Functional GI disease

You may be diagnosed with functional disease/disorder, and you will want, or need, to "digest" this document – it is complex, because the topic is complex, and I think poorly explained by, and poorly understood by, many doctors and patients.

Functional GI disorders (FGID) have now been re-named and better understood as **DGBI - disorders of gut brain interaction.**

The main feature of the diagnosis of functional GI disease is the absence of any abnormality on any clinical laboratory testing (i.e. blood or stool tests), upper or lower endoscopy, including biopsies, or imaging. If there are any abnormalities in those areas, they are usually related to a different diagnosis, and not to the symptoms of functional illness. These non-functional (ie organic) diagnoses may co-exist in the same patient. Some symptoms, such as abdominal pain or diarrhea, can be present and even identical in the nature and severity, in both functional and organic diseases.

The history of the English language, the history of medicine, and of functional GI disorders, as nicely outlined by Professor Drossman here, has included varying degrees of separation or overlap between the mind and body.

Functional GI (gastrointestinal) illnesses are by definition chronic, usually minimum 3-6 months.

The word functional used to be the opposite of organic. New expert opinion suggests that we can finally discard the functional-organic dichotomy that tends to stigmatize these disorders (https://theromefoundation.org/rome-iv/rome-iv-faqs/) . Functional GI disorders are now understood as having abnormalities in mucosal immune dysfunction and the microbiota and the work in biomarkers is likely to be a feature for understanding these disorders in the future. The abnormalities identified by recent research in functional GI diseases are found and described in a new area called neurogastroenterology. This term reflects the structural and physiological components of the biopsychosocial model, while the biopsychosocial model itself represents the clinical research and application. This is the area of research and understanding into the "brain-gut axis" and in some works, the "gut-brain axis", as well. As this research into functional disease continues, there are abnormalities found in some areas, but these are not standard clinical investigations. However, there is a "grey zone" in areas of subtle biopsy findings (histology) and motility tests. 2 fairly classical clinical examples of the "gray zone" include the postinfectious IBS, and the IBD-IBS situation.

It can be a confusing term, especially for those "stuck" in the pejorative dichotomy (the worst indicator of this would be "it is all in your head/mind" - which is clearly not the case). Functional gastrointestinal disorders relates to gastrointestinal function, both sensation and motor function of the GI tract (from esophagus to rectum, and including bile duct, gallbladder and pancreas). Motility disorders are generally considered to be separate from functional GI disorders, although there are exceptions (for example the presence of defined motility disturbances in the anorectal functional disorder group).

The definition of a syndrome (such as irritable bowel syndrome) is important, since it is a collection of symptoms, that may or may not indicate an underlying cause, may transform over time into a specific diagnosis, or may remain nonspecific. In general, a syndrome is considered either incurable or not requiring a cure (for example Down's syndrome). Therefore a syndrome may indicate a treatable disease/disorder, or a disease or disorder with positive diagnostic tests, or a syndrome may not indicate

either treatability or positive tests. A syndrome may cause a person to seek medical advice, which essentially defines them as a patient, or may be experienced without seeking medical advice.

IBS can therefore define a group of symptoms, experienced by people, who are not patients (and a remarkably high percentage of the normal population), and the different situation of an IBS patient, who seeks medical attention for one or more of those symptoms.

Functional GI diseases are described in detail in a book and online resource called "The Rome Criteria" which is now in its fourth edition, hence referenced as "Rome IV". You can access most of the main articles, published originally in the journal Gastroenterology, 2016, directly at https://theromefoundation.org/rome-iv/rome-iv-journal-articles/.

Following are some fairly simple and short "pearls":

1. Functional disease is not associated with any test abnormalities but can have many symptoms, related to sensation (function of nerves) and motility (function of nerves and muscles).

2. Two of the commonest and classic examples of functional GI disease are functional dyspepsia ("upset stomach"), and irritable bowel syndrome.

3. Some patients have only one functional diagnosis, but many patients have more than one – symptoms can overlap and presenting concerns (and the presumed site in the GI tract) can vary over time.

4. Some authors and experts have described functional as equivalent to "visceral hypersensitivity", other experts believe this is a common feature, but not synonymous. Visceral hypersensitivity is the feeling of pain related to normal gastrointestinal function, or increased levels of pain from a condition compared to the levels of pain reported and experienced by people who do not have visceral hypersensitivity.

5. There are 2 components of visceral hypersensitivity, allodynia, and hyperalgesia, each reflects increased intensity of pain (compared to people without hypersensitivity) associated with normal physiological functions of the gastrointestinal tract (allodynia), such as peristalsis, or contraction, or painful conditions arising from the gastrointestinal tract (hyperalgesia), such as infection or distension.

6. Symptoms of functional illness are often experienced by many or most humans (examples would be tightness in the throat with emotion, stomach pains with anxiety, diarrhea with stress, eg exams or public speaking). What makes it a functional illness/disorder is the chronicity (ie number of months or years) and the need to seek medical advice, usually related to the reduced quality of life experienced. The reduction in quality of life has been measured and is very significant.

7. Functional illness requires understanding of the biopsychosocial model, which means we are all affected, to a greater or less degree, by the interactions between our biology, our psychology, and our social environments (past and present). Sometimes we may need to try and treat components of the biopsychosocial environment.

8. It is interesting to conventional physicians when practitioners of alternative, complementary or integrative theories will criticize conventional doctors of not being holistic, and not looking for "root cause(s)". Physicians who deal with functional illness and these patients must bring a holistic approach, to diagnosis and treatment, and there is often no "root cause" of functional illness.

9. There are exceptions to the "root cause" comment above – as an example there is a clearly defined and increasingly understood concept of "post-infectious irritable bowel syndrome", contributing to about 10-15% of all patients with IBS, however the remainder have no defined or obvious preceding infection, and many people recover from gastrointestinal infection without developing IBS – this is a very active and productive area of research.

10. Treatments for functional illness can vary from reassurance (about the absence of organic disease) – many patients just want to know they don't have cancer or IBD (inflammatory bowel disease) - to dietary or lifestyle alterations, all the way to potent drugs or combinations – some proven by RCT (randomized trials) and some never proven, but believed by most experts to play a role. Often these unproven therapies are "orphan drugs" where there is no financial incentive to do the clinical trials, because these drugs are common, cheap, generic and often "over-the-counter".

11. Because functional illnesses are so common, and there is significant suffering and loss of quality of life, including loss of attendance at work, school, and tertiary education, there is increasing interest from large pharmaceutical companies, and many university researchers, to study these diseases and develop treatments.

12. Because these conditions are often called syndromes (a syndrome is a collection of symptoms), some people don't consider them true diseases (eg IBS versus IBD) , but gastroenterologists believe they are equally valid diseases, affecting well-being, requiring diagnosis and treatment, and even if there is no cure, management is required (the same applies to most other GI diseases including IBD).

13. Fortunately functional disease rarely if ever requires surgery (an exception might be colectomy for severe functional constipation, but usually this is a well-defined and rare motility disorder), never requires steroids, and currently is not managed by biologics (the very expensive class of drugs used frequently in IBD).

14. Functional symptoms can worsen significantly after surgery, especially if surgery was intended to treat those specific symptoms.

15. There is increasing interest in dietary management for many functional GI diseases, most classically the low-FODMAP's diet for IBS – here we are beginning to understand the scientific origin of some functional symptoms (pain, bloating, diarrhea, sometimes constipation), but it is rare for functional diseases to be cured by dietary management (as opposed to lactose intolerance, bile acid wasting, and celiac disease).

16. In some of the patients suffering badly from functional illnesses, there is a co-existence of psychological issues or even psychiatric diagnoses – there may be anxiety, depression, hypochondriac thoughts, PTSD (and especially a history of physical or sexual abuse or assault).

17. Some of the psychological and mental health issues may require independent or even simultaneous treatment, and yet the presence of mental health issues in no way invalidates the reality and severity of the gastrointestinal symptoms.

18. Some of the drug treatments for functional illnesses are the same drugs as used for mental health issues. This can lead to confusion for the patient and sometimes the pharmacist or the primary caregiver. A classic example is the use of low-dose tricyclic antidepressants (TCAD's) (amitriptyline, nortriptyline, desipramine etc) for chronic pain and gut "over-activity". We now call these drugs "neuromodulators" and this class can also include the classes of antidepressant and anti-anxiety agents such as SSRI's, and SNRI's. The TCAD's are usually given in much lower doses (eg 10-50 mgs) than the typical anti-depressant doses of the same drugs (eg 100-150 mgs).

19. Some doctors are less sympathetic, or less motivated to see and treat patients with functional illness. Some gastroenterologists are guilty (in my opinion) of ignoring, or downplaying, the severity and urgency of the symptoms. Obviously the chronicity and lack of organic symptoms, absence of abnormalities in lab tests, endoscopy, and imaging tests may mean the condition is not urgent. Most of us went into gastroenterology knowing what types of problems could show up, learning about functional GI disorders, and with an interest and a desire to help people with their symptoms.

20. The health care system (in Ontario, and in Canada) has some intrinsic and serious issues that disadvantage patients with functional illness. There is a shortage of primary care practitioners, and a shortage of gastroenterologists. Some patients will be referred to surgeons for endoscopic tests, and when normal, the likelihood of functional illness is dramatically increased, but the urgency of getting in to see a gastroenterologist is also adversely affected. The ability of the Ontario system to provide second opinion consultations is also very limited. The second opinion rarely if ever discovers a different diagnosis but may sometimes differ in treatment options. On occasion, the physician-patient relationship (in the "first opinion") may be lacking.

21. One of the most important components of the initial assessment, and subsequent management plan, would be the presence (and demonstration/communication) of empathy in the health care provider. Dr Drossman outlines other aspects that are therapeutic, and probab;ly critical, in order to build a therapeutic relationship (p.1275) -see also below, point # 25.

22. Functional illnesses occur in other specialties – examples include chronic headaches, fibromyalgia, pelvic pain and dysfunction, interstitial cystitis, shortness of breath, non-cardiac and non-esophageal chest pain.

23. Some of these illnesses have been called medical unexplained symptoms (MUS). Here there is a stronger overlap with a psychiatric diagnosis, specifically DSM-V, somatic symptom disorder.

24. If a specialist, or even a good primary care provider (family doctor or nurse practitioner) makes a confident diagnosis of functional illness, there is a very low likelihood of that patient developing an organic illness, for at least several years. When patients are diagnosed with an organic illness after some years of functional illness, there is usually no relationship (eg 20-40 years of IBS

symptoms followed by diagnosis of colon cancer at age 65), and the functional illness is likely to persist or may even worsen, after the organic illness is cured or managed.

25. The following link reminds us of ways to improve the patient-provider relationship:

    https://theromefoundation.org/communication-pearls-to-help-the-patient-provider-relationship-and-implement-patient-centered-care/