NHS
Genomic AI Network

Stakeholder Reports

The use of AI and limitations to deployment in the NHS Genomic Medicine Services

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EXECUTIVE SUMMARY

The Genomic AI Network held forums with stakeholders across England, Scotland, Wales and Northern Ireland to ask them where they are currently using AI, where there are gaps in AI use, and what the barriers are to deploying AI in their areas.

Three groups were established through cascade advertising, and participation was voluntary. The groups were advertised based on three areas:

1. Clinical Genetics (20 attendees)
2. Clinical Scientist Reporting and Decision Support Software (20 attendees)
3. Test Ordering and Referrals (18 attendees)

This report is a summary of the three groups.

KEY TAKEAWAYS

AI use is limited by practicality - a trial may be done informally because of what's available based on existing systems, not based on the best tool. Implementation must be bespoke to each individual local system.

Procurement at a Trust or system level is one effective way of getting AI to be used more. Microsoft Co-Pilot is a good example of this.

Tools which can automatically vet referral quality for missing information, automating manual processes such as updating databases or automatic reanalysis are desired.

AI functions often part of a larger package and not a product in isolation. Its utility depends on the existing pipeline.

Clarity and education on the different types of AI would be helpful, and the safety considerations and expectations applicable to each.

There is still hesitation around the effectiveness of AI for complex cases.

The potential benefits of AI in test referrals and ordering are extremely limited at present due to local test ordering systems, non-standardised paper-based referral system, and lack of integration with local EPRs. This impacts data quality of referrals, time and resource, as well as potential incidences where samples and results are lost or unactioned.

CLINICAL GENETICS

Where is AI currently used?

- AI is mostly being used where it is pragmatic to do so.
- For example, where a software or system is already being used, and they release an AI 'add on'. The most common example of this was dictation tools and AVT.
- Alternatively, it is used where there has been procurement/licensing at a wider (Trust, Provider or system level). For example, the use of Microsoft Co-Pilot is popular in Trusts where this has been procured. This is used changing the tone of patient communications, standardising clinic letters, and for enhanced searched, such as literature searches (especially useful for rare conditions).
- There is informal use of non-integrated products, such as ChatGPT, for tasks non-related to direct clinical care, such as student projects or pre-clinic preparation.
- Pilots vary, and stakeholders are doing informal trials (based on opportunistic availability) with dummy data, before embarking on the IG process as this is resource intensive.

Where are the gaps and needs?

- Products where patient information can be used is a gap – there is a want for systems which can integrate with EHRs. There was a tool which was integrated with GP system, EMIS, and there was a want for something equivalent.
- Document summarisation of current and historical records (paper and electronic) would be helpful.
- Manual database entry continues to be resource-intensive.
- Automatic re-analysis of variants, and updates to ClinVar, would be a useful area if such a tool existed.
- There is a need for a tool or system which automatically vets the quality of referrals, not necessarily making triaging decisions, but can flag where there is missing or incomplete information and send back to the referrer automatically.

What are the barriers?

- Information governance in resource intensive, and Trust teams don't have the time.
- Products need to be aligned to individual trusts. The spread of services across different Trusts can cause barriers. Trusts have blocked products before.
- Heterogeneity of EPRs, or lack of EPRs and a paper-based system is a barrier. Similarly, in-house, niche, EPRs is another barrier.
- Processes based on email referrals are a barrier because it is difficult to integrate an AI tool, and the data is unstructured.
- Risk-averse environments mean teams may be less willing and able to try new products and ways of working.

CLINICAL SCIENTIST REPORTING AND DECISION SUPPORT SOFTWARE

Where is AI Currently Used?

- There is current use with various decision support software, which are being evaluated as part of the Rare Disease NoE. Implementations of these still require local validation and adjustments, even if they are off-the-shelf products.
- AI has been used in student research projects of variant interpretation and variant re-prioritisation tools.
- Tools are sometimes chosen based on practical/convenience factors, such as the existing LIMS system, pipelines, or other related workflows.
- Non-clinical outputs are also relevant, for example, Microsoft Co Pilot is popular for PowerPoints, meeting minutes etc.

Where are the gaps and needs?

- There was debate about whether increased HPO terms significantly increases diagnosis.
- Some interest in exploring AI tools in the coding process, but not as developed.
- Not aware of any professional guidance or a reputable source for clinical scientists. There is a need to share knowledge on what is safe and with what caveats, and a recognition of where the responsibility lies on these decisions. This would stop unsafe practice, as well as wasting time on pilots for tools which could never be approved.
- Variant interpretation remains a large bottleneck in clinical scientist reporting. However, this still requires a human to check.
- AI which could help to mine the data that is generated all the time to identify hotspots and variants, and cohort reanalysis, would be extremely useful.

What are the barriers?

- Utility of tools is limited by various factors and dependencies. For example, the existing LIMS systems, availability of phenotype information and HPO terms, and the size of the panels/service requirements all affect how useful a variant interpretation tool is.
- Some of the barriers are not unique to the AI tool specifically, for example, having available phenotype information.
- AI tools in the pathway are often not a single tool available, but part of an existing AI-enabled pipeline, this makes it difficult to do a comparison of just the AI function. Companies are looking to sell the whole pipeline package.
- There is a lack of education, awareness and confidence in what is available, what is being used, and what considerations need to be made, for example information governance
- AI is not always the best or most appropriate solution. For example, clinical reports might be automated or 'smart', but not necessarily a need for AI, as there are existing national standardised templates. There was also hesitancy that AI would be appropriate for very complex cases.
- SDE infrastructure and access is a barrier to utilising data to full potential. There is awareness of a lab's data footprint.

TEST ORDERING AND REFERRALS

Where is AI Currently Used?

- Use of AI was less in this area than in the other stakeholder groups.
- PhenoTips, although not necessarily AI in all definitions, is a tool used in various sites to help guide clinicians, especially useful for HPO terms and gathering information that would be sent with a test order.
- There was a local pilot of an AVT which can help to fill in clinic forms, as well as letter generation and other functions.
- Non-integrated large language model to map HPO terms.

Where are the gaps and needs?

- There is a need for AI for the more mundane tasks, such as letter writing. Especially needed for filling out forms, as some pathways have multiple, complex forms. A tool which could fill out forms using clinic information, scribed consultations or HPO terms would be very useful.
- There is a need for a tool which would check improving the quality of referrals, including the eligibility of a test automatically, before all the forms are filled in. This would help with demand management principles, and saving time. This is especially relevant for mainstreamed clinicians.
- A tool which could track samples and results. This is in response to incidences where samples or results are not shared or actioned, but the team does not know. Currently, there is a reliance on local SOPs for checking missing results and samples and actioning accordingly.
- AI which could be a guide that important information hasn't been missed would be helpful, similar to PhenoTips.
- AI is not necessarily the solution, especially as a lot of issues are system or process based, such as correct data fields or complexity of forms. There is a strong need for a universal system and infrastructure.

What are the barriers?

- There is a lack of central electronic ordering system. This means that labs with different EPRs and LIMs interact on paper-based SOPs, PDFs and emails. Paper-based systems are one of the biggest impediments to adopting AI tools.
- Variance of EPRs within one GLH means that test from different labs need different processes if internal or external ordering.
- Lengthy information governance processes which need to be repeated at each Trust is a barrier.
- Cost and resource.
- AI not commonly used, so there aren't existing exemplars for people to emulate.
- In some Trusts, electronic ordering not possible due to the complexity of genomic tests and the information required. There was discussion as to whether this would be applicable to all types of test, or just some.

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