

Bowel symptoms in Marfan Syndrome

INFORMATION SHEET

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Marfan Trust
Supporting Research into  Marfan Syndrome

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What is Marfan Syndrome?

An inherited disorder of connective tissue that affects many organ systems including the skeleton, eyes, heart and blood vessels.

- Caused by a mutation in the gene for fibrillin-1 on chromosome 15.
- Can affect both men and women of any ethnic group.
- Around 18,000 people in the United Kingdom have Marfan syndrome.

A significant proportion of patients with Marfan syndrome have gastrointestinal disturbances constituting irritable bowel syndrome (IBS). In the general population, IBS commonly first develops in young adults and teenagers.

It is twice as common in women as in men, and is common in Ehlers-Danlos syndrome due to collagen deficiency, which has many similarities to Marfan syndrome (MFS).

At St George's Hospital a medical student project involved 118 Marfan syndrome patients (56 men and 62 women) aged 18-88 years who were asked to fill out a bowel questionnaire. The results were compared with two control groups. All those participating also filled out anxiety and depression questionnaires as these can influence the perception of pain.

MFS patients suffered more abdominal discomfort and IBS than control groups. 40% of the MFS group as compared to 15% of the control group reported symptoms, consisting of both types of IBS, namely diarrhea predominant or constipation predominant. Women with MFS had higher rates of bloating and constipation than men. There was no significant difference in bowel complaints between anxious and non-anxious patients. However, depressed patients reported more symptoms. Medication for Marfan syndrome, for example antihypertensives including beta blockers, did not make any difference to bowel symptoms.

There is no blood test for IBS, however blood tests can rule out more serious conditions such as coeliac disease. The cause of IBS is unknown, but over-activity of the gut, emotional stress, and intolerance to certain foods may play a part. Specific dietary advice and medical management should be sought through the general practitioner, and may include referrals to hospital dietician and gastroenterologist. Careful attention to diet, avoiding foods such as wheat, dairy products, coffee and alcohol may be helpful, as well as eliminating stress and taking regular exercise.

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Useful References

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