Artificial Intelligence in NHS Genomic Medicine Services

October 2024

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NHS Genomic AI Network

NHS Central and South Genomics

NHS South East Genomics

KiTEC

Executive summary

This report, commissioned by the Genomic AI NHS Network of Excellence, explores the current use, opportunities and barriers to adoption of artificial intelligence (AI) tools in NHS England Genomic Medicine Services. The research involved surveys and interviews with the genomics workforce, including AI adopters, researchers, innovators, and managers across England.

The key aims were to understand the landscape of AI in NHS England Genomics Services, including:

- Outlining current and prospective uses of AI tools,
- Identifying impactful areas for AI application and
- Describing opportunities and barriers to AI adoption.

Current use: AI adoption in genomics is currently low, primarily driven by individuals using freely available tools like ChatGPT. Common applications include report and letter writing, bioinformatics pipelines, and variant identification, classification, and prioritisation. Current uses typically involve human-in-the-loop processes with oversight of inputs and outputs. Other use cases may require a higher degree of trust, evidence and capability of the AI and advances in data and computing available to services.

Opportunities: AI holds significant potential to enhance efficiency, particularly in administrative tasks, report writing, and variant analysis. Future applications, building on advances in research and digital infrastructure, could have more transformative impact to services and patients, for example identifying patients when new variants are discovered or improving personalised and precision medicine.

There is a strong desire for increased visibility of AI activities and more training to develop AI literacy and support to improve awareness and engagement. Updating regulations and guidelines to keep pace with AI advancements is essential for ensuring best practices and consistent care across the country.

Barriers: Major barriers include lack of capacity and expertise for staff to scope and implement appropriate tools, lack of capability of IT systems, low system interoperability, inadequate data readiness, and the cost or time required for implementation. The current performance and evidence base for AI tools for genomics services, immature regulation and lack of guidance on the safe qualification and use of these tools were also key concerns.

Leveraging AI's potential can significantly contribute to the mainstreaming of genomics, supporting the realisation of predictive, preventative, and personalised medicine in the NHS. Currently AI is viewed more as an efficiency tool: with improved standardisation and availability of data for clinical decision making it could drive more transformational changes to improve patient outcomes. Improving the quality and particularly the diversity of training data will be essential to improve trust in AI tools and ensure equity of access.

Introduction

The last decade has seen transformational change in the NHS with the introduction of a national genomics service, the completion of the 100,000 Genomes project and the introduction of rapid whole genome sequencing in routine care. The large amount of digital information makes it a prime opportunity to deploy artificial intelligence (AI) to increase efficiency and capability. However, there are challenges in consistency and machine readability of patient records, interoperability and interfacing between systems, ability to synthesise relevant information from multiple sources, and managing efficient flow of data with privacy and security.

The 2022 strategy for accelerating genomic medicine in the NHS[1] proposed a five-year plan to embed genomics across the NHS, delivering equitable testing to improve health outcomes and enable precision medicine and making use of research, innovation and digital capability to develop services. AI is not a panacea, and it is important that it is used in support of wider service and workforce strategies rather than diverting resources. However, use of AI in genomics services has the potential to increase efficiency to reduce backlogs and ease pressure on the workforce, and to increase capability, for example in diagnosis and prognoses. Delivering the prerequisites for AI at scale – curated digital datasets, effective computing infrastructure and technical expertise – may also deliver wider reaching impacts to services.

This report explores the current and future opportunities for AI tools and application in NHS England genomics services, and the infrastructure required to deliver them, from the perspective of NHS staff and experts in the fields of genomics and AI for healthcare. The aims of this piece of work were to:

- outline current and prospective use of AI tools in NHS England genomics medicine services
- identify opportunities for AI adoption
- identify and describe barriers to AI adoption
- identify opportunities for GAIN to support addressing the above.

Information was gathered using a mixture of desk-based research, workforce survey and interviews. Care was taken to include a wide range of perspectives, covering different sectors, job functions and levels of experience. Semi-structured interviews were carried out with representatives from NHS England Genomics Medicine Services, Genomics England, the AI Centre for Value Based Healthcare, the Ada Lovelace Institute, Nuffield Council on Bioethics, NICE and the British Society for Genetic Medicine, secure health data environments as well as former Topol Fellows, Turing-Roche Scholars and clinical and translational researchers.

Interviews discussed the participant's role, experience with AI and perceptions of its use in NHSE genomics services. Those with experience developing, using or implementing AI tools were asked about their motivations, experiences and for a description of the process. Interview responses have been aggregated and themed for this report.

A questionnaire distributed across NHS England genomics services was intended to map the extent of AI use, awareness, readiness and appetite across NHS England. The 125 respondents from included service and programme managers, clinical geneticists, clinical scientists, bioinformaticians, genetic counsellors and GPs with special interest from all regions of England and different seniority levels (see appendix). Key themes were extracted from free-text responses to enable semi-quantification and comparison.

[1] Accelerating Genomic Medicine in the NHS (NHSE, 2022). Available at : www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/

Key drivers for use of artificial intelligence in genomics services

Regardless of AI experience or formal education, the key drivers for wishing to adopt AI tools in the genomics service were focused on increasing efficiency of existing services. It is closely related to points made about preparedness and concerns about AI introduction, namely:

- That AI tools should fit within existing pathways to derive benefit while minimising disruption, complexity and indirect cost.
- Time saving is key for a stretched workforce to enable them to maintain staffing/service at the current level, addressing backlogs, or adjusting to accommodate new service requirements.
- Use of AI to automate repetitive tasks could help to achieve increased efficiency without compromising level of care to the patient. The tasks cited varied by profession but generally included booking tests and results; administrative scheduling and writing letters. This was more divisive as some staff felt a human ability to determine the information and tone needed for each patient was important and were proud of their letters. Others felt happy to use AI for clerical and letter writing tasks with only human checking and editing required.

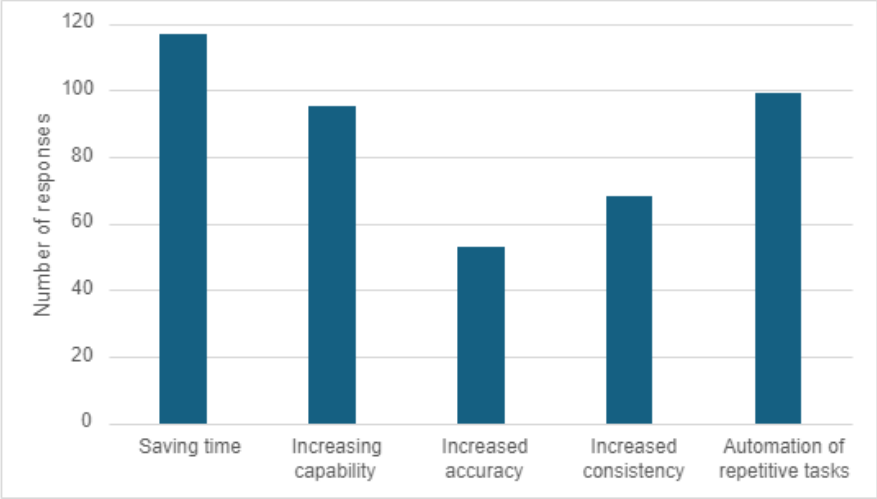


Figure 1: *Key drivers for adopting AI tools in NHS genomics services reported by staff. Multiple responses were allowed. Increased capability included increased compute power.*

Many respondents and interviewees felt that AI may be able to improve consistency of results but would not be able to match the accuracy of a skilled human. Concerns about both errors and a "loss of nuance" in interpretation of results or in writing information for patients meant that respondents were more comfortable with a 'human-in-the-loop' model where experienced staff check outputs and can query and amend them if necessary.

Some respondents noted that they were unsure whether AI was used in some of the software they already used, or alternatively questioned the 'intelligence' or products described as using AI. This has interesting ramifications for both regulation of AI tools that do not quality as medical devices, and for trust and familiarity among the NHS workforce.

AI in use in NHS genomics services

While use of AI[2] was reported in all Genomic Medicine Services, these were largely freely available tools that did not require integration or implementation and could be used by individuals. There was low awareness of the breadth of this individual-level activity taking place, and more generally a strong desire to learn about activity (including planning, evaluation and use) across the country.

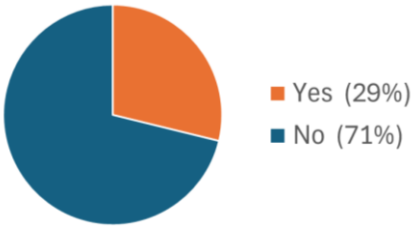


Figure 2: *proportion of respondents reporting current or prior use of AI tools at work. 29% respondents reported using AI; of these, 44% reported only using freely available generative AI chatbots such as ChatGPT, Copilot, Gemini and Claude.*

Over 11% users listed advanced conversational AI assistants like ChatGPT, Claude, Gemini and Co-Pilot as AI tools used at work for tasks including summarising internet-wide information for patients, help with writing code but particularly as a first pass to identify relevant literature and create topic summaries.

Commercially available tools included facial analysis tools such as Gestaltmatcher Face2Gene (FDNA), variant interpretation and prioritisation tools such as SpliceAI (Illumina), VarChat (Engenome) SOPHiA DDM for Genomics (including OncoPortal Plus and Alamut Visual Plus for cancer and rare disease applications, respectively), DeepVariant (Google), and curated genomics information platforms incorporating AI such as Mastermind (Genomenon).

Open-source tools included assistive platforms like Tensorflow and Pytorch which support developers building deep learning models in genomics. Tools such as REVEL and AlphaMissense (which predict the effects or pathogenicity of rare missense variants) and AlphaFold (which predicts a protein's 3D structure from its amino acid sequence), are available as open-source code but were described as commercial tools by respondents.

Those using **in-house developed tools** (including for variant analysis, in bioinformatics pipelines and for searching literature) included a researcher, a bioinformatician and two clinical geneticists as well as a machine learning engineer, and were focused around London and South East England. In-house tools can offer enhanced understanding and transparency, assurance over security and privacy and the ability to custom-build to suit local requirements. However, it requires advanced skill, experience and capacity to develop and maintain and a robust quality management system.

[2] *Artificial intelligence' was not explicitly defined during information gathering to capture as many tools and use cases as possible. Therefore results include some consumer-oriented tools for independent, individual use (i.e. ChatGPT; Bing Copilot) and pieces of software that may incorporate AI/ML algorithms. It is difficult to characterise how substantial the AI/ML component is to the overall software function, particularly where the product is used outside a regulated setting (i.e. does not meet the requirements to be considered AI as a medical device (AIaMD)).*

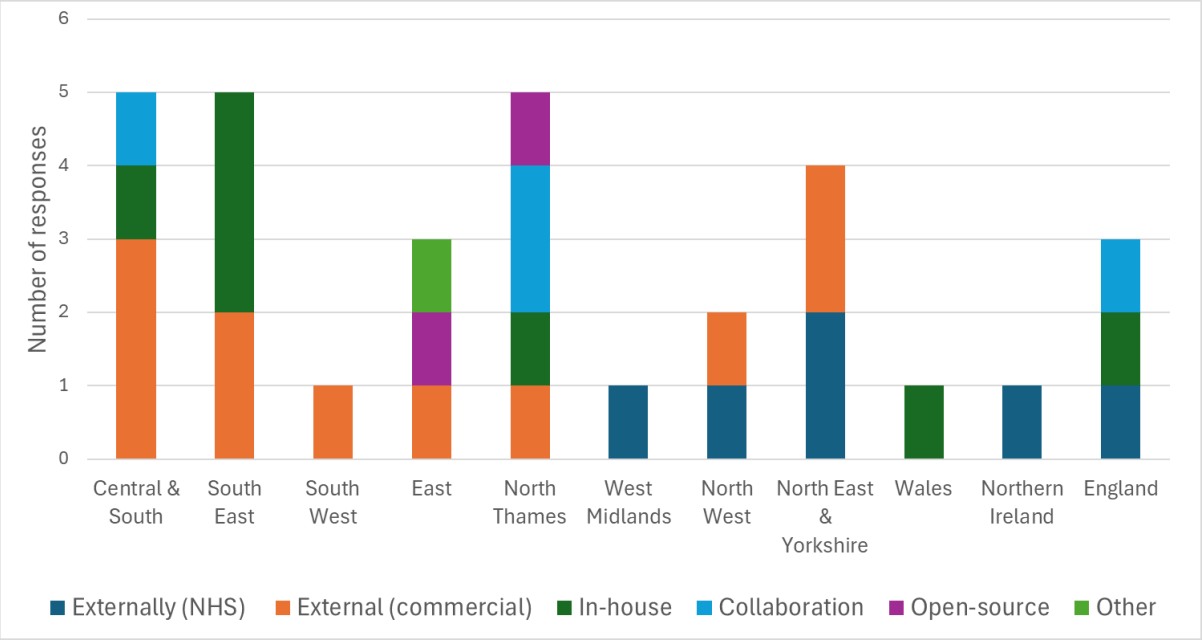


Figure 3: *Source of AI tools reportedly in use across NHS services. In-house developed tools apply to AI developed within the same hospital in which it is used; external NHS source refers to AI developed in another NHS setting.*

Use of AI tools was reported across multiple different staff types. Geneticists and clinical scientists constituted the vast majority of survey respondents and so there may be wider use of AI among other staff types that is not recorded here. The range of roles using AI suggests broad application and utility.

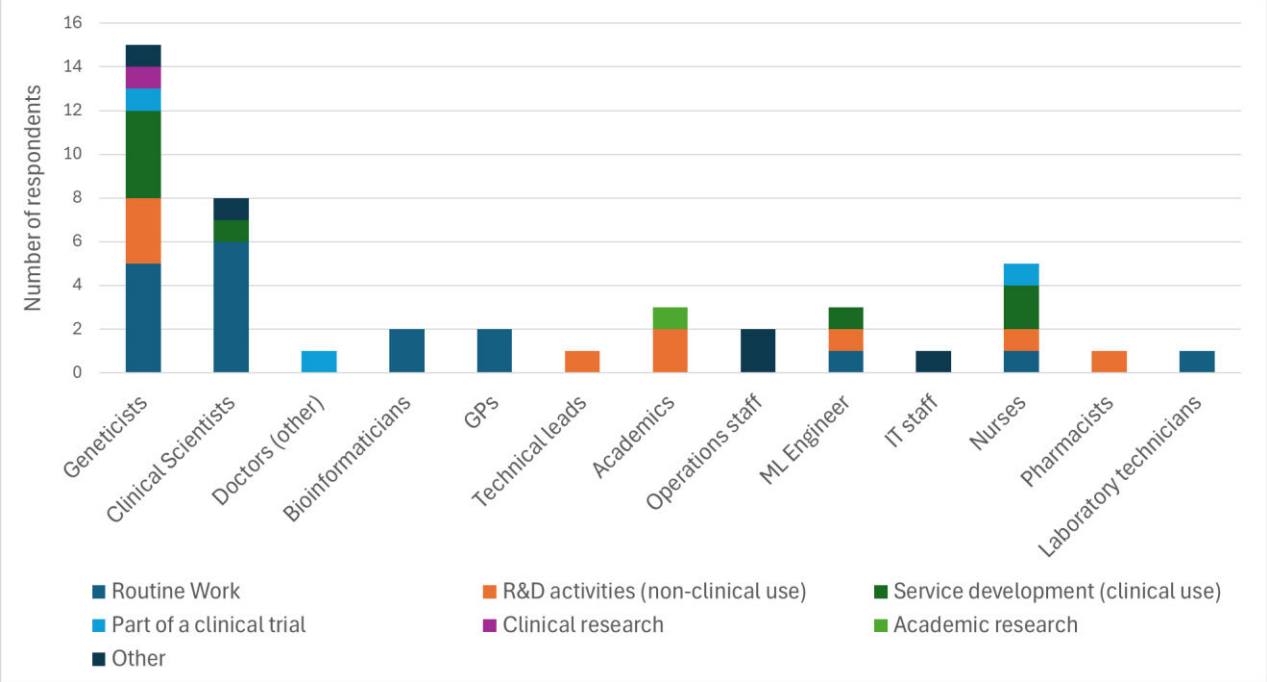


Figure 4: *Use of AI tools in genomics by staff type. 'Doctors' includes medics who did not identify as clinical geneticists. GPs are general practitioners with special interest in genetics and/or genomics. Operations staff includes programme manager, administrators and training professionals not in clinical roles.*

Applications where AI was reportedly in **routine use** included:

- Administration (sending patients appropriate questionnaires based on key words from their enquiry before a GP e-consult; clinical dictation tasks; linking clinical terms to HPO terms for test requests)
- As a writing aid (emails, letters and summarising information for patients)
- To assist engineers and bioinformaticians with coding
- Searching literature and collating and summarising relevant information before targeted searching
- As a variant interpretation aid (SpliceAI), or for prioritisation (Emedgene) or pre-classification of variants
- Variant effect prediction via DECIPHER
- Deep phenotyping

Applications where AI was tested as part of **clinical service development** include:

- Transcribing clinic notes and writing letters (OSLER Tortus)
- As a variant interpretation aid for rare disease
- Facial analysis (Face2Gene, FDNA)

AI was used in **non-clinical research and development** for:

- Filtering mutations
- Classification of copy number patterns
- Identifying biomarkers of cancer prognosis in digital pathology
- For data extraction using natural language processing.

AI tools identified through the questionnaire, during interviews and by targeted searching could broadly be categorised as follows (Table 1):

Writing & administration	**Literature searching**	**Phenotyping**	**Variant analysis**	**Decision support tool**	**Data processing & analysis**	**Bioinformatics pipelines**	**Coding**
OSLER Tortus	Mastermind Genomenon	PhenoScore	SpliceAI	XGBoost	Nucleotide Transformer	NVIDIA Parabricks	Tensorflow
Generative AI for patient resources	*In-house tool in development*	PhenoTips	SOPHiA DDM for Genomics • OncoPortal Plus • Alamut Visual Plus	Visiopharm apps (pathology)	DNABert	Microsoft machine learning	Gitlab co-pilot
Open AI ChatGPT	Open AI ChatGPT	Face2Gene	DeepVariant		HyenaDNA	Huggingface	DeepVariant
Microsoft Copilot	Microsoft Copilot	Eye2Gene	VarChat Engenome		Tensorflow		Pandas ML library
AWS Claude			CADD		IBM SPSS		Pytorch
Google Gemini			REVEL		JASP		DeepAI
Dragon Medical One			Illumina Emedgene		PSPP		Scikit-learn ML library
			Fabric Genomics				
			Exomiser				

Table 1: *AI tools reportedly in use in genomic medicine services, grouped by primary application.*

Potential use cases for AI within NHS genomics services

Maturity and acceptability of AI tools

It may be useful to consider AI tools in terms of technological readiness, system readiness and workforce readiness (including trust and skills). Tools suitable for immediate implementation would have high technological, system and workforce readiness and a low threshold for trust (i.e. processes or their outputs are understandable; errors are infrequent, simple to identify and remedy).

- Technological readiness describes the performance (reliability, sensitivity, specificity), useability and level of evidence for the AI tool in clinical trials or real-world evidence. It also includes practical considerations such as regulatory clearance and DTAC compliance (if applicable).
- System readiness involves the compatibility of the AI tool with IT systems, including both performance and ability to access relevant data, especially if this requires access to multiple systems. It also involves assessment of the data collected and available and whether this is sufficient for the AI tools, privacy and security (how data is accessed, processed and held, for what purpose, and what guardrails are in place).
- Workforce readiness describes the willingness and ability of relevant staff to engage with the AI tool as intended to ensure safe use and support efficiency. Building readiness may involve general awareness or more specific training, careful planning to ensure there is sufficient capacity to scope, plan and deliver AI tools, planning to ensure adoption and deployment do not negatively impact staff, and review of processes and pathways to minimize disruption to busy services.
- Other factors

Trust of the technology is paramount, and relies on evidence for and understanding of the technology itself and its ability to perform as intended in a particular operating environment in different scenarios. Trust could inhibit adoption (taking a risk averse stance) but there is also a risk that over-trust could lead to skills loss if human input is lost in certain tasks or that AI-derived results may be incorrect or used inappropriately. It is therefore important to share knowledge and experience and to build awareness and skills among staff who could procure or use AI tools to ensure they understand the opportunities and limitations of AI; applications where it may or may not be suitable; and so they have the skills and confidence to interrogate outputs of AI tools in use and evidence for tools prior to procurement.

Potential use cases identified by respondents

Respondents were asked which AI tools they used or were aware of and what they considered were the most immediate and impactful application of AI in genomics (figure 5). Immediate use cases were defined as those where AI can be implemented now, and included clinical clerical tasks, variant analysis and other forms of data analysis. Impactful applications with immediate impact were defined as those with potential to deliver meaningful improvement to services or patient outcomes either now (figure 5B) or in five years (5C).

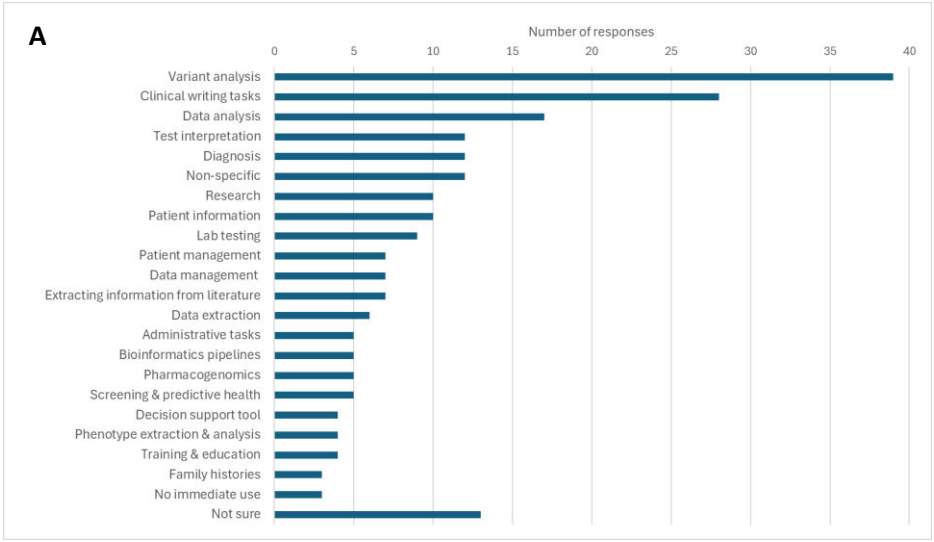
Interestingly, many respondents cited clinical clerical tasks as most immediate and impactful uses for AI, reflecting the large amount of time they consume. Examples of tools ready for immediate application included **generative AI writing assistants** as they are easy to use with minimal knowledge or training in AI; easy to review outputs for accuracy and amend errors and expedite. However, these are unlikely to substantially change pathways. Data access requirements and computational requirements are minimal so existing information governance processes and IT processes and hardware may be used. Many staff reported that writing letters and information sheets were substantial and time-

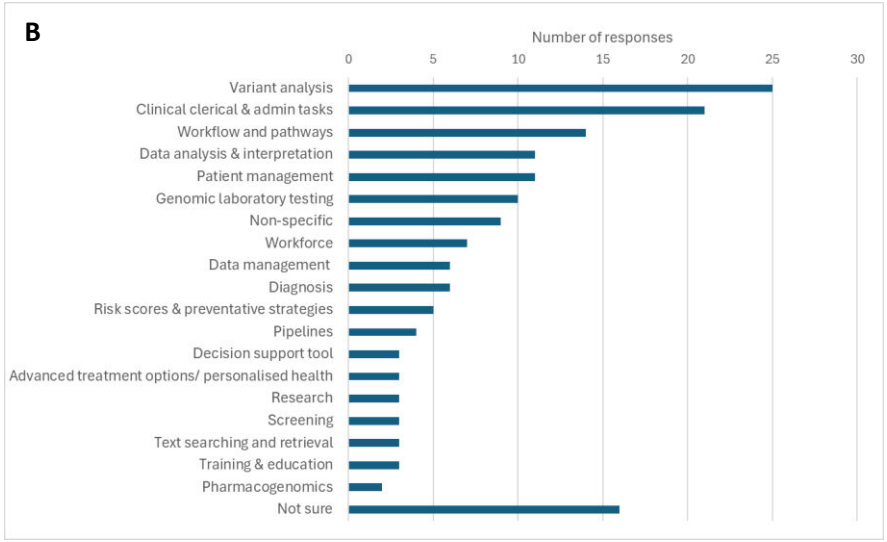
consuming elements of their role and felt AI could save around an hour for each session. Multiplied across staff and departments, this could release a large amount of time for patient-facing activities or service improvement activities.

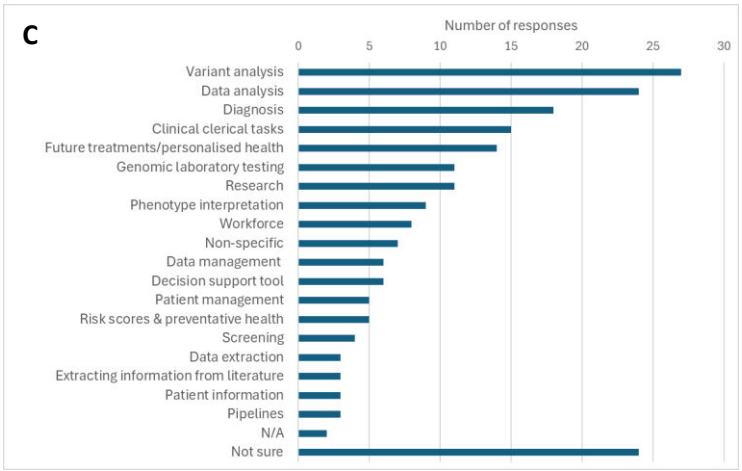
When asked about applications that could see most impact in 5 years, respondents answers moved towards tools likely requiring advances in knowledge, accessibility of data, and performance. The shift in use cases over a 5-year horizon suggests that respondents are optimistic about the potential for AI in genomics.

Awareness of AI use cases in genomics

A quarter of respondents were unsure or unaware of applications for AI in genomics. Other respondents were aware of manufacturers (e.g. Illumina, Quagen) claiming AI functionality on brochures but did not understand what this entailed. Respondents were aware of variant interpretation and analysis tools (such as SeqOne, Exomiser, Revel and Splice, although few named specific tools), the dysmorphology tool Face2Gene, generative AI tools and chatbots, tools for coding and bioinformatics and protein modelling tools such as AlphaFold and AlphaMissense. Increased awareness and knowledge sharing about these tools already in use could support expanded adoption.







Variant analysis includes identification, characterisation and prioritisation.

Clinical clerical tasks refer specifically to tasks performed by clinical or scientific staff such as report and letter writing, whereas administrative tasks include tasks such as requesting forms and tests and scheduling and managing appointments.

Data extraction from databases refers to access and collation of relevant data from disparate sources.

Extraction from literature includes searching for keywords.

Non-specific applications included automation of repetitive tasks and increased efficiency, without description of specific functions.

Figure 5: *Applications of AI with (A) immediate application (i.e. those already in use or ready for deployment now); (B) immediate impact (i.e. applications already in use or ready for deployment with most impact to the service; (C) most potential impact in 5 years (i.e. technologies not yet ready, or with immature infrastructure to support them, that could have significant impact to services in the short-term future).*

Pathways to adoption

The questionnaire results showed a genomic medicine staff have low level of confidence in identifying genomic AI tools, local requirements and the contacts needed to adopt AI. Most of those who were confident in identifying suitable AI products had previous AI education or experience. Confidence appeared closely linked to an individual's role and prior experience in the different area (procurement, identifying tools, contacts etc). Very few respondents had combined confidence in identifying products, requirements and contacts, illustrating the need for resources, a multi-skilled project team, the ability to find the right contacts, and Genomic AI champions.

No formal, standardised pathways to adoption were discovered during this scoping project. Some survey respondents specifically identified a lack of knowledge of how to go about AI adoption, pathways to follow and who to engage. A systematic approach to reviewing system readiness (data infrastructure and interoperability) could be a useful method for other services to identify opportunities to deploy AI (or where AI may not be suitable) and engage with impacted staff in the process.

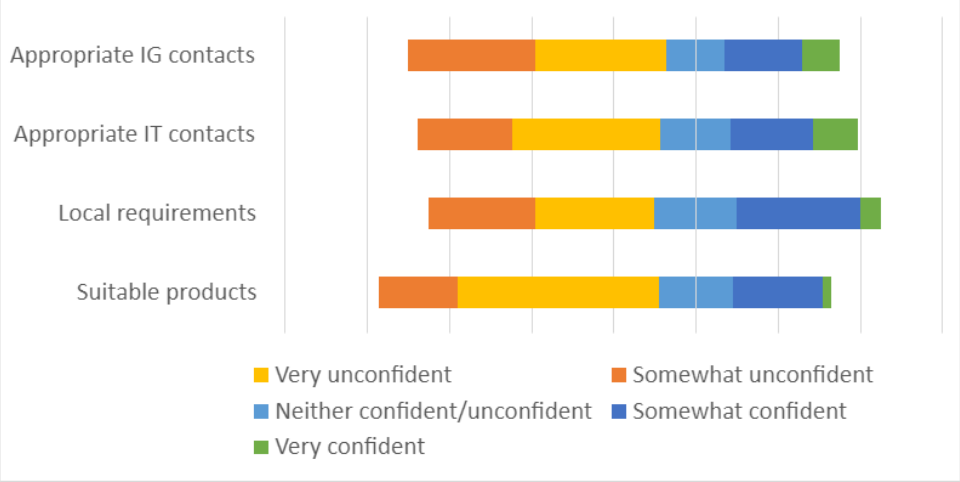


Figure 6: *Level of confidence identifying appropriate local information governance (IG) and information technology (IT) contacts; local requirements (i.e. use cases, specifications, performance criteria) and suitable products as part of adopting an AI technology in the department.*

A summary adoption pathway, using themes from interviewees discussing previously adopted AI tools, is presented in figure 7.

Tips for AI adoption and implementation

When looking into the adoption of AI tools, input from immediate stakeholders will be useful to map the pathway, understand requirements and constraints and understand readiness and willingness to adopt AI. Questions to ask stakeholders may include how the tool will interact with existing software and platforms; whether additional infrastructure or resourcing is required; how the clinical pathway will be affected (including potential changes in staff performing certain tasks, or potential changes or movements in bottlenecks). If available and with support of stakeholders, a trial period may be useful depending on cost and capacity. Stakeholders should remain engaged throughout decision making and procurement to ensure smooth implementation. Validation requirements and quality assurance procedures will vary between AI tools and applications but should be agreed in advance with stakeholders. Pilot testing or small-scale phased introduction can help to identify any issues (e.g. with performance, integration, impact to workflow or staff) before wider roll out. This is an opportunity to adjust, gather feedback, and prepare for potentially larger scale adjustments such as reconfiguring workloads.

Additional notes on implementation of AI tools:

- Monitoring of aspects of suitability, such as impact on the workforce and workflow, should continue throughout the project as more stakeholders become involved and collective understanding of the project develops.
- Processes for data sharing vary across NHS England and can differ between applications. Therefore, early local awareness and ongoing engagement with information governance teams at the relevant Trust is important.
- Costs for AI products can vary according to factors such as purchase model (e.g. outright purchase; subscription; bundle with other products) and scale of projected usage. In some cases, it might be difficult to identify accurate costs so engagement with other teams who have adopted or implemented comparable technologies can be useful in addition to discussion with manufacturers and distributors.
- Advice on pilot study design may be sought from other departments with experience with similar products but should involve locally relevant evaluation criteria and engage the staff who will be affected. There should be no obligation to purchase associated with a pilot trial; however, costs associated with staff to perform the pilot and analyse data should be considered upfront.
- Scoping and adoption of AI can be a time-consuming process involving multiple stakeholders. Although AI may offer time savings it will also cost time to implement and may also take time to validate and perform ongoing QA tasks. This should be considered before decision to adopt, and resources that could help reduce time required (i.e. support from the manufacturer; advice from previous users) identified.
- Trusts may wish to engage with their associated universities or research institutes when implementing AI tools to benefit from access to researchers with appropriate expertise and ability to attract research funding. This may help to ease some barriers noted above by securing additional resource. Larger research centres able to draw on additional resource are well placed support development and translation of AI tools developed in academia or more substantial AI tools that require evaluation and integration.
- Caution should scale with the complexity of the tool, scale, maturity and sensitivity of data it uses; transparency of the process (and whether a human-in-the-loop approach can be used); performance and risk of harm.

Acceptability & trust	Technology readiness	Infrastructure readiness	Potential impact
Ease of use	Defined clinical need	Security and privacy	Impact on patient care
Local evaluation and pilot	Suitability for use case	Governance processes	Efficiency/throughput
Ability to understand outputs and ability to intervene	Acceptable cost	Adequate data storage	Impact on patient experience
QA and validation processes and available guidelines	Robust training and validation	Fit with existing pathways and processes	Impact on performance and capability
Understanding of expectations and limitations with training	Performance at least matches standard care	Integration and reporting with clinical systems	Up- and downstream consequences
Good level of evidence	Guidance on place in care	Adequate compute power	Impact on staff experience
Engagement with and support of staff and patients	Understanding of limitations	Appropriate scale and diversity of training data	Overall impact to workforce

Table 2: *summary of items to consider when reviewing AI Tools, grouped into themes.*

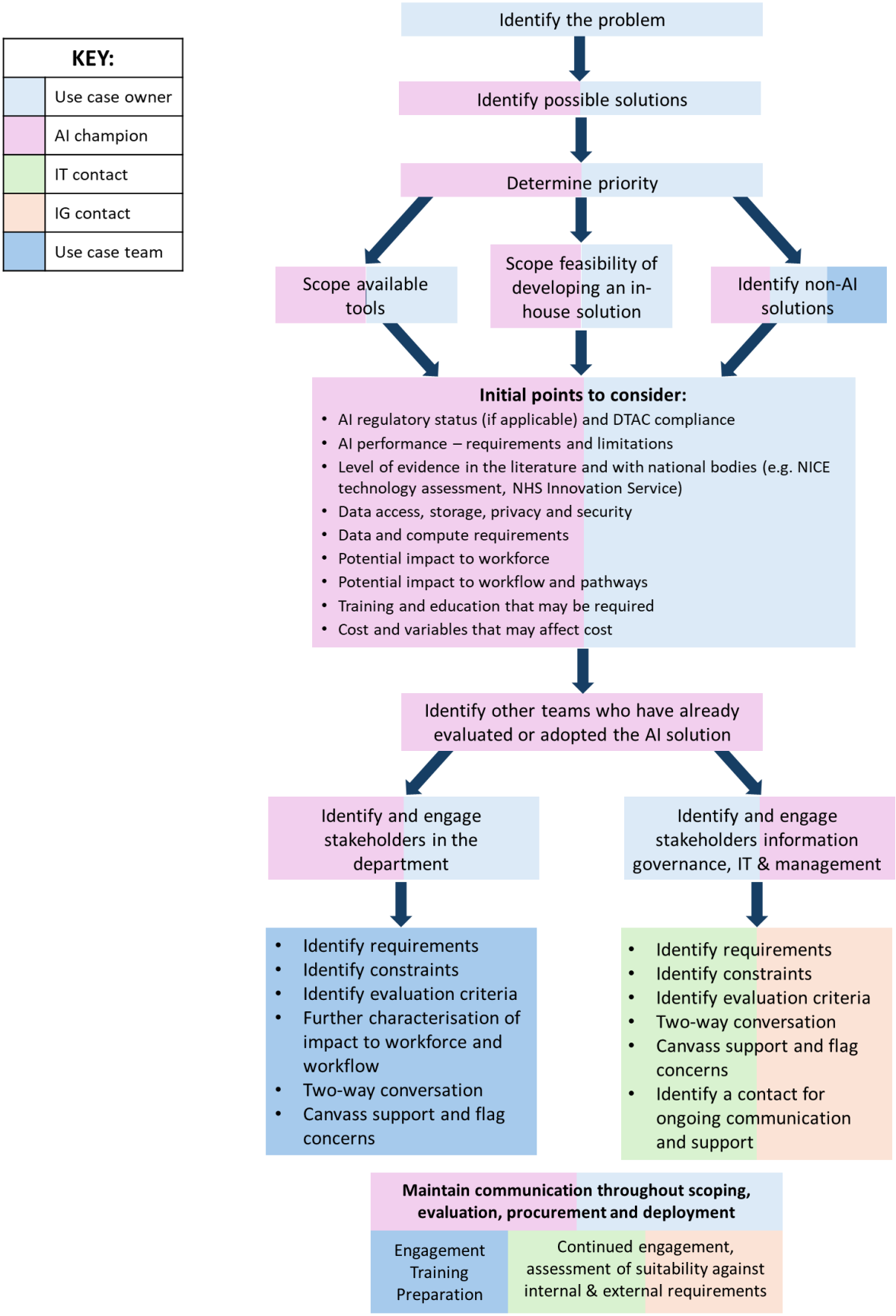


Figure 7: *Summary pathway for adoption of AI in a clinical setting, based on collated insights and feedback from interviewees. The use case team are the staff who work on the process or pathway where AI is being considered.*

Staff preparedness for AI

Very few Genomic Medicine Service staff responding to the survey reported formal training in AI. Informal on-the-job and self-learning were the most common forms of training reported. The need for appropriate education and training was the most cited barrier to adoption by NHS genomics staff surveyed.

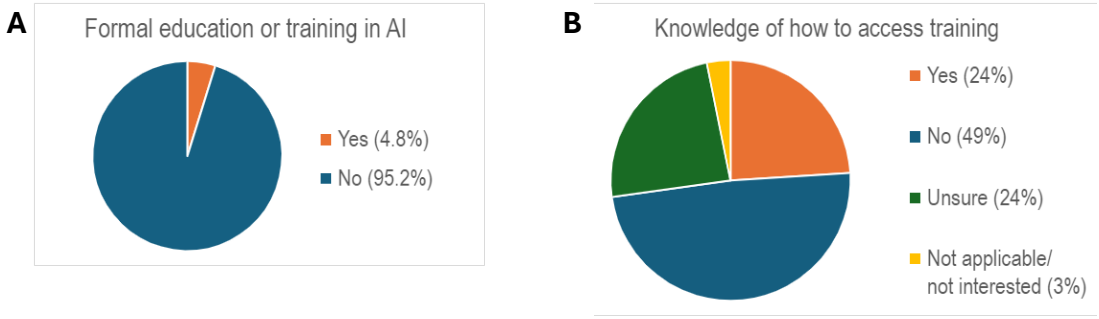


Figure 8: *proportion of survey respondents who had received training or education in AI (A) and proportion of respondents who were aware of where and how to access relevant training in AI (B).*

Lack of training and prior experience were the dominant reasons for feeling unprepared for adoption of AI. Lack of awareness of the tools available and what was already in use in other services was another common reason cited. Other reasons for feeling that genomics is unprepared were issues of interoperability (i.e. with commercial EPR providers), compute requirements, data storage costs, trust and generalisability of AI models, a lack of training and knowledge of available tools.

Those who felt prepared caveated responses with assumptions that training, support, guidance, money for skills acquisition and updated computer systems would be required. One respondent noted that immediate use cases for AI are “no different to any other black box”. Therefore, existing strategies to safely use and monitor other black boxes could be applied to AI tools. Fears around “black boxing” could also be ameliorated by deploying AI in “human in the loop” protocols with careful oversight or inputs and outputs to limit risk.

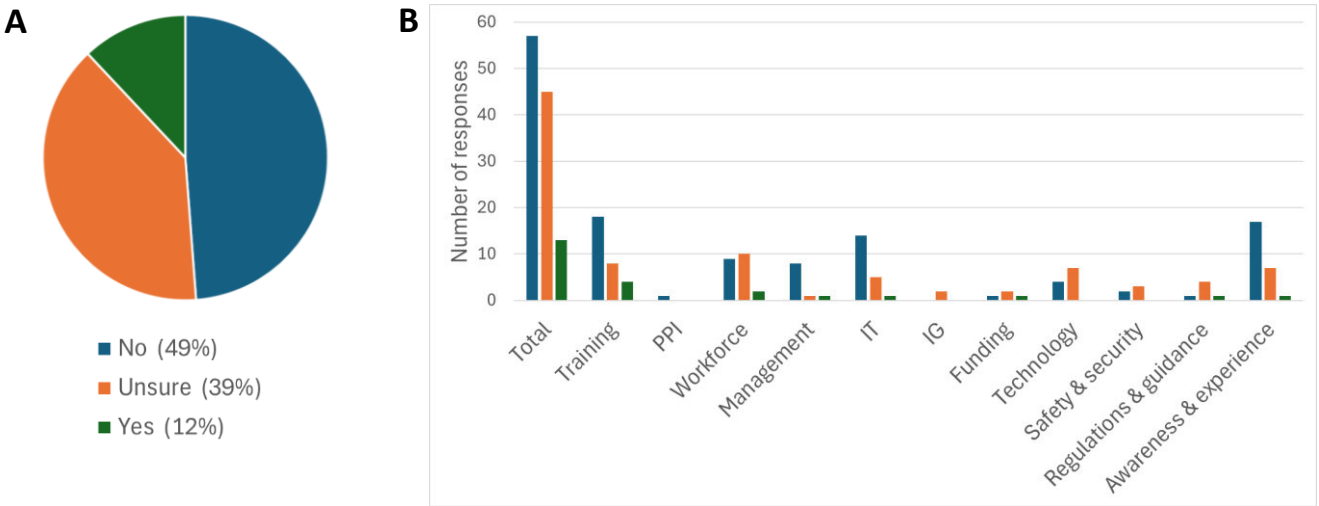


Figure 9: *Feeling of preparedness for introduction of AI to NHS genomics services (A). The reasons for perceived preparedness, collected through an optional free-text field, were categorised by respondents who felt prepared, unprepared, or unsure (B). Explanations for feeling prepared included either existing measures (such as received training or a good awareness of available tools) or hypothetical conditions (such as the need for appropriate training, funding for necessary skills, IT system upgrades, or additional guidelines).*

Provision of training and education resources and a platform to share knowledge, experiences, best practice and learn about available AI tools were the most frequent reasons or requirements cited for preparedness. A skilled team including an AI champion to discuss options with the implementing team, guide the project and communicate with stakeholders was the dominant theme for workforce (excluding training for the team, previously discussed). Respondents also noted the role of management in supporting evaluation and introduction of AI tools and of national-level plans for AI for the genomics medicine services with agreements on IT and software use to promote standardisation.

Barriers to adoption

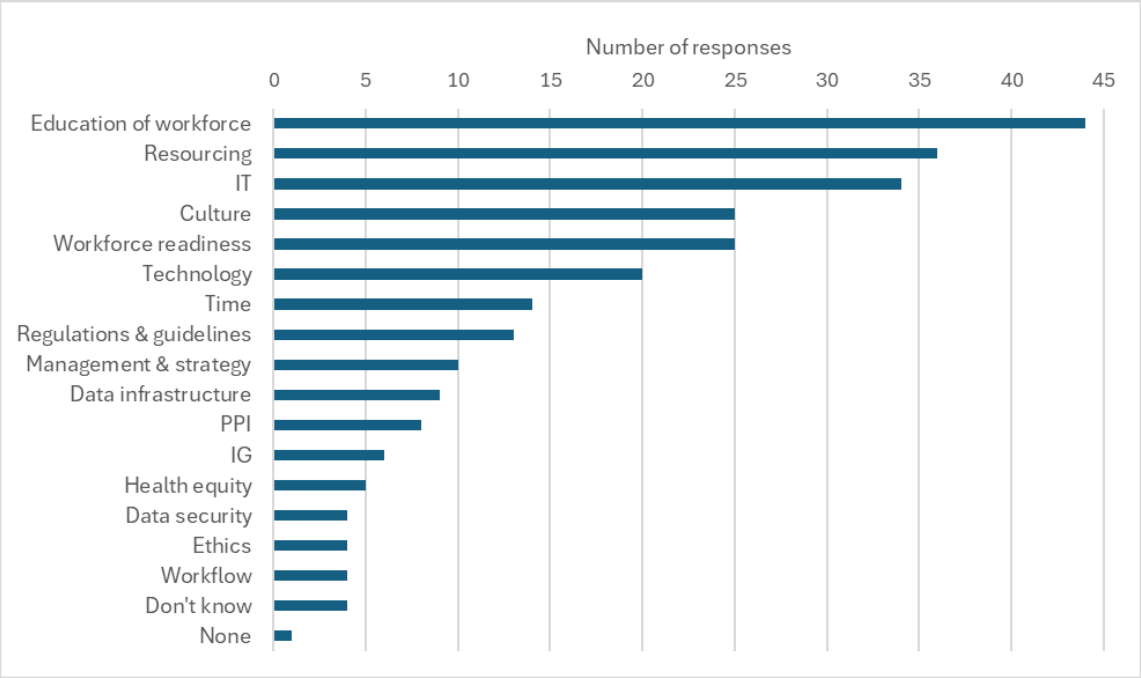


Figure 10: *Reported barriers to adoption of AI technologies in NHS genomics services, grouped and ordered by most reported emerging theme.*

A lack of **education**, training and upskilling was the most common barrier identified. Respondents described a range of training and educational requirements ranging from basic AI literacy to understanding potential applications, limitations, and evaluation criteria; training in the use of specific tools through to formal education programmes.

Resourcing requirements were expressed as a lack of funding (either generally, or for training, hiring to bring in new skills and additional capacity, or to pay for AI tools and required data storage and compute power). This was not expressed as a concern about the cost of AI tools, rather a more general lack of funding to develop the workforce and infrastructure to bring in AI, while maintaining performance in an already stretched service.

IT was frequently cited a barrier to AI adoption. Respondents felt that current IT hardware (physical equipment such as computers and servers) and infrastructure (networks and software) would not be adequate for AI and a lack of interoperability between systems would limit potential AI tools. Lack of compute power and high-performance computing resource were also common themes. Less common but important concerns were lack of data storage (and associated cost and security concerns) as well as physical space to host new IT infrastructure.

Workforce concerns relate to the time and cost of revising pathways; a lack of guidance on standardising pathways nationally, and lack of capacity to engage with AI training, scoping, preparation or implementation. The impact of disruption on staff and patients when introducing new tools was also noted. Respondents felt that there was insufficient staff capacity to scope or implement AI.

A perceived barrier was the potential negative **impact of introducing AI tools to established workflows**, as well as the lack of resource to properly assess and plan for bottlenecks and resource use. Tools that do not fit into existing pathways can incur additional cost, training, review of surrounding pathways and may deviate from existing best practice guidelines. For example, one case study was discussed where a clinical department adopted an AI tool was adopted which was well received by users but fell out of use a short time after implementation because the pathway (including relating administration and validation work) around it had not been revised and maintained. Respondents had concerns about their ability or capacity to properly consider these aspects during scoping and implementation.

Institutional cultural barriers described perceived institutional attitudes of risk aversion, resistance to change, inconsistent messaging and prioritisation and a 'fear of the unknown'.

AI technology itself was cited as a barrier, with some respondents suggesting that AI was not yet ready for clinical deployment in their field. Inadequate reliability, accuracy, and trust or confidence in results meant some respondents felt that AI could lead to mistakes or require additional time to check and troubleshoot. Other facets included a low level of evidence and clinical validation, and a sense that tools were not well matched to clinical need.

There was concern that **regulations & guidelines** would struggle to keep pace with rate of change in the AI landscape and a flexible framework might be required. There was reluctance to adopt AI without adequate regulation in place related to safety concerns and inconsistent use between different operators. However, guidelines and regulations were also referenced as a tool to guide accountability, limit the risk of misuse and provide reassurance that tools met standards of performance and validation, and as a central, external resource to support staff.

Respondents also cited a **lack of time** to learn about AI and to plan for adoption, implementation and reconfiguration of workflows. "Investment of time" to the service is essential to ensure staff have capacity to engage with training and education. Departments need time to investigate suitable tools, engage with stakeholders and distributors and evaluate AI tools before adoption, while maintaining normal service levels.

Data infrastructure and a lack of interoperability between different systems was frequently named as a barrier by those with experience with data science and machine learning. Difficulty moving data around, accessing relevant information and annotating data were all given as barriers.

The main themes of **management & strategy barriers** were bureaucracy and red tape (making it difficult to bring in new technology) and concerns that priorities may not be clear. In some cases, respondents were concerned that existing projects would be prioritized over AI (focussing on short term priorities or stressors); others were concerned that the AI "fad" would take precedent over core priorities to the overall detriment of the service.

PPI barriers included uncertainty around how to explain what AI is and how it was being used to patients and concerns around public perception of AI. Good communication, including the patient voice, would be needed to build trust and acceptance.

Information governance encompassed a lack of familiarity with how to identify and address data governance and confidentiality issues. Many interviewees described genomics as a special case for information governance because of

the intimate nature of genomics data and the potential to identify an individual after normal anonymisation procedures have been applied.

Low data readiness was perceived as a key barrier to those with experience of AI and/or those working in machine learning and data science roles. Extremely low availability of meta data (data which describes the main data being analysed) and difficulty in accessing relevant supporting information (such as relevant pathology data or family records) quickly were discussed during interviews. These were not seen as barriers to implementation of AI, but as barriers that would limit the application and potential impact of AI. Respondents also noted data maturity concerns such as limited diversity in genomic data thus impacting training data sets, and poor understanding of rare disease in non-white populations. Some interviewees noted concerns around potential for regional disparity and inequity of access to accelerated or additional testing and analysis, with some centres (larger centres with more research capacity or links) more ready and able to adopt AI than others.

Ethical concerns were raised relating to health inequity and data bias but also related to patient communication and consent and concerns around black box AI. This includes both concerns about data flow and (mis)use as well as interrogability and understanding the processing of their data by AI tools. Concerns about 'black box' AI tended to be applied to AI in a very general sense rather than specific tools for specific applications in genomics.

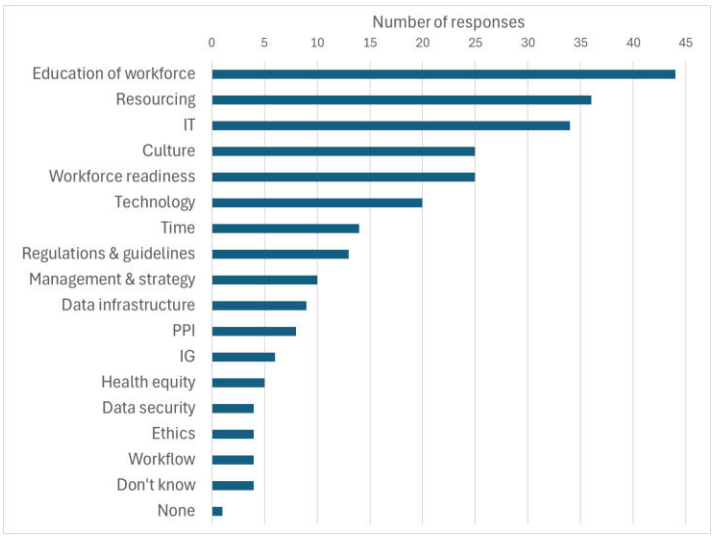
Concerns about adoption of AI

While the overall pattern was consistent, concerns about introduction of AI were expressed differently when respondents were considering national adoption versus adoption in their own department. The primary concern at both local and national scale was patient safety and the risk of inaccurate or hallucinated outputs that could lead to misinterpretation or misdiagnosis. At a local level, this was focused on the patient but at a national level additional themes were introduced such as accountability for potential errors. This was particularly of concern if AI was to be used in place of staff, taking humans out of the loop, rather than as an aid to support staff.

Technology concerns ranged from general mistrust to a concern that the technology was not yet sufficiently mature for use within the respondent's field. Some raised concerns that AI tools were not applicable to clinical scenarios, or would require substantial change to workflows. Loss of accuracy and nuance were also common themes. One interviewee raised concern that, without safeguards or transparency, AI-driven findings could be incorporated into widely used databases such as ClinVar with only a low level of evidence. These findings may be accessed and used without knowledge of origin or evidence base and could potentially be overinterpreted.

Only one respondent raised concerns about the potential ecological impact of the energy required for the increase in data storage and computation. Another aspect of high power consumption discussed at interview was the very high cost of running a currently available AI variant calling tool in pipelines which make its use cost-prohibitive for the performance achieved.

Workforce concerns included both comments on insufficient staff and concerns about job losses and AI replacing skilled professionals. Deskilling of the workforce was a common concern related to overreliance on AI, with fears that a future workforce may lack the skills to identify when an error occurs and remedy it. The role of human sensitivity, care and nuance in genomics was frequently described, with concerns that this could be lost or de-prioritised in favour of efficiency.



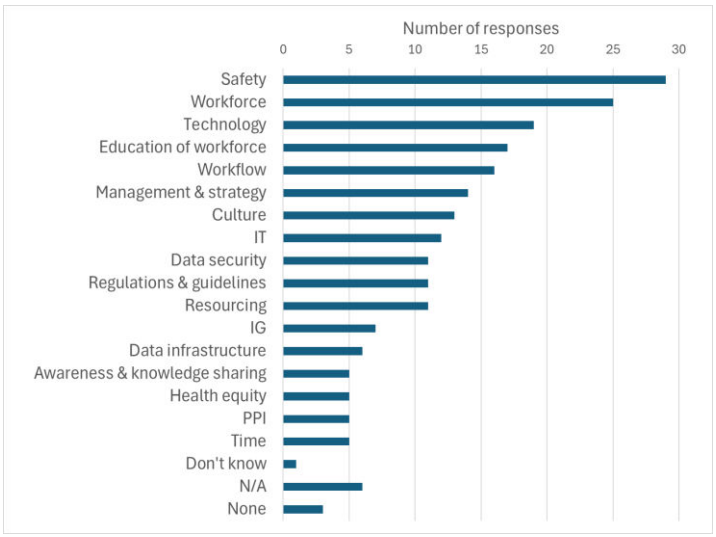


Figure 11: *Concerns expressed by staff regarding introduction of AI to genomics services in the NHS (left) and to the respondent's own department (right).*

Findings and opportunities for GAIN

This section focuses on opportunities to provide resources and address barriers where they are feasibly within the influence of the GAIN network, and relate to the barriers and concerns discussed in the prior sections.

While workforce pressure, resource and IT constraints are well noted they may not be influenced directly. It is therefore important that AI is considered as part of overall strategies to address these recognised issues, and that where AI is adopted there has been careful evaluation of the context and potential impact to the department or Trust. The strong theme among interviewees with significant experience with AI and data science was the need for data maturity to grow the potential applications and impact of AI. This will involve digitisation of paper-based processes, ensuring digital documents are machine readable and use consistent clinical coding, ability to annotate records and ability to link data.

Resources that would be required to support adoption and expansion of AI in NHS genomics services are largely reciprocal to the barriers and concerns with focus on more achievable tasks. Comments on funding and investment focused on training, knowledge sharing and conference attendance and most notably in IT staff and equipment.

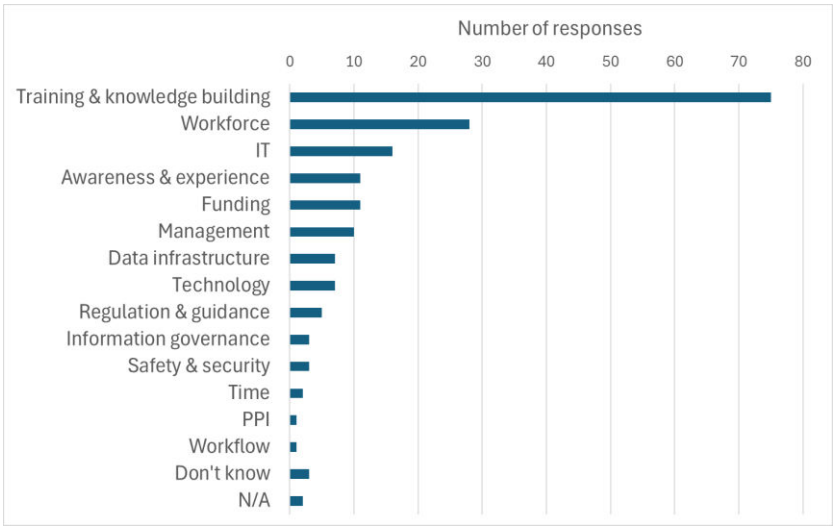


Figure 12: *Resources required to support introduction of AI to NHS genomics services, reported by staff.*

A lack of training and education was consistently cited as a key barrier to adoption of AI tools. Lack of awareness of AI tools available and of potential applications for AI were also frequently discussed. Nearly all respondents, including those who were active in the field, had a low awareness of where AI tools were being used in the NHS outside their own department (however, many were aware or assumed that Genomics England used machine learning algorithms).

Measures the Network could implement to develop awareness, knowledge and knowledge sharing include:

- **Basic training or resources on AI** to build familiarity and sufficient AI literacy across the workforce to understand the strengths and limitations of AI and enable engagement at scale
- **Support identifying problems where AI could offer a suitable solution** (and problems where AI may not be a suitable solution)
- **More detailed training on identifying and implementing suitable AI tools**, including mapping pathways and stakeholders, delivery and surveillance, for individuals and teams seeking to adopt AI for a specific application
- **Access to technical expertise** to support local and regional teams, as well as information to support procurement applications
- **Provision of a forum for knowledge sharing** to agree and consolidate best practice and ensure that AI-enabled processes support safe, efficient and equitable care
- **Description of available AI solutions,** particularly those with regulatory clearance (if applicable), a strong evidence base and that could integrate readily to existing systems and pathways
- **Case studies of AI adoption (successful and unsuccessful)** across NHS England genomics services, including description of the use case, processes followed and lessons learned.

Staff with patient-facing roles may benefit from training in **how to explain AI to patients**, supported by resources for wider patient and public engagement. It may be helpful to root patient communication in specific examples with case studies to build trust and understanding.

It is widely recognised that **existing datasets lack diversity** with over-representation of white European data. There was concern that unrepresentative training and validation data could exacerbate existing health inequalities. While some felt that synthetic data could be used to address this in the short-term, this was felt to be at high risk of error and bias propagation. Instead, investment in engaging with under-represented groups is required to build more diverse, representative datasets that would be used for training Genomic AI tools. 'AI champions' were commonly cited as key factors in successful implementation and more generally in building a culture and awareness of AI. It therefore could be useful to create a person and role specification that could be shared across department to describe more fully what the role entails and the benefit. Communication between 'AI champions' across the country would promote awareness of activity, opportunities for shared learning and peer support.

A minor theme from interviews was **the importance of user-led design**, or development of AI solutions with a clear description of the use case, operating environment and user requirements early in design. The research for this report suggests that most AI tools in use were externally sourced. Therefore, mechanism for identifying use cases and working with AI tool developers to help promote development of tools that fit within existing pathways to help reduce the complexity of implementation and modification, may be helpful.

The same principle could be applied to tools being developed in academia. There is an opportunity both to develop calls for research focused on particular use cases or challenges. Equally, there is an opportunity to provide 'matchmaking' services for research close to clinical translation with an NHS team with a corresponding use case. This could support

national exposure away from the larger research-intensive centres, but support navigating data sharing and evaluation may be required.

While efficiency was the primary driver for adoption of AI, the **principle concern was patient safety**. Many respondents noted a lack of evidence for AI technologies in their discipline and a lack of understanding or guidance on how best to evaluate them. The Genomic AI Network of Excellence could be a useful forum to collate evidence and work with NICE to summarise strength of evidence, gaps in evidence and recommendations for further research.

Regulation and guidance for AI tools was also a strong feature of discussion. The Office for Artificial Intelligence outlined five key principles to inform responsible development and use of AI: safety, security and robustness; appropriate transparency and explainability; fairness; accountability and governance; contestability and redress[3]. This requires regulators such as MHRA to issue guidance on how these principles interact with existing legislation, produce clear and consistent guidance, and monitor their own effectiveness at regulating AI[4]. The principles and strategy align with many of the concerns highlighted in this report. Increased awareness of regulation, legislation and consultation may help support meaningful engagement and assuage more general concerns. However, they are not specific to genomics and may not capture requirements for AI tools that do not meet the threshold for AI as a medical device (AIaMD). Creation of guidelines for safe use and monitoring would provide useful, relevant and actionable information for genomics services.

To promote **awareness of available tools** (and their performance and limitations) for a specific application, it may be useful to include relevant AI in existing guidelines rather than create an additional general document. This could be achieved by liaising with the issuing bodies to identify suitable, relevant guidelines and provide input to future revisions, or by leveraging expertise within the network to develop consensus-based documents on appropriate applications and use that may be referenced by the guidelines. This would require input from experts familiar with the application and tools but could be implemented in a relatively short timeframe and be widely accessible through existing established channels. Awareness and access to any recommendations or guidelines are imperative to ensure uptake and equity across services in England.

Communication within and between services, including dissemination of experience and learning from AI evaluation and implementation would also support increased awareness and knowledge.

[3] Office for Artificial Intelligence (2023). A pro-innovation approach to AI regulation. Department for Science, Innovation & Technology policy paper. https://www.gov.uk/government/publications/ai-regulation-a-pro-innovation-approach/white-paper

[4] Medicines & Healthcare products Regulatory Agency (2024). Software and artificial intelligence (AI) as a medical device. Guidance. https://www.gov.uk/government/publications/software-and-artificial-intelligence-ai-as-a-medical-device/software-and-artificial-intelligence-ai-as-a-medical-device

Next steps

The work presented so far discusses tools that have been identified as being used in NHS genomics services. This may not include tools in development, or early in evaluation or procurement. Further engagement with senior members of all GMSAs and GLHs could be undertaken to identify these, and to gain additional detail on evaluation and procurement processes for commercial and research AI tools.

Secondly, preparatory research identified some relevant open-source and commercially available AI tools that were not mentioned in the questionnaire responses or during interviews. Given the localised and fragmented awareness of AI activity, it is not possible to know whether these tools have not been deployed; have been deployed but not reported; or have been evaluated and subsequently abandoned. Targeted searches of literature and trial databases could subsequently be conducted to complement these results, and to explore reasons for non-adoption where applicable.

Targeted literature searches and additional interviews could be used to identify earlier stage research that may become suitable for translation and adoption in future and assess the increase in technology readiness and IT infrastructure, data required as well as potential impacts to staff and workflows.

The present work suggests a lack of defined pathway for adopting AI tools and a low level of confidence among staff in knowing who to contact in IT and information governance teams and how to navigate those processes. A common theme among survey respondents was a desire for centralised processes and advice and confidence that they would be acting in line with best practice and with others across England.

The research identified early adopters, innovators and expertise both in the NHS and in translational research settings which could support creation of a technical experts group to drive the Network. It also identified barriers and concerns experienced by the workforce. Addressing these issues, with input and engagement from the genomics workforce, should develop awareness and knowledge and ensure activities are done in partnership and will good understanding of application requirements and constraints.

Appendix

A questionnaire was distributed across NHS England genomics services by the Network team. The survey was intended to map the extent of AI use, awareness, readiness and appetite across NHS England. Respondents included service and programme managers, clinical geneticists, clinical scientists, bioinformaticians, genetic counsellors and GPs with special interest from different regions and seniority levels. The interviews were targeted at those with prior experience of AI, either as adopters, developers, translational researchers or those working to develop the supporting infrastructure (such as regulations and ethics, secure data environments and genomics data platforms).

A questionnaire was circulated across NHS genomics services and achieved 125 responses, covering all regions of England, over an 8-week period. The questionnaire is summarised in an appendix to this document. Thematic analysis was applied to the extended free-text responses to enable semi-quantification and comparison.

While the survey was targeted at respondents in England GMS regions, responses from other NHS services and from national bodies were also included. The majority of respondents were at a senior (44.8%) or mid (29.6%) career level. While responses were received from all regions in England, most (40.8%) were collected from the Central and South region.

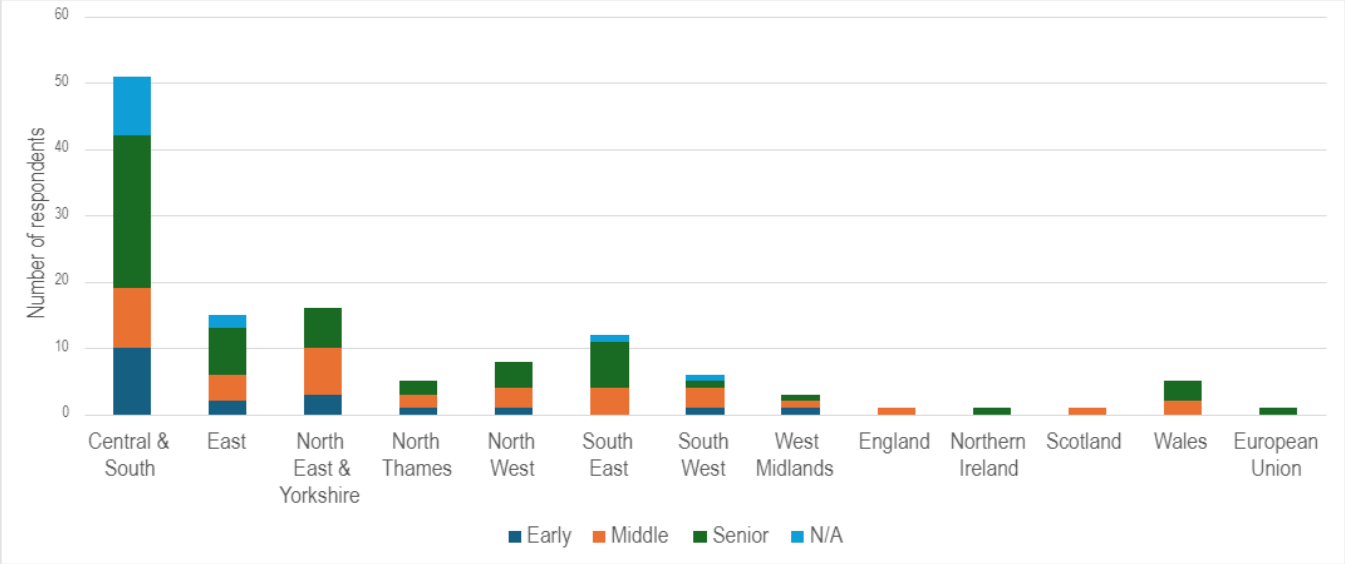


Figure A1: *Summary of questionnaire respondents by region and level of experience. Early career was defined as 1-3 years post-qualification and senior career as holding a consultant or management-level role.*

A selection of survey respondents who reported use of, or particular expertise in AI and machine learning were contacted for interview.