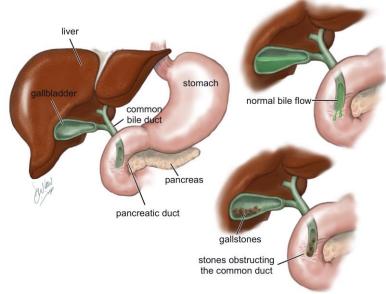
## Gastrointestinal (GI) Series for Individuals with 22q11.2 Differences

## **Cholelithiasis (Gallstones)**

**Cholelithiasis** is the condition of having gallstones. **Gallstones** are little "pebbles" of hardened bile (a digestive fluid) in the gallbladder. Most gallstones do not cause any problems as long as they stay in the gallbladder. However, if they move around, they may cause blockages with serious consequences.

#### Bile and the Gallbladder

- Bile is a digestive liquid that breaks down fats into fatty acid.
  - Consists of water, cholesterol, fats, bile salts (also called bile acids), and bilirubin (a yellow pigment)
  - Made by the liver and stored in the gallbladder
- The gallbladder is a small organ that sits below the liver on the right side of the abdomen. It stores and concentrates bile.
- When you eat, your gallbladder contracts and pushes the bile into the common bile duct. The bile flows into the small intestine to break down fats. The pancreas also uses part of the bile duct to deliver its digestive juices to the intestines.



Left: The location of the gallbladder relative to the liver and pancreas. Top right: Normal flow of bile from the gallbladder to the intestine. Bottom right: Gallstones blocking the duct that carries bile.

#### **Gallstones**

- · Mostly caused by excess cholesterol or bilirubin
- Can also form if bile is left in a gallbladder that contracts poorly
- Start off as collections of cholesterol or bilirubin that sits at the bottom of the gallbladder, but gradually harden into "stones" and get bigger as liquid bile deposits more materials on them
- Usually do not cause any problems if they stay put

If gallstones move out of the gallbladder, they may get stuck at the outlet of the gallbladder or inside the bile duct. These blockages can build up pressure, causing severe pain.

### **Decreasing the risk of gallstones**

To reduce the risk of cholesterol gallstones:

- Eat less fried food.
- Choose mostly plant-based foods and oils
- If obese, lose weight slowly (1-2 lb per week).

## Symptoms of Blockages due to Gallstones

- **Biliary colic** a dull but severe pain in the abdomen's upper right area, lasting one to several hours. This often happens when the gallbladder contracts to release bile after a large meal.
- Pain vaguely located somewhere in the abdomen
- Pain in the right shoulder or arm
- · Nausea or vomiting
- Fever, chills, and high heart rate (if there is acute inflammation)
- **Jaundice** (yellowing of the skin and the whites of the eye) if the yellow pigment bilirubin has entered the blood stream

Get medical help right away if the abdominal pain is severe, the skin or whites of the eyes are yellow, or you have a high fever.

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

## **Cholelithiasis (Gallstones; continued)**

## **Checking for Gallstones**

- Abdominal ultrasound The technician puts a tool on the surface of the tummy. The probe uses high frequency sound waves to take pictures of the inside to check for problems.
- Magnetic resonance cholangiopancreatography (MRCP) –
   The doctor uses large magnets to generate pictures of your tummy area, specifically checking the bile duct.
- Endoscopic retrograde cholangiopancreatography (ERCP) –
   See the next section on Removing Gallstones
- Blood tests may show the complications caused by gallstones.
   E.g. infection, jaundice, and pancreatitis.

### **Complications from Gallstones**

If untreated, the pressure resulting from the blockages due to gallstones can lead to severe inflammation of the gallbladder, liver, pancreas, and the bile duct. You will also have trouble absorbing nutrients from food.

### **Removing Gallstones**

- There are medications that can potentially dissolve gallstones, but they are often inefficient at resolving symptoms.
- Removal of gallstones from the common bile duct using <u>Endoscopic retrograde cholangiopancreatography (ERCP)</u>
  - A long tube is inserted from the mouth down the digestive tract. The tip of the tube is a tiny camera that allows the doctor to check the interior of the digestive tract. A smaller tube is inserted inside the first tube to deliver a dye that lights up under x-ray and tools to remove gallstones in the bile duct.
- · Surgical removal of the gallbladder
  - Removal of the gall bladder is necessary when gallstones cannot be removed via ERCP, patients have gallbladder related symptoms and patients have a chance of re-formation of gallstones. Also, once a gallstone causes blockage, there is a chance of this happening again. Therefore, the entire gallbladder needs to be removed by surgery.
  - The surgery can be a minimally invasive one (<u>laparoscopic</u> <u>cholecystectomy</u>) or an open surgery.
  - After the surgery, you may need to eat a diet with lower fats.
     The digestive system can still function normally after the removal of the gallbladder.

#### **How Common are Gallstones?**

Many people who have gallstones may not know they have them. It is estimated that 10% of adults and 20% of those over 65 years old have gallstones. Women are more at risk of developing gallstones because of their hormones and the way their body deals with excess fat. Gallstones happen in adults with 22q11.2DS but it is not very common.

#### **Need info on Gallbladder Polyps?**

**Gallbladder polyps** are abnormal growth of cells on the inner lining of the gallbladder. They are <u>not</u> the same as gallstones and are <u>not</u> the topic of this info sheet. See the <u>gallbladder polyps</u> <u>webpage</u> from the Cleveland Clinic.

#### References / Resources

# Clinical practice recommendations and research reports

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

   2022

## Websites of medical institutions and foundations

- Gallstones Cleveland Clinic
- Gallstones Mayo Clinic
- Gallbladder and Gallstones GI Society / Canadian Society of Intestinal Research



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.