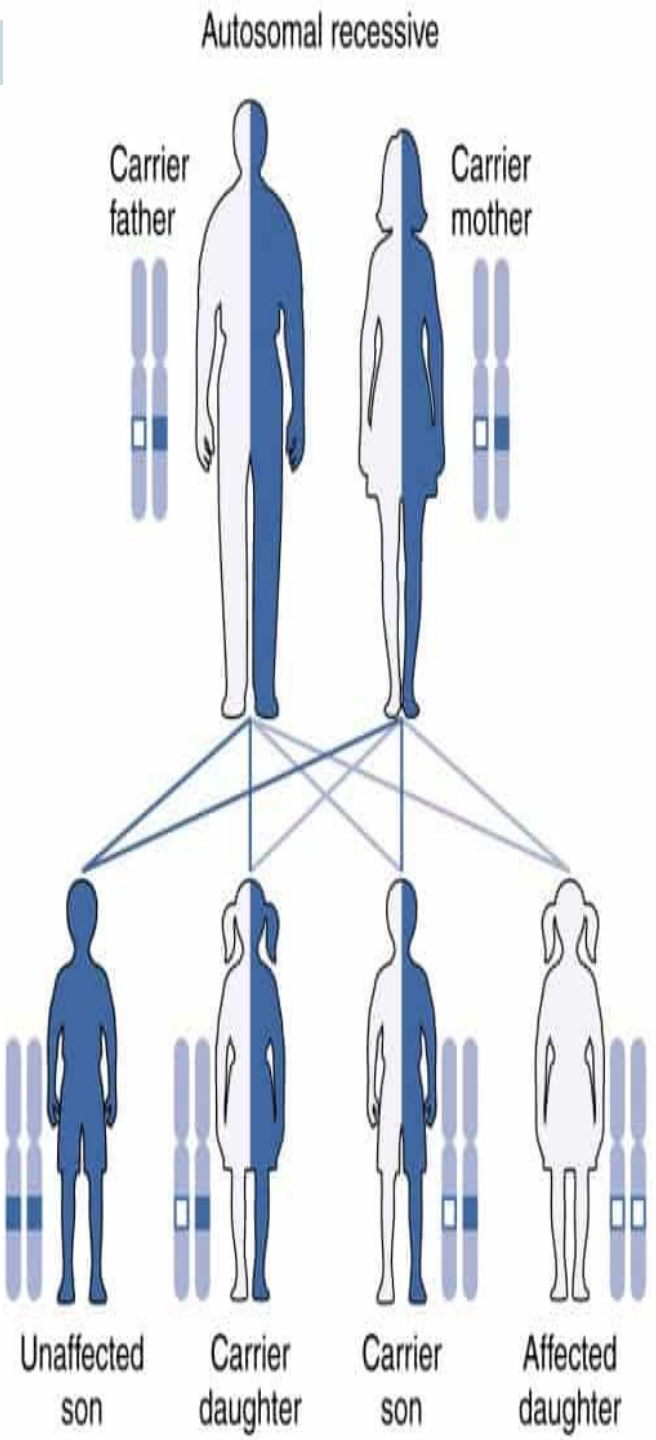
- Four main patterns
- 1 Autosomal Dominant (AD)
- 2 Autosomal Recessive (AR)
- 3 X-linked Dominant (XLD)
- 4 X-linked Recessive (XLR)

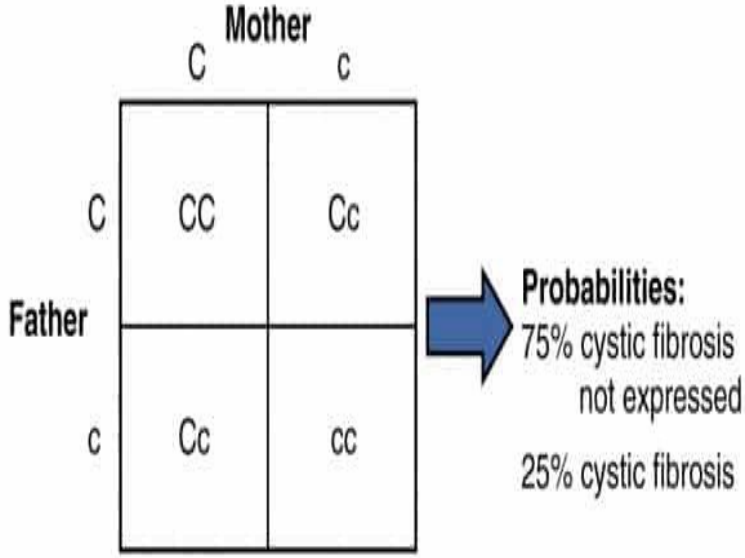
- Autosomal Dominant (AD)
- Features:
- Affected individuals in every generation
- Male = Female
- Affected parent → 50% affected children
- No skipping of generations
- Examples:
- Marfan syndrome
- Polycystic kidney disease



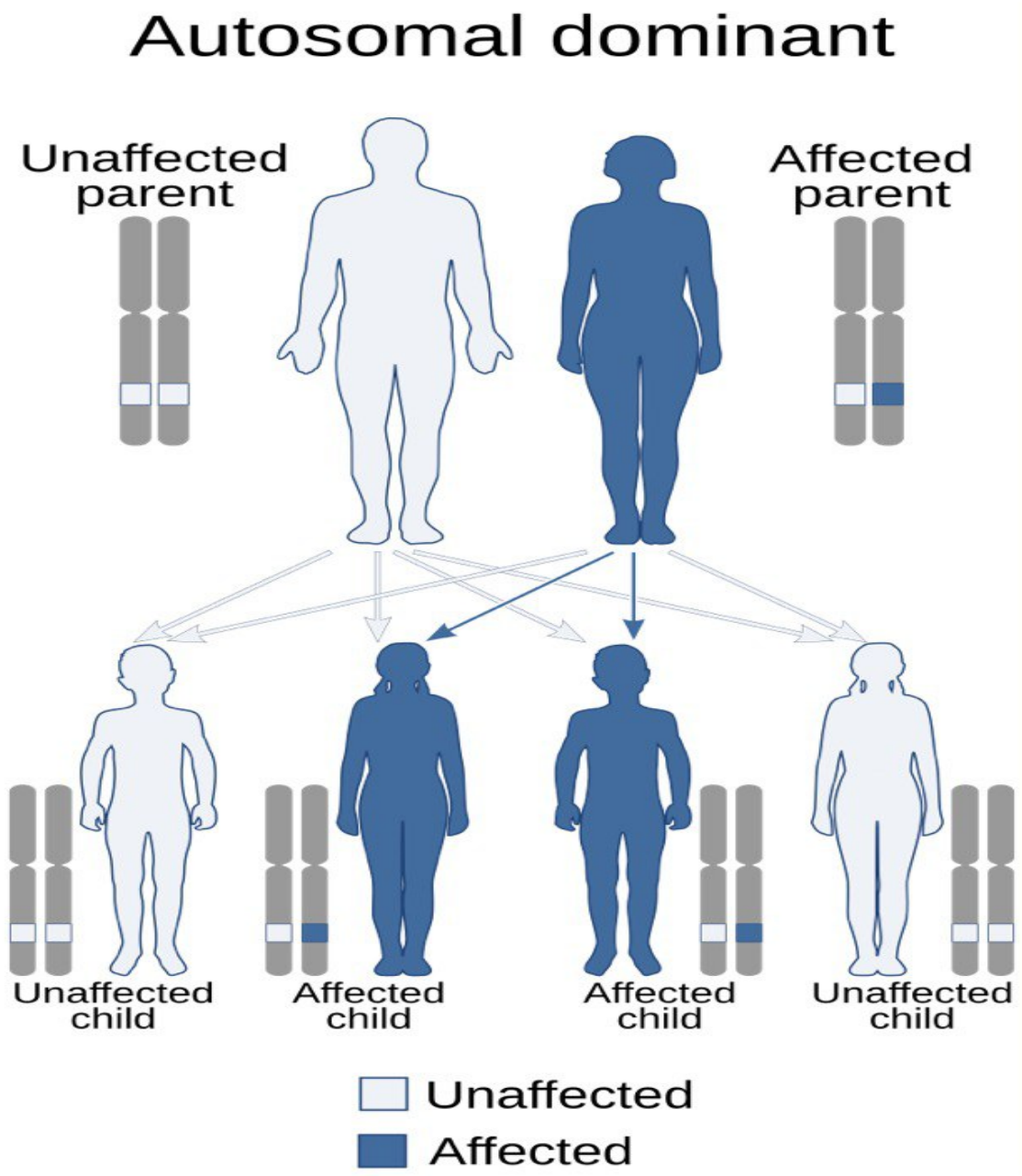
Mendelian Inheritance Patterns

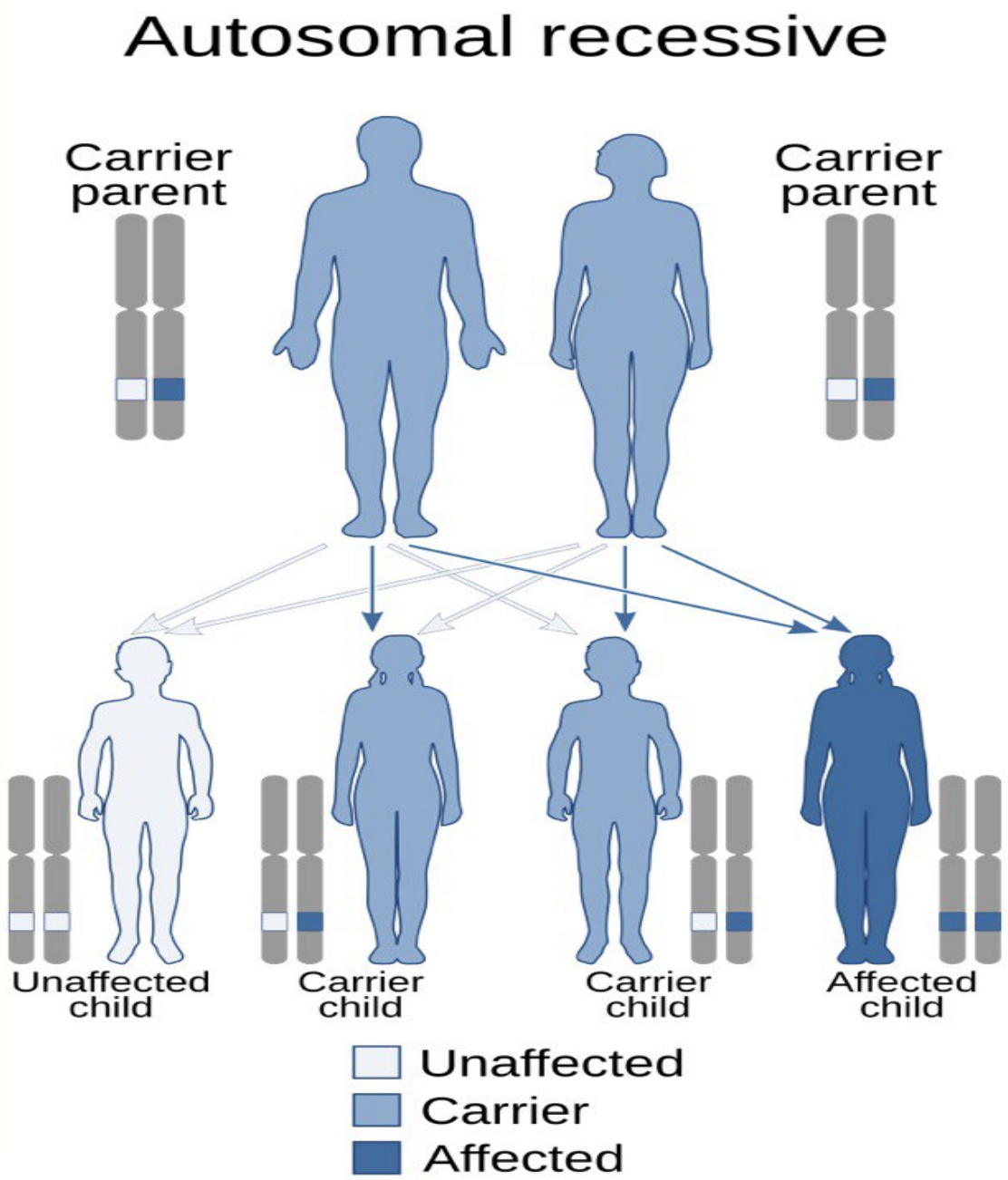
- **Autosomal Recessive (AR)**
- **Features:**
- Often seen in one generation only
- Consanguinity increases risk
- Parents usually carriers, not affected
- Male = Female
- **Examples:**
- Cystic fibrosis
- Sickle cell anemia





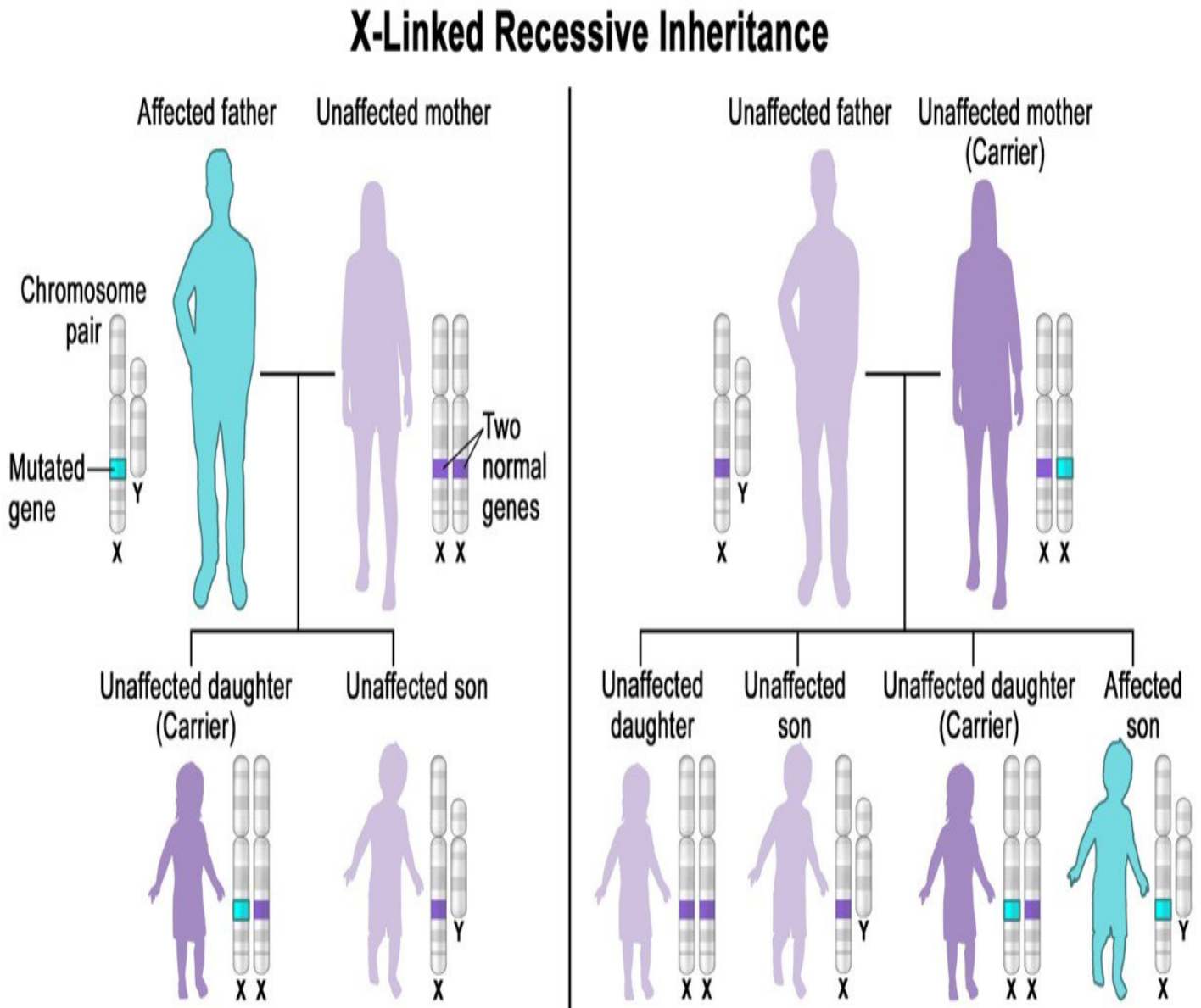
Mendelian Inheritance Patterns





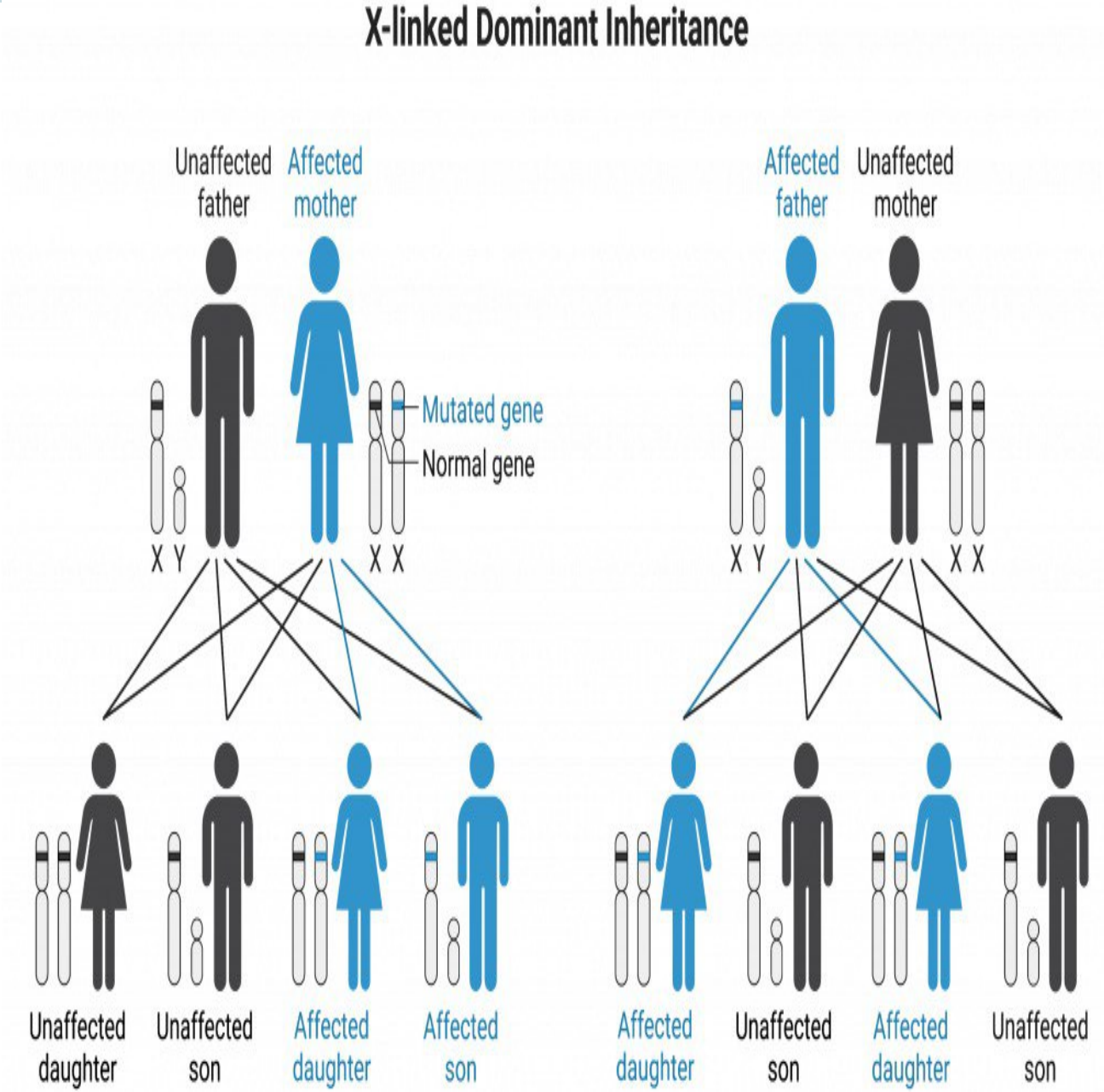
Mendelian Inheritance Patterns

- **X-linked Recessive (XLR)**
- **Features:**
- **Much more common in males**
- **No male-to-male transmission**
- **Affected males → all daughters are carriers**
- **Skipped generations possible**
- **Examples:**
- **Hemophilia A**
- **G6PD deficiency**

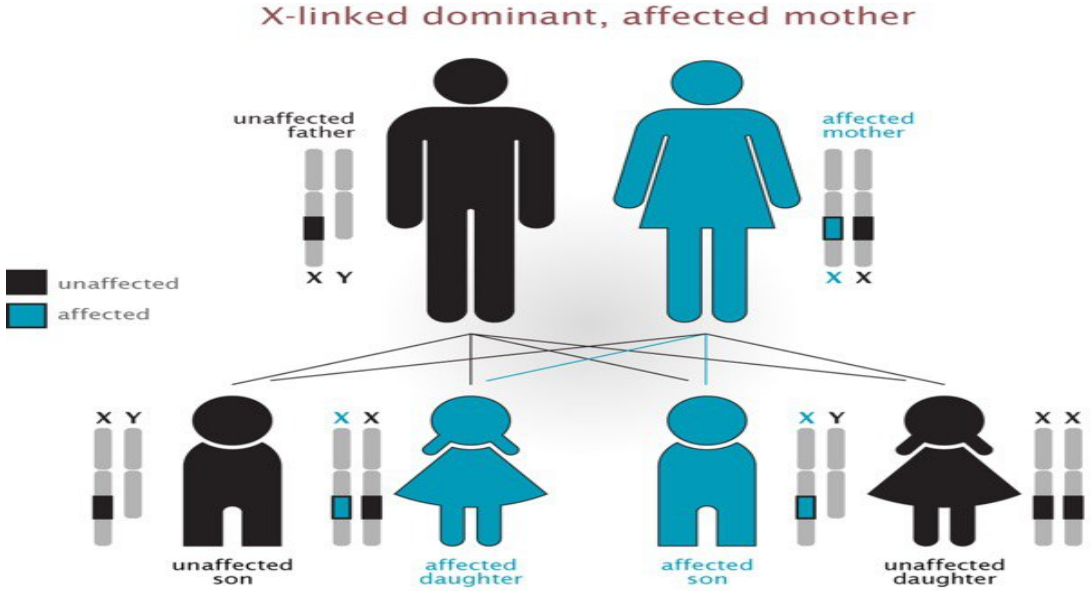


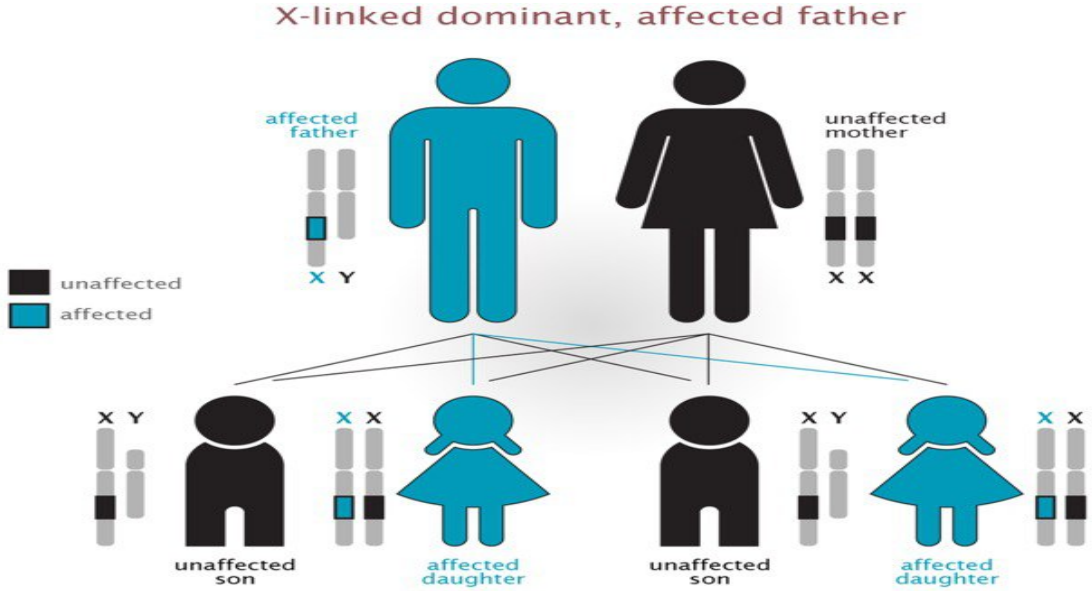
Mendelian Inheritance Patterns

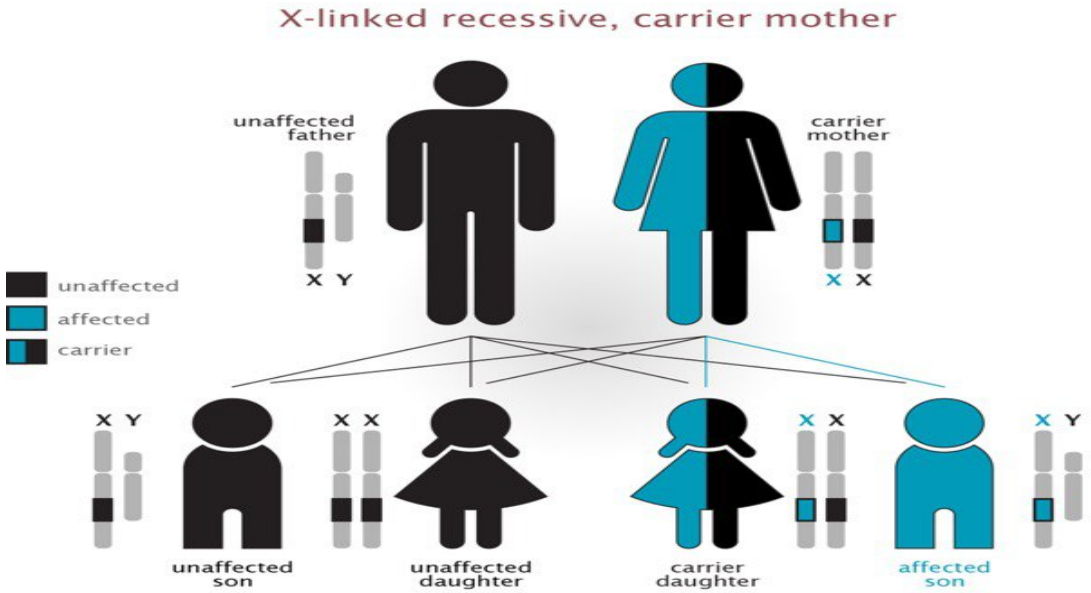
- **X-linked Dominant (XLD)**
- **Features:**
- **Affected fathers → all daughters affected**
- **No male-to-male transmission**
- **Female > Male**
- **Examples:**
- **Fragile X syndrome**
- **Rett syndrome**

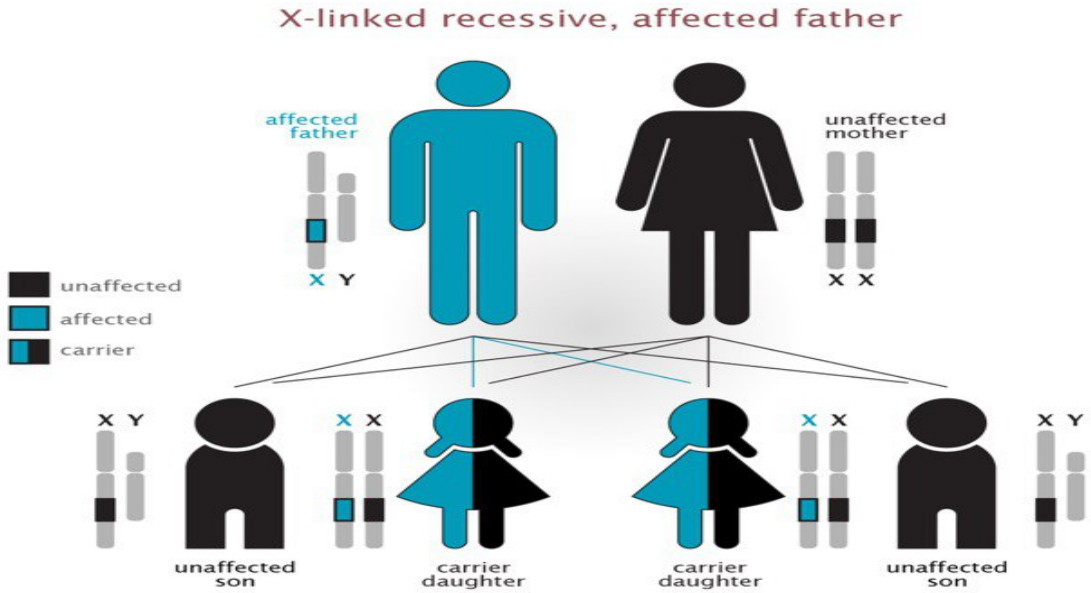


Mendelian Inheritance Patterns



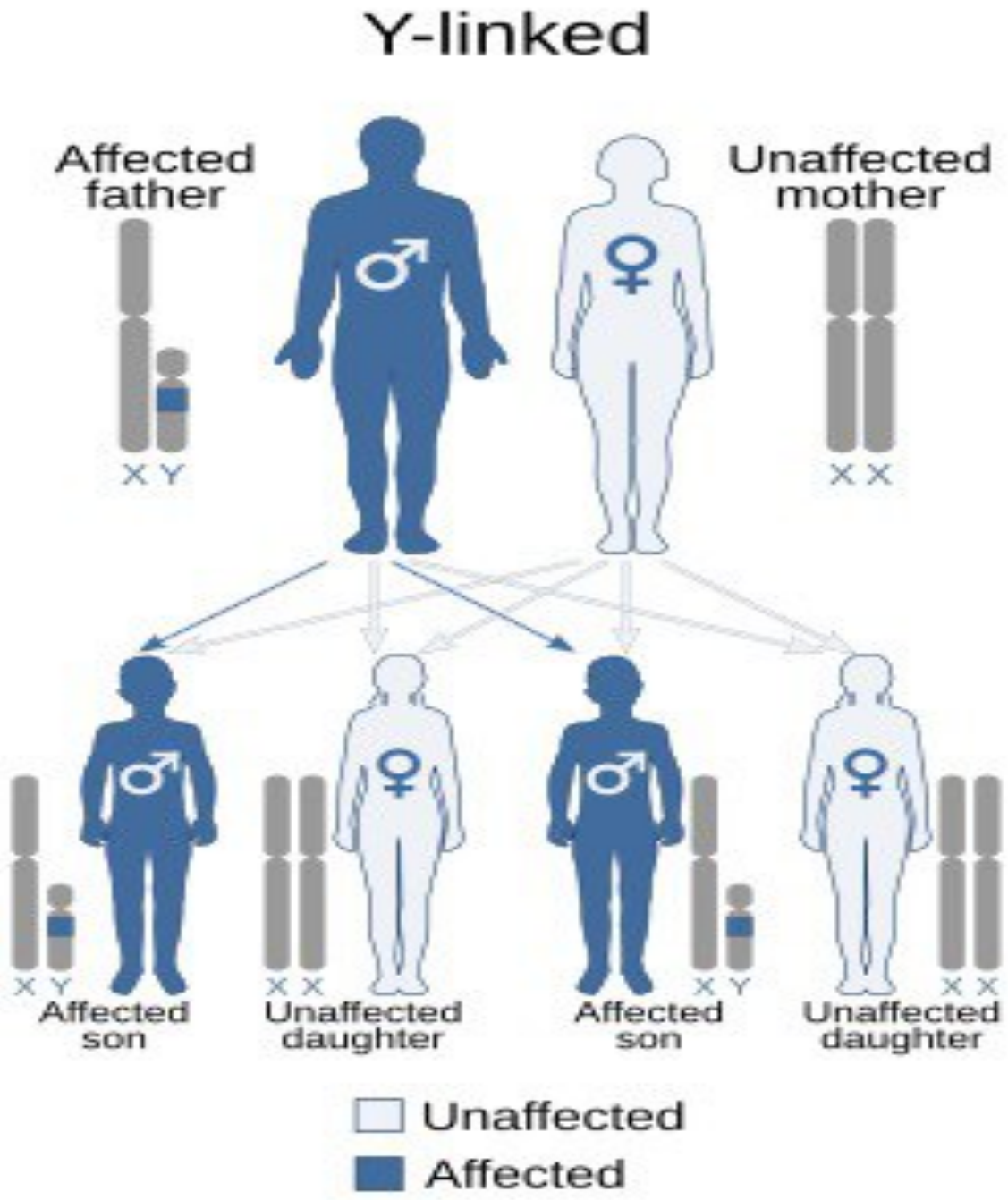






Mendelian Inheritance Patterns

- **Y-linked Inheritance (Holandric)**
- **Features:**
- **Only males are affected**
- **Always transmitted father-son**
- **Very rare**
- **Examples:**
- **Y-linked infertility genes**
- **Hairy pinna**



Co-Dominance

- Key Features
- Both alleles are expressed equally.
- No dominant or recessive relationship.
- Associated DiseaseExample
- ABO Blood Group
- Educational Case Study
- Father = A (IA i)
- Mother = B (IB i)
- Children's outcomes:
- AB
- A
- B
- O

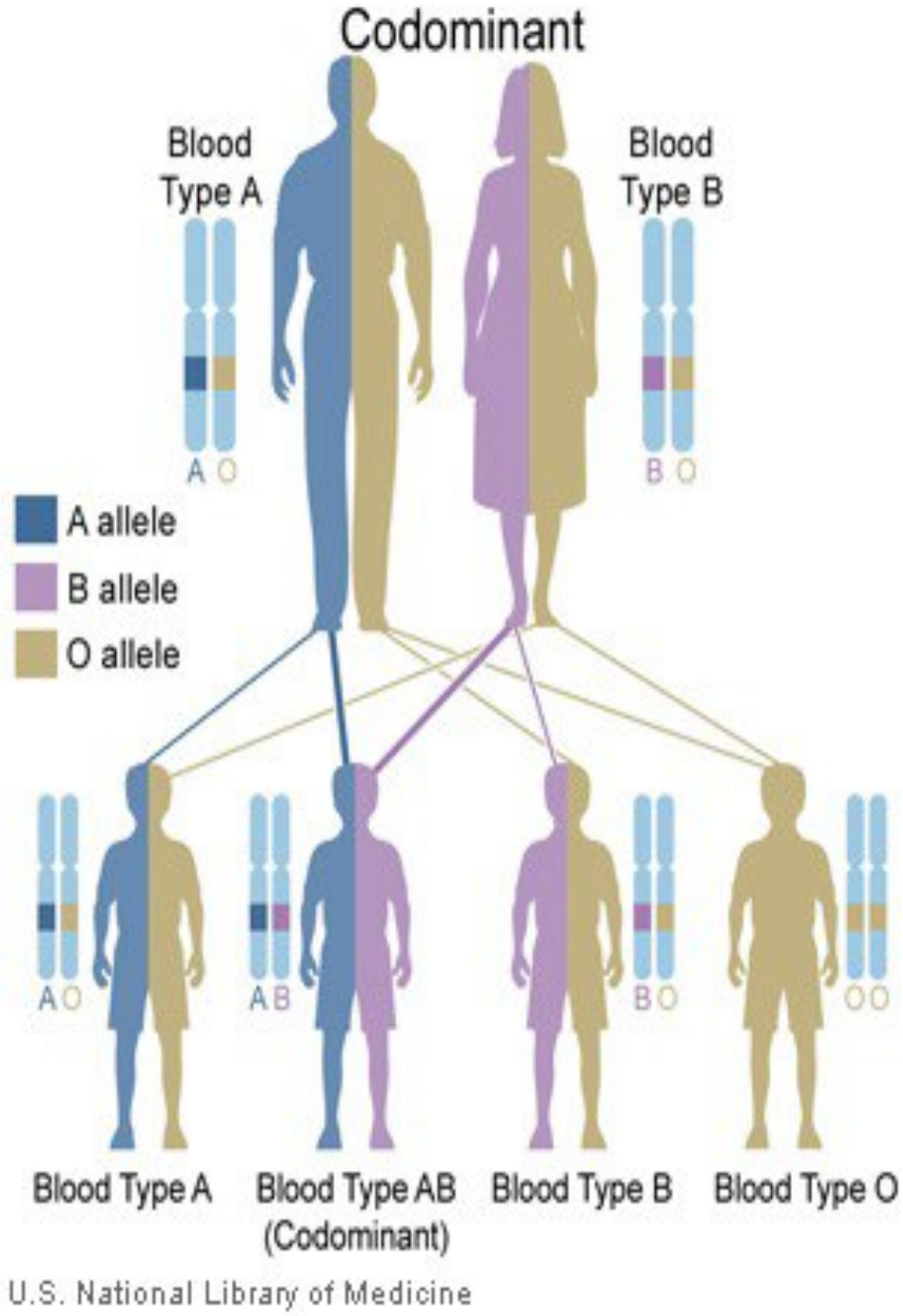
	I^A	I^B	i
I^A	I^AI^A A	I^AI^B AB	I^Ai A
I^B	I^BI^A AB	I^BI^B B	I^Bi B
i	iI^A A	iI^B B	ii O

	I^A	I^B	i
I^A	A	AB	A
I^B	AB	B	B
i	A	B	O

phenotype (Blood type)	genotype
Type A	I^AI^A I^Ai
Type B	I^BI^B I^Bi
Type AB	I^BI^A
Type O	ii

Blood Type Alleles:

- I^A (co-dominant)
- I^B (co-dominant)
- i (recessive)



case study: Draw the pedigree

- **Case 1/ A father is affected with a genetic disorder. The mother is healthy. They have 3 children:**
- **One affected son**
- **One affected daughter**
- **One healthy daughter**

- **Case 2/ A healthy mother has:**
- **Two affected sons**
- **One healthy daughter**

QUIZ

Any questions?