

Fact sheet for MDs: GI Concerns in Adults with Williams Syndrome

Williams syndrome is a relatively rare multi-system genetic syndrome caused by a chromosome 7q deletion. Each patient with GI concerns needs individualized evaluation based on their symptoms. This information sheet alerts providers about certain (potentially life-threatening) conditions more common in Williams syndrome than comparably aged persons in the general population. It is not meant to set standards of care.

If a person with Williams syndrome presents with:

--Abdominal pain	--Constipation/Diarrhea	--Changes in bowel habit
--Involuntary weight loss	--Fever with GI symptoms	--Vomiting

consider the diagnoses listed below:

DDX w/ acute symptoms

- Diverticulitis
- GI Perforation
- Pancreatitis

DDX w/ chronic symptoms

- Celiac disease
- Irritable Bowel Syndrome
- Chronic Constipation
- Diverticulitis
- GERD

Misc considerations

- Rectal Prolapse
- Hemorrhoids
- Somatic Pain (Somatization)
- Vascular Insufficiency

The causes of GI symptoms in adults with Williams syndrome mirror those seen in the general population. However, the diagnoses listed here are more common in Williams syndrome, and you should consider them even though the patient's age or symptoms are atypical.

Bear in mind, there are anecdotal reports of increased pain tolerance so the underlying problem may be more protracted or severe than the patient's demeanor suggests. Finally, the disorders listed above do NOT constitute a complete differential diagnosis (DDX)

Since diverticulitis/bowel rupture occurs in young adults with WS, evaluation of new onset abdominal pain may need to include CBC, ESR, & assessment of bowel integrity (KUB or CT scan with contrast).

*If treatment requires surgery:
An anesthesia consult with a cardiac anesthesiologist is strongly advised. Anesthesia has been associated with unexpected death for people with Williams syndrome*

Many patients with Williams syndrome have high levels of anxiety. The pain they are feeling, and the atmosphere of a health care setting, can heighten this feeling of anxiety.

For additional reading please see: www.williams-syndrome.org
 Partsch CJ, Siebert R, Caliebe A, et al. Sigmoid diverticulitis in patients with William-Beuren Syndrome: relatively high prevalence and high complication rate in young adults with the syndrome. *Am J Med Genet A* 2005;137:52-4.
 Matisoff AJ, Olivieri L, Schwartz JM, Deutsch N. Risk assessment and anesthetic management of patients with Williams syndrome: a comprehensive review. *Paediatr Anaesth* 2015;25:1207-15.