BMJ Commentary 18-04-19

De-diagnosing disease

(763 words)

I tried to placate an angry man in my clinic the other day but I wasn't very successful. The problem wasn't that he had to wait two weeks to get an appointment to see me, it was more profound.
'All I want to know, doctor' said Mr Brown, '*is do I or don't I have a disease?*'.
A recent blood test suggested a diagnosis of pre-diabetes. Our practice systems had swung into action and Mr Brown was trying to deal with the emotional and practical consequences. My explanation that he didn't have a disease as such, more that he was at risk of developing one, a kind of 'pre-disease' so to speak, didn't seem to help.

Making, or excluding, disease diagnoses is what GPs are trained to do. The underpinning methodology is straightforward, in principle at least; panels of experts are convened to develop guidelines which describe diagnostic criteria, based on the best possible research evidence. GPs then apply these criteria to individual patients.

In practice, according to the authors of a recent article in BMJ Evidence Based Medicine (https://ebm.bmj.com/content/ebmed/early/2019/04/11/bmjebm-2018-111148.full.pdf), it's a little more complicated. There is no international consensus governing the development of criteria for how a disease should be defined. In most countries the expert panels comprise mainly disease-focused specialists and their intent is to prevent disease by minimising the risk of under-diagnosis. Their decisions, the authors claim, are often influenced by the pharmaceutical industry. Insufficient attention is given to both the perspective of patients, in particular the unintended consequences of giving them a diagnostic label, and to the degree of influence exerted by vested interests. The problems are less marked in some countries as a consequence of systematic processes introduced by organisations such as the National Institute of Health and Care Excellence in England, but to some degree the challenge is a universal one.

The consequence is a growing number of people being labelled as ‘sick’ without clear evidence that they will benefit from being classified in this way. Too many people are being inappropriately diagnosed and unnecessarily treated for long term conditions such as hypertension, chronic kidney disease and gestational diabetes. Too many people are being given new diagnostic labels such as ‘pre-diabetes’ and ‘pre-osteoporosis’. Over-diagnosis is leading to harm for individual patients, purposeless increased workload for health professionals and inefficiencies for health systems. It’s a grim picture.

The critique is provocative, powerful and engaging to those working in the frontline of healthcare. Disease-mongering feels like a real problem.

So what should be done? The solutions are both simple and radical; new diagnostic labels should be more cautiously applied, perhaps even delayed to allow patients time to consider the benefits and risks of having a disease which might have few or no consequences for their long term health. Established diagnoses should be regularly reviewed. Responsibility for defining the entry criteria for a diseased state should shift. Medical specialists, who have a deep understanding of the pathogenesis, presentation and natural history of individual diseases, should have less influence over how diseases are defined. Medical generalists, who have a deep understanding of how diagnoses affect patients, their families and the health system, and have to deal with the consequences of diagnoses, should have greater influence. Patients, the public and citizen organisations should be central to the process.

Rigid disease definitions should be replaced with ‘thresholds for discussion’ between doctors and patients. Doctors should make greater use of delayed diagnoses and should carry out regular and systematic diagnostic reviews, akin to medication reviews. Having the skill to de-diagnose disease should be a tool in the armory of every doctor. Generalist and specialist clinicians, patients and researchers should work collaboratively in order to fully tease out the risks as well as the benefits of diagnostic labelling and the process should be free from the influence of groups who have commercial interests.

Fundamentally, the medical professional should not have a monopoly on the diagnostic process. The allocation of disease labels should take into account not only the statistical significance of research data underpinning guidelines, and the clinical significance of diseases as determined by doctors, but also the personal significance of disease as experienced by patients. Clinical diagnoses should be judged for their social consequences as well as being used as pathological labels and they should only be used when the benefits clearly outweigh the harms. The parting message of the paper is clear and dramatic: 'the human person should no longer be treated as an ever expanding market place of diseases, benefiting professional and commercial interests while bringing great harm to those unnecessarily diagnosed'.

Martin Marshall

@MarshallProf

Martin Marshall is a Professor of Healthcare Improvement at UCL, Vice Chair of the RCGP and a GP in East London.

Having reviewed the BMJ's Competing Interests Statement I confirm that I have no interests to declare.