Schedule of Accreditation

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Genomics England Limited

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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
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: **Germline** Single Nucleotide Variants (SNVs) including DPYD variants with potential drug toxicity implications Indels Copy Number Variants (CNVs) larger than 2kb in size **Somatic** Single nucleotide variants (SNVs) Indels Copy number variants (CNVs) Structural Variant (SVs) incl. • SSX fusions • 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 **Germline genome alignment** Dragen aligner **Somatic genome alignment** Dragen aligner **Germline variant callers** Dragen variant caller (small variants and copy number variants) **Somatic variant callers** Strelka (SNVs and indels) Canvas (CNVs) Manta (SVs) JuLI (SSX fusions) Pindel (ITDs) **Software and platforms** Bertha Orchestrator (Workflow manager) Open CGA (LIMS) Cellbase (Genomic reference and annotation database) PanelApp (Knowledgebase of gene phenotype association) Interpretation Portal Decision Support Software (DSS)) **SOPs:** GUI-BIO-010 Cancer Genome Analysis Guide SOP-BIO-005 Cancer Bioinformatics Pipeline

Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
Whole human genome sequencing data from an external source (cont'd)	Bioinformatics (cont'd) Identification of variants (as listed below) with potential diagnostic utility in genomes of individuals or families with rare diseases. Single Nucleotide Variants (SNVs) Indels Short tandem repeats (STRs) Copy number variants (CNVs) larger than 2kb in size	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 **Genome alignment** Dragen Aligner **Variant Callers** Dragen variant caller (SNVs/Indels) Dragen/ExpansionHunter (STRs) Dragen (CNVs >2kb) and Manta (CNVs 2-10kb) **Software and platforms** Bertha (Workflow manager) Family selection workflow (assurance check) Genetic v reported (assurance check) Open CGA (LIMS) Exomiser (Variant prioritisation) PanelApp (Knowledgebase of gene phenotype association) Cellbase (Genomic reference and annotation database) Interpretation Portal Congenica (Decision Support Software) Clinical Variant Ark (CVA, Knowledgebase) **SOPs:** GUI-BIO-009 Rare Disease Genome Analysis Guide SOP-BIO-032 Rare Disease Pipeline For GMS
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