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AI in NHS Genomic Medicine Workflows

A Reference Guide to AI Tools, Current Practices, and Existing Guidance and Frameworks in NHS Genomic Medicine Services

This document provides an overview of AI tools in workflows within the NHS Genomic Medicine Service. It outlines ongoing initiatives, current practices, and the application of AI to enhance workflows including genomic data interpretation, diagnostics, and clinical decision-making.

This document should serve as a reference for professionals across the NHS, to stay informed about existing resources and opportunities.

Last updated: October 2025

Responsible use of AI in NHS Genomic Medicine Services

The use of AI within NHS Genomic Medicine Services requires strict adherence to data protection, transparency, and clinical oversight, and well as local policies. The below provides a starting point checklist. Please also speak to your local Trust Governance teams before deploying any AI tools.

🔗 **Data Compliance Checklist** (NHS AI and Digital Regulations Service for health and social care)

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General Guidance for AI Adoption

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The below table details general guidance and resources for the adoption of AI tools in the NHS and in healthcare, which are non-specific to stages of the genomic pathway, but still relevant to those looking to adopt AI in Genomic Medicine.

Name	Author	Areas Covered	Link
Regulations and guidance for adopters	NHS AI Digital Regulations Service	Data compliance checklist for adopters. Guidance on what regulations and best practice principles to follow when adopting your digital health technology.	https://www.digitalregulations.innovation.nhs.uk/regulations-and-guidance-for-adopters/
A buyer's guide to artificial intelligence in health and care	NHS England Digital	AI buyer's guide assessment template to be used in conjunction with the full guide. Questions which must be considered when buying off-the-shelf AI products.	https://digital.nhs.uk/services/ai-knowledge-repository/develop-ai/a-buyers-guide-to-ai-in-health-and-care#document-content
Planning and implementing real-world artificial intelligence (AI) evaluations: lessons from the AI in Health and Care Award	NHS England	Brief overview of the AI Health and Care Award. Evaluation framework for four 'phases' of product-readiness. Key lessons learnt about how to evaluate AI tools and products, and what to consider.	https://www.england.nhs.uk/long-read/planning-and-implementing-real-world-ai-evaluations-lessons-from-the-ai-in-health-and-care-award/#detailed-review-of-lessons-learned-at-each-stage-of-the-evaluation-process
Evidence standards framework for digital health technologies	NICE (National Institute for Health and Care Excellence)	A framework which can be used to evaluate the benefits of for a wide range of digital health technologies (DHTs), including artificial intelligence tools. Provides examples of information that can be used for different areas.	https://www.nice.org.uk/corporate/ecd7
Navigating AI – practical questions for NHS leaders to ask	NHS Providers	Questions for leaders to consider and ask to vendors and clinicians when procuring AI.	https://nhsproviders.org/resources/navigating-ai-practical-questions-for-nhs-leaders-to-ask
Top ten steps to get your genomics data AI-Ready	Wellcome Sanger Institute	Podcast and blog on recommendations for making genomic data AI-ready	Top ten steps to get your genomics data AI-Ready - Wellcome Sanger Institute Blog

The below table details other resources for AI adoption in healthcare.

Name	Author	Areas Covered	Link
AI Knowledge Repository Hub	NHS England Digital	Resources for understanding, developing and spreading and scaling AI. Including NHS case studies, NHS guidance and articles.	https://digital.nhs.uk/services/ai-knowledge-repository
AI Dictionary	NHS AI Lab	Dictionary of key terms related to AI in healthcare.	https://nhsx.github.io/ai-dictionary

Guidance on Involving Patients and the Public in AI Adoption - Coming Soon

NHS Genomic Medicine Workflow Overview

1. Identify patients for genomic Testing

- 1a — Refer to clinic
- 1b / A — Triage to clinic
- 1c / B — Review in clinic
- 1d — Determine genomic test eligibility
- 1e — Order genomic test

2. Generate and process genomic data

- 2a / C — Validate genomic test request
- 2b / C — Test DNA sample
- 2c / C — Process genomic data (variant calling, alignment)

3. Interpret genomic data

- 3a / C — Predict genomic variant effect
- 3b — Review in multidisciplinary team

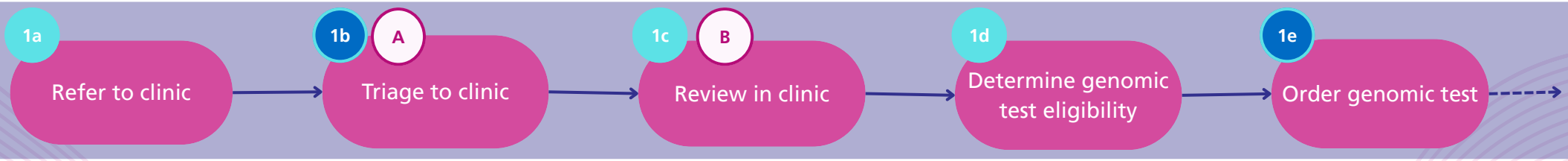
4. Act on genomic results

- 4a / 4b / D — Counsel patient
- 4c / D — Review relatives for cascade testing
- 4d / D — Perform re-analysis
- 4e / D — Match to clinical trials

Key

- Current NHS Deployment
- Use case identified
- Guidance

1. Identify Patients for Genomic Testing



Current NHS Deployments of AI Tools

1a **AI-assisted patient identification.**
MendelScan - Over 50 GP Practices
MendelScan uses AI NLP algorithms to identify patients in primary care who meet genomic testing criteria. MHRA-registered medical device.
Contact: peter@mendelian.co

1c **Ambient scribe technology.**
TORTUS Ambient Scribe - Pilot at Great Ormand Street Hospital (GOSH) (Clinical Genetics)
Trial of TORTUS ambient scribe completed by DRIVE team. TORTUS listens to and scribes consultations to help automate documentation.
Contact: pooja.dasani@nhs.net; Ajith.Kumar@gosh.nhs.uk
B **Guidance: Adopting ambient scribing products in health and care settings - NHS England**

1d **AI-assisted eligibility tools.**
Phenotips - Oxford University Hospitals (OUH) ; Cambridge University Hospitals; East Genomic Medicine Service; GOSH
PhenoTips tools include predictive tools for HPO terms and one-click pedigree assessment for cancer risk.
Contact: Dorothy.Halliday@ouh.nhs.uk; r.sandford@nhs.net; jo.lowry@uhl-tr.nhs.uk; Pooja.dasani@nhs.net; Ajith.Kumar@gosh.nhs.uk

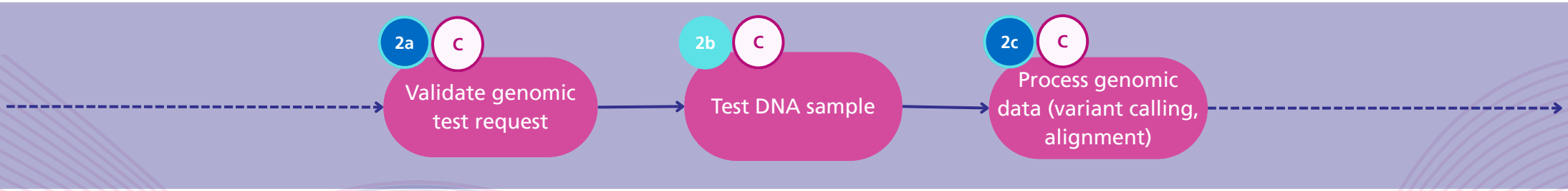
Potential use cases identified

1b **AI-assisted triage tools.**

A **Guidance: https://www.cqc.org.uk/what-we-do/how-we-work-people/digital-triage-health-services**

1e **AI-assisted test order forms.** Tools which read previous documentation to fill out test order forms.

2. Generate and Process Genomic Data



Current NHS Deployments of AI Tools

2b **AI-assisted lab protocols.**

OpenTrons AI - Oxford University Hospitals

OpenTrons AI enables users to create protocols using natural language input. Users can generate either a Python (.py) file or an online protocol designer file (.json) by specifying labware, liquid definitions, and preliminary protocol requirements. The resulting protocol can then be uploaded directly to the OT-2 instrument for execution.

C **Contact: Callum.Spencer@ouh.nhs.uk**

Guidance: In vitro diagnostic medical devices: guidance on legislation - GOV.UK

Potential use cases identified

2a **AI flagging for incomplete test request information.** Tools which flag missing phenotype or other test request information and automatically send back to the referrer.

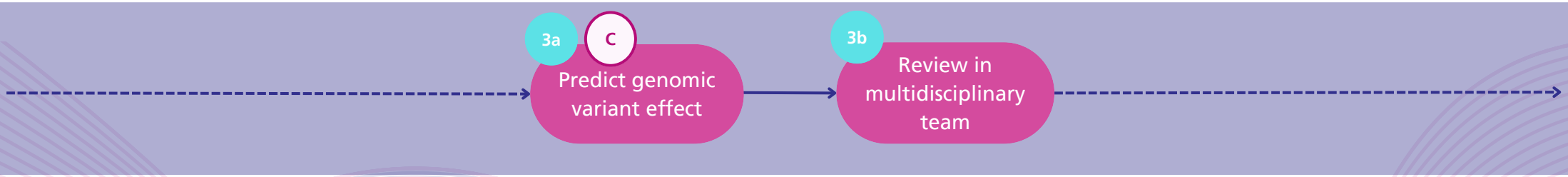
C **Guidance:** In vitro diagnostic medical devices: guidance on legislation - GOV.UK

2c **AI-assisted variant calling.**

DeepVariant. DeepVariant is a data analysis pipeline, identifying genetic variants from sequencing data. It uses a deep learning tool for variant calling.

C **Guidance:** In vitro diagnostic medical devices: guidance on legislation - GOV.UK

3. Interpret Genomic Data



Current NHS Deployments of AI Tools

3a **AI-assisted variant interpretation decision support tools**

Exomiser - Genomics England

- Exomiser finds potential disease-causing variants from whole-exome or whole-genome sequencing data. The Exomiser variant prioritisation framework creates a score using allele frequency and pathogenicity, and phenotpyic similarity.

AI MARRVEL - Great Ormand Street Hospital (GOSH)

- Benchmarking variant effect predictors and variant prioritisation tools.
- **Contact: Henry.Cope@rmh.nhs.uk**

Alpha Missense - Guys and St Thomas' NHS Foundation Trust, GOSH

- Using Alpha Missense for in silico prediction of pathogenicity of missense variants.
- **Contact: eilidh.jackson@nhs.net; Henry.Cope@rmh.nhs.uk**

Splice AI - University Hospital Southampton; GOSH; GSTT

- Variant interpretation. Implemented Splice AI within the lab, for clinical scientists to use on demand as part of their variant interpretation SOP.
- **Contact: manuel.dominguezbecerra@nhs.net; Henry.Cope@rmh.nhs.uk; eilidh.jackson@nhs.net**

Emedgene - Sheffield Children's Hospital; North-East and Yorkshire Genomic Laboratory Hub; North-West Genomic Laboratory Hub; GOSH

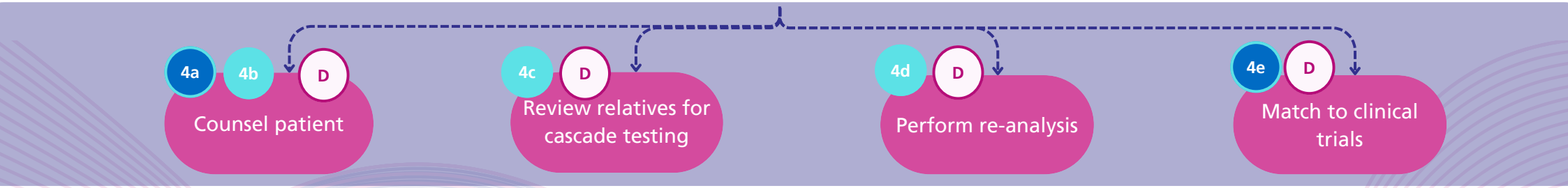
- Using AI to prioritise variants from NGS and WGS sequencing, annotate with relevant data to help clinical scientists triage a short list of likely causative variants for further variant classification.
- **Contact: m.durkie@nhs.net ; Janet.Taylor2@mft.nhs.uk; Henry.Cope@rmh.nhs.uk**

C **Guidance: In vitro diagnostic medical devices: guidance on legislation - GOV.UK**

3b **AI-assisted administrative support. Microsoft Co-Pilot - Across the NHS**

For non-clinical use, such as re-wording materials to be patient friendly, meeting minutes, and transcript summaries.

4. Act on Genomic Results



Current NHS Deployments of AI Tools

D — **Guidance: Explaining decisions made with AI | ICO**

4b — **AI-generated patient information leaflets.**
Manchester University NHS Foundation Trust.
Contact: siddharth.banka@mft.nhs.uk

4c — **See 'Determine test eligibility' (page 7)**

4d — **See 'Predict variant effect' (page 9)**

Potential use cases identified

4a — **AI Genetic Counselling tools.**
Gia counselling agent. Invitae's Gia is a genetic counselling agent which takes patients' information prior to an appointment. It also produces guides and summaries for patients.

4e — **AI-assisted clinical trials matching.** Tools which read a patients' information (e.g. EHRs, test results, phenotypic information) to match them to a clinical trial based on eligibility.

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For enquiries and feedback, please contact: **gstt.genomicainetwork@nhs.net**