The Basics About Mosaic Turner Syndrome

This handout will explain Mosaic Turner Syndrome (TS), a genetic condition that affects growth and development in women and girls.

WHAT IS TURNER SYNDROME?

Our bodies are made up of millions of cells. Typically, each cell has 46 chromosomes. Each chromosome is a package of DNA, which contains our genetic information. Our DNA contains the instructions for our bodies’ growth and development.

Turner syndrome (TS) is a rare genetic condition in which a girl or woman doesn’t have the usual pair of 2 X chromosomes. The cause is a missing or incomplete X chromosome (the chromosome that determines a person’s sex before birth). The missing gene prevents the body from growing and developing normally.

TS affects only women and girls and affects every woman or girl differently. In TS, only some of the cells in the body are missing an X chromosome. About half of girls with TS are “typical.” This means all of their cells are missing an X chromosome.

WHAT IS MOSAIC TURNER SYNDROME?

Mosaic Turner syndrome (TS) is a condition in which cells inside the same person have different chromosome packages. Mosaic TS can affect any cell in the body. Some cells have X chromosomes and some don’t. Every 3 out of every 10 girls with TS will have some form of Mosaic TS.

Did you know?

Turner syndrome and Mosaic Turner syndrome are not anyone’s fault. They don’t happen because of anything the parents did or didn’t do. The loss or change of the X chromosome happens very early in pregnancy by chance.

SIGNS OF TURNER SYNDROME AND MOSAIC TURNER SYNDROME

Signs of Turner syndrome and Mosaic Turner syndrome can be similar:

- Height usually under 5 feet
- Droopy (heavy) eyelids (ptosis)
- Differences in ear shape and position
- Webbed neck (extra skin)
- Arms or feet that are puffy (lymphedema)
- Often, broad chest
- High-arched roof of the mouth
- Teeth that are crowded
- Trouble with certain types of math
- Learning difficulties in school
- Reduced fertility
- Delayed or absent periods

Women and girls with Mosaic TS tend to have fewer signs and health problems than those with typical TS. This is because only some cells are missing the second X chromosome in Mosaic TS. In typical TS, all of the cells in the body are missing the second X chromosome.

RESEARCH GOALS AT MGHFC

More information is needed to care for women or girls with TS or Mosaic TS. We will conduct research at the Turner Syndrome Clinic at MassGeneral Hospital for Children to help us answer important questions. Please speak with us to learn more about research opportunities.