اسم المادة و الكورس

LAB 9 – CHROMOSOMAL ABNORMALITIES (NUMERICAL & STRUCTURAL)

Sawa University	جامعـــــة ســـــــاوة الاهلية
College of health and medical techniques	كليـــــة التقنيـــات الصحيـة والطبيـة
Department of Medical Laboratories	قسم تقنيات المختبرات الطبية
Third Stage	المرحلـــــــــــة الثالثة

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محاضرة رقم 9

الجانب العملي:الوراثة الطبية

Lecture No. 9

Medical genetics-Practical

Learning Objectives

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

1. **Define chromosomal abnormalities and distinguish numerical from structural defects.**
2. **Explain the mechanisms that lead to nondisjunction, breakage, and faulty repair.**
3. **Interpret chromosomal abnormalities clinically using real cases.**
4. **Write correct ISCN nomenclature for numerical and structural changes.**
5. **Correlate genotype → phenotype in common syndromes.**

Introduction

Chromosomal abnormalities are alterations in chromosome number or structure that disrupt normal gene dosage, leading to developmental, physiological, or reproductive abnormalities. They occur during meiosis, mitosis, or due to DNA repair defects.

These abnormalities are classified into:

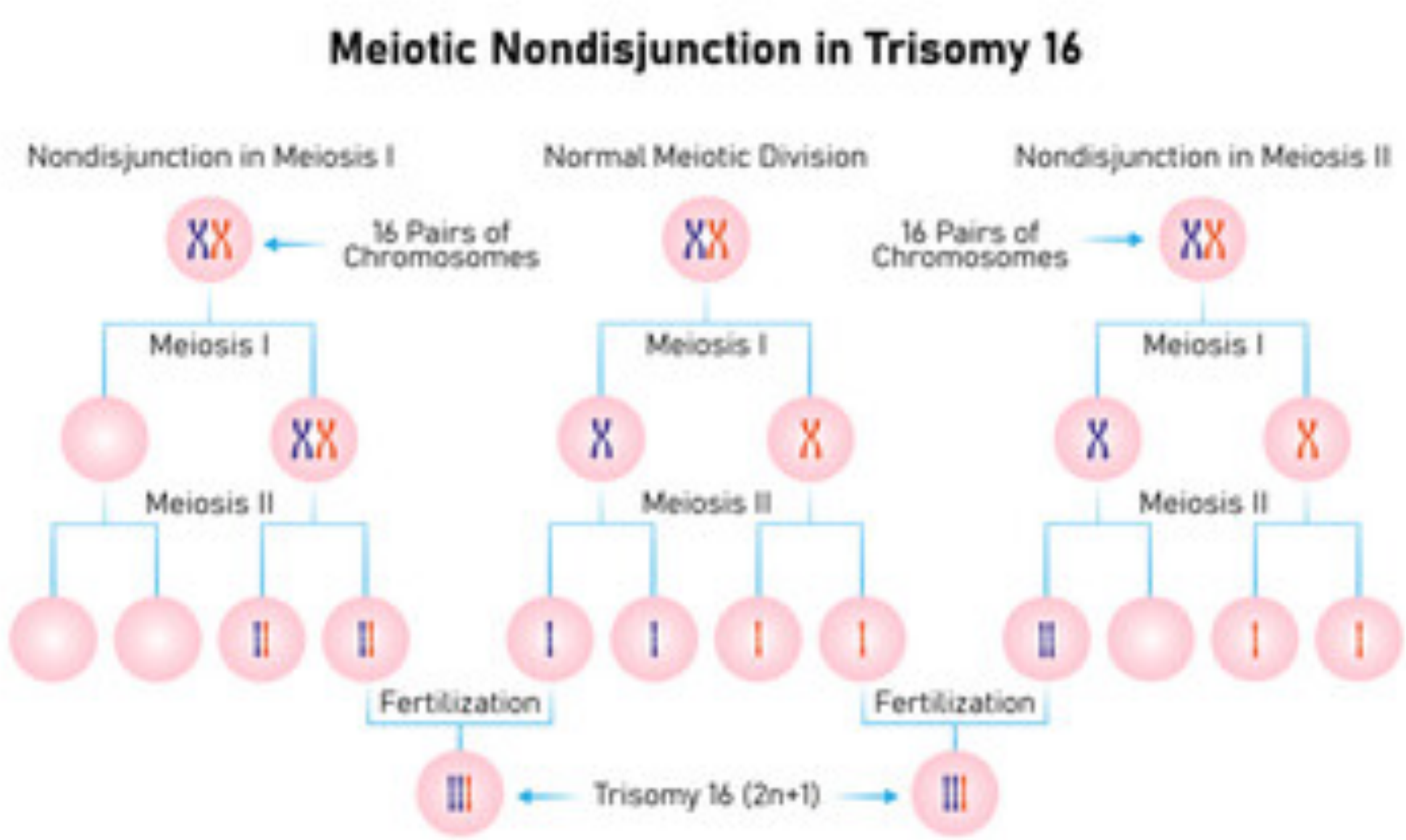
- **Numerical Chromosomal Abnormalities**
- **Structural Chromosomal Abnormalities**

Understanding these abnormalities is essential in clinical cytogenetics, prenatal diagnosis, oncology, and infertility studies.

Causes of Chromosomal Abnormalities

Cause	Mechanism
Nondisjunction	Failure of homologous chromosomes or sister chromatids to separate during meiosis I or II (leading to trisomy or monosomy).
Anaphase Lag	Loss of a chromosome during segregation → monosomy.
DNA Breakage	Ionizing radiation, chemicals → structural abnormalities.
Recombination Errors	Unequal crossing over → duplications or deletions.
Transposable Elements	Cause breaks or rearrangements.
Advanced Maternal Age	↑ nondisjunction risk (especially for chromosome 21 & X).

Causes of Chromosomal Abnormalities



Types of Chromosomal Abnormalities

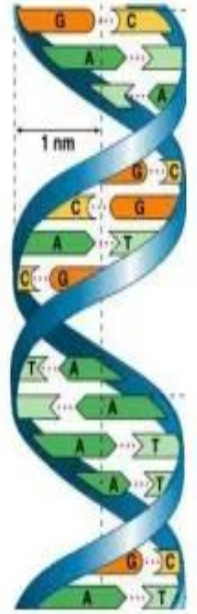
A. Numerical Abnormalities:

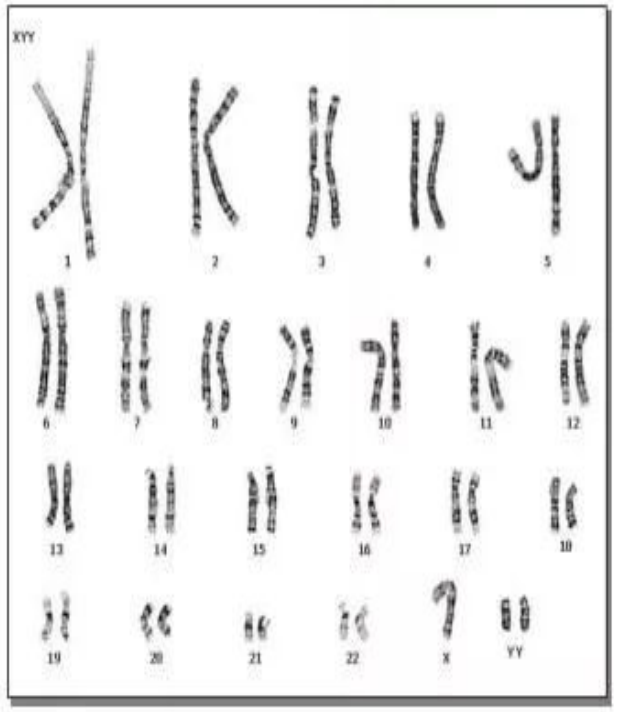
1. **Aneuploidy** (Trisomy / Monosomy)
2. **Polyploidy** (Triploidy / Tetraploidy)
3. **Mixoploidy** (Mosaicism)

B. Structural Abnormalities:

1. **Deletion**
2. **Duplication**
3. **Inversion**
4. **Translocation**
5. – Robertsonian
   – Reciprocal
6. **Ring Chromosomes**
7. **Isochromosomes**
8. **Derivative Chromosomes**

Chromosomal Abnormalities in Humans





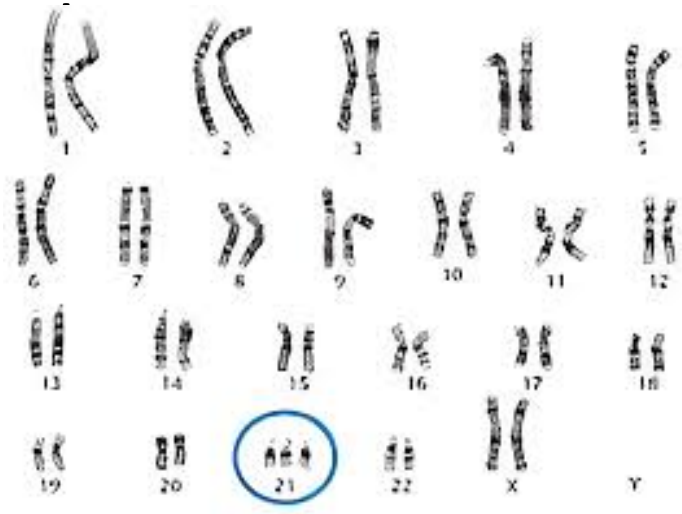
Numerical Chromosomal Syndromes with Cytogenetic Notation

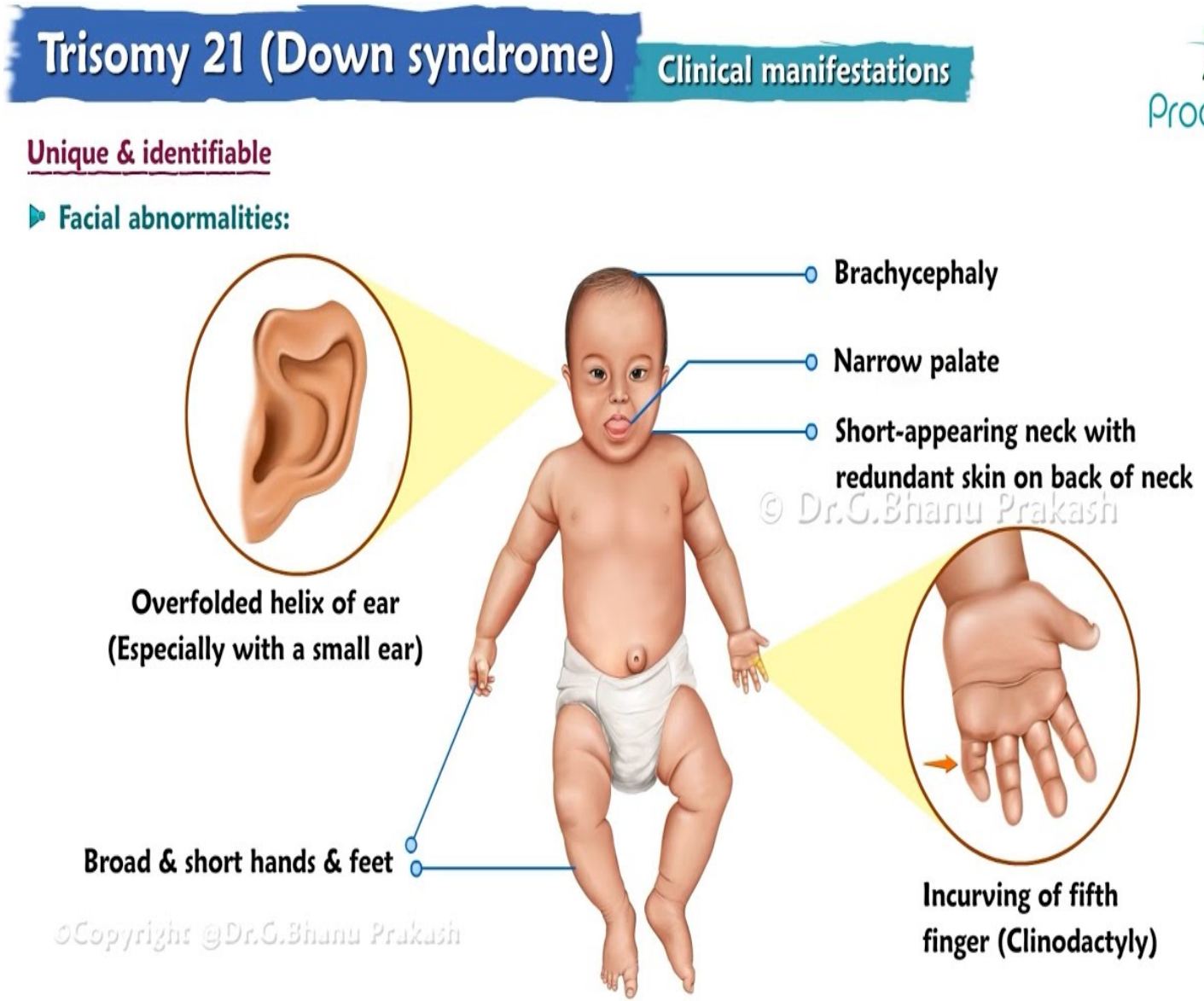
- **1. Down Syndrome (Trisomy 21)**
- **Karyotype /**

47, XX,+21 or 47, XY,+21

- **Cause: Maternal nondisjunction (95%).**
- **Clinical Features**

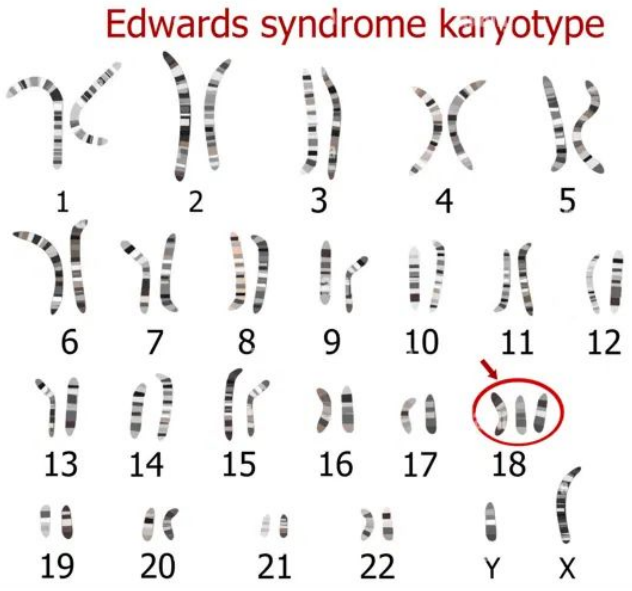
1. **Intellectual disability**
2. **Up-slanting palpebral fissures**
3. **Congenital heart disease (AVSD)**
4. **Single**

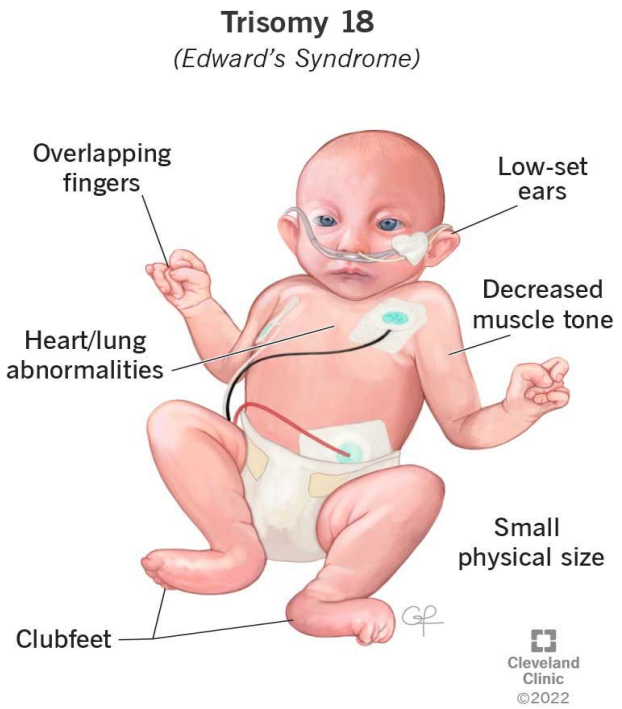




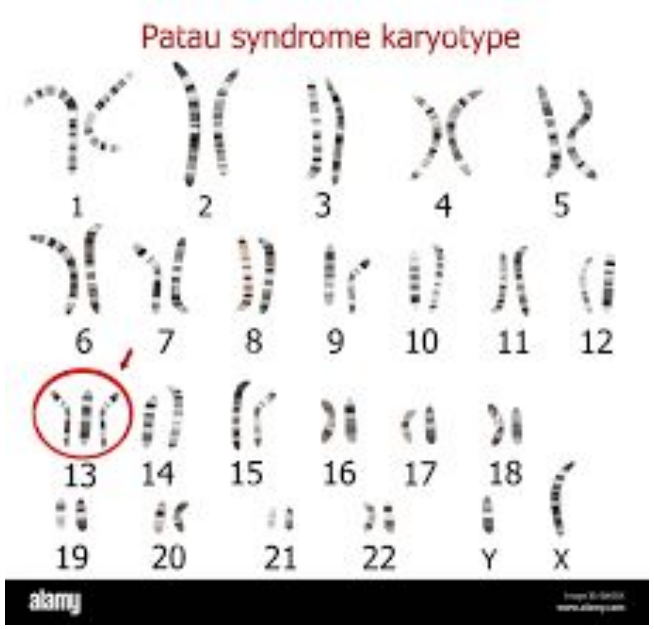
Numerical Chromosomal Syndromes with Cytogenetic Notation

- **2. Edwards Syndrome (Trisomy 18)**
- **Karyotype**
- **47, XX,+18 or 47, XY,+18**
- **Clinical**
- **Clenched fists**
- **Rocker-bottom feet**
- **Severe developmental delay**



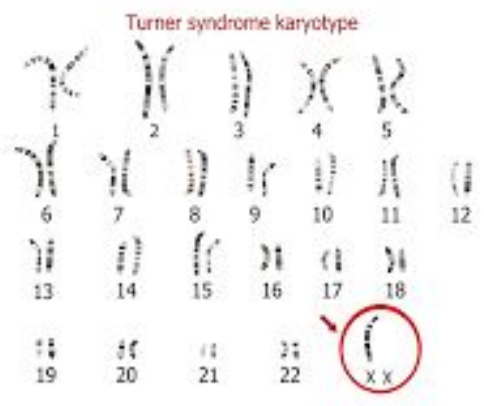


- **3. Patau Syndrome (Trisomy 13)**
- **Karyotype**
- **47, XX,+13 or 47, XY,+13**
- **Clinical**
- **Midline defects**
- **Holoprosencephaly**
- **Polydactyly**



Numerical Chromosomal Syndromes with Cytogenetic Notation

- **4. Turner Syndrome (Monosomy X)**
- **Karyotype** 45, X
- **Clinical**
- **Short stature**
- **Streak ovaries**
- **Webbed neck**
- **Cardiac defects (CoA)**



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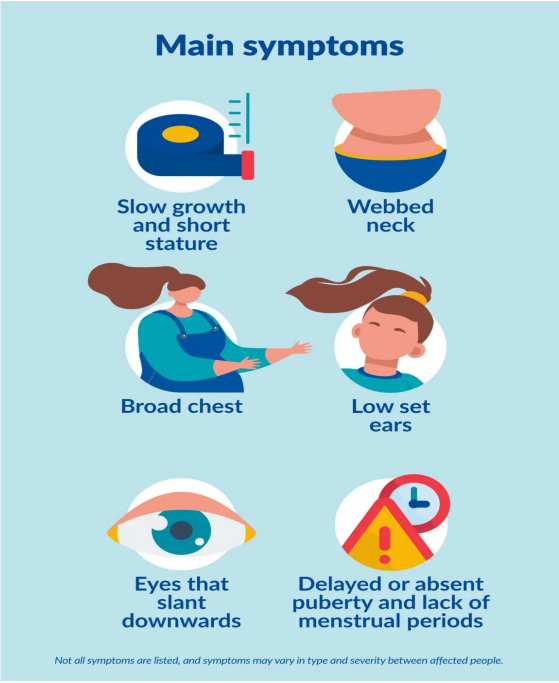
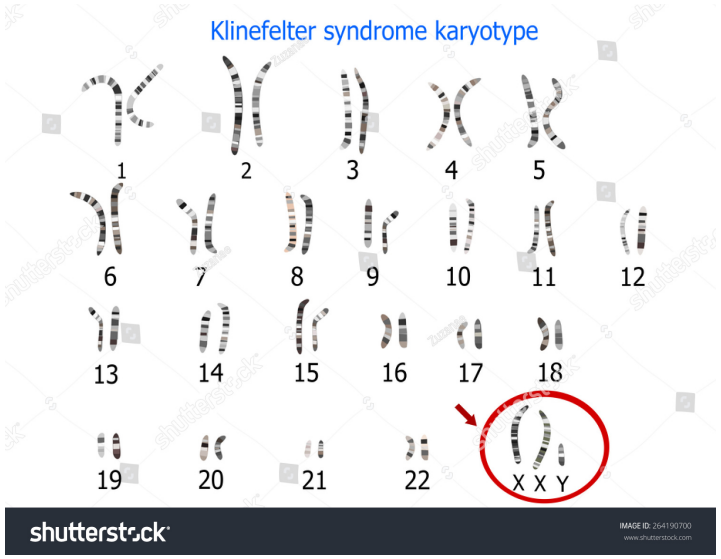
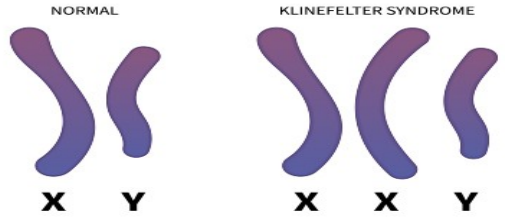


- **5. Klinefelter Syndrome**
- **Karyotype** 47, XXY
- **Clinical**
- **Tall stature**
- **Gynecomastia**
- **Hypogonadism Infertility**





Klinefelter Syndrome Common Symptoms

- *Tall stature*
- *Disproportionately long arms/legs*
- *Small testes*
- *Low testosterone*
- *Enlarged male chest*
- *Sparse body hair*
- *Broad hips*

Numerical Chromosomal Syndromes with Cytogenetic Notation

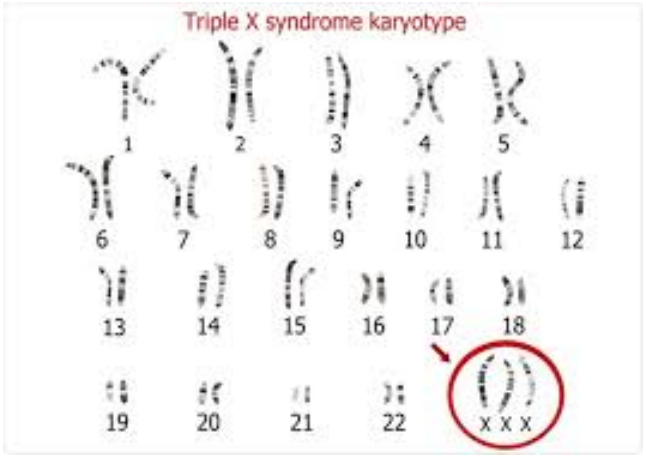
6. Triple X Syndrome

Karyotype

47, XXX

Mild phenotype

; often asymptomatic.

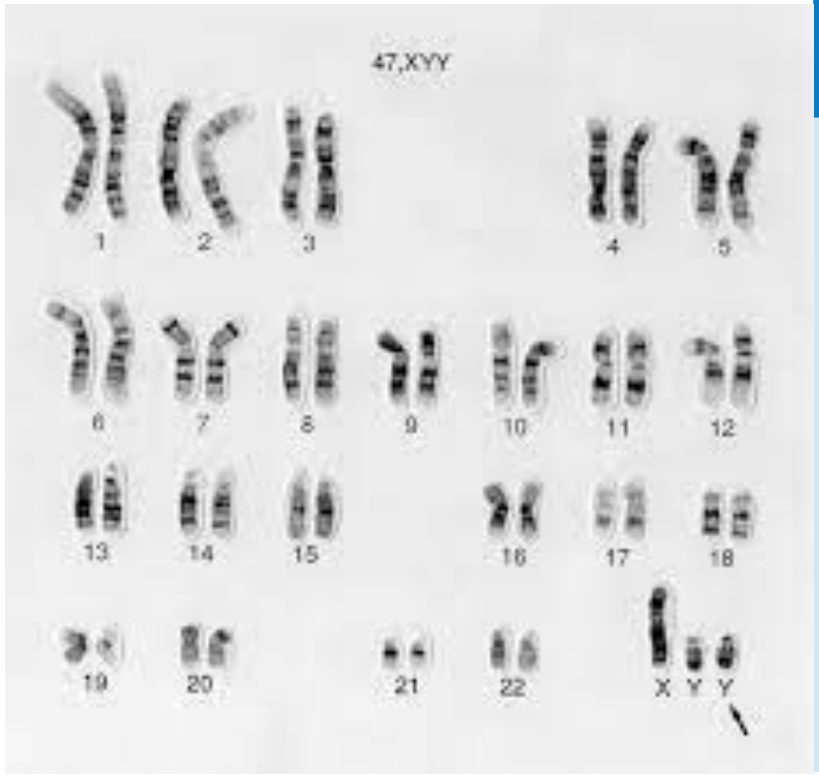


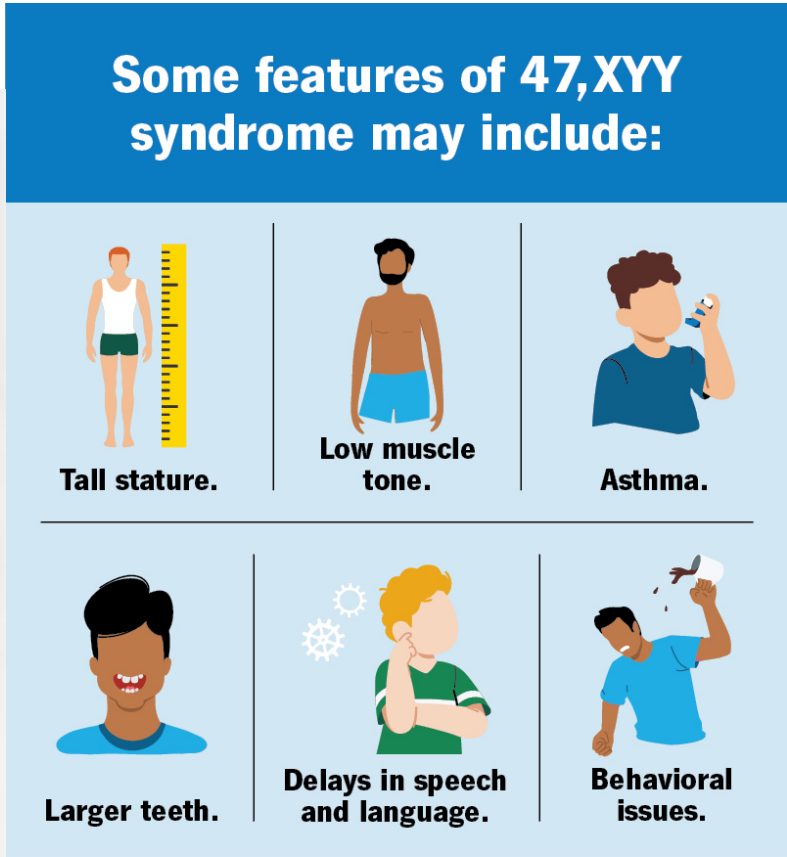
7. XYY Syndrome

Karyotype

47, XYY

Tall male, mild learning difficulties, normal fertility.





Summary Table

Syndrome	Cytogenetic Code	Type
Down	+21	Trisomy
Edwards	+18	Trisomy
Patau	+13	Trisomy
Turner	45,X	Monosomy
Klinefelter	47,XXY	Sex chromosome trisomy
Cri-du-chat	del(5p)	Structural deletion
Wolf-Hirschhorn	del(4p)	Structural
DiGeorge	del(22q11.2)	Microdeletion
Williams	del(7q11.23)	Microdeletion
PWS	del(15)(paternal)	Imprinting deletion
Angelman	del(15)(maternal)	Imprinting deletion

QUIZ

Q1/Case Study:

A newborn has: weak, cat-like cry, a small head, a round face, and feeding difficulty. Chromosomal analysis shows a deletion on the short arm of chromosome 5.

What is the name of this syndrome?

What is the expected ISCN notation?

Q2/Case Study:

A newborn boy presents with: cleft lipextra fingers (polydactyly), very small eyes, and severe developmental problems. The karyotype shows an extra chromosome in the autosomes.

What is the name of this syndrome?

What is the expected ISCN notation?

Any questions?