Joint Hypermobility and Functional bowel disorders
Dr A Fikree and Professor Qasim Aziz

The clinical problem: Functional bowel disorders are a group of gastrointestinal conditions which cause chronic abdominal symptoms but where medical tests (e.g. x-rays, blood tests and endoscopy tests) are all normal. These disorders are very common and account for about 30% of referrals to hospital gastroenterologists.

An example of such a functional bowel disorder is irritable bowel syndrome (IBS). Most people who suffer with functional bowel disorders have mild symptoms e.g. bloating and occasional pain. Others however, can be completely disabled with it – these people cannot leave the house, cannot eat, they rely on artificial nutrition and some end up having sections of their bowel removed just for temporary symptom relief. Unfortunately, the cause for these disorders has not yet been discovered and there is no cure for these conditions.

As medical testing is usually normal, a lot of patients are told that there is nothing wrong with them, or are treated as if they are inventing their symptoms. This causes even more patient distress and leads to high levels of depression in some cases. The research into the cause of functional bowel disease has focussed on factors such as depression, low grade inflammation and bacteria in the gut, and nerve and muscle problems of the gut. Although these are important they do not explain a large proportion of patients with these disorders, which probably means that there is another cause for these problems which has not yet been researched.

This cause may have something to do with connective tissue and this is an area that our group is extremely interested in.

The role of connective tissue: Connective tissue is found throughout the body where it supports and binds all the other organs and tissues, much like scaffolding. It is present in the skin and joints, and is also a very important component of the gut. In patients with the Joint Hypermobility Syndrome a genetic defect of connective tissue results in stretchy skin and flexible joints. Preliminary studies suggest that these patients suffer with unexplained gut symptoms and functional bowel disorders.

Moreover, our group has studied patients with severe functional bowel disorders and found that almost 50% of them had undiagnosed joint hypermobility. This raises the question of whether abnormalities of connective tissue of the gut (which have never previously been looked for) can cause symptoms, and whether this may explain why some people get functional bowel disorders.

Our research: Our group aims to find out what the link is between joint
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hypermobility and bowel disorders, and then to show that the connective tissue in the bowel is abnormal in people with joint hypermobility syndromes and gut symptoms.
In this way, our understanding of functional bowel disorders will be improved and this in turn will lead to the development of better treatments, thus improving quality of life for the millions of people who suffer with these conditions.

We are currently conducting a large study in secondary care in collaboration with Professor Rodney Graham from University College Hospital. In this study all new patients referred to the gastroenterology department are examined on their first visit to the hospital to determine if they have joint hypermobility. These patients then see their respective consultants and are investigated for their symptoms. We then see what diagnosis these patients receive after they have had their investigations and we compare the incidence of joint hypermobility in those who receive a diagnosis of a functional bowel problem (like IBS) and in those who have an organic condition such as a peptic ulcer or inflammation or cancer of the gut. We have so far studied 400 patients but the study requires recruitment of 800 patients to give it sufficient power to be able to provide a result. Our expectation is that we will find a higher incidence of joint hypermobility in the patients with functional bowel problem in comparison to organic conditions which suggests that joint hypermobility is associated with functional bowel disease. This may imply that abnormal connective tissue (which causes hypermobility) may be present in the bowel of these patients and this abnormality, which we do not routinely test, causes the gastrointestinal symptoms in these patients. To test this further we are also investigating bowel tissue obtained from these patients to see if we can detect connective tissue abnormalities there.

Expected outcomes:
If our theory is correct, and abnormal connective tissue is an important cause of abnormal bowel symptoms, this will provide a change in our understanding of the cause of functional bowel disorders. It will lead to a new candidate for research into these disorders, and will result in earlier recognition of these patients with ensuing tailor-made approaches to their treatment, potentially with novel therapeutic agents.

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