The British Student Doctor, 2021;5(1):32-39
doi: 10.18573/bsdj.151
Discussion article

'When you hear hooves, think zebras': further integrating the biopsychosocial model in medical education to help rare disease patients

DISCUSSION

AUTHOR

Lucia Lazzereschi
University of Southampton

Address for Correspondence:
Lucia Lazzereschi
Southampton Medical School
12 University Rd
Southampton SO17 1BJ
United Kingdom

Email: lazzereschi.lucia@gmail.com

Conflicts of interest:
Lucia is a peer reviewer at The BSDJ

Accepted for publication: 20.10.20

ABSTRACT

Diagnostic delay in individuals with a rare disease is, on average, 4.8 years. Their journey to diagnosis, often referred to as a diagnostic odyssey, is plagued by countless investigations and unsuccessful referrals. How can we ensure that healthcare professionals will consider rare diseases when first meeting a patient? The answer may lie in tackling the issue early on when clinicians begin their journey at medical school. The current medical school curriculum approach in the United Kingdom, which is largely based on the biomedical model, causes a hindrance in improving rare disease diagnosis. Bypassing the cultural, social and psychological considerations that a doctor has to address when treating a patient, may contribute to the diagnostic odyssey patients have to go through. Further integration of the biopsychosocial model into medical teaching can improve the diagnostic journey for patients as well as the patient-doctor relationship. The latter could lead to a shorter diagnostic delay by increasing compliance and engagement. Additionally, approaching a patient with the biopsychosocial model could help their wellbeing and mental health during their journey to diagnosis. Integrating rare disease teaching pre-clinically and clinically via student selected units (SSU), paediatrics and primary care placements, will equip future clinicians with an understanding of rare diseases to ultimately reduced diagnostic delay and improve the experience of those living with one.

Rare diseases - of which there are between 6,000 and 8,000 (1) - affect fewer than 5 in 10,000 people. (2) They are often overlooked in medical school education, clinical practice, and research funding. I was not aware of rare diseases until more than half-way through my medical degree, when I met AL (pseudonym). She had been on buprenorphine patches and codeine for osteoarthritis, but they had done little to relieve her pain. When I met her, my initial feeling was surprise; how could she have osteoarthritis in her late twenties? The culprit turned out to be Mucopolysaccharidosis type-I (MPS-I), a rare hereditary metabolic disorder. Individuals suffering from MPS-I lack the enzyme required for glycosaminoglycan degradation, leading to its accumulation in tissues, causing skeletal, neurological and cardiorespiratory problems. (3) AL suffers from osteopenia, osteoporosis, osteoarthritis, kyphosis, carpal tunnel syndrome and cardiovascular issues. These have all impaired her mobility and left her as a wheelchair user. Her limited vision and hearing impairment make it difficult for her to go out alone and have affected her mental health. As misfortune – or luck – would have it, AL suffers from a severe form of MPS-I; she started showing symptoms early on and her diagnosis was not overly delayed. This is imperative. Quick diagnosis in rare diseases has been associated with a significant reduction in morbidity and mortality. (4) Unfortunately, most people affected by rare diseases have a different diagnostic journey, often referred to as a diagnostic odyssey; (5) plagued by an array of misdiagnoses and futile investigations. Diagnostic delay is, on average, 4.8 years long. (6) Recommendations recently published to tackle time-to-diagnosis in rare diseases suggest that primary care healthcare professionals should be equipped with diagnostic tools and flow-chart pathways for rare disease identification. This would increase the likelihood that correct referrals are carried out. (5) Whilst this may accelerate time to diagnosis once a rare disease is considered, the clinician first has to regard it as a differential for the appropriate investigations and referrals to take place. How can we ensure that healthcare professionals will consider rare diseases when diagnosing a patient? The answer may lie in tackling the issue early on when clinicians begin their journey at medical school.

Currently, medical schools in the United Kingdom (UK) adopt one of following types of curricula: (1) traditional - a lecture-based curriculum where there is a clear pre-clinical and clinical divide; (2) problem-based learning where students learn by addressing an open-ended problem found in a trigger material; (7) or (3) a mixture of both. Regardless of the curriculum type, medical schools must comply with the General Medical Council (GMC) requirements, the UK public body in charge of the official register of doctors. (8) Generally speaking, the content of education across medical schools is biomedical in nature, although in varying degrees based on the institution and curriculum type used. This type of model is strictly focused on biological aspects of illness (9) and only superficially addresses the cultural, social and psychological considerations that a doctor has to address when treating a patient. The biomedical model reinforces the paternalistic idea that clinical decisions are made based on what the clinician deems to be in the patient's best interest. Within the model, patients' knowledge is considered poor when compared to the clinician, and it takes a secondary role when making treatment decisions. Perhaps it is these two aspects of the model, if instilled in medical students early on, that contribute to the diagnostic odyssey. This is for two main reasons.

Firstly, an individual who presents with a set of symptoms the clinician has never seen before poses a diagnostic challenge. Clinicians may not refer appropriately and investigations which might prove ineffectual may be performed. (3) Throughout this process, the patient's wellbeing and symptom control may suffer as frequent investigations take a toll on patients' mental health. These risks are typically not prioritised if a pure biomedical model is applied in clinical practice. Medical school education has, in the last decade, introduced elements of the biopsychosocial model in their teaching. This is a holistic and multifactorial approach to clinical practice which includes a strong focus on cultural, social and psychological issues. However, it is often integrated in the clinical years of placement and overshadowed by the practical skills medical students need to hone before becoming junior doctors. (7) Time and time again, medical students are reminded that "when you hear hooves, think horses not zebras." An aphorism coined in the late 1940s, (10) it serves as a reminder to medical students that common conditions are, by default, common. When a patient presents with certain symptoms or signs, you should always consider the most prevalent causes first before considering rarer differentials. Although important when teaching medical students how to approach a clinical consultation, by focusing too much on what is common, and often not considering what is rare, are we doing patients a disservice? I believe the diagnostic odyssey experience in patients with rare diseases would be improved if medical school programmes integrated the 'biopsychosocial' model earlier on by incorporating more behavioural and social sciences alongside pathophysiology and pharmacology. This is because by equipping students with "soft skills" developed through this model, future clinicians will be able to more promptly respond to the emotional needs of a patient who might still not have a diagnosis. Whilst this may not shorten their journey to diagnosis, it will hopefully improve their wellbeing and mental health whilst waiting for one.

A major challenge with achieving this is that the National Health Service (NHS) is a taxpayer-funded system that faces constant pressures due to under-staffing. Delivering holistic care in a system like this is not easy; most clinicians probably do recognise the emotional needs of their patients but responding adequately in a system dictated by short appointment times and ever-increasing workload is challenging. Crucially, it is because of these pressures that further integration of the 'biopsychosocial' model would better enable clinicians to respond to the needs of patients, especially those with rare disease or complex co-morbidities. Training individuals early on to weigh biomedical, social and psychological factors that affect a patient equally in every consultation means that they will have practised identifying which factors are most important to the patient and should be dealt with first. This is key, especially since appointments can be as short as eight minutes in duration. (11)
I wish I had witnessed this first-hand in the case of VL, a 7-year old child I met with Rasmussen Syndrome. It is a rare disease marked

by seizures that increase in frequency and severity due to brain inflammation. This then leads to hemiparesis which with time, continues to weaken and can lead to complete loss of limb function. (12) Conducting the consultation with VL's parents using a 'biopsychosocial' approach might have helped the clinician identify VL's parents' main concerns early in the conversation. It might have prompted the clinician to act on VL's parents' worry that as he grows up and becomes more active, his risk of injury, should he have a seizure or experience limb weakness, may increase. Indeed, a few weeks later he was injured as he was playing in a public playground, exactly as his parents had feared. This risk, which could have been initially mitigated with a referral for Occupational Therapy assessment, was not prioritised when using a biomedical approach as the social issues were not discussed during the consultation. Ultimately, this could have spared VL and his family the physical and psychological pain and engendered greater trust in the healthcare system.

While a biopsychosocial model might not always shorten time to diagnosis directly, it can indirectly shorten a patient's diagnostic odyssey by improving the relationship between the doctor and the patient. A 2019 observational study reported that consultations with conflicting points of views between the patient and the practitioner not only led to mistrust but to unhelpful patient behaviour (non-attendance). (13) This can lead to both patient harm and extended diagnostic delay; especially crucial in the context of rare diseases where symptoms and signs are often harder to diagnose and diagnostic delay is already unacceptably long. (6) Additionally, engagement between the clinician and the patient – which is a key component of the 'biopsychosocial' model – has been shown to improve diagnostic odyssey experience in rare diseases, (14) reinforcing the argument for its integration in the medical school curriculum earlier on.

Secondly, the inherent assumption perpetuated by the biomedical model that patients and family members' knowledge is poor, has to be removed early on. Over the months and years that patients live with a condition, they become experts on how to manage the symptoms they experience. Their expertise can aid diagnosis and reduce diagnostic delay but only if clinicians listen to their patients' concerns and expectations and accept that they may know less than their patient.

The biomedical model perpetuates this bias because it trains medical students to associate success with being able to identify and treat symptoms and signs of a disease, and not the individual as a whole. Treatment often focuses on something tangible that can be measured, like pharmacotherapy. This leads to an overshadowing of social and psychological aspects the patient might be concerned with. Better outcomes could be achieved if the patient's issues are being dealt with holistically and not only from a biomedical point of view. In the case of a rare disease like MPS-I, patients often struggle to use fine motor skills because of the pain they experience. If a piano teacher, still undiagnosed, was presenting to her general practitioner (GP) with arthritic-type symptoms, the clinician might focus on following a biomedical model to diagnosis and prescribe pharmacotherapy for the pain and refer for investigations. If you applied the biopsychosocial model you would also consider the threat to her job security. If she couldn't teach piano, how would she pay for her house and her bills? What effect would that have on her pain, her wellbeing and mental health? Could it indirectly negatively impact any other co-morbidities she may already have? Discussing this could lead to a referral to a link worker for social prescribing (15) which would then put the patient in contact with volunteer organisations and charities that can help with social issues. Applying the biopsychosocial model would not only focus on identifying the cause and treating the problem (the pain), which is an aspect of both the biomedical and biopsychosocial model, but on identifying interim solutions while the diagnosis is being pursued. This will make the investigations to diagnosis more manageable for the patient.

Adoption of a biopsychosocial approach has been extensively researched in chronic illness, autoimmune diseases, functional diseases and in psychiatry. (16-18) Its utilisation has been shown to improve patient trust and satisfaction, medication adherence once diagnosed, promote positive behavioural change and a better quality of life. (16-19) Research has also shown that the adoption of the biopsychosocial model can reduce the amount of diagnostic tests and referrals as the relationship fostered between the doctor and the patient reassures patients that a common ground can be found between both parties. (20) Rare diseases like MPS-I or Rasmussen syndrome are often chronic or progressive, which supports the argument for approaching patients who might have a rare disease using the biopsychosocial model.

Not only does the model lead to better health outcomes physically, but it can also address the non-biomedical aspects of living and the psychological factors that may affect patients' wellbeing. These, if given consideration, can be targeted with an appropriate intervention whilst developing an individual diagnostic and treatment strategy more specific to the array of symptoms and social issues the patient may face during diagnosis. (6) Additionally, patients with rare diseases often experience issues when communicating with medical professionals who do not believe their symptoms (21) or who are themselves frustrated with a lack of diagnosis. This has negative repercussions on both the patient and the length of time to diagnosis. (21) Theoretical knowledge of a disease is not a replacement for years of experience. Involving the patient in decision-making and in addressing what is important for the patient may not only shorten the diagnostic odyssey but may make the journey to diagnosis better and improve the rapport between patients and healthcare professionals.

Not only should the biopsychosocial model be integrated more fully in current medical curricula across the UK but changes should be made to the content of the curriculum itself. Until recently, there was no standardised curriculum across UK medical schools, the topics covered were partly dependent on what was deemed clinically useful, and partly on the healthcare opportunities available in the area. (22) With the introduction of the Medical Licencing

Assessment (MLA) curriculum (23) for the graduating year of 2023-24 this will change. The MLA curriculum outlines a list of conditions that medical students are required to understand by the time they graduate; this knowledge will then be tested in a national exam medical students will be required to sit in order to graduate. Encouragingly, a few rare diseases are mentioned in the MLA curriculum; muscular dystrophies, myeloproliferative disorders and polymyalgia rheumatica. However, two are umbrella terms for groups of conditions – muscular dystrophies, myeloproliferative disorders – and the curriculum does not specify how much time should be spent on each topic and in what format that should be taught. Understandably, as different curricula types employ different teaching methods, this gives medical schools the freedom to integrate the MLA curriculum requirements within their current curriculum without having to re-design a completely new course. However, as no timeframe is specified, this caveat might result in some universities deeming one lecture enough to fulfil the requirement while another could allocate a week of theoretical and clinical work to rare diseases. This would lead to a discrepancy in knowledge between graduating students from different institutions.

It is also important to consider that collectively, rare diseases affect 1 in 17 patients. (2) Tomorrow's doctors must be able to recognise a patient who could be affected by a rare disease, even if they cannot diagnose the rare disease at first presentation. Being able to consider a rare disease can only occur if students have engaged with patients who have experienced the diagnostic odyssey themselves - the signs and symptoms may be different in the patient they encounter as a clinician compared to the patient they met at medical school, but similar diagnostic patterns will be present. (24) Following GMC recommendation, this is already integrated to a varying extent in UK medical schools by having expert patients for conditions that medical students will often see as clinicians. So why not have some rare disease patients as expert patients? (25) After all, who better than someone who has lived through the diagnostic odyssey to explain to medical students their disease and experiences? It is not only beneficial for the patients that medical students will encounter in future, but to the students too, as engagement with expert patients has been reported to increase confidence in clinical decision making. (26) Self-confidence has been shown to be an important resource for effective clinical decisions that improve patient diagnosis and management. (27)

With over 180 rare disease patient organisations present in the UK, integrating rare diseases into medical school teaching via expert patients is a feasible short-term task. (28) It will provide future clinicians with an insight into rare diseases, the struggles of the diagnostic odyssey and an awareness into what living with a rare disease within the NHS is really like. Expert patient integration into the curriculum could then be followed by long-term curriculum changes, for example via student selected units and elective opportunities for students who wish to learn more about rare diseases. Additionally, medical schools could also integrate rare disease teaching in parts of the syllabus where they are commonly encountered, such as paediatrics and primary care. Future clinicians who choose to train in these specialties would therefore already have an understanding of rare diseases and how they may present before they qualify. All of these changes would, in turn, equip future clinicians with an awareness that is currently lacking in clinical practice and help rare disease patients by feeling heard, accounted for and considered. Ultimately, the patient's healthcare experience would be improved. Further integrating the biopsychosocial model and rare diseases teaching in medical education would enable current medical students to be better clinicians and to serve the purpose they set out to fulfil when they enrolled in medical school: care for people in need. People like AL.

.

REFERENCES

1. Dawkins HJS, Draghia-Akli R, Lasko P, Lau LPL, Jonker AH et al. Progress in rare disease research 2010-2016: an IRDiRC Perspective. Clinical Translational Science. 2018;11(1):11-20.

https://doi.org/10.1111/cts.12501

PMCid: PMC5759730. PMid: 28796411

2. Rare Disease UK. What is a Rare Disease? London: Genetic Alliance UK; 2020 [accessed 9th Feb 2020]. Available at: https://www.raredisease.org.uk/what-is-a-rare-disease.

3. Soni-Jaiswal A, Mercer J, Jones SA, Bruce IA, Callery P. Mucopolysaccharidosis I: parental beliefs about the impact of disease on the quality of life of their children. Orphanet Journal of Rare Diseases. 2016;11:96.

https://doi.org/10.1186/s13023-016-0478-z.

PMCid: PMC4942895. PMid: 27406185

4. Polizzi A, Gentile AE, Taruscio D. Competing to raise awareness of rare diseases. Lancet Neurology. 2019;18(8):721-722.

http://doi.org/10.1016/S1474-4422(18)30437-X

PMid:30447970

5. Global Disease Commission. Ending the diagnostic odyssey for children with a rare disease. Zurich: Global Rare Disease Commission; 2018 [accessed 12th Feb 2020]. Available from: https://globalrarediseasecommission.com/Report.

6. Evans WRH. Dare to think rare: diagnostic delay and rare diseases. British Journal of General Practice. 2018; 68(670):224-225.

https://doi.org/10.3399/bjgp18X695957

PMid:29700025 PMCid:PMC5916061

7. Miles S, Kellett J, Leinster J. Medical graduate' preparedness to practice: a comparison of undergraduate medical school training. BMC Medical Education. 2017;17:33.

https://doi.org/10.1186/s12909-017-0859-6

PMid:28166769 PMCid:PMC5295184

8. General Medical Council. Promoting excellence: standards for medical education and training. London: GMC UK; 2016 [accessed 21st Jan 2020]. Available from: https://www.gmc-uk.org/-/media/documents/Promoting_excellence_standards_for_medical_education_and_training_0715.pdf_61939165.pdf.

9. Farre A, Rapley T. The new old (and old new) medical model: four decades navigating the biomedical and psychosocial understandings of health and illness. Healthcare. 2017;5(4):88.

https://doi.org/10.3390/healthcare5040088

PMid:29156540 PMCid:PMC5746722

10. Quote Investigator. When you hear hoofbeats look for horses not zebras. Quote Investigator; 2020 [accessed 15th Jan 2020]. Available at: https://quoteinvestigator.com/2017/11/26/zebras.

11. Oxyoby K. Consultation times. BMJ. 2010;340.

https://doi.org/10.1136/bmj.c2554

12. Orsini A, Costagliola G, Perna D, Esposito MG, Bonofilio L, Striano P et al. Efficacy and tolerability of mycophenolate mofetil in pediatric Rasmussen syndrome. Epilepsy Behav Rep. 2019;13:1000334.

http://doi.org/10.1016/j.ebr.2019.100334

PMid: 32140679 PMCid: PMC7044645

13. Amelung D, Whitaker KL, Lennard D, Ogden M, Sheringham ,. Zhou Y et al. Influence of doctor-patient conversations on behaviours of patients presenting to primary care with new or persistent symptoms: a video observation study. BMJ Quality and Safety. 2020;29:198-208.

http://dx.doi.org/10.1136/bmjqs-2019-009485

PMid:31326946 PMCid:PMC7057803

14. Budych K, Helms TM, Schultz C. How do patients with rare diseases experience the medical encounter? Exploring role behavior and its impact on patient-physician interaction. Health Policy. 2012;105(2-3):154-64.

http://doi.org/10.1016/j.healthpol.2012.02.018

PMid:22464590

15. Moffatt S, Steer M, Lawson S, Penn L, O'Brien N. Link worker social prescribing to improve health and well-being for people with long-term conditions: qualitative study of service user perception. BMJ Open. 2017;7(7):e015203.

http://doi.org/10.1136/bmjopen-2016-015203

PMCid: PMC5541496 PMid: 28713072

16. Stewart M, Brown JB, Donner A, McWhinney IR, Oates J, Weston WW et al. The impact of patient-centered care on outcomes. Journal of Family Practice. 2000;49:796–804.

PMid:11032203

17. Drossman DA. Gastrointestinal illness and the biopsychosocial model. Journal of Clinical Gastroenterology. 1996;22:252–4.

https://doi.org/10.1097/00004836-199606000-00002

PMid: 8771417

18. Covic T, Adamson B, Spencer D, Howe G. A biopsychosocial model of pain and depression in rheumatoid arthritis: A 12-month longitudinal study. Rheumatology. 2003;42:1287–94.

http://doi.org/10.1093/rheumatology/keg369.

PMid:12810932

19. Williams GC, Frankel RM, Campbell TL, Deci EL. Research on relationship-centered care and healthcare outcomes from the Rochester biopsychosocial program: A self-determination theory integration. Journal of Families, Systems and Health. 2000;18:79–90.

http://doi.org/10.1111/j.1525-1497.2004.40901.x

PMCid: PMC1494796 PMid: 15566452

20. Ferrari R. The biopsychosocial model: a tool for rheumatologists. Best Practice and Research Clinical Rheumatology. 2000;14:787–95.

http://doi.org/10.1053/berh.2000.0113

PMid: 11092802

21. Rare Disease UK. The rare disease reality – an insight into the patient and family experience of rare disease. London: Genetic Alliance UK; 2016 [accessed 17th Jan 2020]. Available at: https://www.raredisease.org.uk/media/1588/the-rare-reality-an-insight-into-the-patient-and-family-experience-of-rare-disease.pdf.

22. Lee SWW, Clement N, Tang N, Atiomo W. The current provision of community-based teaching in UK medical schools: an online survey and systematic review. BMJ Open. 2014;4:e005696.

http://dx.doi.org/10.1136/bmjopen-2014-005696

PMid:25448625 PMCid:PMC4256542

23. General Medical Council. Medical Licencing Assessment. London: GMC UK; 2020 [accessed 21st Jan 2020]. Available from: https://www.gmc-uk.org/education/medical-licensing-assessment.

24. Blöß S, Klemann C, Rother A, Mehmecke S, Schumacher U et al. Diagnostic needs for rare diseases and shared pre-diagnostic phenomena: Results of a German-wide expert Delphi survey. PLoS One. 2017;12(2):30172532.

https://doi.org/10.1371/journal.pone.0172532

PMCid: PMC532530 PMid: 28234950

25. General Medical Council. Patient and public involvement in undergraduate medical education. London: GMC UK; 2020 [accessed 21st Jan 2020]. Available from: https://www.gmc-uk.org/-/media/documents/Patient_and_public_involvement_in_undergraduate_medical_education___guidance_0815.pdf_56438926.pdf.

26. Coulby C, Jha V. The role of patient-led education initiatives in medical education. Innovation and Entrepreneurship in Health. 2015;2:33-40.

27. Fry M, MacGregor C. Confidence and impact on clinical decision-making and behaviour in the emergency department. Australasian Emergency Nursing Journal. 2014;17(3):91-7.

http://doi.org/10.1016/j.aenj.2014.03.003

PMid:25113311

28. Genetic Alliance UK. Our Members. London: Genetic Alliance UK; 2020 [accessed 12th Jan 2020]. Available at: https://www.geneticalliance.org.uk/our-members.

bsdj.org.uk


/thebsdj


@thebsdj


@thebsdj

Journal DOI
10.18573/issn.2514-3174

Issue DOI
10.18573/bsdj.v5i1

The **British Student Doctor** is published by **The Foundation for Medical Publishing**, a charitable incorporated organisation registered in England and Wales (Charity No. 1189006), and a subsidary of **The Academy of Medical Educators.**

Cardiff University Press
Gwasg Prifysgol Caerdydd

The British Student Doctor is an imprint of Cardiff University Press, an innovative open-access publisher of academic research, where 'open-access' means free for both readers and writers.

cardiffuniversitypress.org