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 variants with potential drug toxicity implications Indels Copy Number Variants (CNVs) larger than 2kb in size **Somatic variants and variants of uncertain origin (tumour-normal and tumour only)** Single nucleotide variants (SNVs) Indels Copy number variants (CNVs) Structural Variant (SVs) incl. • SSX fusions • Internal tandem duplications (ITDs) • Dux4 rearrangements	Documented in-house methods: Cancer pipeline Flexible scope limited to the application of the methods listed in GUI-BIO-010 and POL-BIO-003 for the agreed boundaries of software updates to the pipeline components, or the addition of new components to the pipeline which supplement existing functionality.

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) Whole human genome sequence data from an external source Identification of variants 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: Rare Disease pipeline Flexible scope limited to the application of the methods listed in GUI-EXT-031 and POL-BIO-004 for the agreed boundaries of software updates to the pipeline components, or the addition of new components to the pipeline which supplement existing functionality.

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 Somatic and Germline regions in cancer patients which contain potentially actionable variants as follows: **Germline variants (tumour normal)** Single Nucleotide Variants (SNVs) including variants with potential drug toxicity implications Indels Copy Number Variants (CNVs) larger than 2kb in size **Somatic variants and variants of uncertain origin (tumour-normal and tumour only)** Single nucleotide variants (SNVs) Indels Copy number variants (CNVs) Structural Variant (SVs) incl. • SSX fusions • Internal tandem duplications (ITDs) • Dux4 rearrangements Pan genomic biomarkers Tumour Mutational Burden	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) Somatic copy number variants (CNVs) using DRAGEN (CNVs) Manta (SVs) JuLI (SSX fusions) Pindel (ITDs) Somatic DUX4 rearrangements using Pelops (DUX4) rearrangements **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 POL-BIO-003 Cancer Genome 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)	<u>Bioinformatics</u> (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-EXT-031 Rare Disease Genome Analysis Guide – Online POL-BIO-004 Rare Disease Pipeline for GMS
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