

The future of diagnosis in general practice

*'One finger in the throat, and one in the rectum, makes a good diagnostician.'*¹

Things have moved on a bit since Sir William Osler's times, though the importance of diagnosis in primary care remains. Indeed, despite the myriad investigations now available in healthcare, Osler's emphasis on examination is still pertinent. Equally important, arguably more so, is good history taking. All these communication skills we've learnt and all the experience we gain from face-to-face contact makes us better diagnosticians. Yes, you know this, but I make no apology for repeating it. In essence, much of diagnosis in primary care happens without any tests at all. This includes diagnosis of the absence of disease, a key and underappreciated skill. However, three separate changes in primary care investigation are current: increasing use of existing investigations; new forms of testing; and new organisation of testing pathways, both in the UK and elsewhere.

INCREASED TESTING

The only normal patient is one you have not tested enough; most medical students hear this quip at some time in their training. The volume of testing in primary care is mushrooming rapidly. For instance, around 10 million thyroid function tests are carried out each year in the UK with enormous variation in frequency across practices,² while in comparison about 30% of the adult German population have thyroid function taken in any one year.³ This testing, arguably over-testing, is inextricably linked with diagnosis, and with over-diagnosis. For example, the median level of thyroid stimulating hormone (TSH) at which treatment is begun has fallen steadily to reach 7.9 mIU/l by 2009,⁴ despite a dearth of evidence of benefit of treating patients with a TSH below 10 mIU/l.⁵ This is not to suggest that testing in primary care is inappropriate, just that it has to be used as a supplement to clinical practice, not

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a replacement for it. Continuing with the aphorisms: 'treat the patient, not the lab results'.

Diagnosis in primary care may be wholly done in-house. Anxiety and (or) depression remain clinical diagnoses (even if supplemented by scoring systems); so does Parkinson's disease, chronic fatigue syndrome, dementia, and many other conditions. For some of these conditions, testing, perhaps even referral, are required to ensure alternative diagnoses are not the cause of the symptoms. In most countries, there is a broad range of imaging available to the primary care clinician, including the exciting possibility of hand-held ultrasound discussed in this month's issue.⁶ GPs can order a brain scan in their patient with headache, find it negative, reassure, and end up with a satisfied patient who consults less.⁷ This is where point-of-care testing has a place, such as direct testing for light chains for myeloma. Remarkably few point-of-care tests have been assessed sufficiently to enter primary care. Diagnostic research is not simple, especially where the target condition is rare, as most are in primary care. Many individuals have to be recruited to get sufficient patients with the disease for the research to be reliable, thus most tests are first assessed in secondary care. Commercial pressures encourage the manufacturer to promote them as fit for use in primary care, despite the paucity of primary care evidence for their use. This is understandable but scientifically unwise, as spectrum bias inevitably weakens diagnostic performance in primary care. Ultimately, this means few definitive point-of-care tests reach general practice.^{8,9} Even so, it is fascinating to consider how different

the current COVID-19 pandemic would have been if a point-of-care test could have been developed for use in the community.

NEW FORMS OF TESTING

However, much of primary care diagnosis, especially for serious conditions, is actually triage rather than definitive diagnosis. It begins with the patient whose symptoms place them at low risk but not no risk: the primary care clinician performs an in-house test and reassesses the post-test risk. Those with a positive test are then generally offered definitive testing, often by a specialist. Here, point-of-care tests may help. The most studied are inflammatory markers, though their value as 'rule-out' tests is not as high as generally thought.⁸ Recent additions to this form of testing are faecal immunochemical tests (seeking haemoglobin in the stool as a marker of possible colorectal cancer),¹⁰ and CA125 for possible ovarian cancer.¹¹ Neither of these tests had primary care evidence to support their introduction, though are being evaluated retrospectively in that setting. All tests, including primary care ones, have a false-negative rate, so have to be seen in a Bayesian fashion, either increasing the likelihood the target disease is present to a point high enough for definitive testing, or lowering it enough to allow reassurance, sometimes supplemented with later review.

How much can computing and artificial intelligence help?¹² Crucially, both are likely to remain adjuncts to diagnosis rather than replace the primary care clinician. 'The robot will see you now' makes for good media headlines though is unlikely to become reality, especially as primary care consultations with a diagnostic element are in the minority. Nonetheless, scoring systems are in daily use, such as QRISK for 10-year prediction of ischaemic heart disease/stroke. A logical extension is for clinical computing systems to prompt clinicians to the possibility of disease by screening the records for features of that disease.¹³ However, even though artificial intelligence

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accommodates missing data — indeed can learn from the simple fact it is missing — it is still likely only to be able to prompt the clinician to the possibility of disease, rather than anything more. Even so, one field where artificial intelligence may bring benefit is in the patient with comorbidity, increasingly the norm in modern primary care. Patients with existing diseases suffer diagnostic delays when they develop new diagnoses, including cancer, and artificial intelligence may bypass some of the cognitive biases all we clinicians have.¹⁴

NEW ORGANISATION OF TESTING

Finally, as many diagnostic tests require hospital facilities, there has been a move in England towards housing them in a single secondary care centre. These Rapid Diagnostic Centres (RDCs), first proposed in *The NHS Long Term Plan*, have an initial focus on cancer.¹⁵ Every region in England now has at least one operating. Though the exact format follows local needs they have much in common, including the target population. The first group of patients chosen for entry are those with symptoms of possible cancer but without a clear clue as to the cancer site (for example, patients with weight loss or abdominal pain). The referring clinician is expected to perform a battery of preliminary tests, including several blood tests, a chest X-ray, and faecal immunochemical testing. The RDC clinical team can then select specific tests, such as CT imaging or endoscopy. The expectation is that at least as many important non-cancer diagnoses will be made as cancer diagnoses. The long term aim is for all cancer diagnostic pathways to go through an RDC, starting with current pathways that appear to be under-performing. In time, RDCs may shift their focus from cancer to diagnosis as a whole and may consider direct patient access, though these aims are some years off. There is some concern that these new pathways for diagnosis may result in ‘deskilling’ GPs, which occurs when a traditional primary care function is re-allocated, though no evidence has been conducted to see if this actually happens — or matters.

Overall, much of primary care diagnosis is unchanged. Basic skills, such as history taking and examination, remain paramount. Where needed, testing is increasingly available, and moving towards being available in primary care or even at home. Anyone with an iWatch, for example, can now do their own electrocardiogram.¹³ More complex tests, such as CT/MRI, are being opened up to primary care use, and a few reliable point-of-care tests are a reality.¹⁶ This is all progress, but does bring the risk of indiscriminate testing, and the possibility of over-diagnosis and over-treatment. Thus,

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a core skill of primary care is knowing when not to test, as well as when to test.

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