Gastrointestinal (GI) Series for Individuals with 22q11.2 Differences

Inguinal Hernias

An hernia happens when a part of the small intestine pushes through a weak area of the abdominal muscles. An inguinal hernia (or groin hernia) is one that protrudes along the inguinal canal, which is the pathway that runs along each side of the pelvis to the sex organs. Inguinal hernias may be felt or be visible as a bulge. They may cause serious complications if not repaired by surgery.

Symptoms

- Bulge under the skin in the groin or scrotum
 - May appear and disappear
 - May get bigger when straining and smaller when relaxed
 - Pain / discomfort / heaviness / pressure / swelling in the groin
- · Crankiness or difficulty feeding

Diagnosing and treating a hernia

A healthcare professional may diagnose a hernia through a physical exam and imaging. The treatment for hernia is surgery.

Incarcerated and strangulated hernias

- Sometimes the contents of the hernia are **incarcerated** or trapped in the abdominal wall, leading to a bowel obstruction (digested food cannot move forward in the intestines).
- An incarcerated hernia can be come strangulated meaning that the blood flow to the trapped tissue becomes cut off. The affected part of the intestine can die.
- A strangulated hernia that is untreated can be life-threatening.
- If these symptoms for incarcerated/strangulated hernia are present,
 seek medical help immediately:
- Nausea and/or vomiting
- Sudden pain that gets worse quickly
- The hernia bulge is red, purple, or dark
- Fever
- Not able to pass gas or have bowel movement

References / Resources

Clinical practice recommendations and research reports

- Updated clinical practice recommendations for managing children with 22q11.2 deletion syndrome – 2023
- Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome – 2023
- Gastrointestinal Features of 22q11.2 Deletion Syndrome Include Chronic Motility Problems From Childhood to Adulthood – 2022

Hernias and 22q11.2DS

In a GI study of 206 adults with 22q11.2DS, 21% had surgery to repair hernias (any type). Hernias that are common in 22q11.2DS include:

- Diaphragmatic an organ in the abdomen pushes up through the diaphragm (the muscle that separates the chest from the abdomen)
- Umbilical an organ in the abdomen pushes outward near the belly button.
- Inguinal an organ in the abdomen pushes outward near the groin.

Direct and indirect inguinal hernia

A **direct hernia** forces through the wall of the inguinal canal. It is usually caused by long-term pressure on or the weakening of the abdominal muscle, and can happen at any point in life.

An **indirect hernia** slides into the inguinal canal from an opening at the top. It happens in newborns if the opening of the inguinal canal did not close all the way when the fetus was developing inside the uterus. It is possible for an inguinal hernia not to be found until adulthood.

Websites of medical institutions and foundations

- Inguinal Hernia (Groin Hernia) | Inguinal Hernia in Babies Cleveland Clinic
- Inguinal Hernia Mayo Clinic
- Inguinal Hernia Johns Hopkins Medicine



The mission of the <u>International 22q11.2 Foundation</u> is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.

This information is brought to you by the Foundation for educational purposes only. It is <u>not</u> intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.