

Turner syndrome

Turner syndrome is due to the deficiency of all or part of one X chromosome in females (45,X).

Epidemiology

- Incidence: 1/2500 live female births
- Sporadic

Genetics

- Most 45, X is due to loss of the paternal X chromosome
- Mosaic forms (42%) may have a milder phenotype
- Short stature in Turner syndrome is due to loss of 'growth' genes on the X chromosome

Clinical presentation

Turner syndrome has a very variable phenotype and presents with diverse clinical features in different age groups:

Pre-natal

- Miscarriage
- Increased nuchal translucency
- Hydrops fetalis

Newborn

- Oedema of hands and feet
- Congenital heart defect

Childhood

- Short stature

Teenage

- Primary amenorrhoea

Adulthood

- Infertility

Physical Signs

- Webbed neck
- Low hairline
- Wide-spaced nipples
- Oedema of hands and feet, especially in infancy
- Short 4th and 5th metacarpals
- Evidence of congenital heart disease: heart murmur, delayed femoral pulses, hypertension
- Short stature

Diagnosis

- Karyotype
- Echocardiogram
- Renal ultrasound scan
- Audiogram
- Complications
- Recurrent otitis media (60%)
- Congenital heart defect (15-50%): coarctation of the aorta, VSD.

- Renal anomalies (30%): horseshoe kidney, agenesis
- Autoimmune disease: hypothyroidism, diabetes mellitus
- Primary amenorrhoea, gonadal dysgenesis
- Infertility
- Osteoporosis (secondary to decreased oestrogen)
- Gonadoblastoma in the presence of a 46,XY cell line in mosaic Turner syndrome
- Intelligence is usually within normal range
- Life expectancy is reduced due to obesity, ischaemic heart disease and aortic dissection.

Treatment

- Removal of testicular tissue if any 46, XY cell line present
- Consider growth hormone therapy
- Commence oestrogens at time of puberty
- Ovum donation

Genetic Counselling

- The recurrence risk for parents of an affected child is negligible.

Surveillance:

- Hearing
- Growth
- Blood pressure
- Autoimmune disease
- Thyroid function
- Aortic root, especially in pregnancy by echocardiogram

Photos

Habitus

Puffy hands/feet

Metacarpals

coarctation