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CLINICAL | OPINION

The challenge of medically unexplained symptoms, overlooked diseases and forgotten patients

Featured image: Forgotten gloves by the kerb, by Andrew Papanikitas, 2022

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Primary care can do so much but not everyone benefits. Some patients with medically unexplained symptoms (MUS) suffer years of referrals and inconclusive tests. Others find themselves overlooked by health care services and feel forgotten. Their voices are unheard, they are unable to find a way forward through the system. Diagnosis may be delayed when the disease is unfamiliar to the doctor,1 or when the physician incorrectly presumes a functional or psychogenic cause while failing to recognise an underlying pathological process. People in this 'no man's land' may receive inappropriate and even harmful treatment.2 They risk mental health problems after being repeatedly told pain is "caused by past trauma or by stress", or that it is "all in their head".3

> *Part of the problem is the system. The current role of hospital clinicians is as skilled purveyors of highly specialist interventions, looking for 'candidates' for their treatments.*

Diagnostic delay may be due to the disease being rare. Many, however, are quite common. The current plight of the forgotten patient is well illustrated by the hypermobility spectrum and Ehlers-Danlos syndromes. Widely taught as being rare, these inherited conditions actually show a prevalence of 194.2 per 100,000 or roughly 10 cases in a practice of 5,000 patients.4 Furthermore, a Rare Diseases Europe survey found that those affected suffer a mean delay in diagnosis of 14 years, often much longer.5 A misdiagnosis was given to 56% of patients, resulting in an inappropriate treatment in 70% of patients. This is perhaps not surprising in view of the diverse problems associated with dysautonomia, mast cell instability and joint hypermobility, which result in initial referral to a range of specialists. Should joint pains and dysarthria predominate, then the pressured physician may not enquire further about other features such as migraine, gastroparesis, intractable hiccups, irritable bowel syndrome, angio-oedema, rashes, gastro-oesophageal reflux, abdominal pain, neuropathy, or non-epileptic seizures. Such diagnostic latency and inappropriate therapy often correlate with deteriorating symptoms. Additionally, with time, the misdiagnosis of a 'psychosomatic' condition becomes more likely.

Part of the problem is the system. The current role of hospital clinicians is as skilled purveyors of highly specialist interventions, looking for 'candidates' for their treatments. Referral implies that others believe the problem falls within their expertise. A negative effect is that they have little to offer individuals that fall outside the categories that convey candidacy. GPs are the last of the generalists but in the face of limited time and resources may struggle to sort patients into credible categories, ready for onward sifting. Unfortunately, even incorrect labels tend to stick. Emergency medicine physicians sometimes use the metaphor of a 'funnel', with diagnoses being excluded until finally one remains. After that process, it is hard to go back to the excluded diagnoses.

A further issue is equality of care. Access is more difficult for the disadvantaged. According to the social model of disability,6 socio-economic factors are relevant. People from privileged backgrounds can choose an environment that fits their needs and afford to search for the appropriate physician.

How might we better address these issues?

1. Concentrate on patients: Their stories are important if we let them speak.7 They are experts in their own experience, increasingly informed by the internet. Often physicians shut down the sharing of new information too early. Patients' narratives, not just specialists' criterion-based checklists, are essential tools in diagnosis.
2. Provide resources for physicians and patients, such as: management algorithms or apps for non-obvious diagnoses; signposting to help patients and families reach appropriate care.
3. Promote a supportive environment for the neurodiverse.
4. Reinvent the 'generalist' in secondary care with time, knowledge, and experience to look beyond the common and routine. The old teaching that ''If it looks like a duck, swims like a duck, and quacks like a duck, then it probably is a duck' should no longer hold in modern medicine.8

5. Embrace doubt: A clinician saying, “I know there is something going wrong, but I am unclear what that ‘something’ is”, is a positive act leaving the door open to further reflect, consult, research, or seek second opinions.
6. Work together: A patient might present with irritable bowel syndrome, migraines, and joint pain, but the proper diagnosis might only emerge if the neurologist, gastroenterologist, and rheumatologist cooperate. Structures must be created to promote such collaborations, and to ensure that diagnoses are recognised by relevant stakeholders including insurance providers.
7. Promote research to discover why some patients are forgotten, diseases missed, or potentially valuable initiatives get dropped. Could a re-examination of symptom clusters using AI reveal new disease entities, potentially amenable to treatment? In absence of a cure, where are the gaps and how might management be optimised?
8. Replicate successful strategies: A few centres in the UK, Germany and elsewhere are leading the way by establishing ‘unexplained symptom’ clinics.

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Our hope from the current pandemic is that there will be a re-evaluation of how we best look after people with atypical health issues. In the UK, the original principles of the NHS included good healthcare freely available to all according to need, rather than ability to pay. If we wish to adhere to these laudable ambitions, providers must understand the reasons why for some, the current system is not working. Research, technological, and organisational initiatives can help. Some of the above will require extra funding. Possibly, of greater importance and relatively inexpensive, is optimising what happens during individual consultations. For this, we need both sufficient time and well-educated, enthusiastic, empathic, and inquisitive physicians with a holistic approach.

Then, hopefully, fewer patients would be forgotten.

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1. This opinion piece is presented on behalf of the Forgotten Patients Group (www.forgottenpatients.org).
2. Other members who have contributed include:

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