Genomic AI Network

Public Perspectives

The use of AI in Genomic Medicine

November 2024

Final

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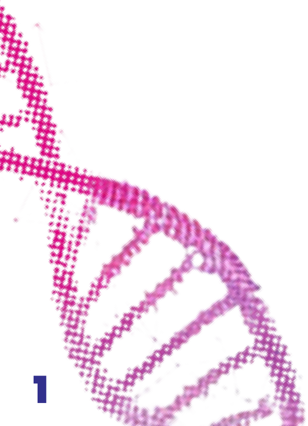
EXECUTIVE SUMMARY

Artificial Intelligence (AI) has the potential to enhance and support clinical decision-making within the NHS, particularly within genetic testing to help improve patient outcomes. However, this comes with a number of social and ethical complexities.

This report summarises the perspectives of over 100 people on the use of AI in genomic medicine. From their feedback we have heard four main themes.

1. Discussions revealed strong concerns around **data responsibility** and **accountability**, especially regarding **transparency and data privacy**. Addressing these concerns is crucial to building trust and patient acceptance of AI in genomics.
2. The importance of **effective, accessible communication**, with participants emphasising that clear information is necessary to make informed choices and **avoid excluding vulnerable groups**.
3. In discussing the role of the clinician, participants highlighted the value of AI as a supportive tool but stressed the **need to preserve the empathetic, human aspects of care.**
4. Finally, AI was recognised as both a help and a hindrance, with AI seen as a **means to improve integrated care** especially for patients with rare diseases, yet possibly limited by wider NHS resource constraints.

These insights will be key in guiding future practices that prioritise patient centred, ethical, and inclusive applications of AI within genomics.



BACKGROUND & AIMS

AI is a powerful tool that is already part of our daily lives.

Already commonplace within social media, financial systems and in our homes through our Alexa and Siri assistants, it's increasingly being used within Genomic Medicine too.

To ensure AI is deployed effectively and responsibly but also to the greatest benefit to the NHS, we have established the new Genomic AI Network (GAIN) Network of Excellence.

The Network aims to build a community of people, professionals and technicians who can harness the power of AI to benefit genomics. A key part of our work is to ensure we are hearing the views and thoughts of the public to help shape and evolve our thinking.

The first step in this work was to:

Understand how patients and the public perceive the role of AI in genomics.

Identify key concerns, hopes and expectations around AI-driven clinical decision support systems.

Work with patients and the public to develop measures and tools that are aligned with patient needs.

HEADLINE FINDINGS

- 100 members of the public across a wide range of ages and demographics shared their thoughts about AI in genomics with us. Twenty-three people have now formed a Public Perspectives Forum to continue this work.
- Communications has a central role in enabling transparency and building trust with public and patients on the use of AI within genomics.
- Questions surrounding consent, data protection, transparency, and patient trust were at the forefront of discussions.
- Many people felt that the use of AI could improve and speed up their care, reduce human error and eliminate bias.
- Can the wider NHS offer the necessary support and treatment needed if AI enables more people to get a genetic diagnosis?

METHODOLOGY

This methodology details our approach taken to reach and listen to over 100 people. We organised 11 conversation-based group sessions across England.

Finding participants:

We worked with a range of patient groups, charities and organisations who helped us to recruit people to take part in conversational group sessions with us. Many thanks are offered to the following organisations who worked with us: Cystic Fibrosis Trust, Huntington's Disease Association, METUPUK, Unique, Genetic Alliance UK, Mendelian, Waldenstrom's macroglobulinemia UK, Cardiac Risk in the Young. We also worked with patient representatives from the Genomic Medicine Service regional teams plus spent time with graduates from the NHS Graduate Scheme.

No prior experience of AI or genomics was required to take part, and everyone was sent some background information to collect their consent prior to the session (Appendix 1).

Session Structure:

The sessions were structured as facilitated discussions, allowing for open dialogue whilst ensuring consistency across the groups on core elements to enable thematic analysis. At the start of the session, participants were also provided with information about the potential of AI and two real life examples of how AI is currently being used within practice. (Appendix 2). During the session, participants were asked a series of questions, including but not limited to:

- How would you feel if your health data was being reviewed by an AI application? For example, how would you feel if your GP called you because AI had flagged your records as potentially benefiting from genetic testing?
- What concerns, if any, do you have about sharing your health data? Would you give NHS permission to use your health data for this purpose?
- How important is it to you to know who has access to your health data when AI is involved?
- How do you feel about doctors using a fully trained and verified AI to help them make a diagnosis?
- AI was used to identify your cousin is at risk of Lynch Syndrome. This result will also affect you and your extended family. How would you feel if AI was used in this way?

Data Collection:

Responses from participants were captured through recordings, transcripts and verbatim notes taken during the session. This ensured that voices were accurately represented during the thematic analysis. To ensure the confidentiality of all participants involved in the session, we shared a pre-session document to explain the purpose of the sessions and consent.

Data Analysis:

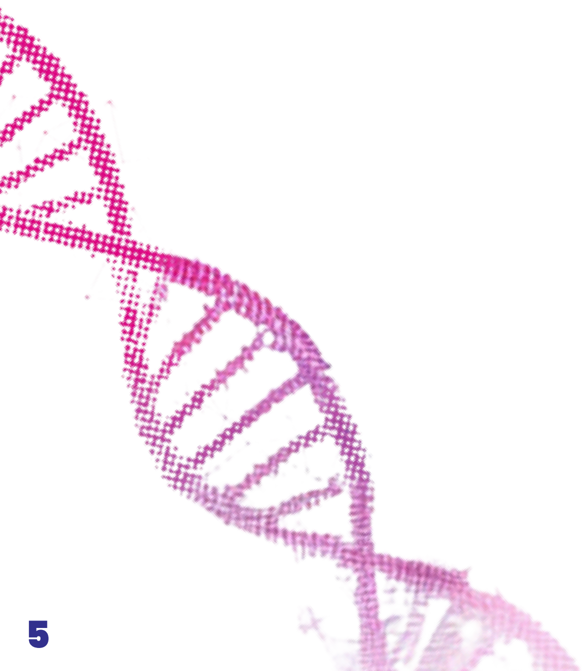
Following the engagement sessions, the data was thematically analysed. Four key themes were derived from this process.

Future Involvement:

All participants were invited to continue to be involved should they wish. 23 people have so far agreed to be part of our new Public Perspectives Forum which will delve into the issues in more detail.

Confidentiality:

All responses from the engagement session utilised within this report have been anonymised. Notes and transcripts have been stored securely in accordance with Guy’s and St Thomas’ NHS Foundation Trust policies.



ANALYSIS

In this section, we will explore the primary insights gathered from the engagement sessions. Four key themes emerged, which offer deeper understanding of participants’ experiences, needs and perspectives.

The four key themes identified are as follows. Click on each box to explore the theme in more detail:

Data Responsibility and Accountability

Communications

The Role of the Doctor

AI: Help or Hindrance?

Based on these themes, we have made a series of recommendations to help shape the future of how AI is integrated within genomics.

DATA RESPONSIBILITY AND ACCOUNTABILITY

With the advancements of both AI and genomics, people talked to us about their concerns about responsibility for, and accountability for data.

As AI systems will increasingly handle sensitive genomic data, questions surrounding consent, data protection, transparency, and patient trust were at the forefront of discussions. The ability to harness AI for the public good weighs against the ethical obligations that the NHS must safeguard patients, prevent misuse of data, and ensure that AI is governed responsibly.

We heard that patient and public mistrust was fuelled by regular data leaks and data breaches reported in the news, with people citing recent examples which have affected the NHS and other healthcare organisations. One person said, “**with all the best data security, determined people and actors will find a way through**.” Many participants were notably concerned about their personal data (such as their name and address) being identifiable beyond those deemed relevant and necessary for their care. Most held expectations that the data would be pseudo-anonymised.

Transparency was raised as a fundamental factor in building trust. A number of representatives highlighted the importance of clarity on who may have access to the data, such as in the form of an audit trail, as one patient quoted:

> “***It is important to know who is accessing it [my data], when and what for, and how long we’re going to be doing it for.***”

However, a number of people recognised that it was not practical to know every single organisation who has access to their data when AI is involved, and felt that this could restrict how the data is leveraged and therefore lessen the potential benefits. This raised discussions about patient choice and consent, with one participant suggesting a ‘**tiered consent model**.’ This would allow individuals to make their own choice about different uses of their data (e.g. for research and/or for commercial purposes). It was also suggested that this should be dynamic, so people can adjust their choices over time as their circumstances or understanding of AI changes. It would be important to reiterate to patients that any decision would not affect their care.

It was noted that there could be a difference in generational opinion about sharing data, with some participants noting that younger populations may not have as many concerns. This should be explored further with age stratification, to confirm to whether this observation/perception is correct. If this is the case, a tailored approach to communicating how data will be shared is important to ensure we reach all generations in the most appropriate manner.

AA tension between public good and profit-oriented private interest was evident, as several concerns were raised about the commercialisation of data and where data may be used for purposes beyond the remit of patient care. Following the question, "What concerns, if any, do you have about sharing your data?" alarm bells were raised about the possibility of data being shared beyond the NHS, and landing in the hands of health insurance companies, mortgage lenders and future employers:

> *I don't want this in the hand of insurance companies... [and] ... the worries about insurance companies ruling you out of many more conditions and using data to penalise you.*

This extended to concerns about how sharing data may impact on their children, which is especially relevant when thinking about the nature of genomic data.

Stories were recounted of previous cases where people had shared their data as part of clinical trials and research, and this had been shared with companies without their consent and/or knowledge. Participants across the board shared their concerns about their data being "**misused**", "**abused**" or "**compromised**" by private companies, and leaned towards having their data stored and managed "in-house."

In contrast, a number of representatives thought that there was a valuable opportunity for the NHS to work with the private sector and use this '**valuable data asset**' to generate income to improve NHS services. Participants raised that public messaging should be transparent about this and highlight the greater good that could come from it.

> *"Sometimes the greater advances come from involving the private sector. The NHS can't afford to be paying hand over fist, there has got to be joint shared outcomes."*
>
> *"We are in a unique position, we shouldn't undersell ourselves. Commercially, how much is that data worth and can we actually improve NHS services by sharing it?"*

In order to tackle apprehensions, several points were raised about the importance of protecting and safeguarding data. Furthermore, if the NHS worked with private companies, participants felt it would be important for those companies to have an established presence online, in order for the public to have confidence in them.

However, some participants shared that they already had faith in the NHS to keep their data safe, with many noting that they already share their data within the NHS. Others voiced that provided the safeguarding measures are in place to protect patient data, they could ultimately let go of any apprehensions if it was of benefit to both their own health and wider public health, linking in with the theme of improved knowledge leading to improved treatment. They felt that by sharing their data they would be taking a proactive approach, mirroring the shift in the NHS towards preventative care.

Participants would like to see clear information about how AI systems are being governed. This extends further to the importance of having a strong framework or committee in place with accountability and responsibility for overseeing the use of AI, ensuring compliance with ethical standards and addressing public concerns.

Recommendations

- Further research into generational attitudes about data sharing is needed to inform a tailored communication approach.
- The Genomic AI Network to escalate patient concerns to NHS England about the need to strengthen the UK's concordat with the insurance industry, to ensure that genetic information is not exploitative or used unfairly.
- Ensure that any commercial use of genomic data using AI prioritises patient benefit, with clear boundaries set on how private companies can access and use this data. Ensure this is communicated to the patient.
- The Public Perspectives Forum should work with both data and consent experts to design a tiered consent model, where patients can opt in or out of different uses of their data (e.g., for research and/or for commercial purposes), allowing them to make more informed decisions. Ensure that these are dynamic and accessible so all patients can adjust their choices over time as their circumstances or understanding of AI changes.
- The NHS Genomic AI Network to discuss how to effectively and efficiently govern the application of AI in genomics and roles and responsibilities. This should include but not be limited to; auditing systems, ensuring compliance with ethical, legal and patient safety standards and address wider public concerns that may arise.

COMMUNICATIONS

To ensure patients feel informed, respected, and empowered in making decisions about the use of AI and Genomics, communications have a central role in enabling transparency and building trust with public and patient groups on the use of AI within genomics.

AI was seen as a "**divisive**" topic amongst participants, and there was a broad spectrum of opinions on the use of AI more generally and societal implications of this. A small group of people raised that AI has been around for a while, and is already commonly used within the NHS, with several people describing instances when their GP had used AI or technology to assist with diagnosis. However, within the discussions the negative perception and reputation of AI was frequently raised, with it being described as a "**stained word**," with "**a lot of bad press out there at the moment**". Representatives highlighted the importance of communicating that AI was solely a support tool, and that there would be clinical oversight over any potential flag. (This is discussed further in the later section regarding the role of the doctor.)

In the same vein, to combat misunderstanding and misrepresentation, people called for more education and information about the capabilities of AI and how it would be used to inform clinical decision making within the field of genomics, and ensure that they have enough knowledge to make "**informed decisions**." This links to the theme previously discussed about how AI can benefit the patient.

> ***"I support a lot of families who are in the undiagnosed and rare community. I think a lot of them would probably want more education on what AI actually does in this respect, because there's a lot of misunderstanding and misrepresentation of it."***

Furthermore, a number of participants voiced the importance of working with religious leaders and ethics committees so there isn't a "**stumbling block**" in getting the message across to all communities, ensuring that religious and cultural taboos are not broken. One participant mentioned that whilst some people may find AI taboo, if they know both the service and AI is on their side, they may be more willing to share more information. With regards to consent and patient choice, people suggested taking measures to ensure that patients who were not proficient in English or have accessibility requirements, could make informed decisions. There was a range of views as to what sort of model would be the most appropriate and beneficial to ensure that communities were not excluded (e.g. opt in or opt out model, tiered consent).

Taking a tailored approach is particularly pertinent to ensure data is representative of our diverse population, and consequently so that we do not exacerbate health inequalities, especially for those who may be prone to certain illnesses such as sickle cell and thalassemia.

Following discussion of the question “How would you feel if your GP called you because AI had flagged your records as potentially benefitting from genetic testing?” it was highly contended amongst participants across all groups how they would like to be informed about potential genetic screening. Participants discussed the logistical roll out and how this message would be translated front-facing. Across the board it was raised that it is important that they were offered a consultation, and were largely against the message being delivered “untold” or solely via a text or letter, as this could raise “anxieties.” The language used should be carefully considered if a letter is to be used in conjunction with a consultation, to mitigate against any apprehensions.

There were various perspectives as to who this consultation should be with. One patient group highlighted that they were notorious for having a lack of trust in their GP, and suggested that consultants would be best placed to deliver the information, especially if they were already in the midst of treatment. However, other groups highlighted the value of the GP as a first point of contact in helping the individual make informed decisions as to whether they would like to progress with testing. Further ideas included integrating a GP within the Genomic Medicine Service or having genetic counsellors deliver the news, however the availability of genetic counsellors was raised as a bottleneck.

In all cases, people widely agreed that the choice should rest with the patient as to whether they engage with the use of AI in genetic testing, and whether they would like their data to be shared. This should be respected, and patients’ preferences must remain a central focus in all communications and engagement efforts throughout the implementation process. By fostering transparency and building trust, communication efforts can bridge knowledge gaps, address concerns and clarify misconceptions that surround the use of AI in genomics.

Recommendations

- The Genomic AI Network to continue to engage within patient and public dialogues regularly to build trust and understanding about how AI systems are being used to benefit genomics medicine to alleviate any concerns or misconceptions.

- The Public Perspectives Forum to develop accessible materials (both online and offline) explaining the benefits of AI, with a focus on its role in improving diagnostics, treatment, and patient outcomes.

- The Public Perspectives Forum to work with religious leaders & ethics committees to understand more about the barriers that different communities may face in engaging with AI and Genomics.

- The Genomic AI Network to explore whether an opt in, or opt out consent model would be most suitable to ensure the public can make informed decisions about participating in genetic testing. Draw on learnings from similar models in other parts of the health care system such as the organ donation model, and communication and engagement measures that were used to help ensure that as many people as possible take informed decisions.

- The Public Perspectives Forum to develop a process map for how patients and the public will be informed about the possibility that AI has flagged them as potentially benefiting from genetic testing, including clinical responsibility in delivering this message.

THE ROLE OF THE DOCTOR

> ***I have to have regular brain scans and I would much rather an AI look at those scans than a busy brain surgeon.***

When bringing AI into the clinical setting, many participants questioned what the new role of the clinician would be when delivering care. A number of people expressed that AI could help with current resource strains on clinicians who are **"busy", "under pressure" and "aren't going to be poring through records themselves".** There was a sense that information and subsequent diagnoses are missed due to capacity constraints, and participants were **"aware that your doctor is a human and makes mistakes"**. In this vein, a number of participants saw AI as a tool which doctors can use to enhance their practice by sifting through information quicker, and that it is the **"equivalent to lots of people looking at something instead of it potentially being missed".**

Whilst some participants followed the sentiment that AI could be a helpful tool to enhance what the doctor already does for earlier and more accurate diagnosis, there was a vast range of opinions regarding the role of the doctor more broadly when AI is involved. At one end of the scale, participants discussed how, beyond helping with capacity, AI could play a more central role. For example, one participant said they would be **"more confident to ask an AI for a diagnosis"** by themselves without the initial intervention of a doctor. Some of this sentiment comes from current dissatisfaction with doctors and GPs. Multiple participants expressed existing concerns of bias amongst medical professionals, and the idea that AI may reduce that bias and can screen patients more anonymously. One participant described how people with their genetic disorder generally have **"bad relationships with GPs"**, and do not always trust them. Other points of dissatisfaction came from the lack of information sharing and integrated care, for which AI was seen as a solution:

> ***"We are being failed at the moment and it is discriminatory … we have to be braver and move forward to see change."***
>
> ***"I would prefer AI. At the moment I go to three hospitals and none of the doctors know about my records. It must cost the NHS and absolute fortune. I would be absolutely over the moon."***

In this way, the experiences people have with their healthcare providers influences their opinions about, and reception to, the role of AI in their care.

For others, the idea that an AI could play a more prominent role than a doctor, or replace the doctor, is frightening. Many participants hoped that there would be the opportunity for a **"second opinion"**, **"double check" or "backup plan"**, and that somewhere in the process a clinician was still there.

> ***My concern would be false positives and false negatives. This is where the nuanced expertise of the human workforce comes in.***

Part of this fear came from not knowing or trusting that AI was fully accurate, or at least equal to a doctor. In these instances, people wanted to know that clinicians would not be **"just taking what the AI said as gospel"**, rather using their "clinical judgement". Although this seems like an opposite sentiment to that which sees AI as potentially more capable than a human, the fear is the same; not trusting one or the other to be accurate enough. Building this trust in the competency of AI is therefore key to acceptability. In a similar vein, many participants expressed a fear of not knowing who would be ultimately responsible for clinical decisions, outputs, or errors. Here, a large part of building this trust is making the role of AI tangible. This links back to the section on how to communicate the benefits of AI.

> ***"If you lead with AI it becomes scary, but if you say it's a tool it makes it easier to picture."***

It is crucial to understand that participants have broader, long-term worries about the role of the doctor aside from individual interactions. Multiple participants discussed how they worry that because of AI, the clinical workforce will de-skill and become redundant:

> ***My long-term concern is that we might be encouraging a reduction of personal clinical experience to be confident in making those diagnoses …. the human input and clinicians' ability to see things independently is not something I would like to see eroded to be honest***

The question then is what do patients fear losing? What is the human element which an AI cannot bring, which they feel may be at risk? The most obvious answer is patients fear a doctor being unable to complete the service: in an analogy to optometry, one participant shared that **"when that camera breaks, you still need to be able to give that patient a service. What happens when the AI breaks - can you still provide a service?"**

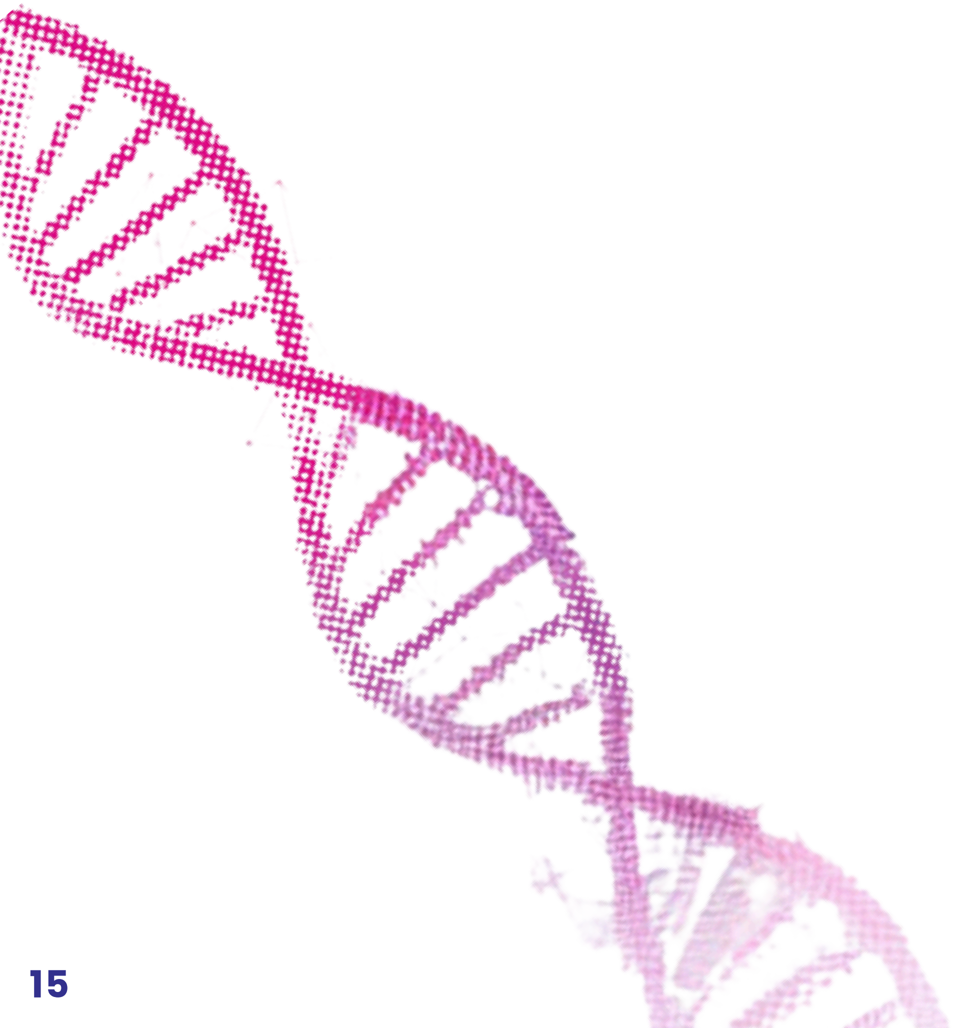
However, this does not fully address the needs related to quality of care. In addition to practically being able to provide a service, there was a clear need from patients and public to have a high quality of care, and they feared this may be taken away with AI. For many participants, essential to this quality of care was the **"human touch"**, where doctors know you as a person, and treat you as such. This is expressed well by the three participants below:

> ***"Will they be focusing on the AI rather than me? They want to do diagnostic tests even though I am in front of them. Look at me!"***
>
> ***"We've had a series of locum GPs who are there to deal with current complaints, but they don't know you from Adam or Eve."***
>
> ***"Straightaway in my head I'm thinking are you going to end up with doctors who put symptoms in and see what comes up, losing the personal touch or personal conversation."***

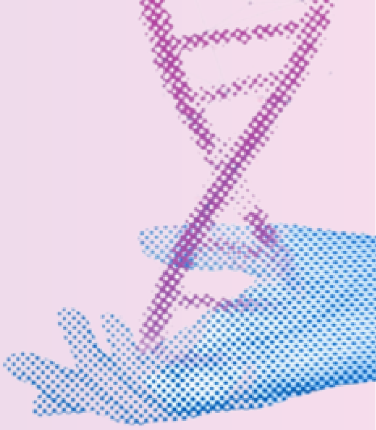
Understanding this demand and the need for quality in the care delivery experience, outside of numerical performance and targets, is essential to AI acceptability.

Recommendations

- The NHS to develop a process for monitoring and improving rates of false positives and false negatives. Communicate this with the public.
- Consider making it clear to the public what the responsibilities of the doctor, or specialist clinician, are when using an AI tool (e.g. that the doctor is still making final diagnosis decisions). This may look different for each tool which is developed, but will alleviate public concerns.
- Training must be provided for clinicians and healthcare professionals to use each AI tool with case study examples to ensure equitable implementation. Clinicians should understand the public fear that they will lose the 'human element' so that they can proactively reassure their patients.



AI: HELP OR HINDRANCE?



With the impact of AI being unclear and unknown, perspectives on how effective it would be for different purposes were not unanimous: some saw it as a help to genomic medicine and the NHS, whilst others saw it as a hindrance limited by the wider context.

Help: Improving Treatment Through Increased Knowledge from AI

At the start of each session, a few ‘real life’ examples were given to participants which helped make the use of AI more tangible: MendelScan which is already scanning GP records for signs of rare diseases for example. Many participants, however, could easily imagine benefits beyond this. There was a general agreement that improved knowledge (through the integration of AI into care), had the potential to improve treatment because it could identify more things, and identify them earlier.

> ***“I think knowledge is power. I see it as another screening. The more screening that is done the more likely you are to get things early.”***
>
> ***“All I can see is that this would’ve sped up the process.”***

> ***“I have a rare cancer that perhaps if it was identified earlier I wouldn’t have had the journey I’ve had. I think it is fundamentally important that if medical science improves we should use it. It will help peoples’ lives and it is the quality of life that is important.”***
>
> ***“When I was diagnosed with WM, my haematologist also looked back at my historical blood tests and said to me that blood tests dating three years prior at routine GP appointments were borderline abnormal, but because it was borderline no action was taken. It was another three years that the disease progressed to symptomatic. I could see potentially that by using AI with some haematology knowledge this may have been picked up three years ago. It can only seem like a good thing for me.”***

At an individual level, the idea that AI could be used to increase the level of knowledge was especially important to patients with rare diseases, those who have undergone a “diagnostic odyssey” or “diagnostic delay” trying to understand their symptoms. felt that an AI tool screening records could have improved their diagnosis. A participant pointed out that in the diagnostic odyssey process, an AI which is able to rule out certain diseases is also useful, even if it cannot diagnose. As well as diagnosis, other patients saw how this could benefit them in their daily life.

Two patients with Cystic Fibrosis explained:

> ***“They are now using AI to try and see when I am about to have an exacerbation before I know …If there’s something that can alert me, for me it’s all about starting treatment as soon as possible”***
>
> ***“In terms of the outcome as a patient that’s fantastic to understand and anticipate what’s coming … I certainly would embrace the idea of having more information and being better able to predict”***

Across the Cystic Fibrosis group, the South East Genomics Patient Involvement Group, and Metastatic Breast Cancer group, there was a general understanding that **“it may be impossible for a human to understand all the symptoms in rare diseases”**, especially when they have not encountered them locally or in their own clinical practice. Currently, patients felt they were at the mercy of “luck” as to whether your doctor has seen your symptoms before, raising significant inequality concerns. All participants recognised the benefit of using AI to identify rare diseases.

> ***In my family circumstances, there is so much information to remember for different appointments. Each specialty has their own bit that they pick and choose the highlights for them. This [AI] would be less chance of something being missed. I know specialties have niches, but I have been in appointments where they’ve told me something is completely irrelevant, then into another that says they’re all linked. I would hope AI could catch more.***

As well as increasing knowledge of the individual to improve treatment, participants in the rare disease groups pointed out that AI may help increase wider knowledge through research, and improve treatment more broadly. Many had already shared their data for research purposes:

> ***“Do I trust it? Well I have to if I want to take part in research and help researchers find a cure for Huntington’s”***
>
> ***“There’s a difference if you have health conditions. Almost everybody with Cystic Fibrosis already gives permission for their records to go on the registry.”***
>
> ***“In my world of amyloidosis, the more that is shared, the more people can sort things out.”***

For some, they were more inclined to share their data for research than for personal treatment. For some, they would even consider sharing their data for research even if they could not treat the disease at the moment.

> ***I always give data for trials and I’m used to it. It feels easier for me to give my data away to improve someone else’s health, but more difficult to allow AI to tell me if there’s something wrong with me. It’s a really weird feeling for this to be switched.***

As shown above, understanding that people do and will feel confliction towards the use of AI depending on what the benefit is and to whom, is really important when thinking about how to implement it in the general public. The benefits which people respond to may vary, and some may not want the personal benefits, but are willing to share data with AI for the benefit of the wider population.

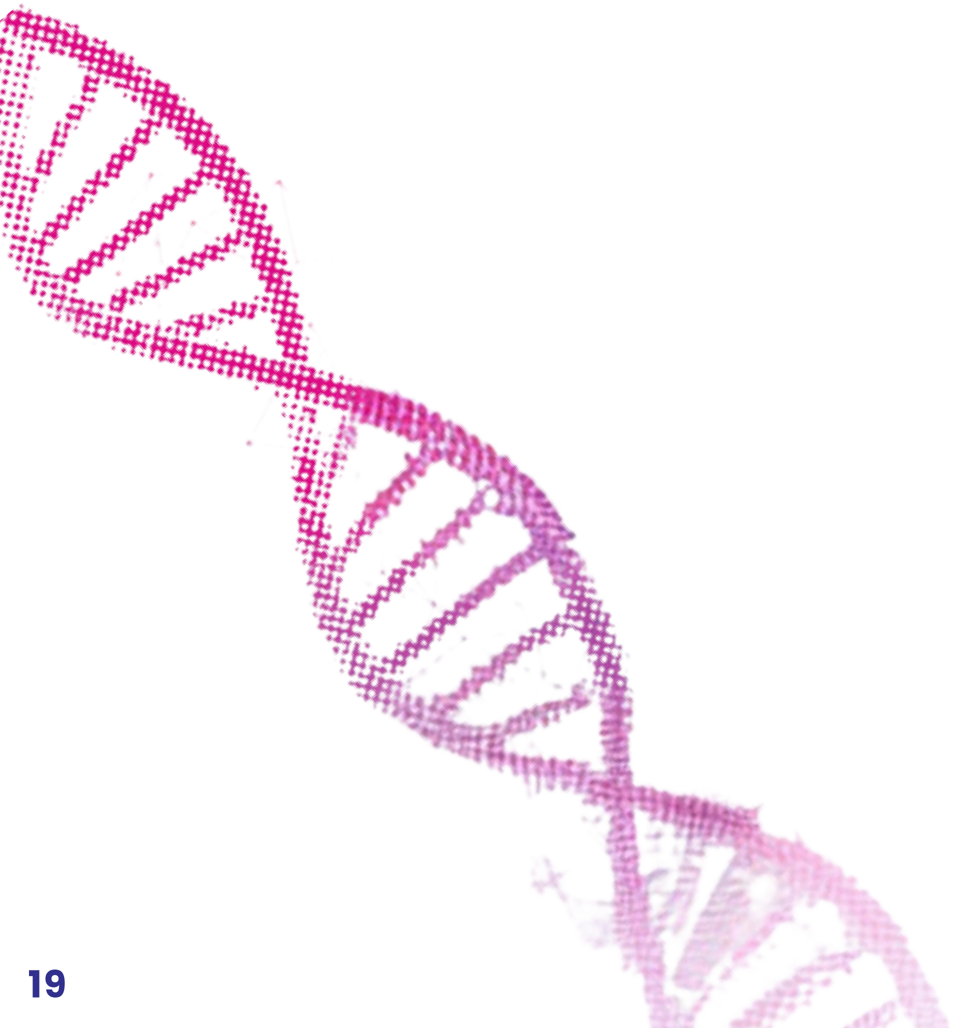
Outside of the rare disease space, patients also see another personal benefit from the increased knowledge from AI in prescription management – that AI could calculate and "come up with the best cocktail to recommend for the patient straight away". There was anecdotal experience of having potentially dangerous prescriptions, and doctors already using a form of AI to check medications for drug-drug interactions. Another perceived benefit was the ability of AI to better integrate care and share knowledge across services. Multiple participants expressed frustration that records, and information were not shared, and saw AI as a solution to scan records from multiple sources, communicate, and "join the dots", because "it shouldn't be all down to the patient". Some extended this to "flagging patterns within families", "tracking back" through old records, and tapping into "unmined data" such as smartwatches as well. This links to a greater desire for care which looks at the whole person, as explored when looking at the role of the doctor as well. AI may be able to complement person-centred care in this way.

> ***"There are some young people who die before their symptoms are picked up, and it is only with hindsight that the whole file is reviewed"***
>
> ***"I just want someone to join the dots, and maybe AI will do that."***

However, it should be noted that the participants in the rare disease group may be particularly receptive to the idea of increasing knowledge to increase treatment. On the other hand, other participants expressed that they would not necessarily want to know if they were genetically predisposed to conditions which they could not treat, and that if AI picked that up without their consent, this would not be of benefit. Having not experienced these conditions, they may also be less receptive or understanding of the benefit, and concerns such as data privacy or financial implications discussed earlier in the report may hold more weight. This is important to consider when communicating the benefit of AI to the wider population. Benefits such as further integrated care or broader improved treatments in the future, may need to be emphasised more than individual care opportunities.

Recommendations

- The Genomic AI Network should explore the use of AI tools to ‘backtrack’ previous records. If deemed successful, the public should be provided with examples about how this approach has benefited patients.
- The Genomic AI Network, supported by the Public Perspectives Forum should work with Rare Disease charities to build further trust and communicate together the benefits of AI to reduce diagnostic odysseys.
- The Genomic AI Network to consider different communication/ patient opt in options for research and clinical trials compared to personal treatment.



Hindrance: Wider Limitations

Participants repeatedly raised the concept that the use of AI in genomic medicine cannot be looked at as an independent entity. Rather, it needs to be understood within the wider NHS context: **"this to me isn't just a genomic question, it is an NHS question"**. Participants raised important questions about the wider NHS capacity to support expedited and increased diagnoses. As previously mentioned, at a practical level, participants had concerns about clinical "bottlenecks", ability to integrate processes across the wider NHS and how to equitably educate clinicians and primary care. There was also significant concern that diagnoses may increase the need for genetic counselling and psychological support:

> ***"There are individuals who may be very scared about the fact that the NHS has not yet got the capacity."***
>
> ***"I experienced health anxiety for the first time around last Christmas and it was very unpleasant. One of the reasons it was unpleasant is because the NHS wasn't in a fit state to support my mental health in a timely manner"***
>
> ***"It's making sure that when that [AI flagging a record] happens people have the right access to information and support. Is there a helpline? Who am I meant to contact? Is there a leaflet?"***
>
> ***"When I think of the practicalities it is important to think about the infrastructure. It is not data at the end of it, it's people who have other things to consider. What is the supporting infrastructure?"***
>
> ***"You already feel guilty and isolated that you have something you could pass on. I think a lot of counselling is needed. It needs to be done very gently with clear pathways to follow."***

The wider support which may be required in the health and care system also extends to people's families. This is especially important as people may not always come from a "cohesive" family unit, and there is concern that inappropriate use of AI, such as sharing information between family members, could disrupt family dynamics and relationships.

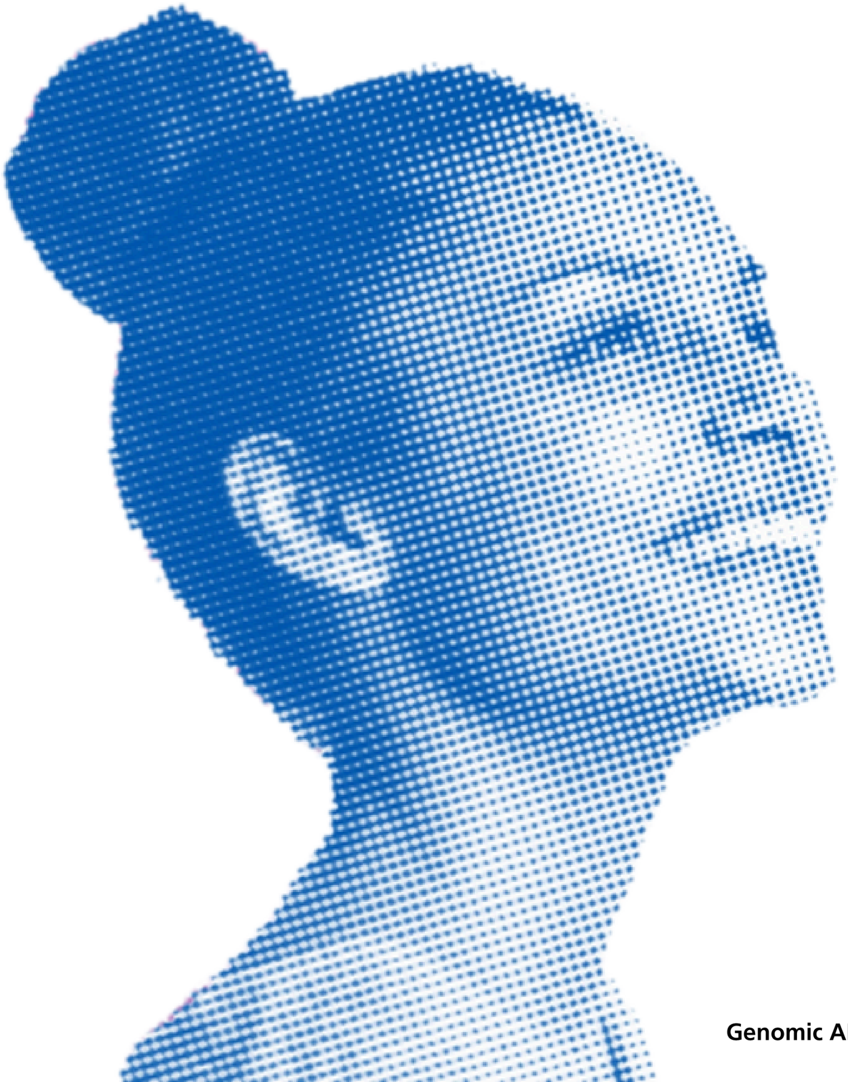
Another limitation of the wider system discussed was bias in the data being used to build and train the AI. For some participants, this concern came from a belief and experience that doctors, and the wider health system are already biased, so will this be exacerbated if that is the data used to train the AI. There were also concerns that it would not be implemented equitably: "do all GPs have access to this?". On the other hand, other participants saw the potential of AI that "where we can use any kind of automated process, we can help to remove prejudices." In a separate group another participant asked that, if there was an opt out system, would these people potentially miss out on treatment? The reasons for opting out may be related to wider health inequalities, such as age, geography, digital disengagement and disability. For example:

> ***I think it's a great idea but it's only going to be as good as the geographical mapping. I work between England and Wales NHS systems and they don't talk to each other. In my case, I would've been diagnosed earlier.***

As well as this, many people expressed with cascade testing that two children in similar situations have completely opposite reactions; this may be unexpected or unpredictable. Linking back to the earlier section around consent, whilst people may need an opt out system to accept the use of AI more generally, it is imperative that people's access to the services should be continually reviewed with the consideration of data bias and equitability. Implementation of AI therefore should be used to reduce existing biases, and the data used to train the systems should not be assumed to be unbiased.

> **“If it's not the right group to start where you're taking your reference points, then it's not going to be appropriate for our multicultural society or if people have disabilities.”**

Beyond limitations at the individual treatment level, one participant mentioned that by being in the U.K., the AI consent and access systems may be only as good as the U.K. standard GDPR laws, and these may not have caught up with the AI technology that is available. Others expressed concerns around the environmental impact of AI. One participant also discussed how the data could be used to generate income effectively for the NHS (although others were fearful of this). All these points are a reminder that implementing AI is not an isolated process, and the wider context of the NHS must be considered and communicated effectively.

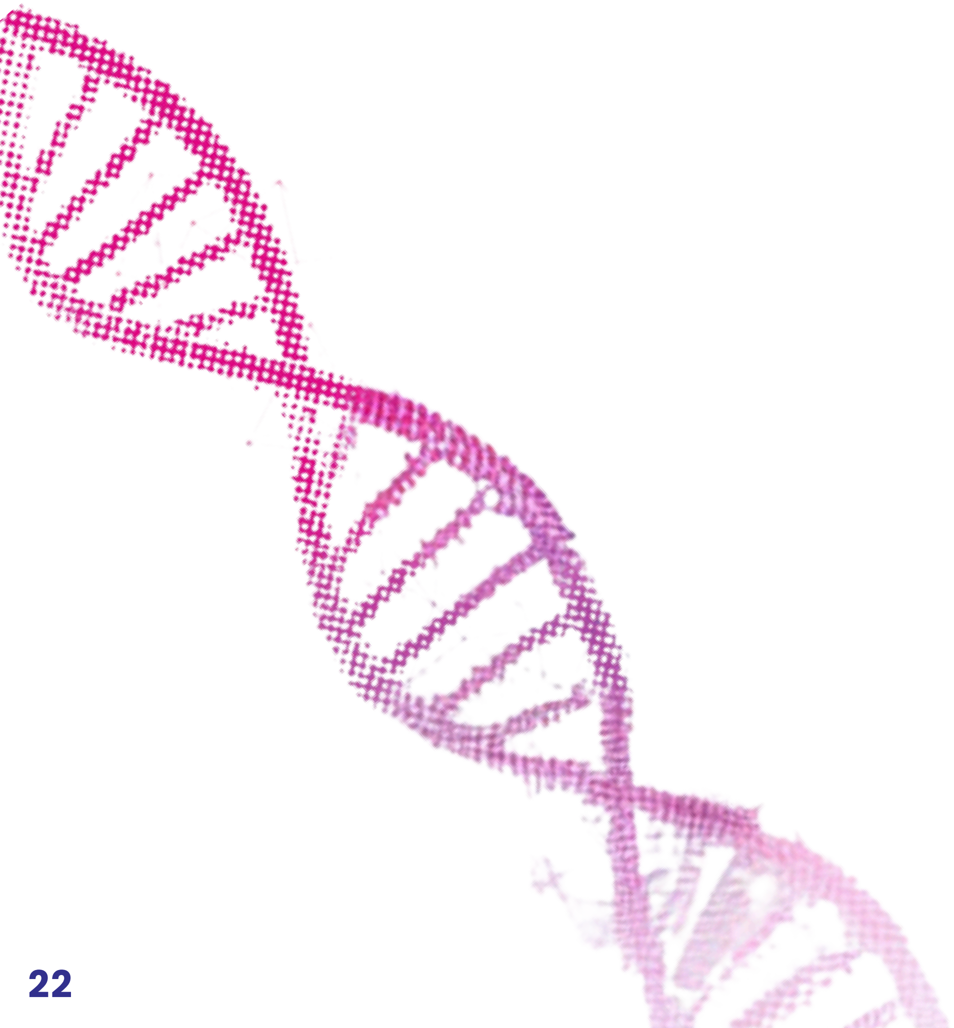


Recommendations

- With each AI tool, NHS England will need to assess how it will increase demand for other NHS services, e.g. by increased diagnoses of rare diseases, and ensure that a demand and capacity assessment is done. This means that related elements of patients' care (such as psychological support or further screening pathways) are available.

- NHS England must ensure that implementation of AI tools works to address current health inequalities and biases, and not increase them. For example, ensure they are implemented geographically and continually review who has opted in or out, and whether this has affected their access to care.

- The healthcare system must not treat the implementation of AI in a vacuum. Assess the wider impacts and limitations of U.K. laws and regulations around data protection, climate impact and income generation. Consider addressing these concerns explicitly with the public in further consultation.



CONCLUSION

This report has identified four main themes following extensive discussions with over 100 members of the public.

Overall, perceptions of the use of AI in genomic medicine were positive, however, there were notable differences in opinion. When implementing AI in Genomics, we recommend an increased understanding into the wider, contextual factors affecting people's health and care experiences. This insight should inform how to implement the tools as equitably as possible, and to tailor communications accordingly as this is essential to increase acceptability.

The first theme **'Responsibility and Accountability of Data'** addresses the widespread concerns people had about data privacy with a new AI tool.The report has uncovered that a lot of this anxiety comes from not knowing about the safeguarding and governance measures in place, and that transparency is crucial for building trust and acceptability. It is important to note that opinions on how readily people would be willing to share data and how patients responded in their acceptance of the NHS using this data as an asset in partnership with the private sector varied greatly. Participants discussed how these fears (and their variances and nuances) may come from socio-cultural, demographic, socio-economic or other factors. There are strong discourses of AI as "stained" or "taboo".

The second section, '**Communications**', discussed how creating tailored, educational and accessible communications around the use of AI is crucial for overcoming these fears and anxieties. This report has identified a risk that if communications are not cascaded so patients feel they can make "informed decisions", the use of AI tools may exclude groups who are least likely to be willing to share their data due to preconceptions of AI, and they may also be the ones who are experience existing health inequalities and whose care could benefit the most.

Other anxieties stemmed from uncertainty about what the '**role of the doctor**' would be with these new AI tools, and fears that doctors may de-skill over time, and/or clinical interactions and care would lose the essential 'human element'. Here, we learnt that patients and the public continue to value a doctor who sees and treats you as a person with empathy and compassion. Many participants were more receptive when they saw that AI could be a tool, and not a full replacement. Others, who may not have good relationships with their primary care providers, saw the benefit of AI if it could be leveraged to free up time for a clinician to focus on those quality-of-care elements. In this respect, communicating to patients the roles and responsibilities of the doctor and how this will change for the benefit of the patient, and continued focus on quality of care, is essential as part of the implementation of any AI tool.

The final section of the report, **‘AI: Help or Hindrance’**, highlights the different ways that participants viewed how AI could aid their care, but also concerns that it would be limited by the wider NHS. Many participants saw AI as a route to increased knowledge, and therefore improved treatment. Part of this was seeing AI as a route to better integrated care, as opposed to current dissatisfaction with fragmented care and siloed services. Participants also saw benefit to increasing data for research purposes. This was particularly pertinent to patients with rare diseases, who had undergone “diagnostic odysseys”. However, as noted in previous sections, the benefits which are most convincing to people vary, and may need to be communicated in different ways. When discussing the potential benefits, concerns arose in relation to the wider NHS, notably capacity and psychological support, which may come from an increased rate of diagnoses with AI. In this way, it is essential to consider the implementation of AI tools not in a vacuum, but address the wider NHS and how it fits into patients’ and their families’ holistic care over many years.

The report findings and recommendations must be central to the new Public Perspectives Forum which is being established. Their role will be to ensure the considerations raised in this report continue to be discussed at all levels within the NHS Genomic AI Network and ultimately in the implementation of AI tools within the NHS.

LIMITATIONS

The following limitations are acknowledged to the findings in this report:

1. Demographic data of participants (e.g. age, gender, location, ethnicity) was not systematically captured. This was a practical decision as we were not in direct contact with participants prior to the sessions. Concerns were raised that it may have led to overwhelming representation or exclusion of people in certain categories. Therefore, we are not able to stratify opinions based on demographic data to gain specific insights into health inequalities and acceptability.
2. The discussions may have been in some part skewed towards the session design, specifically the examples given of MendelScan, Lynch Syndrome and cardiac record screening, as well as the members of the GMS present. This was also not uniform across every session. Whilst this was essential in providing prompts for discussion, we do not know how this may have impacted on discussions being biased or skewed.
3. The sample was focused on patient groups across charities and the Genomic Medicine Service. They noted themselves that they could be a more receptive audience to the use of AI for better treatment due to their extensive and personal experiences with the NHS. A broader range of participants, such as religious groups, community groups, schools and universities, clinical groups and many more should be invited for further discussion.

SUMMARY OF RECOMMENDATIONS

Data Responsibility and Accountability

- Conduct further research into attitudes of different generations on data sharing, and tailor communication for younger and older populations depending on the outputs.
- Consider lobbying avenues for the strengthening of the UK's concordat with the insurance industry, to ensure that genetic information is not exploitative or used unfairly.
- Ensure that any commercial use of patient data prioritises patient benefit, with clear boundaries set on how private companies can access and use this data. Ensure this is communicated to the patient through data privacy information.
- Explore designing tiered consent models, where patients can opt in or out of different uses of their data (e.g., for research, for commercial purposes), allowing them to make more informed decisions. Ensure that these are dynamic so patients can adjust their choices over time as their circumstances or understanding of AI changes.
- Work to create a dedicated governance body who will have oversight over the application of AI in genomics and roles and responsibilities not limited to, but including: auditing systems, ensuring compliance with ethical, legal and patient safety standards and address wider public concerns that may arise.

Communications

- Continue to engage with patient and public dialogues regularly to build trust, with clear information about how AI systems are being used to alleviate any concerns or misconceptions (e.g. data privacy, use of AI as a support tool).
- Provide accessible materials (both online and offline) explaining the benefits of AI, with a focus on its role in improving diagnostics, treatment, and patient outcomes.
- Work with religious leaders and ethics committees to understand more about the barriers that some communities may face in engaging with AI and Genomics.
- Explore whether an opt in or opt out consent model would be most suitable to ensure the public can make informed decisions about participating in genetic testing. Draw on learnings from similar models in other parts of the health care system such as the organ donation model, and communication and engagement measures that were used to help ensure that as many people as possible take informed decisions.
- Following further engagement with patients, develop a process map for how patients and the public will be informed about potentially being flagged by the NHS for genetic testing, including clinical responsibility in delivering this message.
- If patients are informed via letters or text, ensure that the tone is appropriate so as not to cause fear and anxiety amongst patients.

SUMMARY OF RECOMMENDATIONS

Role of the Doctor - Specialist Clinician

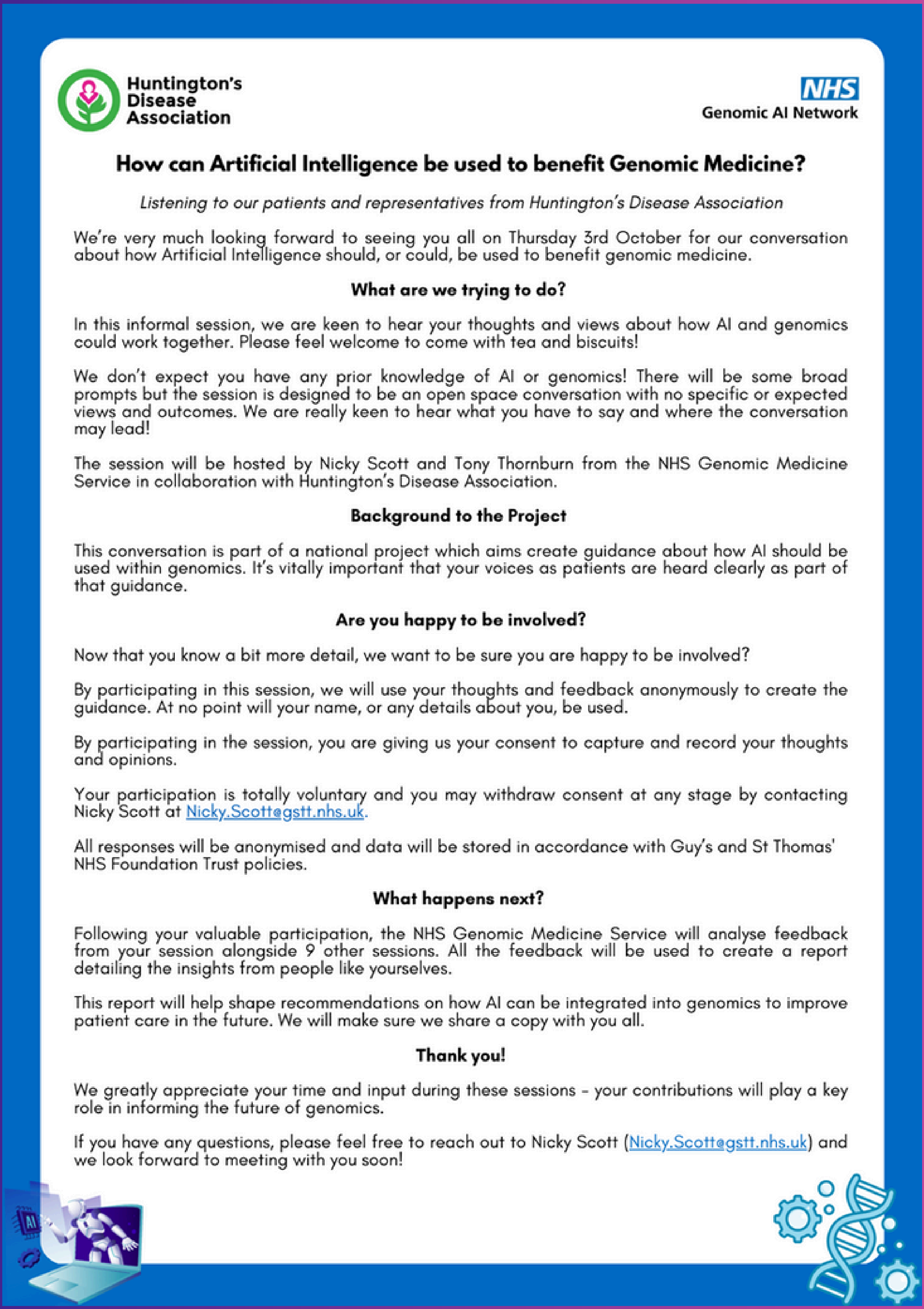
- Develop a process for monitoring and improving rates of false positives and false negatives. Communicate this with the public.
- Consider making it clear to the public what the responsibilities of the doctor are when using an AI tool (e.g. that the doctor is still making final diagnosis decisions). This may look different for each tool which develops, but will alleviate public concerns. Doctors should be trained to use the tool in the same ways with case study examples to ensure equitable implementation.
- Use the above to acknowledge and address the fear of losing the 'human element'. Consider that the 'human touch' is essential for quality of care and patient experience in addition to activity and clinical outcomes.

AI: Help or Hindrance?

- Where possible, use AI tools to 'backtrack' previous records. Provide the public with examples where this has helped new diagnoses or re-diagnoses to make the impact tangible and clear of sharing old records.
- Leverage the acceptability and positive outlook of patients in the rare disease space to communicate the benefits of using AI. Rare diseases where there are only a handful of cases is a good example of how AI can bring knowledge which a clinician would not plausibly be able to.
- Acknowledge that it may be more acceptable to some people to the use of AI if they believe it has a positive impact on wider research at the population level, more-so than at the personal level (where they may be more fearful). Consider communicating and highlighting the use of AI in research and clinical trials. Consider allowing people to opt in sharing data for these purposes, but not personal treatment.
- With each AI tool, assess how it will increase demand for other NHS services, e.g. by increased diagnoses of rare diseases, and ensure that a demand and capacity assessment is done. This means that related elements of patients' care (such as psychological support or further screening pathways) are available.
- Ensure that implementation of AI tools works to address current health inequalities and biases, and not increase them. For example, ensure they are implemented geographically and continually review who has opted in or out, and whether this has affected their access to care.
- Do not treat the implementation of AI in a vacuum. Assess the wider impacts and limitations of U.K. laws and regulations around data protection, climate impact and income generation. Consider addressing these concerns explicitly with the public in further consultation.

APPENDICES

APPENDIX 1



Huntington's Disease Association

NHS Genomic AI Network

How can Artificial Intelligence be used to benefit Genomic Medicine?

Listening to our patients and representatives from Huntington's Disease Association

We're very much looking forward to seeing you all on Thursday 3rd October for our conversation about how Artificial Intelligence should, or could, be used to benefit genomic medicine.

What are we trying to do?

In this informal session, we are keen to hear your thoughts and views about how AI and genomics could work together. Please feel welcome to come with tea and biscuits!

We don't expect you have any prior knowledge of AI or genomics! There will be some broad prompts but the session is designed to be an open space conversation with no specific or expected views and outcomes. We are really keen to hear what you have to say and where the conversation may lead!

The session will be hosted by Nicky Scott and Tony Thornburn from the NHS Genomic Medicine Service in collaboration with Huntington's Disease Association.

Background to the Project

This conversation is part of a national project which aims create guidance about how AI should be used within genomics. It's vitally important that your voices as patients are heard clearly as part of that guidance.

Are you happy to be involved?

Now that you know a bit more detail, we want to be sure you are happy to be involved?

By participating in this session, we will use your thoughts and feedback anonymously to create the guidance. At no point will your name, or any details about you, be used.

By participating in the session, you are giving us your consent to capture and record your thoughts and opinions.

Your participation is totally voluntary and you may withdraw consent at any stage by contacting Nicky Scott at Nicky.Scott@gstt.nhs.uk.

All responses will be anonymised and data will be stored in accordance with Guy's and St Thomas' NHS Foundation Trust policies.

What happens next?

Following your valuable participation, the NHS Genomic Medicine Service will analyse feedback from your session alongside 9 other sessions. All the feedback will be used to create a report detailing the insights from people like yourselves.

This report will help shape recommendations on how AI can be integrated into genomics to improve patient care in the future. We will make sure we share a copy with you all.

Thank you!

We greatly appreciate your time and input during these sessions - your contributions will play a key role in informing the future of genomics.

If you have any questions, please feel free to reach out to Nicky Scott (Nicky.Scott@gstt.nhs.uk) and we look forward to meeting with you soon!

NHS
Genomic AI Network