Schedule of Accreditation

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United Kingdom Accreditation Service

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Accredited to ISO 15189:2022

Sheffield Children's Hospital NHS Foundation Trust

Issue No: 009 **Issue date:** 14 January 2025

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Testing performed at the above address only

DETAIL OF ACCREDITATION

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE and FLUIDS Whole Blood, Bone Marrow, FFPE, Slide sections, Sperm, Saliva, Buccal swabs, Fetal Blood, Products of conception, Tissue, Fibroblasts, Lyophilised Cells, Amniocytes, Dried bloodspots, Buccal Swabs, Foetal Blood	<u>Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer</u> Forwarding of human tissue and fluids to partner laboratories in the NEYGLH or other specialist centres, receipt of test results data for in-house analysis and reporting.	
MLPA data files received from an external source within the NEYGLH	Detection known pathogenic variants and copy number changes [Definitive list in 407.103]	**Multiplex Ligation Probe Amplification (MLPA) – Data analysis only:** MLPA Analysis using MRC Holland Coffalyser data analysis software SOP 401.071

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE and FLUIDS (cont'd) Sanger sequencing data files received from an external source within the NEYGLH	<u>Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer</u> (cont'd) Detection and confirmation of known and unknown nucleic acid sequence variants and confirmation of SNVs and Indels and fusion genes [Definitive list of known targets listed in 401.336]	**Sanger Sequencing – Data analysis only** Analysis using mutation surveyor and variant interpretation and reporting using variant classification procedure SOPs, 601.235, 601.314 402.005
Real time PCR data files received from an external source within the NEYGLH	Quantitative analysis of known gene fusions events SNVs and indels (for the purpose of end point genotyping) [Definitive list in 407.102]	**Real Time PCR – Reporting only:** Results only SOPs: 401.094, 601.313, 601.037, 401.312, 401.313
ddPCR data files received from an external source within the NEYGLH	Detection of nucleic acid sequence variants - SNVs and Indels [definitive list in 407.446]	**ddPCR using Droplet Generator – Reporting only:** Results only SOPs: 401.283, 401.286
Fragment length data files received from an external source within the NEYGLH	Detection of fragment length size, deletions, known mutations, and repeat expansions [Definitive list in 407.100, 407.101]	**Fragment length analysis – Data analysis only:** GeneMapper software SOPs 401.026, 401.347, 401.036

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE and FLUIDS (cont'd) Next Generation Sequence data files received from an external source within the NEYGLH	<u>Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer</u> (cont'd) Sequencing of large gene panels for genetic variants (SNVs/indels/CNVs/gene fusions) [Definitive list in 413.004]	Next Generation Sequencing **– Data analysis only:** Analysis using NEYGLH or in-house pipeline and variant interpretation and reporting including variant classification procedures SOPs 401.058, 401.047, 402,005, 401.341, 401.394; 401.387, 401.398
SNP array data files received from an external source within the NEYGLH	Detection of microscopic chromosomal imbalance (gains and losses) expressed as changes to copy number	**SNP Array – Data analysis only:** Analysis and interpretation of genetic imbalances using BlueFuse Multi, the 'Heidelberg' classifier and web-based UCSC genome browser SOPs 401.340, 401.346, 401.388
Karyotyping image files received from an external source within the NEYGLH	Karyotyping Detection of chromosomal rearrangements or aberrations arising from: Prenatally detected Disorders Developmental disorders Reproductive medicine disorders Haematological/Oncology disorders Chromosomal breakage disorders	**Microscopic analysis chromosomes – Data analysis only:** SOPs 501.010, 401.356, 501.032, 501.044, 501.028 501.051

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE and FLUIDS (cont'd) FISH image files received from an external source within the NEYGLH	<u>Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer</u> (cont'd) Detection of chromosomal aberrations in the diagnosis of: [Definitive list 506.008 and as Probe Master Software] Haematological malignancy Bone marrow failure syndromes, Non haematological malignancies and constitutional disorders, Solid tumours and companion testing Using: Amplification probes Break apart probe Break apart rearrangement probe Centromeric Hetro chromatin Deletion/ Duplication probe Dual fusion rearrangement probe Duplication/ Deletion/Break Apart Probe Hetrochromatin Probe Sub Telomere	**Fluorescence in situ hybridisation (FISH) – Data analysis/Reporting only:** Results only
QF-PCR data files received from an external source within the NEYGLH	Rapid analysis of common trisomy's: [Definitive list in 401.006].	**QF-PCR – Data analysis only:** **Newcastle kit** **Leeds kit** Analysis by Genemapper Software SOPs 401.006, 401.026
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