Turner Syndrome in Pediatrics

NATIONWIDE CHILDREN'S®

When your child needs a hospital, everything matters.

What is Turner Syndrome?

Turner Syndrome results from a chromosomal abnormality in children who are assigned female at birth. "Classic" Turner Syndrome results from a complete loss of an X chromosome in all cells (karyotype 45,X). Other variations exist including a "ring" X chromosome, missing only a part of an X chromosome, or mosaicism in which some cells may have two copies of the X chromosome but others do not. Turner Syndrome from any of these causes can result in a wide range of possible conditions that could affect the patient's health if untreated, and that may develop over their lifetime.

Identifying Turner Syndrome

Children assigned female at birth with a completely missing X chromosome tend to present with the "textbook" Turner Syndrome phenotype and may be more easily identifiable. Some characteristic features may include:

- Shield-shaped chest
- Widely spaced nipples
- Webbed neck
- Edema of hands/feet
- Lowset hairline
- Short stature

Mosaic Turner Syndrome results in a wide range of phenotypes and may not include all of the characteristic features listed above. On their own, many of the following conditions are common in children without Turner Syndrome. This can make Turner Syndrome difficult to identify and diagnose. However, when taken in whole, the following constellations of characteristics are reasons a primary care pediatrician might order diagnostic blood work:

- Infant: lymphedema, coarctation of aorta, hypoglycemia
- Toddler to early elementary years: learning disabilities, hearing difficulties or recurrent acute otitis media, short stature (height below the 3rd percentile, or significantly shorter than midparental height estimate)
- Adolescent/teenage years: delayed or absent puberty (such as absence of breast development by 13 years or no menses by 15 years), secondary amenorrhea, premature ovarian insufficiency.

How is it diagnosed?

Turner Syndrome is diagnosed by karyotype through a simple blood test. As cell-free fetal DNA and other prenatal diagnostic testing has become more advanced and more common, Turner Syndrome is being diagnosed prenatally more often. However, diagnosis via karyotype can happen at any age, and many children are still being diagnosed later in life.

Post Diagnosis

Once a diagnosis of Turner Syndrome has been confirmed, patients should be screened for possible associated conditions and complications. These may include cardiac abnormalities (such as bicuspid aortic valve and hypertension), renal anomalies (such as horseshoe kidney), autoimmune diseases (such as hypothyroidism, diabetes and celiac disease), and learning difficulties. While it's sometimes necessary to refer patients to individual subspecialists, Nationwide Children's Hospital Turner Syndrome Clinic brings many subspecialties together to comprehensively care for children with Turner Syndrome in one place (see next page). This clinic can also help facilitate referrals to other subspecialties not yet included in this clinic, such as cardiology.

Endocrinology and gynecology screenings are time sensitive for children with Turner Syndrome in order to optimize outcomes. If needed, pubertal induction and growth can be assisted with hormone replacement therapy.

While many conditions associated with Turner Syndrome can be successfully managed with treatment, fertility loss is not one that can currently be reversed. For patients with Turner Syndrome who desire pregnancy and maintain fertility, or who choose assisted reproductive technology, pregnancy still carries a high cardiac risk. Nationwide Children's Hospital Turner Syndrome Clinic can provide nuanced counseling about safe family building. Genetic counseling and psychologic screening for patients with Turner Syndrome and their families are also provided.

With early diagnosis and comprehensive treatment, children with Turner Syndrome can look forward to full and healthy lives.

Nationwide Children's Hospital Turner Syndrome Clinic

Nationwide Children's Hospital is excited to announce the start of a new multidisciplinary clinic for patients with Turner Syndrome. This clinic will provide comprehensive care for children of all ages with Turner Syndrome including:

- Monitoring and treatment of growth and pubertal development
- Genetic counseling
- Gynecology care and fertility discussion
- Anxiety/depression screening
- Hearing screening
- Opportunities to participate in research to improve clinical care

Current subspecialists in the Turner Syndrome Clinic include:

- Endocrinology
- Gynecology
- Genetics
- Nephrology
- Psychology
- Audiology
- Social Work
- Specialty Pharmacy

Turner Syndrome Clinic Leadership

Jennifer M. Ladd, MD, MSc
Endocrinology

Chelsea A. Kebodeaux, MD
Pediatric and Adolescent Gynecology

To learn more or to refer a patient, contact our team at (614) 722-4425 or visit NationwideChildrens.org/Turner-Syndrome-Clinic

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