



# Rare Disease and Genomic Educational needs of UK General Practitioners

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## Background

Genomics is having an increasing impact across medicine, including primary care. Rare diseases are at the forefront of these genomic advances, with 80% having a genetic basis<sup>2</sup>. To realise the full potential of these technologies, knowledge of its application will be required in all areas of medicine, including primary care. To optimise the educational resources for GPs, the largest group of clinicians working in the NHS, we need to understand their educational needs and where they currently access genomic information.

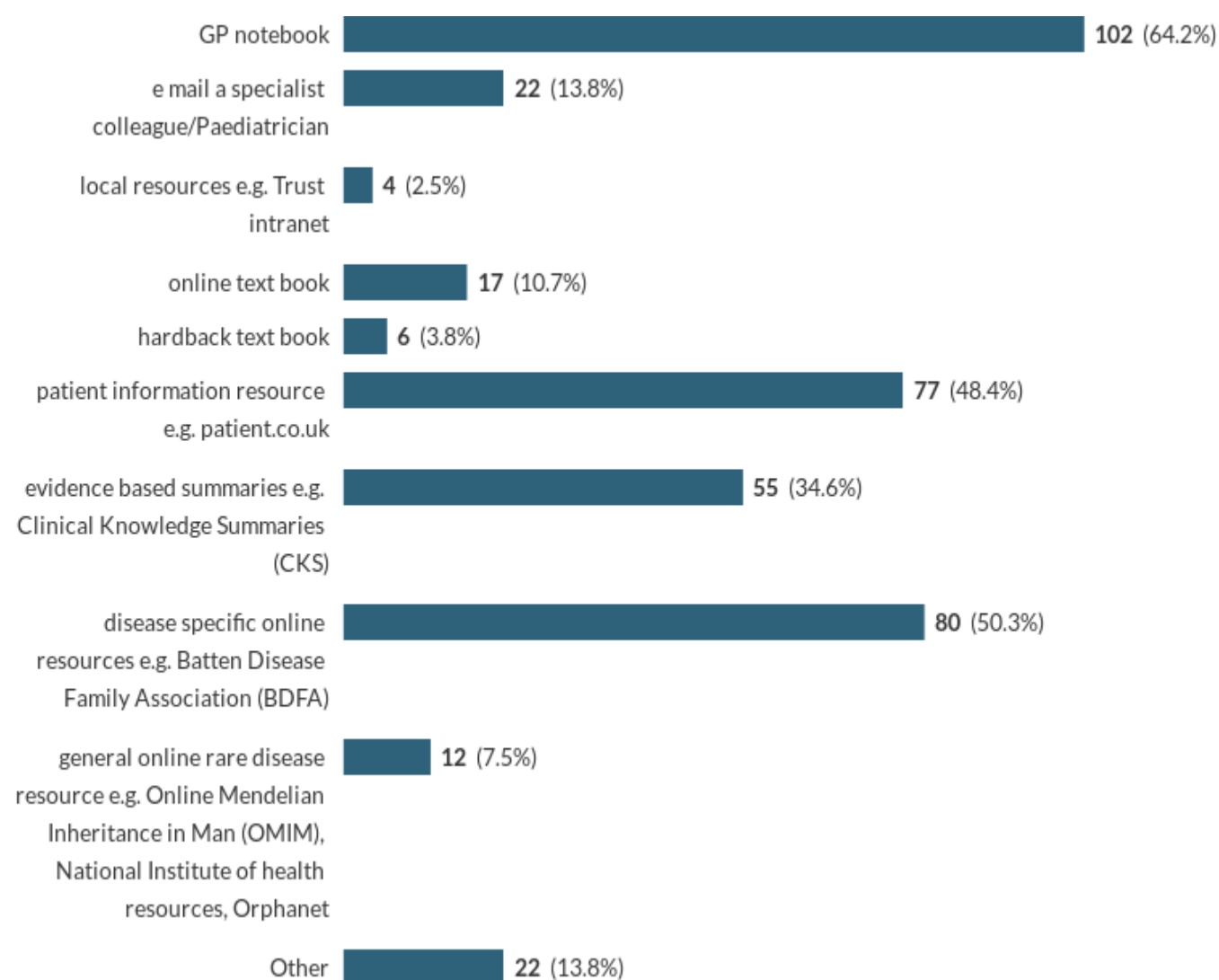
## Methods

159 UK GPs completed the survey in response to an open invitation, between September 2017 and September 2018. Questions were in response to 4 clinical genomic scenarios including a rare disease scenario, with further questions exploring direct to consumer testing, how one collects a family history and their preferences and format for educational materials.

*'A 28-year-old lady attending for preconception advice, as her sister has a child with Batten disease. (Batten disease, also known as CLN3 is a rare fatal inherited degenerative neurological disorder of childhood).'*

## Results

- “Just in time” (readily accessible at the moment they are needed and deemed relevant) internet based resources in trusted and familiar places (e.g. GP notebook and NICE CKS) were the most popular, with disease specific pages (e.g. charities) and internet search engines also highlighted. (chart 1)
- Few respondents (4%) would access rare disease specific internet resources (Orphanet, OMIM).
- Referral/communication to local specialist or genetic service was overwhelmingly the most popular response when deciding on the next step to explore preconception testing.
- A large proportion (41%) had not heard of Genomics England's 100,000 genome project.
- For genomic continuous medical education over 70% of respondents preferred online learning.
- Most (48%) would use an internet search engine to understand the value of commercial direct-to-consumer (DTC) genetic testing. However (41%) said they would contact/refer to a local specialist.
- When collecting a family history, various approaches were declared, listing relatives the most popular with some utilising local templates.



**Chart 1** Where GPs would look for information about Batten disease during a consultation (Multi-answer: respondents were asked to mark all that applied)

## Key Messages

- Ensure that “Just in time” rare disease resources are readily accessible near the top of search engine results.
- Ensure information about rare diseases in primary care resources, such as GP notebook, is accurate.
- There is a need for a standardised and encodable method of collecting a family history in the electronic health record.
- 41% of GPs had not heard of Genomics England's 100,000 genome project, despite widespread publicity.
- Direct-to-consumer testing (DTC) poses a workload risk to clinical genetics with 41% of GPs contacting or referring a patient with a DTC test query.

## References

1. Evans, W.R.H., Tranter, J., Rafi, I. *et al.* How genomic information is accessed in clinical practice: an electronic survey of UK general practitioners. *J Community Genet* (2020).
2. Institute of Medicine Committee on Accelerating Rare Diseases Research and Orphan Product, D. (2010). In M. J. Field, & T. F. Boat (Eds.), *Rare Diseases and Orphan Products: Accelerating Research and Development*. Washington (DC): National Academies Press (US)