Name of Tool	Company/Developer	Description of tool & benefits	Stage of genomics impacted	NHS Deployment Contact Name	NHS Deployment Key Contact Details	Tool/Company URL
Datum	Datum	Provides pharmacogenomics recommendations, details on phenoconversion, genotype implications, clinical considerations, AI-based chat using LLM and precision guided recommendations.	Pharmacogenomics	Aris Saoulidis	aris.saoulidis@nhs.net	http://datum.bio
GenePy	University of Southampton	Scoring to estimate gene pathogenicity using next-generation sequencing data	Variant Interpretation	Sarah Ennis		
MendelScan	Mendelian	Captures disease features from electronic health records across a patient population for early disease diagnoses	Patient Identification	Peter Fish		
Face2Gene Gestaltmatcher	FDNA	Patient identification for genetic testing, rare disease matching using facial phenotype descriptors.	Patient Identification			www.face2gene.com/gestaltmatcher
SpliceAI	Illumina	Annotates genetic variants with their predicted effect on splicing	Variant Interpretation			emea.illumina.com
NHS Deployment	**University Hospital Southampton NHS Foundation Trust**	**N/A**	**Variant interpretation, classification and prioritisation**		**gstt.genomicainetwork@nhs.net**	
NHS Deployment	**Guy's and St Thomas' NHS Foundation Trust Synnovis**	**N/A**	**Variant interpretation, classification and prioritisation**		**gstt.genomicainetwork@nhs.net**	
VarChat	Engenome	Automates ACMG guidelines and prioritizes variants to highlight candidate diagnoses.	Variant Interpretation			https://varchat.engenome.com/
SOPHiA DDM Platform	SOPHiA GENETICS	Analyzes complex data to provide precision medicine to patients worldwide, connects network of healthcare institutions globally	Variant Interpretation			https://www.sophiagenetics.com/technology/
OncoPortal Plus	SOPHiA GENETICS	Evidence-based decision support software intended as an aid in the interpretation of variants	Variant Interpretation			sophiagenetics.com/platform/oncoportal-plus/
Alamut Visual Plus	SOPHiA GENETICS	Reduce variant analysis time with curated data and robust predictors in a single workflow	Variant Interpretation			https://www.sophiagenetics.com/platform/alamut-visual-plus/
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
Mastermind	Genomenon	Comprehensive source of published genomic literature. Simplifies complex genetic data into actionable insights for patient diagnosis and precision medicine development	Pharmacogenomics			Mastermind Genomic Genetic PLG
GenomicIO	Tensorflow	Data Handling	Data Processing & Transformation			
Pytorch	MetaAI	Open source deep learning framework built to be flexible and modular for research, with the stability and support needed for production deployment.	Other			

		Rare Exome Variant Ensemble Learner				
REVEL	NCBI	An ensemble method for predicting the pathogenicity of missense variants on the basis of individual tools	Variant Interpretation			
AlphaMissense	Google DeepMind	Analyses related protein sequences and the structural context of variants to estimate how likely a variant is to be pathogenic	Variant Interpretation			
NHS Deployment	**Guy's and St Thomas' NHS Foundation Trust Synnovis**	**N/A**	**Variant interpretation, classification and prioritisation**		**gstt.genomicainetwork@nhs.net**	
AlphaFold	Google DeepMind	3D Protein Modelling	Genomic Sequencing		alphafold@deepmind.com	AlphaFold Protein Structure Database
DECIPHER	Decipher Genomics	DECIPHER helps the clinical community share and compare human genome variants and phenotypes in a database of tens of thousands of patients worldwide.	Variant Interpretation			https://www.deciphergenomics.org/
Emedgene	Illumina	Variant interpretation software streamlines tertiary analysis workflows for rare disease genomics and other germline research applications	Variant Interpretation			https://emea.illumina.com/products/by-type/informatics-products/emedgene.html
NHS Deployment	**Sheffield Children's NHS Foundation Trust**	**N/A**	**Variant interpretation, classification and prioritisation**		**gstt.genomicainetwork@nhs.net**	
C.R.E.O.L.A	OSLER Tortus	Clinical Review Of LLMs and AI Evaluation of LLM models prior to their deployment	Other			https://tortus.ai/creola/
PhenoScore	Dingemans et al	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			
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			
NHS Deployment	**Nottingham University Hospitals**	**N/A**		**Professor Dick Sandford Jo Lowry**	**r.sandford@nhs.net jo.lowry@uhl-tr.nhs.uk**	**https://www.eastgenomics.nhs.uk/**
NHS Deployment	**Cambridge University Hospitals**	**N/A**		**Professor Dick Sandford Jo Lowry**	**r.sandford@nhs.net jo.lowry@uhl-tr.nhs.uk**	**https://www.eastgenomics.nhs.uk/**
NHS Deployment	**University Hospitals of Leicester**	**N/A**		**Professor Dick Sandford Jo Lowry**	**r.sandford@nhs.net jo.lowry@uhl-tr.nhs.uk**	**https://www.eastgenomics.nhs.uk/**

CADD	University of Washington	Combined Annotation Dependent Depletion, tool for scoring the deleteriousness of single nucleotide variants, multi-nucleotide substitutions as well as insertion/deletions variants in the human genome	Variant Interpretation	https://cadd.gs.washington.edu/
CADD-Splice	University of Washington	CADD for prediction of splicing effects	Variant Interpretation	https://cadd.gs.washington.edu/
Exomiser	Wellcome Sanger Institute	Finds potential disease-causing variants from whole-exome or whole-genome sequencing data.	Variant Interpretation	
Visiopharm APPs	Visiopharm	APPs designed to solve specific problems using our tissue image analysis software covering all tissue, stain and modality types APP Center can significantly reduce your development time by providing a base for building and refining your own workflows.	Other	https://visiopharm.com/
Nucleotide Transformer	H Dalla-Torre	The Nucleotide Transformer model accurately predicts diverse genomics tasks after fine-tuning.	Other	
DNABert	Yanrong Ji	A novel 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
HyenaDNA	arXiv	Genomic foundation model pretrained on the human reference genome	Variant Interpretation	
IBM SPSS	IBM	Software platform offers advanced statistical analysis, a vast library of machine learning algorithms, text analysis, open-source extensibility	Other	
NVIDIA Parabricks	NVIDIA	GPU-accelerated software suite for secondary genomic analysis	Variant Interpretation	https://www.nvidia.com/en-gb/clara/genomics/
Fabric GEM	Fabric Genomics	Fabric GEM utilizes AI to enable accurate and near-instant identification of disease-causing genes in WGS & WES	Variant Interpretation	https://fabricgenomics.com/fabric-gem/