What is Turner Syndrome?

Turner syndrome (TS) was discovered in 1938 and defined as a chromosomal condition describing females* with common features, physical traits, and medical conditions caused by the complete or partial absence of the second sex chromosome. TS affects people differently, so each person has unique health needs and characteristics. Early diagnosis is vital to treat TS-related health issues and health maintenance is necessary throughout life.

*Although TS is defined as a female condition, our organization understands males with a 45 X cell line may be diagnosed with TS.

- Short Stature (under 5’)
- Unexplained delayed puberty/menstruation
- Cardiac issues & Kidney irregularities
- Non-functioning ovaries/reduced fertility
- Numerous moles
- Droopy eyelids, down-turned eyes, low-set, prominent ears, narrow roof of mouth
- Neck webbing
- Hearing loss (both conductive and sensorineural)
- Recurrent middle ear infections
- Hip dysplasia
- Upturned fingernails/toenails
- Educational and behavioral difficulties
- Slower Processing speed
- Repetitive thoughts
- Anxiety/Depression
- Excellent verbal skills/vocabulary
- Educational and behavioral difficulties
  - Slower Processing speed
  - Repetitive thoughts
  - Anxiety/Depression

Some Health Issues Related to Turner Syndrome

- SLOWER PROCESSING SPEED, REPETITIVE THOUGHTS, ANXIETY/DEPRESSION ARE ALSO COMMON.
- Scoliosis, osteopenia, and osteoporosis
- Celiac/Chron's Disease
- Reduced Fertility
- Educational Difficulties
- Hearing Loss
- Thyroid Disease
- Hypertension, Heart Abnormalities
- Kidney and Liver Concerns, Type I and II Diabetes
- Hip Dysplasia
- Lymphedema (swelling)

How Common is Turner Syndrome?

- Down Syndrome: 1 in 700
- Turner Syndrome: 1 in 2,000 - 4,000
- Cystic Fibrosis: 1 in 3.5k

TS can be diagnosed

- During fetal life
- In infancy
- During childhood
- During late pre-teen period (8-12 years)
- Late adolescence/adulthood.

A simple blood test is used to determine the karyotype (chromosome makeup).

Turner syndrome is a variable condition. Not every person will be affected in the same way or to the same degree.