

# Schedule of Accreditation



Organisation Name	Children's Health Ireland
Trading As	CHI at Crumlin: Department of Clinical Genetics Laboratory
INAB Reg No	417MT
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Accreditation Standard	EN ISO 15189
Standard Version	2022
Date of award of accreditation	17/04/2024
Scope Classification	Genetics
Services available to the public <sup>1</sup>	No

<sup>1</sup> Refer to document on interpreting INAB Scopes of Accreditation

Sites from which accredited services are delivered		
(the detail of the accredited services delivered at each site are on the Scope of Accreditation)		
	Name	Address
1	Head Office	Department of Clinical Genetics, Crumlin, Dublin, Dublin

# Scope of Accreditation

Head Office

Genetics

Category: A

Medical pathology field - Test	Test/assay	Specimen Type	Technique	Equipment	Method (CE/Non-CE/In house developed/based on standard method)	Std. Ref & SOP
1071 Cytogenetics - .25 Fluorescent in situ hybridisation	BCR/ABL1 Plus, dual fusion	Blood, Bone marrow, Human tissue, Slides (unstained)	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
	CBFA2T3/GLIS2, dual fusion		Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
	CBFB, break apart		Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-

				SlideMake, LP-CYT-Reporting	
	CBFB/MYH11, dual fusion	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
	CHD5/160H23	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
	Chromosome 10 enumerater probes	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
	Chromosome 11 enumerater probes	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems,LP-CYT-SlideMake, LP-CYT-Reporting
	Chromosome 12 enumerater probes	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
	Chromosome 15 enumerater probes	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-

			microscope, Metasystems		Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
Chromosome 16 enumerater probes	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems		Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
Chromosome 17 enumerater probes	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems		Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
Chromosome 18 enumerater probes	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems		Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
Chromosome 19 enumerater probes	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems		Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
Chromosome 2 enumerater probes	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems		Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
Chromosome 20 enumerater probes	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss		Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-

			microscope, Metasystems		GrantWBath, LP-CYT- Matasystems, LP-CYT- SlideMake, LP-CYT- Reporting
Chromosome 3 enumerater probes		Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHPRocedure LP-CYT-FISHAnalysis, LI- CYT-Thermocrite, LI-LAB- GrantWBath, LP-CYT- Matasystems, LP-CYT- SlideMake, LP-CYT- Reporting
Chromosome 4 enumerater probes		Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHPRocedure LP-CYT-FISHAnalysis, LI- CYT-Thermocrite, LI-LAB- GrantWBath, LP-CYT- Matasystems, LP-CYT- SlideMake, LP-CYT- Reporting
Chromosome 5 enumerater probes		Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHPRocedure LP-CYT-FISHAnalysis, LI- CYT-Thermocrite, LI-LAB- GrantWBath, LP-CYT- Matasystems, LP-CYT- SlideMake, LP-CYT- Reporting
Chromosome 6 enumerater probes		Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHPRocedure LP-CYT-FISHAnalysis, LI- CYT-Thermocrite, LI-LAB- GrantWBath, LP-CYT- Matasystems, LP-CYT- SlideMake, LP-CYT- Reporting
Chromosome 7 enumerater probes		Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHPRocedure LP-CYT-FISHAnalysis, LI- CYT-Thermocrite, LI-LAB- GrantWBath, LP-CYT- Matasystems, LP-CYT- SlideMake, LP-CYT- Reporting
Chromosome 8 enumerater probes		Manual/microscope	Thermobrite, Grant JB1 water	Based on standard method	LP-CYT-FISHPRocedure LP-CYT-FISHAnalysis, LI-

			bath, Zeiss microscope, Metasystems		CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
	Chromosome 9 enumerater probes	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHPRocedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
	Chromosome X enumerater probes	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHPRocedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
	Chromosome Y enumerater probes	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHPRocedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
	CKS1B/CDKN2C	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHPRocedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
	CRLF2, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHPRocedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting

D13S319/D13S1825/D12Z3	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
D15S10/D15Z1/PML Angelman	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
D22S75	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
D22S75/SHANK3	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
DEK/NUP214, dual fusion	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
del(13q)	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting

del(5q)/monosomy 5	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
del(7q)/monosomy 7	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
ELN/D7S486/D7S552	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
ERG/EWSR1, dual fusion	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
ETV6, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
ETV6/RUNX1, dual fusion	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting



EWSR1, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
FGFR1, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
FIP1L1-CHIC2-PDGFR, DelF	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
FLI1/EWSR1, dual fusion	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
FOXO1, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
FUS, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting

IGH, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
IGH/BCL2, dual fusion	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
IGH/CCND1, dual fusion	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
IGH/FGFR3, dual fusion	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
IGH/MAF, dual fusion	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
IGH/MYC, dual fusion	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting

KMT2A, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
MECOM, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
MLL/MLLT10, dual fusion	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure, LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems,, LP-CYT-SlideMake, LP-CYT-Reporting
MPO/TP53	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis,LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
MYC	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
MYC, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting

MYCN	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
NUP214/DEK, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
NUP98, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
NUP98/NSD1, dual fusion	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
PAX3, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
PAX7, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting

PBX1/HLF/TCF3, dual fusion	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
PDGFRB, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
PML/RARA, dual fusion	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
RUNX1/RUNX1T1, dual fusion	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
SHOX/DYZ1	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
SNRPN	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting

SRY	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
SS18, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
STS (Xp22)	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
Subtelomere probes (locus specific)	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
TCF3, break apart	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
TERT	Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting

	TLX3, break apart		Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
	TP53		Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
	XIST		Manual/microscope	Thermobrite, Grant JB1 water bath, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-FISHProcedure LP-CYT-FISHAnalysis, LI-CYT-Thermocrite, LI-LAB-GrantWBath, LP-CYT-Matasystems, LP-CYT-SlideMake, LP-CYT-Reporting
1071 Cytogenetics - .99 Miscellaneous tests	Chromosome analysis by G-band staining	Blood, Bone marrow, Human tissue	Manual/microscope	Hood, Incubator, Cell counter, Centrifuge, Olympus BX51 microscope, Zeiss microscope, Metasystems	Based on standard method	LP-CYT-Tissues, LP-CYT-HONCLabProcedure, LI-CYT-CellCounter, LI-CYT-HONCSetUpandHarvest, LP-CYT-SlideMake, LP-CYT-HONCAnalGuide, LI-CYT-ConstBloodInst, LP-CYT-ConstBlood, LP-CYT-ConstGbandAnal, LP-CYT-ConstGbandGuide,LP-CYT-Gbanding, LP-CYT-Reporting, LP-LAB-Hoods, LP-LAB-IncubatorsOven, LP-LAB-Centrifuges, LP-LAB-Metasystems
1075 Molecular genetics - .05 Screening for unidentified pathogenic variant(s)	Detection of whole genome genetic imbalance using Array Comparative Genomic Hybridisation (aCGH)	DNA	Microarray based Comparative Genomic Hybridisation (aCGH)	Ozone hoods, Oven, Agilent SureScan Microarray Scanner, Thermocycler,	Non-CE, Based on standard method	LP-CYT-CghLabelHyb, LP-CYT-CghProcess, LI-LAB-Nanodrop, LP-CYT-CGHAnal, LP-CYT-Reporting, LP-CYT-ConcPlus, LP-LAB-Hoods,

				NanoDrop, Concentrator, Vortex, Centrifuge, Pipettes, Agilent CytoGenomics Software		LP-CYT-Surecycler8800, LP-LAB-Pipettes, LP-LAB-Vortex, LP-LAB-Centrifuges, LP-LAB-IncubatorsOven, LP-CYT-CGHALLAnal
1075 Molecular genetics - .06 Assay for identified pathogenic variant(s)	Angelman/Prader Willi syndrome (AS/PWS)		MS-MLPA assay kits by MRC-Holland- Kit ME028-C1	Biorad tetrad and Bioer thermocyclers, ABI 3130xl/3500xl Genetic Analysers	MLPA Kits: Version C1 Non -CE, Version D1 CE marked ABI 3130xl/3500: Non-CE	LI-MOL-MS-MLPA, LI-MOL-MLPAGeneMarker, LP-MOL-CheckingMS-MLPAdata LP-MOL-PWS&Asreport
	Cystic Fibrosis (CF)		Multiplex PCR by X-Tag method (Luminex) Cystic Fibrosis 39 Kit v2	Luminex 200 System, Biorad tetrad and Bioer thermocyclers	Non-CE	LP-MOL-Luminex xTag CF39Kit, LP-MOL-LumxTagCF39dataCheck, LP-MOL-CFReporting, LP-MOL-CF2ndCheck
	Fragile X		Expansion PCR and sizing , Amplidex PCR CE kit (Asuragen)	Bioer thermocycler, Biorad tetrad thermocycler, MiniAmp thermocyclers, ABI 3130/3500xl Genetic Analyser	Expansion PCR and sizing: Based on standard method Amplidex PCR kit (Asuragen): CE ABI 3130xl/3500: Non-CE	LP-MOL-FRAXPCR&sizing LP-MOL-FRAXAsuragen LP-MOL-FraxReporting
	Spinal Muscular Atrophy (SMA)		PCR Testing (via MLPA). MLPA assay kits by MRC-Holland- Kit P021-B1	Biorad tetrad and Bioer thermocyclers, MiniAmpl thermocyclers, ABI 3130xl/3500xl Genetic Analyser	MLPA kit: Non -CE (as do not use Coffalyser) ABI 3130xl/3500: Non-CE	LI-MOL-MLPA, , LI-MOL-MLPAGeneMarker, LI-MOL-CheckingMLPAdata, LP-MOL-SMAReporting
1075 Molecular genetics - .10 DNA sequencing	Breast Cancer - hereditary BRCA1 & BRCA2		Sanger sequencing	Biorad tetrad, Bioer and MiniAmp thermocyclers, ABI 3500xl Genetic Analyser	Sanger sequencing: Based on standard method ABI 3500: Non-CE	LI-MOL-SangSeqGen, LI-MOL-ABI3500XL, LP-MOL-BRCAPredMgt, LI-MOL-BRCAPCR, LP-MOL-BRCAReporting, LI-MOL-BRCAAnalysis



1075 Molecular genetics - .11 DNA extraction	DNA extraction (Cytogenetics & Molecular Genetics)	Whole blood	Magnetic bead technology	Chemagic 360	Non-CE	LP-LAB-ChemDNAextract
	DNA extraction (Cytogenetics)	Blood, Bone marrow, Human tissue	Qiagen Midi kit technique	Hood, Shaking Waterbath, Vortex, Pipettes, Centrifuge, Concentrator, NanoDrop	Non-CE, Based on standard method	LP-CYT-CghDNAExtract, LP-CYT-NeoplasticDNAExtract, LP-LAB-Hoods, LP-CYT-StuartWBath, LI-LAB-Nanodrop, LP-LAB-Centrifuges, LP-LAB-Pipettes, LP-CYT-ConcPlus, LP-LAB-Vortex
	DNA extraction (Molecular Genetics)	Whole blood, Dried Blood Spot	Magcore tissue extraction (Magnetic particle technology)	Magcore Plus II Instrument	CE marked	LI-MOL-MagcoreExtract
		Whole blood, Dried Blood Spot, Human tissue	EZ1 DNA extraction (Magnetic bead technology)	EZ1 Advanced, EZ1 Advanced XL	Non-CE (EZ1 Advanced) CE marked (EZ1 Advanced XL)	LP-MOL-EZ1SolidTissue; LP-MOL-DNAExtCFDBS, LP-MOL-EZ1DNAExtract
		Whole blood, Human tissue	Phenol/ Chloroform	Fumehood, Centrifuge	Based on standard method	LP-MOL-PNDExtraction
1075 Molecular genetics - .99 Miscellaneous tests	Linked STR Markers for Uniparental Disomy (UPD), for Chromosomes 15	DNA	Linked marker PCR	Bioer thermocycler, Biorad tetrad thermocycler, MiniAmp thermocycler, ABI 3130xl/3500xl Genetic Analysers	Linked marker PCR: Based on standard method ABI 3130xl/3500: Non-CE	LP-MOL-UPD, LP-MOL-LinkedMarkerPCR, LP-MOI_UPDRepdataandcheck,