Genomic AI Tool Log

NHS Genomic AI Network

Name of Tool	Company/ Developer	Description of tool & benefits	Stage of genomics pathway impacted	NHS Deployment Contact	Tool/ Company URL
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		https://ai.marrvel.org
AION	Nostos Genomics	AI-driven variant interpretation platform AION automatically identifies likely pathogenic variants.	Variant Interpretation		https://www.nostos-genomics.com/
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		www.sophiagenetics.com/platform/alamut-visual-plus
AlphaFold 3	Google DeepMind	AlphaFold is an AI system developed by Google DeepMind that predicts a protein's 3D structure from its amino acid sequence.	Genomic Sequencing	alphafold@deepmind.com	https://deepmind.google/technologies/alphafold
AlphaMissense	Google DeepMind	Machine-learning model which classifies missense variants. Analyses related protein sequences and the structural context of variants to estimate how likely a variant is to be pathogenic.	Variant Interpretation		https://github.com/google-deepmind/alphamissense
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	Pharmacogenics		https://www.deepgenomics.com/AI-Platform/
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		https://cadd.gs.washington.edu/

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CADD-Splice	University of Washington	CADD for prediction of splicing effects.	Variant Interpretation		https://cadd.gs.washington.edu/
Congenica 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		https://genomics.congenica.com/exomiser
Datum	Datum	Provides pharmacogenomics recommendations, details on phenoconversion, genotype implications, clinical considerations, AI-based chat using LLM and precision guided recommendations.	Pharmaco genomics	aris.saoulidis@nhs.net	http://datum.bio
DeepVariant	Google	Analysis pipeline that uses a deep neural network to call genetic variants from next-generation DNA sequencing data	Variant Interpretation		https://github.com/google/deepvariant
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		https://github.com/jerryji1993/DNABERT
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		https://emea.illumina.com/products/by-type/informatics-products/emedgene.html
Exomiser	Genome Diagnostics	Uses atificial 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		https://github.com/exomiser/Exomiser

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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		https://fabricgenomics.com/fabric-gem/
Face2Gene Gestaltmatcher	FDNA	Patient identification for genetic testing, rare disease matching using facial phenotype descriptors.	Patient Identification	info@fdna.com	www.face2gene.com/gestaltmatcher
Galactic AI	Biorelate	Uses AI to connect together causal insights from multiple sources and uncovers relationships not previously described in the literature.	Variant Interpretation		https://biorelate.com/solutions/data-scientists
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 counselor.	Other		https://www.invitae.com/providers/gia-chatbot
Helical AI	Helical	Platform for Bio Foundation Models - Leverage biological data to accelerate drug discovery with the most powerful use cases.	Pharmaco genomics		https://www.helical-ai.com/
HyenaDNA	Stanford University	HyenaDNA is a genomic foundation model that leverages advanced deep learning techniques to analyze DNA sequences with unprecedented context lengths and single nucleotide resolution.	Variant Interpretation		https://github.com/HazyResearch/hyena-dna
Lucid AI	Lucid Genomics	AI powered platform for variant detection, prioritization, interpretation and data sharing.	Variant Interpretation	achim@lucid-genomics.com	https://www.lucid-genomics.com/
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 disease.	Variant Interpretation		https://www.genomenon.com/

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MendelScan	Mendelian	Captures disease features from electronic health records across a patient population for early disease diagnoses	Patient Identification	peter@mendelian.co	https://www.mendelian.co/
Nucleotide Transformer	InstaDeep	AI tool designed for genomic data analysis, leveraging transformer-based deep learning models. Used to analyze DNA sequences and predict molecular phenotypes	Variant Interpretation		https://github.com/instadeepai/nucleotide-transformer
NVIDIA Parabricks	NVIDIA	Leverages NVIDIA GPUs and AI to speed up the analysis of next-generation sequencing (NGS) data.	Variant Interpretation		https://www.nvidia.com/en-gb/clara/genomics/
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		sophiagenetics.com/platform/oncoportal-plus/
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		https://www.diagnostics.ai/
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		https://phenotips.com/
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		https://github.com/Illumina/PrimateAI-3D

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NHS
Genomic AI Network

NHS Deployments of commercially available Genomic AI Tools

Name of Tool	NHS Trust	Description	Stage of genomics pathway impacted	NHS Deployment 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,	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	Variant Interpretation	m.durkie@nhs.net 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
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 &	Using PhenoTips - A digital tool for taking the genetic family history of patients, which can improve the	Clinical Decision	r.sandford@nhs.net jo.lowry@uhl-tr.nhs.uk
Phenotips	Oxford University Hospitals NHS Foundation Trust	Harnessing PhenoTips' cloud-hosted Genomic Health Record to digitse, integrate and standardise genomic care	Data Processing & Transformation	
Splice AI	University Hospital Southampton NHS Foundation Trust	Using SpliceAI in variant interpretation	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

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