# Schedule of Accreditation

**issued by**

# United Kingdom Accreditation Service

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

**10170**

**Accredited to ISO 15189:2022**

## Genomics England Limited

**Issue No:** 013 **Issue date:** 06 May 2025


**Level 21**
**One Canada Square**
**Canary Wharf**
**London**
**E14 5AB**

**Contact: Stephen Street-Howard**
**Tel: +44 (0)808 2819 535**
**E-Mail: Stephen.Street-Howard@genomicsengland.co.uk**
**Website: https://www.genomicsengland.co.uk/**


**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><br><br>Identification of Somatic and Germline regions in cancer patients which contain potentially actionable variants as follows:<br><br>**Germline**<br>Single Nucleotide Variants (SNVs) including variants with potential drug toxicity implications<br>Indels<br>Copy Number Variants (CNVs) larger than 2kb in size<br><br>**Somatic variants and variants of uncertain origin (tumour-normal and tumour only)**<br>Single nucleotide variants (SNVs)<br>Indels<br>Copy number variants (CNVs)<br>Structural Variant (SVs) incl.<br>• SSX fusions<br>• Internal tandem duplications (ITDs)<br>• Dux4 rearrangements | Documented in-house methods:<br><br>Cancer pipeline<br><br>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)<br><br>Whole human genome sequence data from an external source<br>Identification of variants with potential diagnostic utility in genomes of individuals or families with rare diseases<br><br>Single Nucleotide Variants (SNVs)<br>Indels<br>Short tandem repeats (STRs)<br>Copy Number Variants (CNVs) larger than 2kb in size | Documented in-house methods:<br><br>Rare Disease pipeline<br><br>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)<br><br>Identification of Somatic and Germline regions in cancer patients which contain potentially actionable variants as follows:<br><br>**Germline variants (tumour normal)**<br>Single Nucleotide Variants (SNVs) including variants with potential drug toxicity implications<br>Indels<br>Copy Number Variants (CNVs) larger than 2kb in size<br><br>**Somatic variants and variants of uncertain origin (tumour-normal and tumour only)**<br>Single nucleotide variants (SNVs)<br>Indels<br>Copy number variants (CNVs)<br>Structural Variant (SVs) incl.<br>• SSX fusions<br>• Internal tandem duplications (ITDs)<br>• Dux4 rearrangements<br><br>Pan genomic biomarkers<br>Tumour Mutational Burden | Documented in-house methods:<br><br>Alignment, Variant calling, QC steps and checks and identification of potentially clinical actionable variation from WGS data from cancer samples<br><br>**Germline genome alignment**<br>Dragen aligner<br><br>**Somatic genome alignment**<br>Dragen aligner<br><br>**Germline variant callers**<br>Dragen variant caller (small variants and copy number variants)<br><br>**Somatic variant callers**<br>Strelka (SNVs and indels)<br>Somatic copy number variants (CNVs) using DRAGEN (CNVs)<br>Manta (SVs)<br>JuLI (SSX fusions)<br>Pindel (ITDs)<br>Somatic DUX4 rearrangements using Pelops (DUX4) rearrangements<br><br>**Software and platforms**<br>Bertha Orchestrator (Workflow manager)<br>Nextflow<br>Workflow configuration settings for the Genie workflow selection service (WSS)<br>Open CGA (LIMS)<br>Cellbase (Genomic reference and annotation database)<br>PanelApp (Knowledgebase of gene phenotype association)<br>Interpretation Portal<br>Decision Support Software (DSS)<br><br>**SOPs:**<br>GUI-BIO-010 Cancer Genome Analysis Guide<br>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) | Bioinformatics (cont'd)<br><br>Identification of variants (as listed below) with potential diagnostic utility in genomes of individuals or families with rare diseases.<br><br>Single Nucleotide Variants (SNVs)<br>Indels<br>Short tandem repeats (STRs)<br>Copy number variants (CNVs) larger than 2kb in size | Documented in-house methods:<br><br>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<br><br>**Genome alignment**<br>Dragen Aligner<br><br>**Variant Callers**<br>Dragen variant caller (SNVs/Indels)<br>Dragen/ExpansionHunter (STRs)<br>Dragen (CNVs >2kb) and Manta (CNVs 2-10kb)<br><br>**Software and platforms**<br>Bertha (Workflow manager)<br>Nextflow<br>Workflow configuration settings for the Genie workflow selection service (WSS)<br>Family selection workflow (assurance check)<br>Genetic v reported (assurance check)<br>Open CGA (LIMS)<br>Exomiser (Variant prioritisation)<br>PanelApp (Knowledgebase of gene phenotype association)<br>Cellbase (Genomic reference and annotation database)<br>Interpretation Portal<br>Congenica (Decision Support Software)<br>Clinical Variant Ark (CVA, Knowledgebase)<br><br>**SOPs:**<br>GUI-EXT-031 Rare Disease Genome Analysis Guide – Online<br>POL-BIO-004 Rare Disease Pipeline for GMS |
| END | | |