Schedule of Accreditation

issued by

United Kingdom Accreditation Service

2 Pine Trees, Chertsey Lane, Staines-upon-Thames, TW18 3HR, UK

10170

Accredited to ISO 15189:2012

Genomics England Limited

Issue No: 004 **Issue date:** 08 December 2021

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Testing performed at the above address only

DETAIL OF ACCREDITATION

Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
MATERIALS/PRODUCT Tested Whole human genome sequencing data from an external source	<u>BIOINFORMATICS</u> Identification of Somatic and Germline regions in cancer patients which contain potentially actionable variants as follows: Single Nucleotide Variants (SNVs) Indels Copy Number Variations (CNVs) Structural Variant (SVs) Internal Tandem Duplications (ITDs)	Documented in-house methods: Alignment, Variant calling, QC steps and checks and identification of potentially clinical actionable variation from WGS data from cancer samples Software and platforms: Bertha (Workflow manager) Open CGA (LIMS) Cellbase (Genomic reference and annotation database) Panel App (Knowledgebase of gene phenotype association) Interpretation Portal Decision Support (BSVI) SOPs: BER-SOP-0001 Cancer Bioinformatics Pipeline BER-SOP-0007 GMS Cancer Bioinformatics Pipeline

Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
		Dragen Alignment Dragen variant caller (Small variants) Strelka (Somatic small variant caller) Pindel (ITDs) Canvas/Dragen CNV (CNVs) and Manta (SVs) Clinical Variant Ark (CVA, Knowledgebase) Starling for germline small variants
MATERIALS/PRODUCT Tested (cont'd)	BIOINFORMATICS (cont'd)	

Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
Whole human genome sequencing data from an external source	Identification of variants (as listed below) with potential diagnostic utility in genomes of individuals or families with rare diseases. Single Nucleotide Variants (SNVs) Indels Copy Number Variations (CNVs) Short tandem repeats (STRs)	Documented in- house methods: Alignment, Variant Calling, QC steps and checks and identification of potentially clinically actionable variation from WGS data from affected individuals with rare disease and their family members Software and platforms: Platypus (variant caller) Dragen Alignment Dragen variant caller (Small variants) Canvas/Dragen CNV (CNVs) and Manta (SVs) ExpansionHunter (STRs) Bertha (Workflow manager) Family selection workflow (assurance check) Genetic v reported (assurance check) Open CGA (LIMS) Variant Interpretation Pipeline Panel App (Knowledgebase of gene phenotype association) Cellbase (Genomic reference and annotation database) Interpretation Portal Decision Support (Congenica/Fabric) Clinical Variant Ark (CVA, Knowledgebase) SOPs: BER-SOP-0002 - Rare Disease Pipeline BER-SOP-0006 Rare Disease Pipeline for GMS (Genomics Medicine Service) IP–SOP-0001 Interpretation pipeline
MATERIALS/PRODUCT Tested (cont'd)	<u>BIOINFORMATICS (cont'd)</u>	

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Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
Whole human genome sequencing data from an external source	Identification of germline variants with potential drug toxicity implications (as listed below) in genomes of cancer patients. DPYD variants: c.1905+1G>A c.1679T>G c.2846A>T c.1129-5923C>G c.1236G>Aii	Documented in-house methods: Variant calling and identification of PGx variants in cancer patients Software and platforms: Dragen variant caller (Small variants) Strelka (Somatic small variant caller) Starling for germline small variants SOP: CUR – SOP -0001 Curation of Pharmacogenetics for genome analysis BER-SOP-0001 Cancer Bioinformatics Pipeline BER-SOP-0007 GMS Cancer Bioinformatics Pipeline Bertha, Interpretation Portal and CVA
END		