



DOWN SYNDROME PROGRAM
www.massgeneral.org/downsyndrome

Celiac Disease and Down Syndrome

Celiac disease is a chronic digestive condition where the body reacts against a protein called gluten. Gluten is found in foods such as wheat, barley and rye. The body produces an immune reaction against gluten that affects a person's ability to digest nutrients. Read this handout to learn about how celiac disease is diagnosed in people with Down syndrome.

HOW COMMON IS CELIAC DISEASE?

- In the typical population, up to 1 out of 100 people may have this condition.
- In people with Down syndrome, up to 10 out of 100 people may have this condition.

WHAT ARE COMMON SYMPTOMS IN CELIAC DISEASE?

- Diarrhea
- Constipation
- Stomach pain or bloating
- Growth failure
- Anemia
- Behavioral problems
- Personality changes such as irritability

HOW IS CELIAC DISEASE DIAGNOSED IN PEOPLE WITH DOWN SYNDROME?

At the MassGeneral Hospital Down Syndrome Program, we follow the guidelines of the American Academy of Pediatrics. We screen people with Down syndrome for celiac disease if they have any of the symptoms listed above.

- The screen involves ordering a blood test.
- It is important to remember that this blood test is only a screen. If the result comes back positive, more testing is needed to confirm the diagnosis.

WHAT HAPPENS IF THE BLOOD TEST IS POSITIVE?

If the blood test results are positive, your doctor will call you to talk about next steps. Usually, they will suggest that the person have an upper endoscopy.

- An upper endoscopy is a procedure that uses a lighted, flexible camera to see the inside of the digestive tract. This procedure is performed under anesthesia by a gastroenterologist specialist.
- We will help you stay comfortable through the whole procedure.
- During the procedure, the gastroenterologist will examine the intestine and take a biopsy to see whether signs of celiac disease are present. A biopsy is a sample of tissue taken so that a doctor can look at it closely.

IF THE BLOOD TEST IS POSITIVE, DO I HAVE TO HAVE AN INTESTINAL BIOPSY?

Almost every blood test that doctors order has a chance to be what we call a "false positive." This means that there are some people that will have their blood tests come back "positive," but they DO NOT, in fact, have celiac disease. An intestinal biopsy will make clear whether you have celiac disease and or not. This also makes clear whether you need to have dietary treatment.

WHAT IS THE TREATMENT?

Once we have the final diagnosis, the best treatment for celiac disease is a 100% gluten free diet. Our MassGeneral Hospital Down Syndrome Program and the MassGeneral Center for Celiac Research will provide you with more information on gluten-free diets and celiac disease.