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
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) Short tandem repeats (STRs)	Documented in-house methods: QC steps and checks for aligned output of clinically interpreted genetic variants from WGS of cancer samples Software and platforms: Bertha (Workflow manager) Open CGA (LIMS) Cellbase (Genomic reference and annotation database) Panel App (Knowledgebase of gene phenotype association) Decision Support (BSVI) SOPs: BER-SOP-0001 Cancer Bioinformatics Pipeline

Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
MATERIALS/PRODUCT Tested (cont'd) Whole human genome sequencing data from an external source	<u>BIOINFORMATICS (cont'd)</u> 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: QC steps and checks and variant calling for aligned output of clinically interpreted variants from WGS of affected individuals with matched family counterparts Software and platforms: Platypus (variant caller) 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) Decision Support (Congenica/Fabric) SOPs: BER-SOP-0002 - Rare Disease Pipeline
END		