

Celiac Disease in Down Syndrome  
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Individuals with Down syndrome have an increased risk to develop celiac disease. Celiac disease is also called “gluten enteropathy” or “celiac sprue.” It is an autoimmune disorder caused by a reaction to the “gluten,” or protein in grains, including wheat, barley and rye. The symptoms are highly variable. The classic presentation is with abdominal distension, bloating, gas, diarrhea, abnormal appearing stools, increased appetite, weight loss or poor weight gain, and fatigue or weakness. However, some people do not have the classic symptoms, and may be misdiagnosed as having irritable bowel syndrome with intermittent diarrhea and constipation, or may have other non-bowel related problems, such as skin, teeth and tongue changes. We can test for celiac disease by checking certain autoantibodies in the blood. The test that is recommended for screening is the tissue transglutaminase antibody (tTG), which is an IgA antibody. Since IgA deficiency is relatively common, it is suggested that a total IgA level also be checked to avoid a false negative tTG result. If the tTG is elevated, the next step is referral to a gastroenterologist for a biopsy of the small bowel. The typical abnormality on the biopsy is loss or flattening of the normal villi lining the bowel surface and inflammation with influx of white blood cells; this leads to malabsorption, or inability of the bowel to absorb nutrients from food. Celiac disease is both a genetic and an environmental disorder. Certain people are at risk to develop the condition based on their genetic background, and then become symptomatic when exposed to gluten in the diet. Specific HLA types (human leukocyte antigens, which are genetic markers on white blood cells) are more frequently observed in people with celiac disease. The HLA markers DQ8 and DQ2, encoded by genes on chromosome 6, are the most common types found in people who develop celiac disease. In one study, nearly 100% of children with Down syndrome and celiac disease had the HLA DQ2 marker, which suggests that the genetic influence from having an extra chromosome 21, in combination with a specific genetic variation on at least one other chromosome, leads to the development of celiac disease.

The only treatment for celiac disease is removal of gluten from the diet. The diet involves strict avoidance of wheat, barley, rye, most oats and all products made from these grains. The protein in corn and rice is safe for people with celiac disease. Although it can be a difficult diet to follow, there is usually a dramatic response to treatment, with recovery of the intestinal abnormality and improved symptoms. In the general population, about 1 in 100 people has celiac disease, but in Down syndrome, the risk is about 1 in 10 to 1 in 20, or a 5 to 10 fold increased risk. Many people with Down syndrome with celiac disease do not have typical symptoms, and therefore, screening at 2 to 3 years of age is recommended by the American Academy of Pediatrics healthcare guidelines for Down syndrome. In order for the tTG blood test to be accurate, the child must have eating foods containing gluten for at least several months. If your child is older and has never been screened, we still recommend that the blood test be done at least one time. Since we opened the Down Syndrome Center one year ago, we have identified two children with biopsy-proven celiac disease through screening.