Published on *My46* (https://www.my46.org) 01/20/2022

My46 Trait Profile

Turner syndrome

Turner syndrome is a genetic condition that affects females and is caused by a missing or incomplete X chromosome.

Characteristics of Turner syndrome

Turner syndrome is a genetic condition that affects approximately 1 in 2,500 females. Features of this condition can vary by system, but the most common concerns include short stature, learning disabilities, heart defects, and infertility. Short stature occurs in virtually all individuals with Turner syndrome (average adult height is 4 feet 7 inches). Individuals with Turner syndrome can have learning disabilities that can include issues with spatial perception and mathematics, while language skills can be normal. Individuals with TS can have heart defects and are evaluated accordingly, with some needing to be treated with surgery. Infertility is common in Turner syndrome, although spontaneous pregnancy, or pregnancy with the assistance of fertility specialists, can occur. Other medical issues can include kidney problems, hormone problems, high blood pressure, hearing loss, and vision issues.

Diagnosis/Testing

Turner syndrome is a chromosomal condition and is most often diagnosed by karyotype (chromosome analysis). Most individuals have two sex chromosomes, either two X chromosomes (most females) or one X chromosome and one Y chromosome (most males). Females with Turner syndrome have a single X chromosome and are missing all or part of the second sex chromosome in all or some cells in the body.

Management/Surveillance

Management of Turner syndrome requires a multidisciplinary approach to care as there are many differences in the type, severity, and number of medical problems individuals with Turner syndrome can have resulting in a difference in the treatments required. Ongoing surveillance of current or possible medical issues is important. Management includes blood draws to monitor specific chemical levels in the blood (like hormone levels and indicators of kidney function), imaging to view the heart and kidneys, monitoring of blood pressure, and appropriate educational supports and evaluations. Hormone replacement therapy (e.g., estrogen to promote normal sexual development; growth hormone to treat short stature) is often recommended.

Although infertility is common in Turner syndrome some women are able to get pregnant. For women who do become pregnant, close cardiology involvement is recommended throughout the pregnancy and shortly after delivery. Adult women with Turner syndrome should be monitored for the development of osteoporosis, high blood pressure, and diabetes.

Testing for Y chromosome material should be done in any individual with Turner syndrome. For individuals that have some Y chromosome material, abdominal ultrasounds are important since there is an increased risk of gonadoblastoma (a type of tumor) and surgery could be recommended.

Mode of inheritance

Turner syndrome is not inherited, but rather is a de novo or chance occurrence.

Risk to family members

For parents of a child with Turner syndrome, the risk to have another child with Turner syndrome is not increased.

The chance of having another child with Turner syndrome has no relationship to the mother's age.

Special considerations

None

Resources

Turner Syndrome Society of the United States
http://turnersyndrome.org/
Genetics Home Reference: Turner syndrome
http://ghr.nlm.nih.gov/condition/turner-syndrome
Turner – know your body! (Editor: CH Gravholt, PhD, MD)
http://np.netpublicator.com/netpublication/n75088268
Medical Home Portal: Turner syndrome
http://www.medicalhomeportal.org/diagnoses-and-conditions/turner-syndrome/description

References

Bondy, CA. et al. (2007)."Clinical Practice Guideline: Care of girls and women with Turner syndrome: a guideline of the Turner Syndrome Study Group." Journal of Clinical Endocrinology and Metabolism 92(1): 10-25.
Davenport, ML. (2010)."Approach to the patient with Turner syndrome." Journal of Clinical Endocrinology and Metabolism 95(4): 1487-1495.
Frias, JL. et al. (2003)."Health supervision for children with Turner syndrome." Pediatrics 111(3): 692-702.

Created: 04/2013 **Created by:** Tomi Toler, MS, CGC
Updated: mm/yyyy **Edited by:** Seema Jamal, MSc, LCGC