TURNER SYNDROME PROJECT

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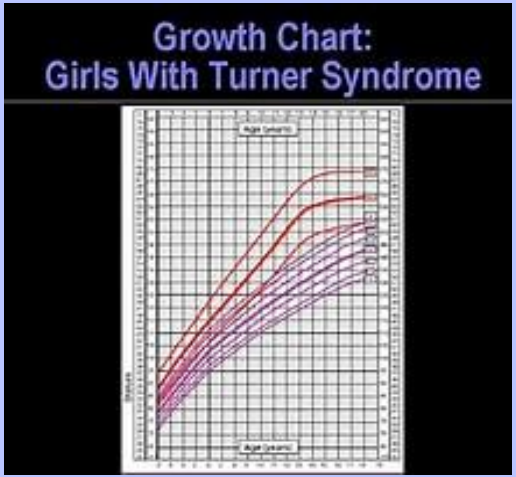
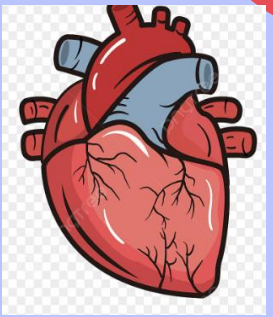
Meet Sara: A Girl with Turner Syndrome

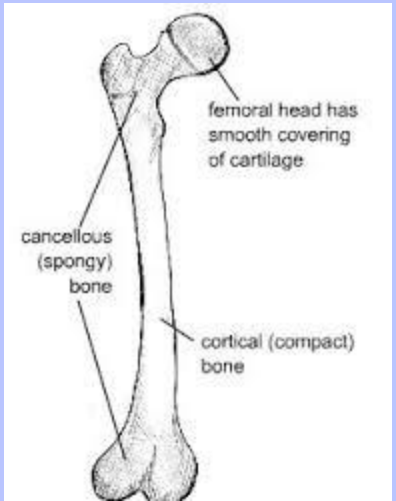
- Sara is a 10 year old girl who has always been the shortest in her class
- She gets tired easily and has frequent ear infections
- Her doctor hears a heart murmur and worries about her slow growth
- A karyotype test shows 45,X, which confirms Turner syndrome

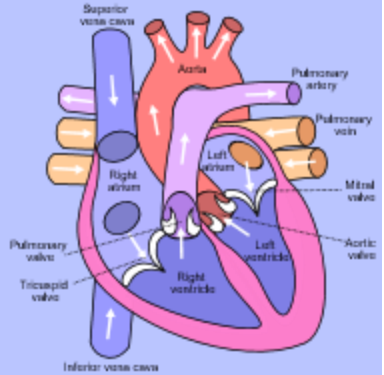


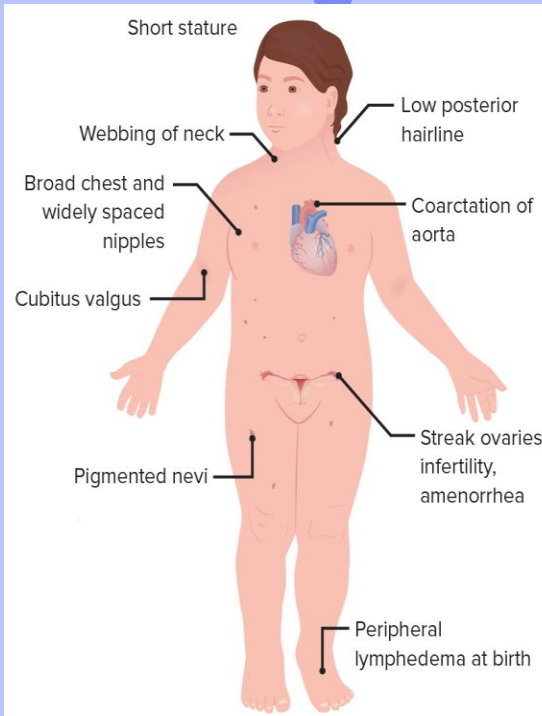


Clinical Features and Complications

- Short stature and slow growth
- Webbed neck, low set ears, broad chest, swelling of hands and feet at birth
- Delayed puberty, no periods, infertility
- Heart problems such as bicuspid aortic valve and coarctation of the aorta
- Kidney differences, thyroid disease, frequent ear infections and hearing loss
- Long term: high blood pressure, aortic enlargement, osteoporosis, diabetes, high cholesterol

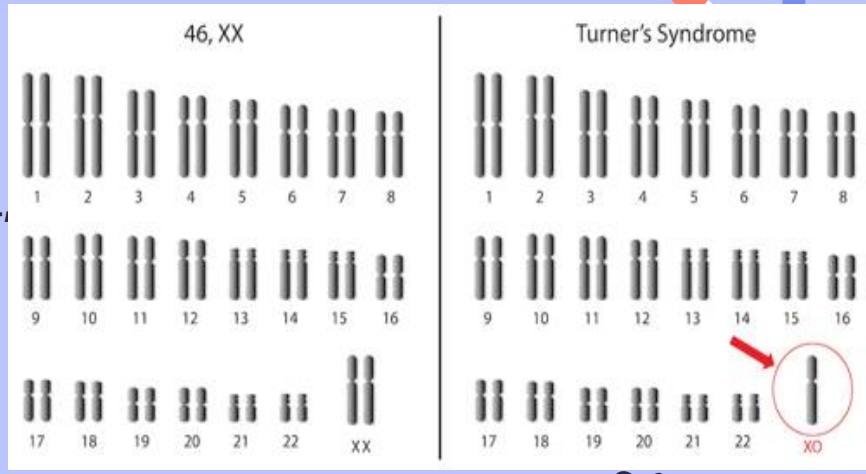






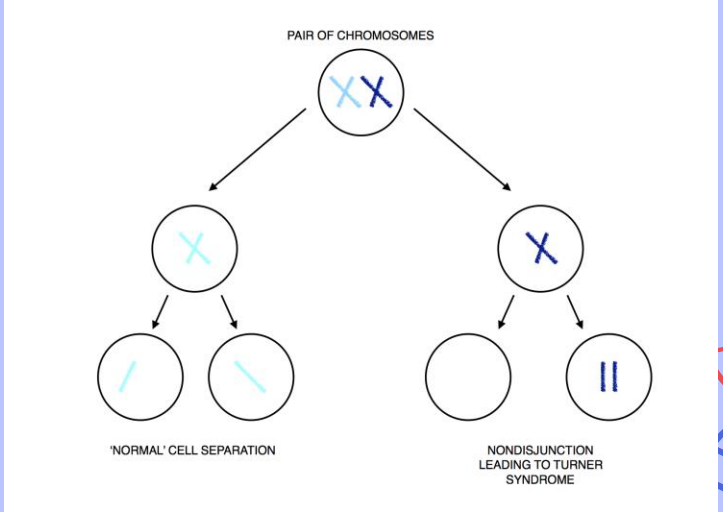
What Causes Turner Syndrome?

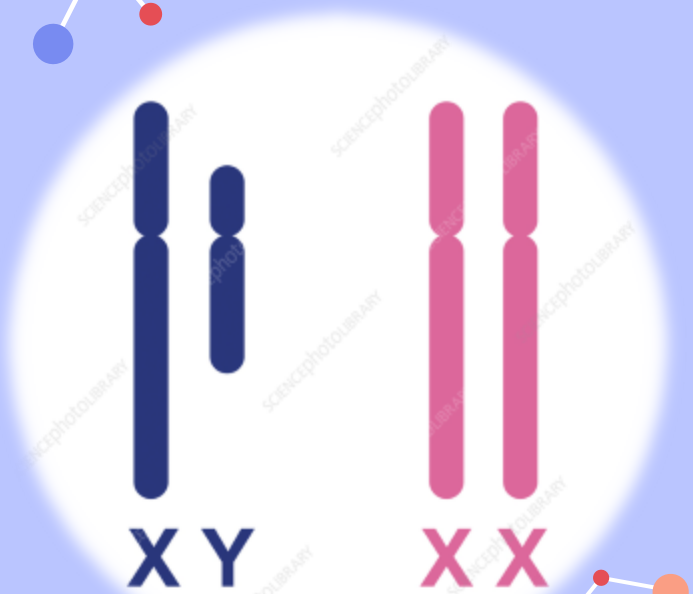
- Turner syndrome occurs when all or part of one X chromosome is missing
- Most common pattern: 45,X (monosomy X)
- Mosaic cases: 45,X / 46,XX (some cells normal, some missing X)
- Can also involve structural changes in the X chromosome
- Happens by chance during meiosis or early embryo cell division
- Not caused by anything parents did



Inheritance and How It Occurs

- Turner syndrome is usually not inherited
- Does not follow a Punnett square pattern like many single gene disorders
- Most cases are random, one time chromosome errors
- Parents of a child with Turner syndrome usually are not at higher risk in future pregnancies

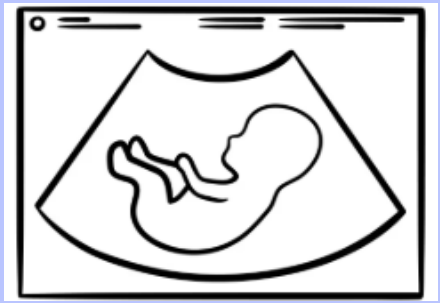




Risk Factors

- No known lifestyle or environmental risk factors
- Not strongly linked to older maternal age
- Main “risk” is being biologically female
- Most cases appear without any known cause

How Doctors Diagnose Turner Syndrome



- ❑ Prenatal clues: ultrasound may show cystic hygroma, swelling, or heart defects
- ❑ Prenatal tests: chorionic villus sampling (CVS) or amniocentesis with karyotype
- ❑ After birth: swelling of hands and feet, webbed neck, heart murmur, short height
- ❑ In childhood: slow growth, frequent ear infections, learning differences in math or spatial tasks
- ❑ In adolescence: no puberty or irregular periods

❑Confirmed by karyotype blood test

Treatment Options

1- No cure, but many treatments improve health and quality of life.

2- Growth hormone injections to increase final height

3- Estrogen and later progesterone therapy to start puberty and protect bones

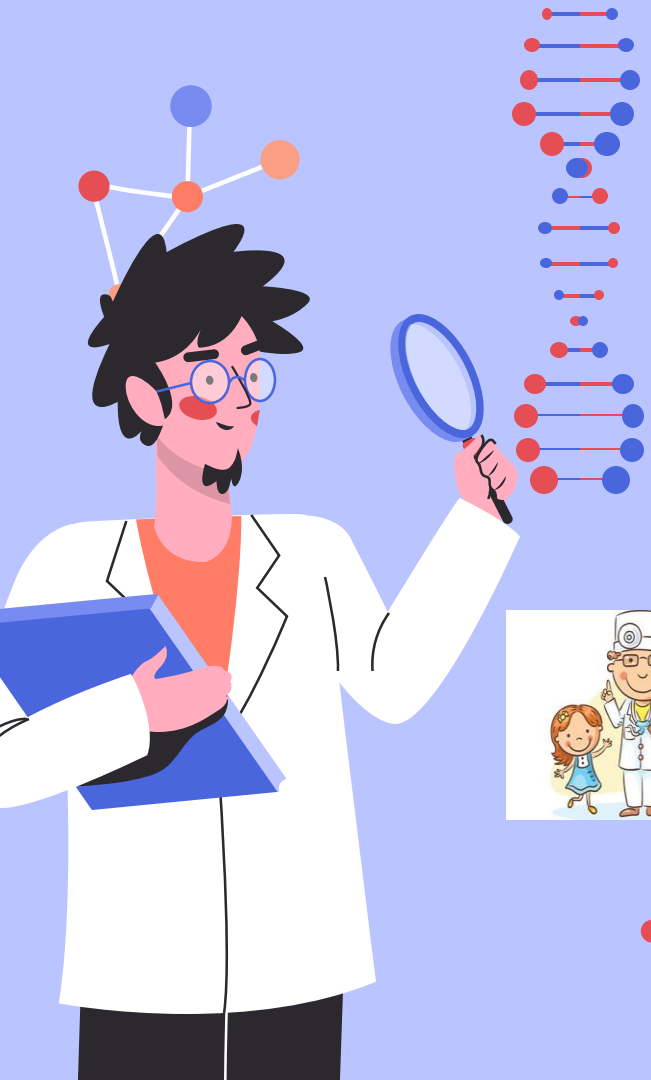
4- Regular heart, kidney, thyroid, hearing, and blood pressure checks

5- Calcium, vitamin D, and weight-bearing exercise for bone health

6- Fertility options such as IVF with donor eggs in selected patients

7- Counseling and school support for learning and emotional needs

Research and Support



- Turner Syndrome Foundation (TSF) and Turner Syndrome Society of the United States (TSSUS)
- Provide education, support groups, and advocacy
- Research on early heart detection and ways to protect the aorta
- Studies on best timing and dose of growth hormone and estrogen therapy
- Research on safe fertility options and pregnancy in Turner syndrome
- Long term registries help track health outcomes over time

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