



Free informational handouts for educators, parents, and students

Turner Syndrome

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Turner Syndrome, or TS, is a genetic syndrome that occurs in approximately one out of every 2,500 girls. This chromosomal condition is caused by a missing X chromosome. It was

discovered in 1938 by Dr. Henry Turner in the United States and is named after him.



TS is not caused by any actions of the mother before or during pregnancy. It is also not caused by a parent's ethnicity, age, diet, or any other factors. It occurs as a random event in the early stages of

fetal development.



TS can have many possible characteristics. It is variable, meaning each girl with TS will have unique symptoms, physical traits, and medical needs. The most common physical trait

of TS is short stature, which becomes noticeable by about age 5. It is common for girls with TS to take growth hormone injections to help them grow as tall as their bodies will allow. Other physical characteristics include down-turned eyelids, low-set and protruding ears, a small jaw, a high and narrow roof of mouth, a short and thick neck, delayed puberty, puffy hands and feet (lymphedema), turned up nails, and short roots of teeth. Health concerns can include reduced fertility, chronic middle ear infections, hearing loss, heart/liver/ kidney defects, autoimmune disorders (e.g. celiac disease or underactive thyroid), developmental delays, difficulty with spatial relationships (i.e. driving or riding a bike), difficulty with nonverbal communication skills, and difficulty with executive functioning. These symptoms and characteristics can vary depending on the unique genetic makeup of each girl with TS.



TS can be diagnosed during fetal life or at any time up into late adolescence and early adulthood. However, with increasing awareness in the medical community, the age of diagnosis is

decreasing. Diagnosis is made after a genetic blood test, called karyotyping. Usually, karyotyping alone is enough, but sometimes skin cells or cheek cells from a swab can be examined. These tests determine if one of the X chromosome pairs is missing the full or partial set.



TS is not necessarily treated by one doctor, but by specialists who can help with the individual concerns that may arise for each girl with TS. Treatment can include growth hormone therapy,

estrogen replacement therapy, heart surgery, hearing evaluations, in vitro fertilization, social and psychological testing, and connection to a strong social support system. These treatments can possibly come from a team of doctors, audiologists, psychologists, speech-language pathologists, and others who share similar experiences and concerns.

Resources:

"About Turner Syndrome," accessed January 27, 2020, https://www.turnersyndrome.org/about-turnersyndrome

Special Books by Special Kids. "Living with Turner Syndrome (Missing a X Chromosome)." YouTube. January 23, 2020. Accessed January 27, 2020. https://www.youtube.com/watch?v=DhypDYP3OI4

"Turner Syndrome," accessed January 27, 2020, https://ghr.nlm.nih.gov/condition/turner-syndrome#

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