FREE ARTICLE

RARE DISEASE IN UK PRIMARY CARE: WHY “MORE EDUCATION” ISN’T ENOUGH

A research-backed review of what it really takes to reduce diagnostic delays for rare immune and genetic conditions in UK general practice.

68,705

patients screened in UK pilot study

using MendelScan EHR tool

>90%

of patients & carers reported anxiety

or low mood (UK survey)

12–14%

had a care coordinator

of adults with rare disease in UK

Key figures from recent UK evidence on rare disease in primary care [6,17,18]

Primary care's role in early recognition

Primary care is central to earlier diagnosis of rare immune and genetic diseases through longitudinal contact, first-contact access, and care coordination.[1–3] When GPs are actively involved, the evidence consistently shows shorter diagnostic delays, smoother care transitions, fewer emergency admissions, and better quality of life for patients.[1–4]

Digital case-finding tools such as MendelScan can be embedded directly into UK primary care electronic health records (EHRs) to flag patients who may have a rare disease. In a UK pilot study involving 68,705 patients, 227 were flagged by the tool; after clinical review, nine were judged to represent a reasonable possible diagnosis and were investigated further.[6] A qualitative evaluation in UK general practices found that GPs saw this approach as feasible and potentially valuable for reducing inequity in diagnosis, though time, data-sharing concerns and workload were identified as barriers to wider rollout.[5–7] Similar artificial intelligence (AI)-based approaches for primary immunodeficiencies and other rare conditions show promise for earlier detection in primary care.[7,8]

How education impacts recognition

Genetics and genomics in the context of rare disease present a genuine challenge in primary care; surveys suggest that specialist resources such as Orphanet are underutilised and that GPs would welcome more support in managing genomic queries.[3,9–11] Targeted education does improve knowledge and confidence; however, the picture is more nuanced than simply 'more training'.

FIGURE 1

Impact of targeted genetics education formats on primary care practice

Educational approach	Observed impact	Ref.
Focused primary immunodeficiency (PID) education programmes	Correct answers increased from 58%→79%; better recognition of warning signs and syndromic primary immunodeficiencies	[12,8]
Genetics/genomics continuing medical education (CME) for GPs	Consistent gains in knowledge and confidence, but modest, patchy change in practice	[10,11]
Just-in-time online resources	Preferred source for genomic queries; EHR-linked tools viewed as optimal	[9,10,13]

Systematic reviews show that education alone is insufficient; enablers such as clear referral pathways, decision-support tools, protected time, and reimbursement are also needed to translate improved knowledge into lasting change in day-to-day primary care practice.[1,2,10–13]

What the evidence says about system-level change

The case for going beyond education is not just theoretical. A large UK survey of 760 adults with rare diseases and 446 carers found that only 12–14% had a care coordinator, just 32–33% were seen at a specialist centre, and only 10% of adult patients had a care plan in place; a mere 2–5% had all three.[17] These are not gaps in GP awareness — they are gaps in the system itself.

The mental health burden tells a similarly stark story. A UK survey of over 1,200 patients and 564 carers found that more than 90% reported anxiety or low mood, and 36% of patients had experienced suicidal thoughts.[18] The authors found that limited access to psychological support was a key driver, suggesting that better factual knowledge alone will not resolve these outcomes.

● **KEY TAKEAWAY**

Education improves knowledge — but without integrated digital tools, clear referral pathways, and structural support for GPs, that knowledge rarely translates into faster diagnoses for patients with rare diseases.

What works best in the UK context

The strongest evidence points to a combination of approaches working together. Embedding digital rare-disease case-finding tools into EHRs[5–7] moves the system from passively waiting for a classic presentation to proactively identifying patients who may have been missed. Concise, primary-care-focused genomic resources such as GeNotes and the Rare Disease Education Hub[13–15] give GPs access to the right information at the right moment.

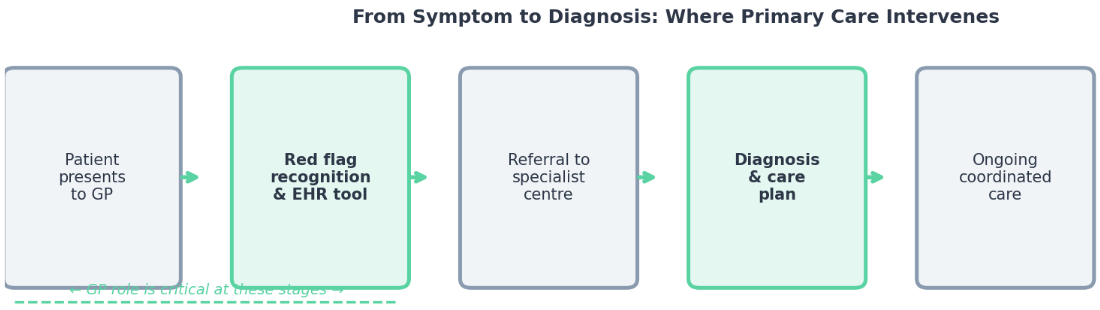
Pathway templates tailored for rare genetic disorders — including those for neurofibromatosis type 1 (NF1) and achondroplasia established by the NHS North East and Yorkshire Genomic Medicine Service — provide primary care with a defined and practical framework for coordinating and managing transitions in patient care.[13] Multi-modal online genomics education, specifically tailored to GP roles,[9,11,16] demonstrates optimal effectiveness when structured around authentic clinical scenarios rather than condition-by-condition factual recall.

Conclusion

Primary care is well placed to spot early red flags for rare diseases, and targeted education clearly improves knowledge and confidence. But the research suggests that education alone is not enough. The lengthy process of diagnosis will only improve when clinical expertise is supported by digital tools, defined referral pathways, coordinated care, and structures that enable GPs to apply their knowledge. That is a system-level challenge, not just an educational one.

FIGURE 2

From symptom to diagnosis — where primary care intervenes in the rare disease pathway



References

1. Vidic N, et al. Integrated Care for People Living With Rare Disease: A Scoping Review on Primary Care Models. *J Primary Care Community Health.* 2025;16. doi:10.1177/21501319241311567
2. Ferreira R, et al. The utilisation of primary health care system concepts positively impacts the assistance of patients with rare diseases. *J Global Health.* 2023;13. doi:10.7189/jogh.13.04030
3. Byrne N, et al. The role of primary care in management of rare diseases in Ireland. *Ir J Med Sci.* 2020;189. doi:10.1007/s11845-019-02168-4
4. Lodh R, et al. Health care utilisation and education outcomes of children with rare diseases: a born in Bradford cohort study. *Eur J Pediatr.* 2023;182. doi:10.1007/s00431-023-05225-4
5. Yahia R, Rafi I, Evans W. Implementing a digital rare disease case-finding tool in UK primary care. *Br J Gen Pract.* 2024;74 suppl 1. doi:10.3399/bjgp24x738105
6. Buendia O, et al. Is it possible to implement a rare disease case-finding tool in primary care? A UK-based pilot study. *Orphanet J Rare Dis.* 2021;17. doi:10.1186/s13023-022-02216-w
7. Evans W, et al. Opportunities and challenges for identifying undiagnosed Rare Disease patients through analysis of primary care records. *J Community Genet.* 2024;15. doi:10.1007/s12687-024-00742-7
8. Soler-Palacín P, et al. New tools for diagnosis of primary immunodeficiencies: from awareness to artificial intelligence. *Front Immunol.* 2025;16. doi:10.3389/fimmu.2025.1593897
9. Evans W, et al. How genomic information is accessed in clinical practice: an electronic survey of UK GPs. *J Community Genet.* 2020;11. doi:10.1007/s12687-020-00457-5
10. Paneque M, et al. A systematic review of interventions to provide genetics education for primary care. *BMC Fam Pract.* 2016;17. doi:10.1186/s12875-016-0483-2
11. Dunlop K, et al. Building capacity for genomics in primary care: a scoping review. *Front Med.* 2025;12. doi:10.3389/fmed.2025.1577958
12. Hariyan T, et al. Evaluation of awareness about primary immunodeficiencies among physicians before and after an educational program. *PLoS ONE.* 2020;15. doi:10.1371/journal.pone.0233342
13. Evans W, et al. Enhancing care coordination for neurofibromatosis type 1 in primary care. *J Community Genet.* 2025;16. doi:10.1007/s12687-025-00811-5
14. Baynam G, et al. Global health for rare diseases through primary care. *Lancet Glob Health.* 2024;12(7). doi:10.1016/s2214-109x(24)00134-7
15. Briscoe S, et al. Evidence of inequities experienced by the rare disease community. *Orphanet J Rare Dis.* 2025;20. doi:10.1186/s13023-025-03818-w
16. Griffiths R, Lewis C. Mainstreaming genomics in the NHS in England. *BMJ Paediatr Open.* 2025;9. doi:10.1136/bmjpo-2024-003286
17. Walton H, et al. Experiences of coordinated care for people in the UK affected by rare diseases. *Orphanet J Rare Dis.* 2023;18. doi:10.1186/s13023-023-02934-9
18. Spencer-Tansley R, et al. Mental health care for rare disease in the UK. *BMC Health Serv Res.* 2022;22. doi:10.1186/s12913-022-08060-9