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

GI Issues in 22q11.2 Deletion Syndrome

Gastrointestinal (GI) problems are common among individuals with 22q11.2 deletion syndrome (22q11.2DS). About 91% of individuals with 22q11.2DS experience long-term GI symptoms some time in their lives. Autoimmune diseases and birth defects related to the GI system are rarer.

GI conditions in 22q11.2DS vary in types, severity, and the way they affect the quality of life of the patient. Different individuals often have different experiences. Many GI issues that are problematic in childhood get better as the child grows up, but some symptoms persist or can even start in adulthood.

Topics covered in this GI Series

This series provides information on many of the GI conditions that affect individuals with 22q11.2DS.

- GI Issues in 22q11.2 Deletion Syndrome (this sheet)
- The Digestive System (Gastrointestinal Tract)
- Feeding Difficulties
- Swallowing and Dysphagia
- Constipation
- Gastroesophageal Reflux Disease (GERD)
- Esophageal Dysmotility
- Gastroparesis
- Cholelithiasis (Gallstones)
- Nausea and Vomiting
- Autoimmune Issues in the GI System
- Inguinal Hernias
- Nonalcohol Fatty Liver Disease (NAFLD)

GI Conditions and 22q Differences

Only a few studies that systematically look at GI problems in individuals with 22q11.2 deletion syndrome (22q11.2DS). The largest and most detailed one examined the GI-related medical records of 206 patients aged at least 17 years.

Gastrointestinal Features of 22q11.2 Deletion Syndrome Include Chronic Motility Problems From Childhood to Adulthood

As of February 2024, there are no known GI studies related to 22q11.2 duplication syndrome (22q11.2DupS).

Recommendations for GI-Related Routine Assessments

Assessments and Management (children)	At Diagnosis	Annual/Biennial	0-1y	1-5y	6-12y	13-18y
• Physical examination	☑	☑	☑	☑	☑	☑
• Nutritional assessment, feeding, swallowing, GERD, constipation, and growth	☑	☑	☑	☑	☑	☑
• Dental evaluation (measure saliva secretion rate from 6 years)				☑	☑	☑

Assessments and Management (adults)	At Diagnosis or Initial Assessment	At Follow-up (Every 1-2 years)
• Comprehensive history-taking (including family history), systems review, and medication review	☑	☑
• Nutritional assessment; diet and exercise counseling	☑	☑
• Body mass index (BMI), resting heart rate, blood pressure	☑	☑
• 22q11.2DS-relevant laboratory tests	☑	☑
• Abdominal ultrasound	Perform if not done in recent years, or if gallstones or fatty liver may be happening	
• Dental assessment	☑	☑
• Gastroenterology	☑	☑

References / Resources

- 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

22q International 22q11.2 Foundation

The mission of the International 22q11.2 Foundation 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 not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.

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