

Gastrointestinal Health of People with Down Syndrome, Part 2 From a Primary Care Perspective

Brian Chicoine, MD

Medical Director, Advocate Medical Group Adult Down Syndrome Center
Faculty, Family Medicine Residency, Advocate Lutheran General Hospital

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Please note:

- This presentation is intended for families, caregivers, health care professionals, and service providers of individuals with Down syndrome.
- The information in this presentation is provided for educational purposes only and is not intended to serve as a substitute for a medical, psychiatric, mental health, or behavioral evaluation, diagnosis, or treatment plan by a qualified professional.
- We recommend that you bring specific questions about an individual with Down syndrome to their medical and/or therapy providers.

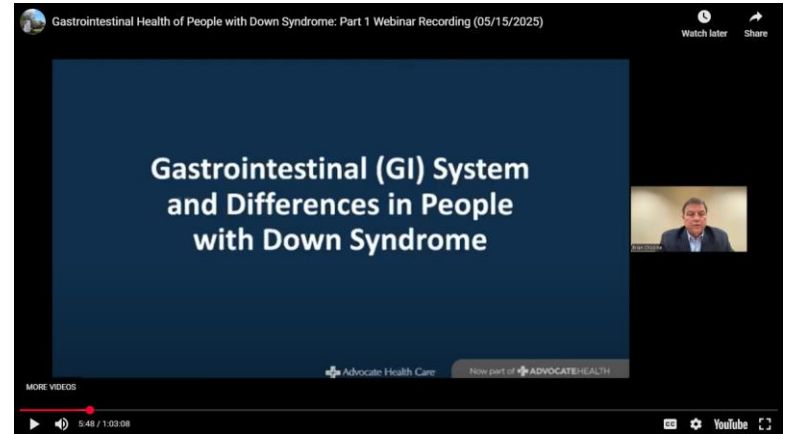


Brian Chicoine, MD

Outline

Part 1

- Introduction
- Differences in the GI tract of people with Down syndrome
- How to optimize GI health
- Common GI symptoms
- GI testing



[Recording of Part 1](#)

Outline

Part 1

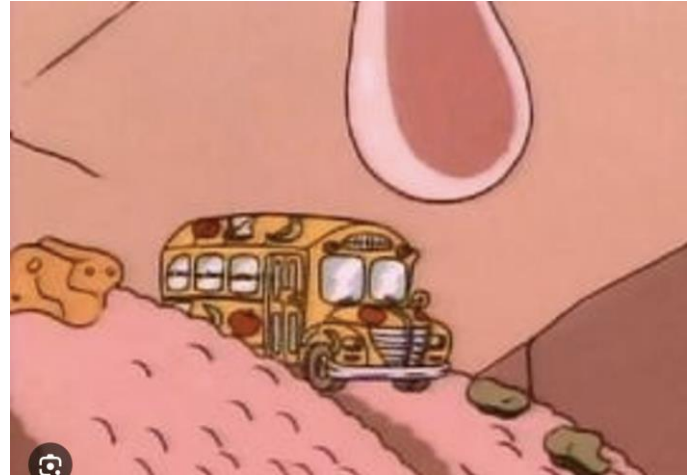
- Introduction
- Differences in the GI tract of people with Down syndrome
- How to optimize GI health
- Common GI symptoms
- GI testing

Part 2

- A trip through the GI tract
 - Swallowing dysfunction
 - Celiac
 - Inflammatory bowel disease
 - Constipation
 - And several others



Fantastic Voyage (1966)
image from mptvimages.com



Magic School Bus (2015)
Image from Daily Motion

GI Conditions

Treatment

For each condition, in addition to the treatment discussed, we encourage the recommendations discussed in Part 1 regarding optimizing GI health.

Mouth and throat

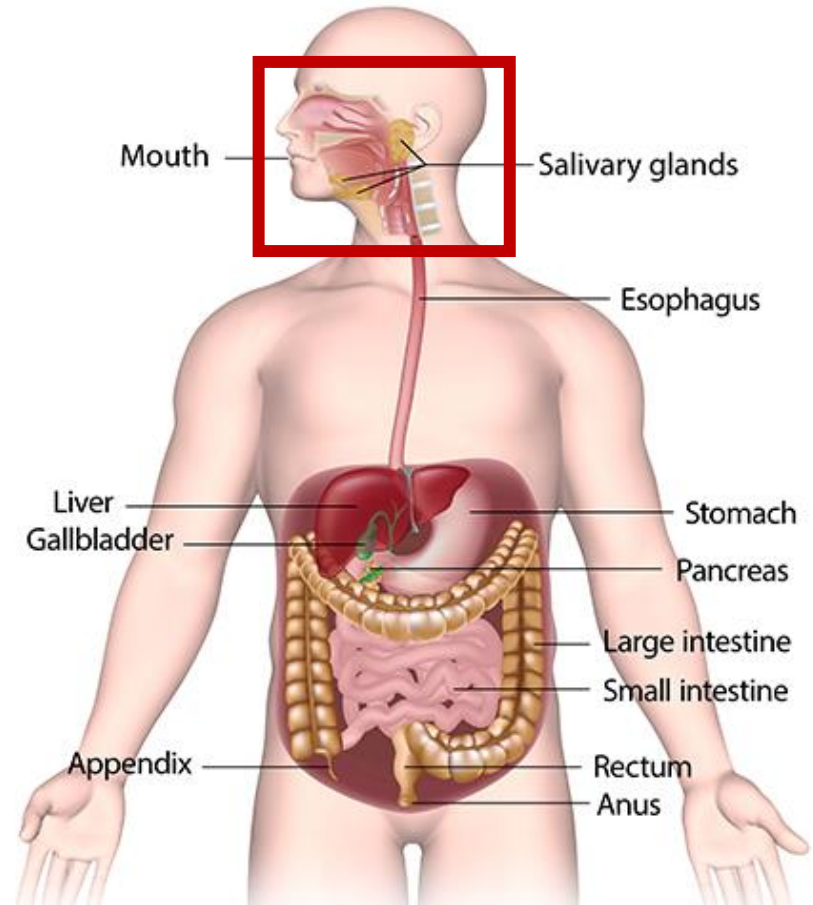


Image from the [National Institute of Diabetes and Digestive and Kidney Diseases](#)

Dental disease

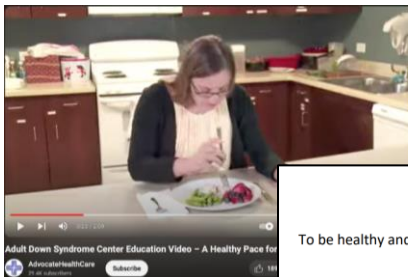
- Malocclusions
 - Missing teeth
 - Gum disease
 - Difficulty chewing

 - Cavities – less common
- Prevention and Treatment
 - Dental hygiene
 - Dental appointments
 - Braces?

Dysphagia





- Swallowing dysfunction
- Causes
 - Differences in anatomy and physiology
 - Eating too fast
 - Not chewing well
 - GERD, esophageal conditions
- Symptoms
 - Choking
 - Gagging
 - Eating refusal or eating selectivity
 - Aspiration pneumonia
- Diagnosis
 - Speech therapy
 - Video swallow study
 - Esophagram
 - Upper endoscopy (EGD)
- Treatment
 - Eat slower, chew thoroughly
 - Treat underlying conditions
 - Swallowing therapy
 - Diet change
 - Feeding tube (e.g., PEG)

Resources











Healthy Pace for Eating

To be healthy and enjoy my food, I should...

 <p>Take a bite.</p>	 <p>Put the fork down.</p>	 <p>Chew 5 to 10 times.</p>	 <p>Swallow the food.</p>	 <p>Take a small drink.</p>	 <p>Pick up your fork and start all over.</p>
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[Healthy Pace for Eating Video & Visual](#)

Getting a Swallow Study

<p>My doctor wants me to get a swallow study.</p> 	<p>It will show the doctor where my food and drinks go after I put them in my mouth.</p> 	<p>A swallow study takes pictures of me eating and drinking.</p> 	<p>I will sit in a chair while I eat and drink.</p> 
<p>There will be other people in the room while I eat.</p> 	<p>A family member or caregiver will be right next to me.</p> 	<p>I can watch a video or listen to music if that will help!</p> 	<p>It will not hurt.</p> 

[Getting a Swallow Study Story](#)

Esophagus

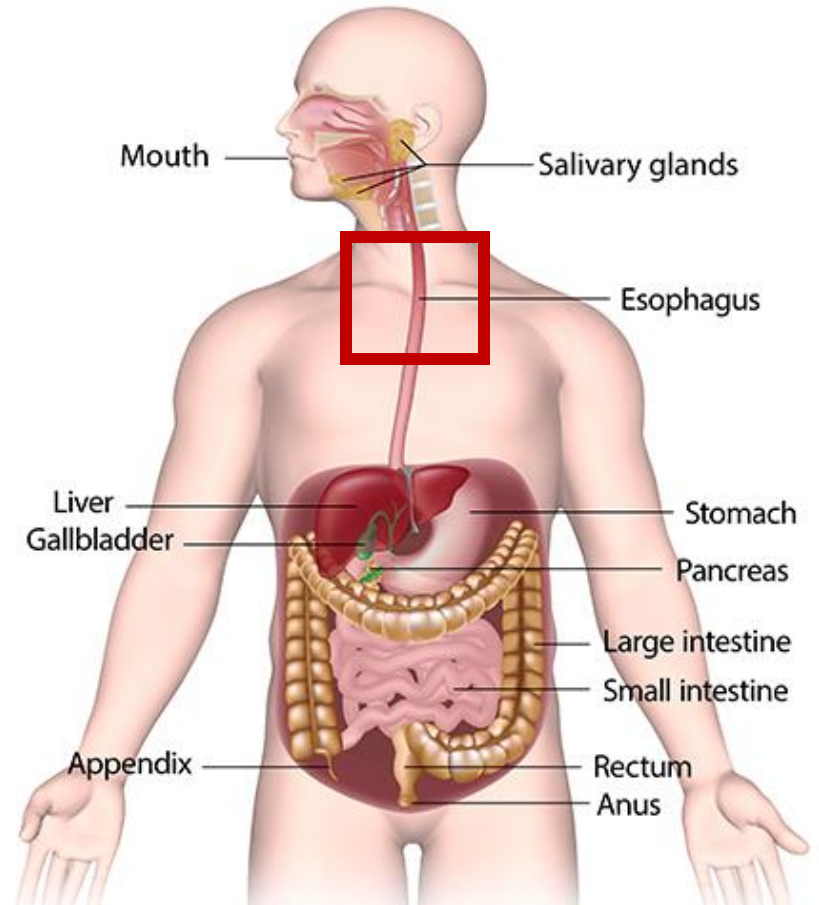


Image from the [National Institute of Diabetes and Digestive and Kidney Diseases](#)

Achalasia

- Improper contraction of muscles in esophagus and sphincter
- Causes
 - Genetic
 - Autoimmune
 - Infectious (viral)
- Symptoms
 - Difficulty swallowing
 - Regurgitation
 - Chest pain
 - Weight loss
- Diagnosis
 - Esophagram
 - Upper endoscopy (EGD)
 - Manometry
- Treatment
 - Balloon dilatation
 - Botox injections
 - Surgical myotomy

Gastroesophageal reflux disease

- Also referred to as GERD, heartburn, laryngopharyngeal reflux
- Backward flow of stomach acid into the esophagus
- Risk factors
 - Obesity
 - Sleep apnea
 - Overeating
 - Consuming caffeine, spicy foods, carbonated beverages
 - Lying down after eating
- Symptoms
 - Chest pain
 - Sore throat
 - Regurgitation of digested food
 - Abdominal pain
 - Hoarseness
 - Bronchospasm (asthma)
 - Difficulty swallowing
 - Aspiration (breathing food into the lungs)
 - Chronic cough
 - Loss of enamel on the teeth

Gastroesophageal reflux disease

- Possible complications
 - Barrett's esophagitis
 - Scarring of the esophagus
- Diagnosis
 - Clinical diagnosis
 - Upper endoscopy (EGD)
 - Upper GI (barium)
- Treatment
 - Avoid overeating
 - Reach/maintain ideal body weight
 - Limit caffeine and carbonated drinks
 - Avoid lying down for an hour (or more) after a meal
 - Avoid clothing that constricts the abdomen
 - Elevate the top of the bed
 - Treat sleep apnea

Gastroesophageal reflux disease

- Treatment
 - Take medications
 - Antacids
 - H2 blockers (e.g., ranitidine)
 - Proton pump inhibitors (PPI) (e.g., omeprazole)
 - Metoclopramide

Case example

23-year-old man with Down syndrome

- Having difficulty eating
- Gagging, choking, complains of difficulty swallowing
- Review eating habits
- Video swallow study
- Swallowing therapy
- Esophagram, upper endoscopy

Stomach

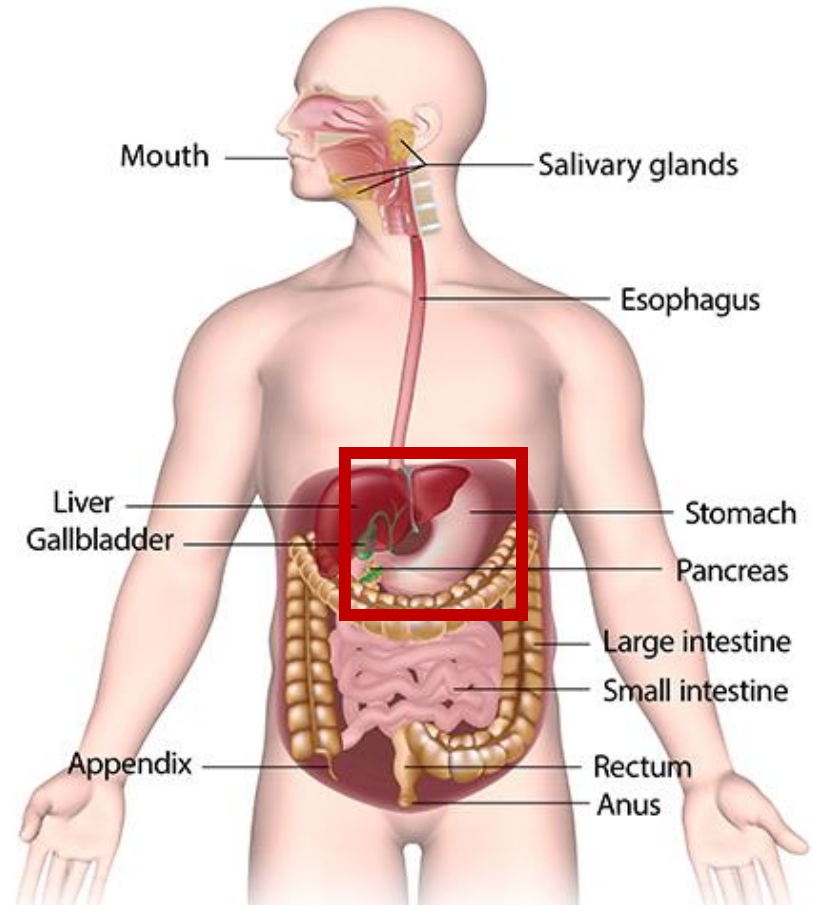


Image from the [National Institute of Diabetes and Digestive and Kidney Diseases](#)

Ulcers

- Erosion of tissue that can occur in the esophagus, stomach, and small intestine
- Contributing factors
 - Helicobacter pylori (H. pylori) infection
 - Non-steroidal anti-inflammatory medication and aspirin
 - Bisphosphonates (e.g., alendronate)
- Symptoms
 - Abdominal or chest pain
 - Resistance to eating
 - Vomiting
 - Symptoms associated with blood loss
 - Dark stools
 - Blood in the stool
 - Vomiting blood
 - Fatigue
 - Syncope (passing out)

Ulcers

- Diagnosis
 - Blood tests (CBC, H. pylori)
 - Stool tests (hemocult to test for blood, H. pylori)
 - Endoscopy (EGD)
- Treatment
 - Antibiotics
 - Medications
 - Antacids
 - H2 blockers (e.g., ranitidine)
 - Proton pump inhibitors (PPI) (e.g., omeprazole)
 - Metoclopramide

Vitamin B12 deficiency

- Found in foods such as beef, poultry, fish, eggs, and milk
- Contributing factors
 - More common in people who have celiac disease (which is more common in people with Down syndrome)
 - Parietal cells, intrinsic factor
 - Autoimmune
- Symptoms
 - Poor appetite
 - Anemia
 - Fatigue
 - Weakness
 - Numbness or tingling in the hands or feet
 - Difficulty with balance
- Can also contribute to depression, cognitive impairment, and dementia.

Vitamin B12 deficiency

- Diagnosis
 - Blood tests
- Prevention and Treatment
 - Diet
 - Supplements

Small intestine

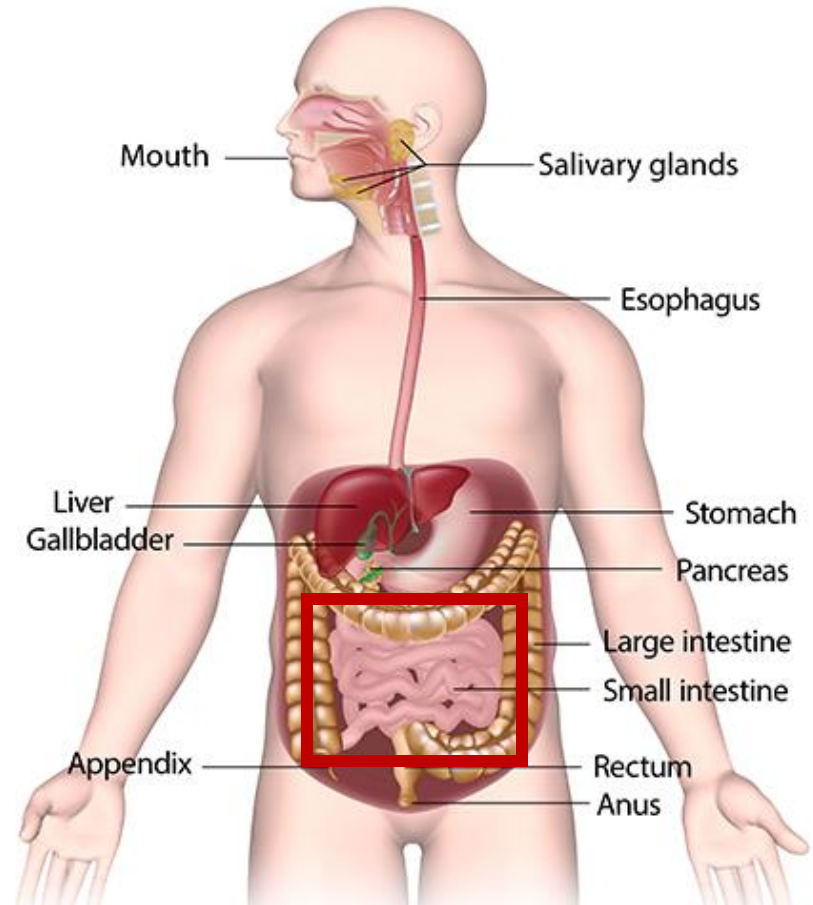


Image from the [National Institute of Diabetes and Digestive and Kidney Diseases](#)

Celiac disease

- Sensitivity to gluten that causes an inflammatory response
- Damages villi
- Affects absorption of nutrients
- Causes
 - Genetic predisposition
- Symptoms
 - Diarrhea
 - Bloating
 - Weight loss
 - Mood and behavior change
 - Many other symptoms

Celiac disease

- Diagnosis
 - History and physical exam
 - Blood test
 - Anti-tissue transglutaminase IgA
 - Total IgA
 - Genetic testing
 - Endoscopy (EGD) with biopsy
- Treatment
 - Gluten-free diet
 - Supplements
 - Dietician consult
- Screening
 - Guidelines for adults with Down syndrome: annual assessment for GI and non-GI signs and symptoms of celiac disease
 - Guidelines for children with Down syndrome: review for symptoms at each health supervision visit

Lactose intolerance

- Inability to digest lactose (a sugar in milk and dairy)
- Cause
 - Deficiency in lactase (enzyme in the small intestine)
- Symptoms
 - Bloating
 - Gas
 - Diarrhea
 - Abdominal discomfort
- Diagnosis
 - History and physical exam
 - Elimination diet
 - Hydrogen breath test
 - Lactose tolerance test
- Treatment
 - Eliminate lactose in the diet
 - Lactase supplements
 - Probiotics
 - Lactase-treated foods (e.g., lactase-treated milk)

Inflammatory bowel disease

- Inflammation of the small and/or large intestines
- Causes
 - Autoimmune
- Symptoms
 - Diarrhea
 - Abdominal pain
 - Weight loss
 - Other
- Crohn's disease
- Ulcerative colitis
- Diagnosis and treatment
 - Referral to gastroenterologist
 - Immunomodulating medications
 - Diet

Irritable bowel syndrome

- Can affect the small and/or large intestines (bowel)
- Causes
 - Gut brain interaction
 - “Functional”
 - Interaction between gut and nervous system
- Symptoms
 - Pain
 - Vomiting
 - Diarrhea
 - Constipation
 - Nausea
 - Bloating
 - And others

Irritable bowel syndrome

- Diagnosis
 - Rome IV Criteria
 - Recurrent abdominal pain on average at least 1 day/week in the last 3 months, associated with two or more of the following criteria:
 - Related to defecation (having a bowel movement)
 - Associated with a change in frequency of stool
 - Associated with a change in form (appearance) of stool
- Rule out other causes
- Multiple sub-types depending on symptoms
- Treatment
 - Lifestyle
 - Medications
 - Other therapies
 - Stress reduction
 - Psychological counseling
 - Probiotics

Case example


34-year-old woman with Down syndrome

- Developed worsening diarrhea with some abdominal discomfort over several months
- Evaluation: history and physical, labs
 - Celiac disease, lactose intolerance, food sensitivity, constipation, inflammatory bowel


Resources

What is Celiac Disease?


I have celiac disease.



That means I should not eat foods with gluten.



Gluten is a part of some foods.



Page 1 of 8
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Eating Gluten-Free


 Usually Gluten-Free I can usually eat these foods!	 Might Have Gluten I should check before eating these foods.	 Usually Have Gluten I should only eat these if they say gluten-free.
  		

Page 1 of 1
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Before Going Out to Eat


I can look for a restaurant that has gluten-free options.



I can use my computer or phone to look for restaurants.



I can use apps such as [Find Me Gluten Free](#) to find restaurants with gluten-free options.



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Celiac Disease Visuals

Gall bladder

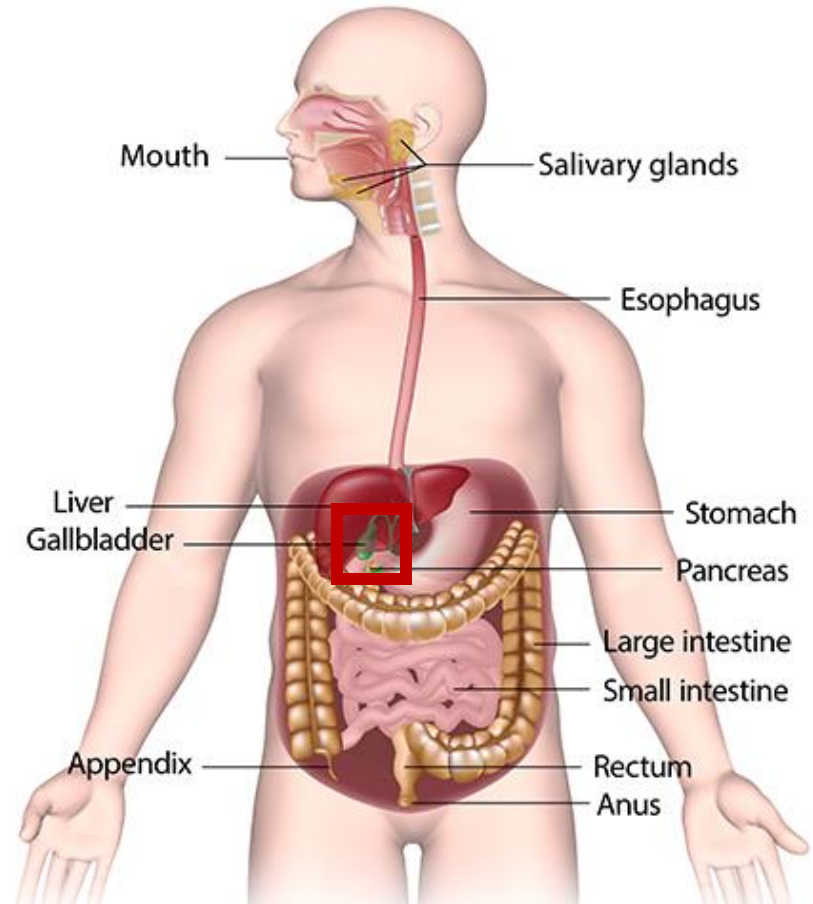


Image from the [National Institute of Diabetes and Digestive and Kidney Diseases](#)

Gallstones

- Also called cholelithiasis
- Hard, pebble-like lumps that form in the gallbladder. Usually made of cholesterol or bilirubin.
- Symptoms
 - Abdominal pain
 - Vomiting, nausea
 - Diarrhea
 - Indigestion, bloating
 - Fatty food intolerance
 - Mood changes
- Diagnosis
 - Blood tests (liver)
 - Imaging (ultrasound, CT scan, HIDA scan, MRCP)
 - ERCP (endoscopy that specifically looks at the duct to the gall bladder)
- Treatment
 - Low fat diet
 - Surgery
 - Medications

Liver

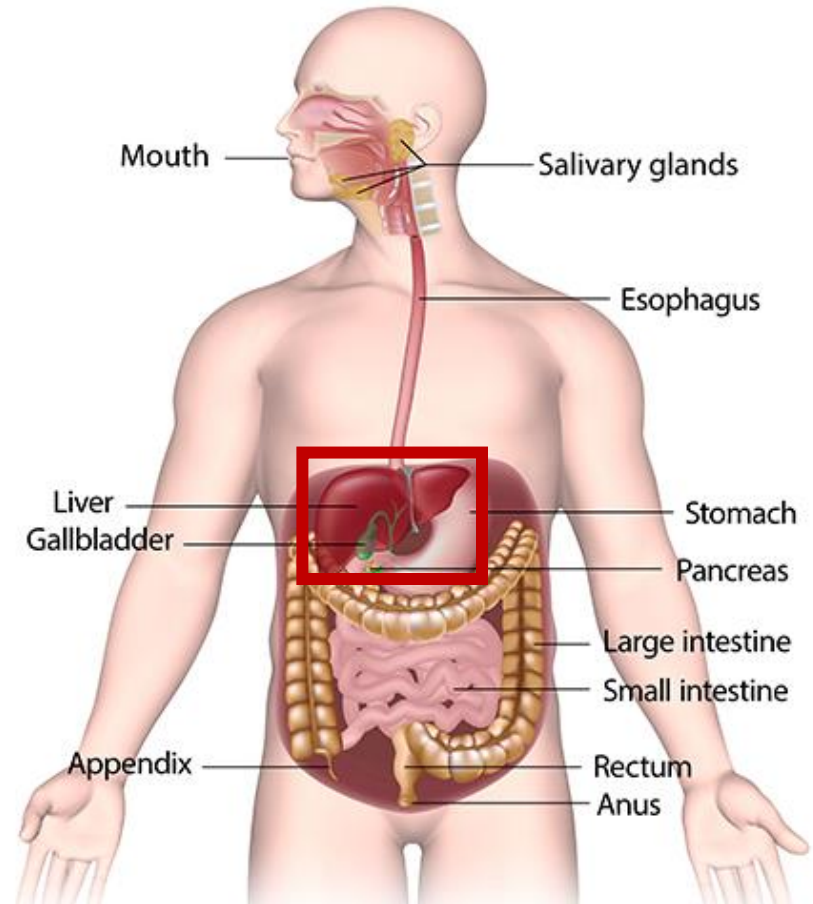


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Fatty liver

- Also called metabolic dysfunction-associated steatotic liver disease (MASLD)
- Build-up of fat in the liver
- Causes
 - Alcohol, obesity, metabolic disorders, some medications, toxins, gut flora, obstructive sleep apnea
- Symptoms
 - It is usually asymptomatic earlier in course
 - Symptoms of liver disease (e.g., fatigue, jaundice, abdominal pain)
- Diagnosis
 - Blood tests
 - Imaging
 - Ultrasound, CT, MRI
 - Additional specific ultrasound tests (e.g., elastography)
- Treatment
 - Diet
 - Medications
 - Treat diabetes

Hepatitis

- Inflammation of the liver
- Causes
 - Viral infections
 - Other causes
- Symptoms
 - Previously described GI symptoms
 - Jaundice (yellow skin)
 - Dark urine
 - Joint pain
- Diagnosis
 - Blood testing
 - Imaging (e.g., ultrasound)
 - Occasionally liver biopsy
- Treatment
 - Depends on cause
 - Rest, healthy diet, fluids
 - Antiviral medications
 - Immune suppression (autoimmune hepatitis)
 - Liver transplant

Pancreas

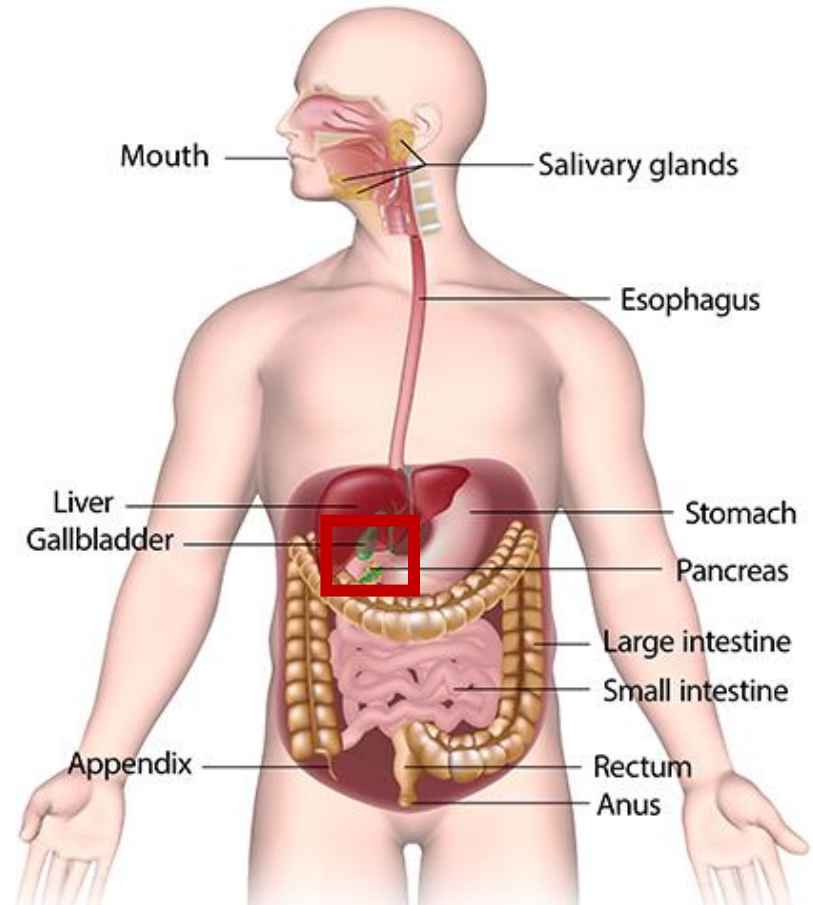


Image from the [National Institute of Diabetes and Digestive and Kidney Diseases](#)

Type 1 diabetes (endocrine)

- Endocrine condition
- Causes
 - Autoimmune
- Symptoms
 - Earlier symptoms may be fatigue, sense of ill health
 - Increased urination, increased thirst, weight loss
- Screening
- Diagnosis
 - Blood testing
- Treatment
 - Insulin
 - Diet

Exocrine pancreatic insufficiency

- Enzyme insufficiency
- Causes
 - Decreased secretion of pancreatic enzymes
 - More common in people with annular pancreas
 - Annular pancreas more common in Down syndrome
- Symptoms
 - Poor digestion/absorption
 - Oily, bulky stools (steatorrhea), chronic diarrhea, weight loss or poor weight gain, and a voracious appetite.
- Diagnosis
 - Stool testing
 - Blood testing
- Treatment
 - Medications (replace enzymes)
 - High fat, high calorie diet
 - Supplements (vitamins and minerals)

Large intestine

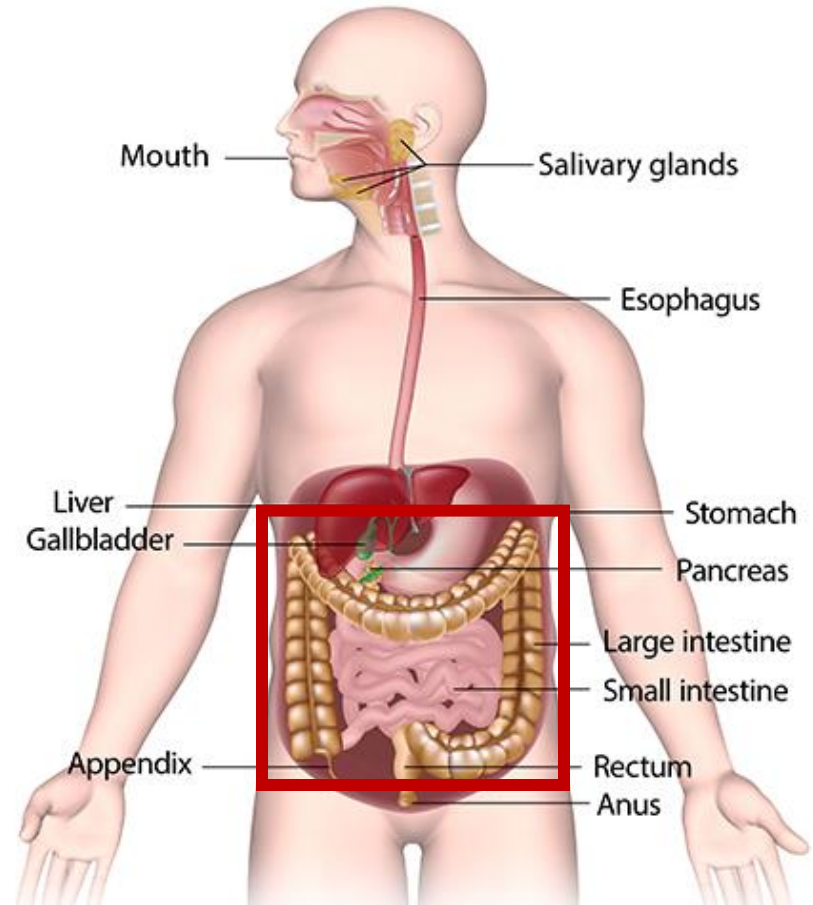


Image from the [National Institute of Diabetes and Digestive and Kidney Diseases](#)

Constipation

- Change in the frequency, size, consistency, and/or ease of bowel movements (BMs)
- Causes
 - Smooth muscle tone
 - Autonomic nervous system (possibly fewer nerve cells)
 - Skeletal muscle tone
 - Diet, hydration
- Symptoms
 - Infrequent, hard, or difficult to pass BMs
 - Diarrhea
 - Fecal impaction
- Diagnosis
 - History and physical
 - X-ray
 - Colonoscopy
 - Celiac testing
- Treatment and prevention
 - Fluids
 - Exercise
 - Fiber
 - Medications

Colon cancer

- Causes
 - Family history (genetics)
 - Diet (red processed meats, low fiber)
 - Lack of physical activity
 - Obesity
 - Smoking
 - Excess alcohol consumption
- Symptoms
 - Constipation
 - Symptoms of anemia (e.g., fatigue, pallor)
 - Blood in stool, dark stools
 - Additional symptoms as it progresses
- Diagnosis
 - History and physical
 - Blood testing
 - Imaging
 - Colonoscopy
- Screening
 - Is it indicated in people with Down syndrome?
- Treatment
 - Surgery, medications, diet

Eating refusal

- Consider possible gastrointestinal cause
 - Often seems to start with a gastrointestinal cause
 - Eating is painful or causes pain
 - An “event”- e.g., choking episode
- May have psychological components
 - Avoidant/restrictive food intake disorder (ARFID)
 - Obsessive compulsive component

Case example

20-year-old man with Down syndrome

- Has become very selective in what he will eat and often refuses to eat at all
- Evaluate for physical cause
- Assess for other symptoms

Takeaways

- There are differences in the frequency of GI conditions in people with Down syndrome compared to those without Down syndrome.
- They may present with GI symptoms or non-GI symptoms.
- Due to limits in communicating symptoms (or specifics of symptoms), it is important to consider GI conditions as contributing factors when a person has a change in function or behavior.
- Effective treatment of GI conditions includes optimizing GI health (addressed in Part 1) and specific treatments for each condition.

Resources



Articles

[Atlantoaxial Instability](#)

[Autonomic Nervous System](#)

[Celiac Disease](#)

[Connective Tissue](#)

[Constipation](#)

[Eating Refusal](#)

[Gallstones](#)

[Gastroesophageal Reflux Disease](#)

[Gastrointestinal Bleeding](#)

[Global Medical Care Guidelines for Adults with Down Syndrome](#)

[Healthy Snacking](#)

[Hernias](#)

[Irritable Bowel Syndrome](#)

[Lactose Intolerance](#)

[Pain](#)

[Swallowing Problems \(Dysphagia\)](#)

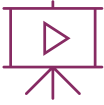


Articles

[Use of Probiotics](#)

[Vitamin B12 and Folate](#)

[Weight Management in Adults with Down Syndrome](#)



Videos and visuals

[Celiac Disease Visuals](#)

[Getting a Swallow Study](#)

[Guides to Healthy Eating and Drinking](#)

[Healthy Eating at Buffets Video and Visual](#)

[Healthy Lifestyle Activity Book](#)

[Healthy Pace for Eating Video and Visuals](#)

[Hydration Video and Visuals](#)

[Tips for Being Physically Active Video](#)

[Tips for Eating Health Meals Visual](#)

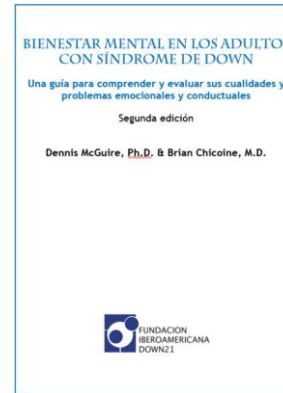
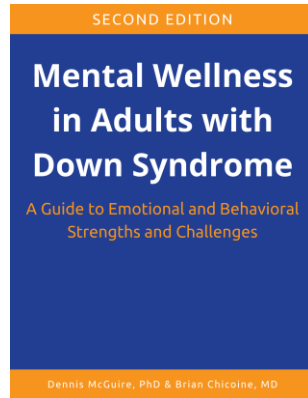
[Using the Toilet Visual](#)

[Visuals and Videos About the Food Groups](#)

[What to Drink Instead of Pop/Soda Visual](#)

Resources

- [Mental Wellness in Adults with Down Syndrome: A Guide to Emotional and Behavioral Strengths and Challenges](#)
 - Available as a free PDF in English and Spanish



Resources for health professionals

- [Down Syndrome Medical Interest Group \(DSMIG-USA\)](#)
 - Project ECHO
 - Webinars
 - Resources
 - Membership

Welcoming Participants to the Down Syndrome Medical Interest Group-USA Project ECHO

All health care professionals and trainees are invited to participate in the Down Syndrome Medical Interest Group-USA (DSMIG) Project ECHO.

This is a case-based opportunity for health care professionals to:

- Learn about health care for children and adults with Down syndrome.
- Earn FREE continuing education credits.
- Present a de-identified case and learn with a panel of experts.

Health care professionals from any discipline welcome.



Non-DSMIG members welcome!

Find out more here: dsmig-usa.org or contact us at info@dsmig-usa.org

 **DSMIG-USA**
Down Syndrome Medical Interest Group

Resources



References

- Biagi E, Candela M, Centanni M, et al. Gut microbiome in Down syndrome. *PLoS One*. 2014;9(11):e112023. doi:10.1371/journal.pone.0112023
- Bull MJ, Trotter T, Santoro SL, et al. Health supervision for children and adolescents with Down syndrome. *Pediatrics*. 2022;149(5):e2022057010. doi:10.1542/peds.2022-057010
- Cai S, Lin J, Li Z, et al. Alterations in intestinal microbiota and metabolites in individuals with Down syndrome and their correlation with inflammation and behavior disorders in mice. *Front Microbiol*. 2023;14:1016872. doi:10.3389/fmicb.2023.1016872
- Mitsuhashi C, Kado N, Hamada M, Nomura R, Kozai K. Characterization of the unique oral microbiome of children with Down syndrome. *Sci Rep*. 2022;12(1):14150. doi:10.1038/s41598-022-18409-z
- Tsou AY, Bulova P, Capone G, et al. Medical care of adults with Down syndrome: A clinical guideline. *JAMA*. 2020;324(15):1543-1556. doi:10.1001/jama.2020.17024

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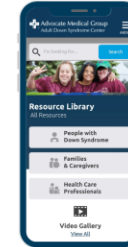
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FREE HEALTH RESOURCES

for people with Down syndrome, families and caregivers, and professionals



Resource Library



Find information on aging, puberty, mental health, self-talk, weight management, Alzheimer's disease, social skills, and more.

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