NHS
Genomic AI Network

Genomic AI Tool Log

To view the list of Genomic AI tools being deployed in NHS Trust, go to page 4

Name of Tool	Company/ Developer	Description of tool & benefits	Stage of genomics pathway impacted
AI MARRVEL	MARRVEL	Uses random-forest machine-learning classifier trained on over 3.5 million variants from thousands of diagnosed cases. Incorporates expert-engineered features into training to recapitulate the intricate decision-making processes in molecular diagnosis. Improves rate of accurate genetic diagnosis.	Variant Interpretation
AION	Nostos Genomics	AI-driven variant interpretation platform AION automatically identifies likely pathogenic variants.	Variant Interpretation
Alamut Genova	SOPHiA GENETICS	Alamut Genova integrates AI-driven features to interpret complex genomic variants. The software combines analytical capabilities of the SOPHiA AI platform with advanced visualization tools, enabling precise identification and exploration of variants.	Variant Interpretation
CADD	University of Washington	Combined Annotation Dependent Depletion, combines a variety of predictive features in a machine-learning algorithm to predict the deleteriousness of genetic variants.	Variant Interpretation
CADD-Splice	University of Washington	CADD for prediction of splicing effects	Variant Interpretation
Congenic AI	Congenica	Congenica AI uses machine learning to classify and rank all variants so you can focus on those that matter the most.	Variant Interpretation
DeepVariant	Google	Analysis pipeline that uses a deep neural network to call genetic variants from next-generation DNA sequencing data	Variant Interpretation
DNABert	Yanrong Ji	A pre-trained bidirectional encoder representation to capture global and transferrable understanding of Pre-trained bidirectional encoder representation to capture global and transferrable understanding of genomic DNA sequences based on up and downstream nucleotide contexts.	Variant Interpretation
Emedgene	Illumina	Variant interpretation software streamlines tertiary analysis workflows for rare disease genomics and other germline research applications. Powered by Explainable AI (XAI) and automation, save 50-75% time per sample.	Variant Interpretation
Exomiser	Genome Diagnostics	Uses artificial intelligence and machine learning to help interpret genetic data and finds potential disease-causing variants from whole-exome or whole- genome sequencing data.	Variant Interpretation
Fabric GEM	Fabric Genomics	Fabric GEM utilises AI to enable accurate and near-instant identification of disease-causing genes in WGS and WES	Variant Interpretation

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If you have feedback or any questions, please email: gstt.genomicainetwork@nhs.net

Name of Tool	Company/ Developer	Description of tool & benefits	Stage of genomics pathway impacted
Galactic AI	Biorelate	Uses AI to connect together causal insights from multiple sources and uncovers relationships not previously described in the literature.	Variant Interpretation
Eye2Gene	UCL & Moorfields Eye Hospital	Uses AI on retinal scans to predict which inherited retinal disease the patient has, and specifically which gene is likely to be affected. Once validated, Eye2Gene can help make decisions about patient care.	Patient Identification
HyenaDNA	Stanford University	HyenaDNA is a genomic foundation model that leverages advanced deep learning techniques to analyse DNA sequences with unprecedented context lengths and single nucleotide resolution.	Variant Interpretation
Lucid AI	Lucid Genomics	AI powered platform for variant detection, prioritization, interpretation and data sharing. Full secondary and tertiary analysis workflow for SNVs, SVs, InDels and Repeat Expansions covering short and long reads.	Variant Interpretation
Mastermind	Genomenon	Leverages AI-driven technologies to process and interpret vast amounts of genomic literature, facilitating the identification and understanding of genetic variants associated with diseases	Variant Interpretation
Nucleotide Transformer	InstaDeep	AI tool designed for genomic data analysis, leveraging transformer-based deep learning models. Used to analyse DNA sequences and predict molecular phenotypes.	Variant Interpretation
NVIDIA Parabricks	NVIDIA	Leverages NVIDIA GPUs and AI to speed up the analysis of next-generation sequencing (NGS) data.	Variant Interpretation
OncoPortal Plus	SOPHiA GENETICS	Uses machine learning for genomic data analysis, primarily used in cancer genomics to assist clinicians in identifying and interpreting genetic variants that may be relevant to cancer diagnosis and treatment	Variant Interpretation
Primate AI - 3D	Illumina	Deep-learning network trained on 4.5 million common genetic variants from 233 primate species. Accurately quantifies missense variant pathogenicity in humans, improving discovery of genes affecting clinical phenotypes.	Variant Interpretation
REVEL	Illumina	Rare Exome Variant Ensemble Learner: An ensemble method for predicting the pathogenicity of missense variants on the basis of individual tools. Utilizes a machine learning model to predict the functional impact of rare genetic variants.	Variant Interpretation
SOPHiA DDM Platform	SOPHiA GENETICS	SOPHiA DDM™ for Genomics uses advanced AI algorithms to detect, annotate, and pre-classify genomic variants from complex and noisy NGS sequencing data. Knowledge sharing across the SOPHiA GENETICS network supports faster, more confident decision-making for Rare & Inherited Disorders.	Variant Interpretation
SpliceAI	Illumina	Leverages AI to annotate genetic variants with their predicted effect on splicing	Variant Interpretation

Name of Tool	Company/ Developer	Description of tool & benefits	Stage of genomics pathway impacted
Tensorflow Genomics IO	Google	Open-source machine learning framework, leverages deep learning techniques for tasks such as DNA sequence analysis, variant calling and prediction of genetic diseases.	Variant Interpretation
PhenoScore	Radboud University Medical Center	Open source, AI-based phenomics framework that combines facial recognition technology with Human Phenotype Ontology (HPO) data analysis to quantify phenotypic similarity at both the level of individual patients as well as of cohorts.	Variant Interpretation
BigRNA	Deep Genomics	Accurately predicts the tissue-specific regulatory mechanisms of RNA expression and the binding sites of proteins and microRNAs, plus the effects of variants and candidate therapeutics.	Pharmacogenomics
Datum	Datum	Provides pharmacogenomics recommendations, details on phenoconversion, genotype implications, clinical considerations, AI-based chat using LLM and precision guided recommendations.	Pharmacogenomics
Helical AI	Helical	Helical is the first open-core Platform for Genomics Bio Foundation Models solving the hardest problems in drug discovery (target identification, biomarker discovery, patient stratification, sequence design). With new models being released at an incredible pace, Helical's focuses on flexible model selection (i.e. the best model and infrastructure for each use case) and the toolkit to operation Bio Foundation Models (i.e. fine-tuning, RLHF, in-silico perturbations) to close the gap between models and applications.	Pharmacogenomics
Face2Gene Gestaltmatch	FDNA	Patient identification for genetic testing, rare disease matching using facial phenotype descriptors.	Patient Identification
MendelScan	Mendelian	Captures disease features from electronic health records across a patient population for early disease diagnoses. MHRA-registered medical device.	Patient Identification
Gia	Invitae	Genetic Information Assistant: A HIPAA-compliant clinical chatbot, Gia facilitates virtual conversations with patients, including intake of family history and automatic delivery of results. Gia can walk patients through genetic test results or schedule a genetic counsellor.	Other
PCR.AI	Diagnostics AI	Leverages artificial intelligence to improve accuracy, standardization, and efficiency in PCR testing, which is crucial for various applications, including infection detection and genetic analysis.	Data Processing & Transformation
PhenoTips	Phenotips	PhenoTips is a centralised, cloud-based database for family history and associated information that integrates with the existing Electronic Patient Records	Clinical Decision
VarChat	Engenome	Automates ACMG guidelines and prioritizes variants to highlight candidate diagnoses.	Variant Interpretation

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NHS Deployments of commercially available Genomic AI Tools

Name of Tool	NHS Trust	Description	Stage of genomics pathway impacted	NHS Deployment Key Contact Details
AI-MARRVEL	Great Ormond Street Hospital NHS Foundation Trust	Benchmarking Variant Effect Predictors & Variant prioritisation tools	Variant Interpretation	Henry.Cope@rmh.nhs.uk
AlphaMissense	Guy's and St Thomas' NHS Foundation Trust & Synnovis	Using AlphaMissense for in silico prediction of pathogenicity of missense variants	Variant Interpretation	eilidh.jackson@nhs.net
AlphaMissense	Great Ormond Street Hospital NHS Foundation Trust	Benchmarking Variant Effect Predictors & Variant prioritisation tools	Variant Interpretation	Henry.Cope@rmh.nhs.uk
Emedgene	Sheffield Children's NHS Foundation Trust - Sheffield Diagnostic Genetics Service, NEY GLH	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	Variant Interpretation	m.durkie@nhs.net
Emedgene	North West Genomic Laboratory Hub (NW GLH)	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	Variant Interpretation	Janet.Taylor2@mft.nhs.uk
Emedgene	Great Ormond Street Hospital NHS Foundation Trust	Benchmarking Variant Effect Predictors & Variant prioritisation tools	Variant Interpretation	Henry.Cope@rmh.nhs.uk
Splice AI	University Hospital Southampton NHS Foundation Trust	SpliceAI is a deep learning-based tool that predicts the impact of genetic variants on splicing. Developed by Illumina, it analyses sequences to identify potential splice site disruptions, aiding in variant interpretation for genomic research and clinical diagnostics.	Variant Interpretation	manuel.dominguezbecerra@nhs.net
Splice AI	Guy's and St Thomas' NHS Foundation Trust & Synnovis	Using SpliceAI in variant interpretation	Variant Interpretation	eilidh.jackson@nhs.net
Splice AI	Great Ormond Street Hospital NHS Foundation Trust	Benchmarking Variant Effect Predictors & Variant prioritisation tools	Variant Interpretation	Henry.Cope@rmh.nhs.uk
Phenotips	Oxford University Hospitals NHS Foundation Trust	Harnessing PhenoTips' cloud-hosted Genomic Health Record to digitise, integrate and standardize genomic care	Data Processing & Transformation	
Phenotips	Cambridge University Hospitals	Used PhenoTips to improve family history taking. Provides data capturing, document storage, and improved diagnoses for the oncology and dysmorphology departments	Clinical Decision	
Phenotips	East GMSA & GLH - University Hospital Leicester NHS Trust & Nottingham University Hospitals	Using PhenoTips - A digital tool for taking the genetic family history of patients, which can improve the process for diagnosing cancers and rare conditions	Clinical Decision	r.sandford@nhs.net jo.lowry@uhl-tr.nhs.uk