### Human chromosomes

- There are 46 chromosomes (23 homologous pairs) in each somatic cell
- 22 pairs of autosomes
- 1 pair of sex chromosomes
  XX = Female, XY = Male
- Karyotype chromosomes are arranged according to shape and size

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#### Nondisjunction and chromosomal disorders

•Nondisjunction – failure of chromosomes to separate and segregate into daughter cells

•Nondisjunction may occur during meiosis 1 or meiosis 2

•Abnormal number of chromosomes may result







#### **EXAMPLES OF NON-DISJUNCTION**

#### ABNORMALITIES OF SEX CHROMOSOME NUMBER IN HUMANS

Sex Chromosomes	Syndrome	Origin of Nondisjunction	Frequency in Population
XXY	Klinefelter syndrome (male)	Meiosis in egg or sperm formation	<u>1</u> 2,000
XYY	None (normal male)	Meiosis in sperm formation	2,000
XXX	Metafemale	Meiosis in egg or sperm formation	1,000
XO	Turner syndrome (female)	Meiosis in egg or sperm formation	<u>    1</u> 5,000

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#### AN EXTRA COPY OF CHROMOSOME 21 CAUSES DOWN SYNDROME

This condition is called **trisomy 21**. Person with this condition suffers from the Down syndrome.

Characteristic facial features:

- Round face
- Flattened nose bridge
- Small, irregular teeth
- Short stature
- Heart defects



# Klinefelter's syndrome, 47, XXY

It is the most common sex chromosome disorder and the second most common condition caused by the presence of extra chromosomes

Symptoms:

- Language impairment
- Lanky, youthful build or rounded body type
- Low levels of Testosterone and small testicles / Infertile

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# Turner's Syndrome (X)

#### Common symptoms: Short stature Characteristic facial features Short stature Low hairline Fold of skin Constriction swelling of the hands and fee ۲ Shield-shaped of aorta thorax Poor breast Broad chest and widely • development Widely spaced nipples spaced nipples Elbow Shortened deformity metacarpal IV Low hairline • Small Rudimentary finger nails ovaries Gonadal streak Low-set ears • (underdeveloped gonadal Reproductive sterility structures) Brown spots (nevi) Increased weight, obesity No menstruation ulletSmall fingernails ۲

- Characteristic facial features
- Webbed neck

## What genetic disorder is this?

