**Genetics and Genomics**

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**Identify a genetic disease or disorder.**

Turner syndrome is a genetic disorder that is near and dear to my heart because I was diagnosed with Turner Syndrome. It is a chromosomal problem that affects development in females, only disrupting the X chromosome. Within the genetic makeup of a female, typically both X chromosomes are perfectly structured; however, in people like me with Turner Syndrome, one X chromosome is just as it should be, but the other X is either altered in some way or missing completely (Mayo Clinic, 2022). Mosaic Turner Syndrome is the type that I have, in which some of the cells contain partial X chromosomes and the other cells are absent of an X chromosome. There are several variations of the disease, so severity and presentation differ among those with this type of genetic makeup.

**Identify all signs and symptoms of this disease or disorder.**

Turner Syndrome affects the development of the entire body, so signs and symptoms are very broad and unique. Heart problems and congenital defects, such as issues with the aorta and valves, increase the risk of serious health conditions (Mayo Clinic, 2022). As an example, I have a bicuspid aortic valve, meaning that instead of my aortic valve having the normal three segments, mine has two, causing regurgitation. Hearing loss is common due to gradual nerve function loss or constant middle ear infections from an undeveloped eustachian tube (Mayo Clinic*,* 2022). I have had to have three sets of tubes placed in my eardrums due to infections to prevent me from completely losing my hearing. Skeletal issues and problems with the growth and development of the bones often occur, such as spinal curvatures and brittle bones (Mayo Clinic*,* 2022). A DEXA scan discovered that I have osteopenia, a precursor to osteoporosis which is the classic diagnosis for weak and brittle bones. Short stature is another common sign, as I am only four feet and ten inches tall. A high palate and recessed jaw are usual occurrences (Mayo Clinic, 2022). I had an overbite that caused me to need a complete upper and lower jaw reconstruction at age fifteen. I also have a flat foot, or pes planus, which is a normal skeletal sign of Turner Syndrome (NIH, *Turner syndrome - about the disease,* n.d.). Vision problems, such as strabismus and nearsightedness are seen; as an example, I am nearsighted and have astigmatism with cataracts. Autoimmune disorders are typical, such as dermatographia like I have where my overactive mast cells cause hives to develop from things like pressure and heat (Mayo Clinic, 2022). Learning disabilities with spatial concepts, math, and attention can be present, although most women with Turner Syndrome have normal intelligence. Math has been a struggle for me all my life. Mental health can also be challenging for those diagnosed with Turner Syndrome, especially in social situations (Mayo Clinic, 2022). I know I have social anxiety and can become easily depressed over small things that happen. Infertility is a touchy symptom of Turner Syndrome, as only a very small number can become pregnant with the help of fertility treatment (Mayo Clinic, 2022). I have been diagnosed as infertile, and even if I were to become pregnant, I would be at high risk of pregnancy complications, such as an aortic dissection. Hyperconvex fingernails, as I have, are a classic symptom, which is where the fingernails grow upwards in a curve instead of straight (NIH, *Turner syndrome - about the disease,* n.d.). Atypical scarring is common, such as keloids as I tend to develop when my skin gets cut. I also have a low posterior hairline, which is a sign of this genetic disorder (NIH, *Turner syndrome - about the disease,* n.d.). Other signs and symptoms of Turner Syndrome, which I do not have, include a horseshoe-shaped kidney, high blood pressure, webbed neck, flat chest; missing or hyperactive joints, hip dysplasia, celiac disease, obesity, low-set ears, a broad chest with widely spaced nipples, high gonadotropin levels, and a wide thorax (NIH, *Turner syndrome - about the disease,* n.d.). Before birth, a large collection of fluid might be seen on the back of the neck in an ultrasound.

**How is this disease or disorder diagnosed?**

Diagnosing Turner Syndrome is extremely difficult, as it is a very rare condition. It starts with talking to the primary care provider and getting a genetic lab workup if the signs and symptoms are present (Mayo Clinic, 2022). A buccal smear or skin sample could be requested as well (Mayo Clinic, 2022). Prenatally, if the mother is at risk of bearing a child with chromosomal abnormalities, a cell-free DNA screening can be performed using a blood sample from the mother to see if Turner Syndrome is present (Mayo Clinic, 2022). Chorionic villus sampling between eleven and fourteen weeks involves taking a small piece of tissue from the developing placenta to assess the genetic composition of the baby (Mayo Clinic, 2022). Amniocentesis after fourteen weeks is another possibility, which involves taking a sample of amniotic fluid from the uterus and assessing the genetics of the fetus (Mayo Clinic, 2022). To diagnose and treat specific conditions that underlie Turner Syndrome, specialists such as endocrinologists, cardiologists, nephrologists, and reproductive specialists make up the treatment team.

**Identify the most effective treatment for this disease or disorder. Identify any experimental treatments currently available.**

Turner Syndrome has many ways in which it affects the body, so treatment is specified to the symptoms that are currently present. Growth hormone therapy is a primary option for treating this genetic disorder. It is typically given as a daily injection of recombinant human growth hormone to increase height and bone growth (Mayo Clinic, 2022). This, however, was not a possibility for me as I was diagnosed later in my adolescent years. Growth hormones do not work effectively after the bone plates have fused and puberty has occurred. Estrogen therapy, which I use, is prescribed to promote breast development and enhance the size of the uterus (Mayo Clinic, 2022). It also assists bone mineralization. Therapy continues until the typical age of menopause (Mayo Clinic*,* 2022). Other treatments for Turner Syndrome involve specialists; for example, I have a cardiologist watching my bicuspid valve and heart function in case I need an aortic valve replacement, I have a fertility specialist treating my low egg count and post-menopausal reproductive system, I have an ENT working on my hearing, I have an ophthalmologist monitoring my cataracts, and I have an orthodontist and dentist working together to make sure my teeth and jaw remain properly aligned.

**What are the causes or risk factors related to this disease or disorder?**

Females inherit an X chromosome from each parent, but in Turner Syndrome, one X is missing or changed in some way. Monosomy is an X completely missing, mosaicism is an error in the cell division causing some copies to have both X chromosomes and others to only have one, and in some cases, there is one copy of the X chromosome with some Y chromosome material (Mayo Clinic, 2022). The loss or change within the X chromosome occurs randomly from an issue with the sperm or egg or with no explanation during early fetal development (Mayo Clinic, 2022). Inheritance does not seem to be a risk factor.

**Who is most commonly affected by this disease or disorder?**

Turner Syndrome is not a well-known disorder as fewer than 50,000 women in America have been diagnosed with it. It only affects approximately one in two thousand five hundred births (NIH, *How many people are affected or at risk of Turner Syndrome?*, n.d.). Only females are at risk of Turner Syndrome. There are no known environmental risk factors, such as race or ethnic factors that play a role in the development of Turner Syndrome (Crenshaw & Bondy, 2019).

**Resources**

Crenshaw, M. & Bondy, C., NORD (National Organization for Rare Disorders). (2019, March 1). *Turner syndrome*. Retrieved September 26, 2022, from https://rarediseases.org/rare-diseases/turner-syndrome/

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