Full A-Z List of Genetic Tests

SHEFFIELD DIAGNOSTIC GENETICS SERVICE

You can search for a test by typing into the 'Find' facility in the toolbar above and pressing enter. Click the 'Find next' icon to locate multiple entries within the document.

***Gene content for Next Generation Sequencing (NGS) panels can be referenced at:**
https://www.sheffieldchildrens.nhs.uk/sdgs/next-generation-sequencing/

FISH tests listed under "F" starting with constitutional FISH tests listed by chromosome number 1-22,X,Y, followed by oncology FISH tests listed A-Z by gene name.

** Our turnaround times are listed here based on the recommendations of the Association for Clinical Genetic Science (http://www.acgs.uk.com/media/949852/acgs_general_genetic_laboratory_reporting_recommendations_2015.pdf) and are in calendar days. These may be altered locally based on specific Service Level Agreement or clinical urgency.

Test	Specimen Type	Volume	Turnaround Time**	Notes/Comments
Achondroplasia, hypchondroplasia and thanatophoric dysplasia	Blood	0.5-5ml	2-6 weeks	
Acute Lymphoblastic Leukaemia (ALL) + FISH	Bone marrow/ leukaemic blood	0.25-1ml BM in 5-10ml of transport medium **OR** 1ml BM/VB in Li Hep	14 days	
Acute Myeloid Leukaemia (AML) (+/- FISH)	Bone marrow/ leukaemic blood	0.25-1ml BM in 5-10ml of transport medium **OR** 1ml BM/VB in Li Hep	14 days	
Adrenoleukodystrophy (ALD) (X-linked)	Blood	0.5-5ml EDTA	2-6 weeks	
ALK Breakapart FISH (2p23) for lung cancer – FISH only	PETS 2x4µm sections on slides		7 days	
Alpha thalassaemia	Blood	0.5-5ml EDTA	2-4 weeks	
Alport Syndrome sequencing panel	Blood	0.5-5ml EDTA	2-6 Weeks	
Alveolar rhabdomyosarcoma – FISH only	PETS 2x4µm sections on slides		14 days	
Anaplastic large cell lymphoma, ALK positive – FISH only	PETS 2x4µm sections on slides		21 days	
Androgen Insensitivity Syndrome (Testicular Feminisation)	Blood	0.5-5ml EDTA	2-6 weeks	
Antithrombin Deficiency	Blood	0.5-5ml EDTA	2-6 weeks	
Aneuploidy Detection QF-PCR	Amniotic fluid / chorionic villus	10-20ml sterile universal / 3-4 fronds sterile universal in CVS culture medium	3 days	
Aneuploidy FISH test	Amniotic fluid sample	2-5ml in sterile universal	2-3 days	
Apolipoprotein E (APOE)) / Type III Hyperlipoproteinaemia – E2 allele	Blood	0.5-5ml EDTA	4 weeks	
Array CGH (see microarray)	-	-	-	
Ataxia Next Generation Sequencing panel (Please see Hereditary Ataxia panel)	-	-	-	
Ataxia telangiectasia/ATR-Seckel syndrome prenatal testing	Amniotic fluid / chorionic villus	10-20ml sterile universal / 3-4 fronds sterile universal in CVS culture medium	3 days	Known mutations only, in collaboration with Institute of Cancer Studies, Birmingham

Bernard-Soulier syndrome (*GP1BA, GB1BB, GP9*)	Blood	0.5-5ml EDTA	2-6 weeks	
BCR-ABL1 Quantitation (RQ-PCR)	Blood / Bone marrow	0.5-5ml EDTA	14 days	Send to Lab immediately
BCR-ABL1 Tyrosine kinase domain (TKD) analysis (Sanger sequencing)	Blood / Bone marrow	0.5-5ml EDTA	21 days	Send to Lab immediately
Beta thalassaemia	Blood	0.5-5ml EDTA	2-4 weeks	
Bladder cancer –FISH only	PETS 2x4µm sections on slides		21 days	
Blooms syndrome	Blood	3-4ml Li Hep	28 days	Inform lab prior to sample dispatch
Bone marrow failure Next Generation Sequencing panel* - Diamond-Blackfan anaemia - Dyskeratosis congenital - Fanconi anaemia - Myelodysplastic syndrome - Severe congenital neutropenia - Thrombocytopenia with absent radii	Blood	0.5-5ml EDTA	6-12 weeks	*see website for panel content. Sub-panels available
BRAF (V-raf murine sarcoma viral oncogenes homolog B1) mutations affecting codon 600 in melanoma and colorectal cancer	PETS 8x10µm sections in universal		7 days	
Hereditary Breast & Ovarian Cancer *(BRCA1, BRCA2 & PALB2)* Next Generation Sequencing* & MLPA	Blood	0.5-5ml EDTA	6 weeks full screen, 2 weeks predictive	*see website for panel content
Breast Cancer (Her2) - FISH only	PETS 2x4µm sections on slides		7 days	Contact Lab prior to referral
Hereditary Breast & Ovarian Cancer Extended Gene Panel*	Blood	0.5-5ml EDTA	12 weeks	*see website for panel content
Bruck syndrome (*PLOD2*)	Blood	0.5-5ml EDTA	2-6 weeks	
Burkitt lymphoma –FISH only	PETS 2x4µm sections on slides		21 days	
CALR Exon 9 mutation screen	Blood/Bone marrow	0.5-5ml EDTA	2 weeks	
Carnitine Acylcarnitine Translocase (CACT) Deficiency	Blood or Fibroblasts	0.5-5ml EDTA	2-6 weeks	
Carnitine Palmitoyl Transferase Type2 (CPT2) Deficency - c.338C>T p.Ser113Leu common mutation - Full sequencing	Blood or Fibroblasts	0.5-5ml EDTA	2-6 weeks	
Cartilage-associated protein (*CRTAP*) -autosomal recessive OI.	Blood	0.5-5ml EDTA	2-8 weeks	
Cerebral AD Arteriopathy with Subcortical Infarcts & Leukoencephalopathy (CADASIL)	Blood	0.5-5ml EDTA	2-6 weeks	
CGH/microarray –see microarray	-	-	-	
Chimerism post bone marrow/stem cell transplant (Sex matched/ Powerplex)	Blood/Bone marrow	0.5-5ml EDTA (see below if lineage-specific analysis required)	7 days	

Chimerism post bone marrow/stem cell transplant (Sex mis-matched (FISH))	Blood/Bone marrow	2-3ml Li Hep (see below if lineage-specific analysis required)	7 days	
Chimerism if lineage-specific analysis required (MACS separated fraction e.g. CD3+)	Blood only	0.5-1ml EDTA (IN ADDITION TO ABOVE)	7 days	
Chimerism Pre transplant DNA storage (donor or recipient)	Blood preferable but other samples can be processed e.g. buccal swabs	0.5-5ml EDTA	-	
Chromosome – Adult (with or without FISH)	Blood	2-3ml Li Hep	28 days	
Chromosome – Child (with or without FISH)	Blood	1-2ml Li Hep	28 days	
Chromosome – Neonate	Blood	0.5 – 1ml* Li Hep	10 days	* smaller samples can be attempted but may reduce the likelihood of a successful result
Chromosome (with or without FISH) PRENATAL	Amniotic Fluid sample	10-20ml sterile universal	14 days	
Chromosome (with or without FISH) PRENATAL	CVS	3-4 fronds sterile universal in CVS culture medium	14 days	
Chromosome (with or without FISH) PRENATAL	Fetal blood cordocentesis	0.5-1ml Li Hep	10 days	
Chromosome (with or without FISH) POSTNATAL	Cord blood	1-3ml Li Hep	10 days	
Chromosome (with or without FISH) FETAL LOSS	Placental biopsy at cord insertion site, fetal membrane, villi and cord biopsies	<1cm cubed Sterile tissue culture medium pots	28 days	See our "additional information" page for information regarding sending samples
Chromosome (with or without FISH)	Skin biopsy	1-2mm cubed Sterile tissue culture medium pots	2-3 weeks	
Chromosome (with or without FISH)	Solid Tumour Biopsy	<1cm cubed Universal with 5-10ml of transport medium	21 days	
Chronic Lymphoproliferative Leukaemia (CLL) –FISH	Bone marrow/ leukaemic blood	Universal with 5-10ml of transport medium OR BM/VB Li Hep	21 days	
Chronic Myeloid Leukaemia (CML) Karyotyping &/or BCR-ABL1 FISH (**at diagnosis**)	Bone marrow /leukaemic blood	0.25-1ml BM in 5-10ml of transport medium **OR** 1ml BM/VB in Li Hep	7 days	If blood ONLY then FISH result will be issued and bone marrow sample requested for karyotyping
Chronic Myeloid Leukaemia (CML) Karyotyping &/or BCR-ABL1 FISH (**monitoring**)	Bone marrow /leukaemic blood	0.25-1ml BM in 5-10ml of transport medium **OR** 1ml BM/VB in Li Hep	14 days	
Collagen 6 related myopath panel	Blood	0.5-5ml EDTA	2-6 Weeks	
Colorectal Cancer (HNPCC/FAP) Extended Gene Panel*	Blood	0.5-5ml EDTA	12 weeks	*see website for panel content
Connective Tissue Disorders Next Generation Sequencing Panel	Blood	0.5-5ml EDTA	12 weeks	*see website for panel content Sub-panels available
Congenital thrombotic thrombocytopenic purpura	Blood	0.5-5ml EDTA	2-6 weeks	

(ADAMTS13 deficiency)				
Crigler-Najjar Syndrome types I and II	Blood	0.5-5ml EDTA	2-6 weeks	
cfDNA testing for EGFR mutations (Relapse or diagnostic)	Peripheral blood	10ml PB in Streck/Paxgene preservative tubes	7 days	Preservative tubes can be supplied by the laboratory if required. Contact lab prior to sending
Cutis Laxa sequencing panel	Blood	0.5-5ml EDTA	2-6 weeks	
Cystic fibrosis – 50-mutatIon panel	Blood or Dried bloodspots	0.5-5ml EDTA	2 weeks	
Cystic fibrosis – newborn screening - 4-mutation and 50-mutation panels	Dried bloodspots	0.5-5ml EDTA	4 days	
Cystic fibrosis – newborn screening NGS pilot study - NGS panel (332 mutations)	Dried bloodspots	0.5-5ml EDTA	4 days	
Dementia Next Generation Sequencing Panel*	Blood	0.5-5ml EDTA	12 weeks	* see website for panel content
Dentatorubral-pallidoluysian atrophy (DRPLA)	Blood	0.5-5ml EDTA	4 weeks	
Dermatofibrosarcoma protuberans – FISH only	PETS 4x4µm sections on slides		21 days	
Diamond Blackfan Anaemia (*RPS19*)	Blood	0.5-5ml EDTA	2-8 weeks	
Diamond Blackfan Anaemia (dosage testing by MLPA)	Blood	0.5-5ml EDTA	8 weeks	
Dopa-responsive dystonia (Segawa syndrome), dominant	Blood	0.5-5ml EDTA	2-8 weeks	
DYPD	Blood	3-5mL EDTA	3 days	
Dystonia and Parkinsonism Next Generation Sequencing Panel*	Blood	0.5-5ml EDTA	12 weeks	*see website for panel content
Dystonia 1 (DYT1) or Idiopathic Torsion Dystonia, dominant	Blood	0.5-5ml EDTA	4 weeks	
Dystrophia myotonica (DM)	Blood	0.5-5ml EDTA	4 weeks	
EGFR (exons 18-21)	PETS 8x10µm sections in universal		7 days	EGFR testing form required please contact laboratory
Ehlers Danlos Next Generation Sequencing Panels* (Vascular, Classic and Kyphoscoliotic)	Blood	0.5-5ml EDTA	8 weeks full screen, 2 weeks predictive	*see website for panel content
Ehlers-Danlos Syndrome Classical (*COL5A1* and *COL5A2*)	Blood	0.5-5ml EDTA	2-6 weeks	
Ehlers-Danlos Syndrome - Hypermobile (*TNXB*)	Blood	0.5-5ml EDTA	2-6 weeks	
Ehlers-Danlos Syndrome - KMH (*FKBP14*)	Blood	0.5-5ml EDTA	2-6 weeks	
Ehlers-Danlos Syndrome - Kyphoscoliotic (*PLOD1*)	Blood	0.5-5ml EDTA	2-6 weeks	
Ehlers-Danlos Syndrome - Musculocontractural (*CHST14*)	Blood	0.5-5ml EDTA	2-6 weeks	
Ehlers-Danlos Syndrome - vascular (*COL3A1*)	Blood	0.5-5ml EDTA	2-6 weeks	
Ehlers-Danlos Syndrome - arthrochalasic (*COL1A1* and *COL1A2*)	Blood	0.5-5ml EDTA	2-6 weeks	
Endometrial stromal tumours – FISH	PETS 4x4µm sections on slides		14 days	

Episodic ataxia type 1	Blood	0.5-5ml EDTA	2-8 weeks	
Episodic ataxia Next Generation Sequencing panel	Blood	0.5-5ml EDTA	8 weeks	
Ewings sarcoma and rearrangements of EWSR1 associated with clear cell sarcoma, extraskeletal myxoid chondrosarcoma and desmoplastic small round cell tumour – FISH only	PETS 2x4µm sections on slides		21 days	
Factor V deficiency (F5)	Blood	0.5-5ml EDTA	2-6 weeks	
Factor XIII Deficiency	Blood	0.5-5ml EDTA	2-6 weeks	
Factor XI Deficiency (Haemophilia C)	Blood	0.5-5ml EDTA	2-6 weeks	
Familial Adenomatous Polyposis Coli (FAP) & MUTYH Gene Panel (*APC & MUTYH* sequencing and MLPA)*	Blood	0.5-5ml EDTA	6 weeks full screen, 2 weeks predictive	*see website for panel content
Familial Amyotrophic Lateral Sclerosis with or without Frontotemporal Dementia 42 gene Next Generation Sequencing Panel*	Blood	0.5-5ml EDTA	12 weeks	*see website for panel content
Familial hemiplegic migraine Next Generation Sequencing panel	Blood	0.5-5ml EDTA	8 weeks	*see website for panel content
Familial hypercholesterolaemia (APOE, LDLR, APOB, PCSK9 and LDLRAP1 sequencing and LDLR MLPA	Blood	0.5-5ml EDTA	2-6 weeks	
Familial motor neurone disease / amyotrophic lateral sclerosis with or without frontotemporal dementia (ALS/FTD) *C9orf72* gene	Blood	0.5-5ml EDTA	4 weeks	
Familial motor neurone disease / amyotrophic lateral sclerosis (ALS) *SOD1* gene	Blood	0.5-5ml EDTA	2-8 weeks	
Familial Porencephaly (*COL4A1 & COL4A2*) by Next Generation Sequencing	Blood	0.5-5ml EDTA	6 weeks full screen, 2 weeks predictive	*see website for all CTD NGS panels & content
Familial Thoracic Aortic Aneurysms Next Generation Sequencing Panel	Blood	0.5-5ml EDTA	12 weeks	*see website for all CTD NGS panels & content
Fanconi Anaemia (Chromosome Breakage Studies)	Blood	3-4ml Li Hep	28 days	Inform lab prior to sample dispatch
Fanconi Anaemia by Next Generation Sequencing (16 genes)	Blood	0.5-5ml EDTA	12 weeks	*see website for panel content
Fatty acid metabolism disorders Next Generation Sequencing Panel	Blood	0.5-5ml EDTA	12 weeks	* see website for panel content. Sub-panels available
Fibrinogen disorders (FGA, FGB, FGG)	Blood	0.5-5ml EDTA	2-6 weeks	

FISH **CONSTITUTIONAL FISH TESTS** *In Chromosome Order 1-22,X,Y*	**Gene**	**Comments**
1p36.33 microdeletion syndrome (inc. hypertrichotic osteochondrodysplasia)	CEB108/T7 and D1Z2	Terminal and interstitial deletions detected
2q37.3 Brachydactyly-mental retardation microdeletion syndrome (inc. Albright hereditary osteodystrophy (AHO)-like metacarpal/metatarsal shortening)	D2S447	
4p16.3 Wolf-Hirschhorn microdeletion syndrome	WHSC1	

5p15.3 Isolated Cat Cry microdeletion syndrome (ICS) and 5p15.2 Cri Du Chat microdeletion syndrome (CDC)	FLJ25076 (ICS) and CTNND2 (CDC) respectively	
5q35 Sotos microdeletion syndrome	NSD1	
7q11.23 Williams microdeletion syndrome	ELN	
7q11.23 microduplication syndrome (inc. speech delay, ADHD)	ELN	
8q12.1-12.2 CHARGE microdeletion syndrome (inc. ocular coloboma, heart defects of any type, atresia of the choaneae, retardation, genital and ear anaomalies)	CHD7	
8q23.3-8q24.1 Langer-Giedion microdeletion syndrome (inc. trichorhinophalangeal syndrome type 1 and multiple cartilaginous exostoses)	TRPS1 and EXT1 respectively	
9q34.3 Kleefstra microdeletion syndrome (inc. craniofacial features, hypotonia, obesity, microcephaly and speech delay)	D9S325	
15q11.2 Prader-Willi microdeletion/ microduplication syndrome	SNRPN	For 1st line test see molecular genetic referral
15q11.2 Angelman microdeletion syndrome	D15S10/UBE3A	For 1st line test see molecular genetic referral
16p13.3 Rubenstein-Taybi microdeletion syndrome (inc. short stature, talon cusps, patellar dislocation, broad thumbs and big toes)	CREBBP	
17p13.3 Miller-Dieker microdeletion syndrome	LIS1 (PAFAH1B1)	
17p11.2 Smith-Magenis microdeletion syndrome	RAI1	
17p11.2 Potocki-Lupski microduplication syndrome (inc. neonatal hypotonia, sleep apnea, hyperactivity, structural cardiovascular abnormalities)	RAI1	
17q11.2 NF1 (Von Recklinghausen) microdeletion syndrome	NF1 (RP1-4C23)	Home grown
17q21.31 microdeletion syndrome (inc. neonatal hypotonia, developmental delay and speech delay)	MAPT	
22q11.2 DiGeorge/VCFS microdeletion or microduplication syndrome	TBX1	
22q13.3 Phelan-McDermid microdeletion syndrome (inc. neonatal hypotonia, absent or delayed speech)	N85A3	N85A3 is the control sequence for TBX1
Xp22.3 or Yp11.32 Leri-Weill Dyschondrosteosis inc. short stature and madelung deformity (heterozygous microdeletion syndrome) or Langer Mesomelic Dysplasia inc. severe short stature and skeletal abnormalities (homozygous microdeletion syndrome)	SHOX	Located in the PAR1 pseudoautosomal regions of both X and Y chromosomes
Xp22.3 Kallmann microdeletion syndrome (inc. hypogonadotrophic hypogonadism and anosmia)	ANOS1	
Xp22.3 Steroid Sulphatase Deficiency microdeletion syndrome inc. X-Linked Ichthyosis	STS	
Xq13.2 X inactivation centre deletion	XIST	Critical for the determination of phenotypic severity of abnormal X chromosomes
Yp11 Swyer microdeletion syndrome (XY female) and detection of unbalanced t(X;Y) leading to XX with male phenotype	SRY	Sex determining region
XCEN/YCEN/18CEN/13q14/21q22.13-q22.2 Sex chromosome aneuploidy, Edward, Patau and Down syndrome	DXZ1/DYZ3/D18Z1/RB1/D21S342, D21S341,D21S259	Common aneuploidy detection
FISH **ONCOLOGY FISH TESTS** ***A-Z by gene name***	**Location or Rearrangement**	**Comments**
ABL1 breakapart	9q34.12	All variants
ABL2 breakapart	1q25.2	All variants
AFF1 (MLLT2) / KMT2A dual fusion	4q21-22/11q23	

ALK breakapart	2p23	All variants
ALK/EML4 dual fusion	inv(2)(p21p23)	
BCL2 breakapart	18q21	All variants
BCL6 breakapart	3q26.2	All variants
BCR/ABL1/ASS1 dual fusion	t(9;22)(q34;q11)	Tricolour –Complex deletion rearrangement pattern monitoring possible
BLADDER PANEL single locus D3Z1/D7Z1/CGKN2A/D17Z1	3CEN/7CEN/9p21/17CEN	3, 7 and 17 aneuploidy detection and 9 short arm deletion detection
CBFB breakapart	16q22	All variants
CBFB/MYH11 dual fusion	inv(16)(p13q22) and t(16;16)(p13;q22)	
CCND1 breakapart	11q13	All variants
CCND1/D11Z1 single locus	11q13/11CEN	CCND1 amplification detection
CDKN2C(p18)/CKS1B amplification in MM single locus	1p32.3/1q21	CDKN2C deletion / CKS1B
CERVICAL PANEL single locus hTERC/MYC/D7Z1	3q26.2/8q24/7CEN	Detection of hTERC and/or MYC gain
CDKN2A/CEN9 single locus	9p21/9CEN	Short arm deletion detection
CHD5/1qter single locus	1p36	Short arm deletion detection
CLL PANEL single locus MIX 1 - ATM/TP53 MIX 2 – D12Z3/D13S319/13q34	 11q22.3/17p13.1 12CEN/13q14.3/13q34	 Deletion detection Aneusomy 12 and aneusomy 13 or heter/homozygous long arm deletion detection
CRLF2	XP22.3 and Yp11.3	
D1Z2 single locus	1p36	Short arm deletion detection
D8Z2 single locus	8CEN	Aneusomy 8 detection
D12Z3 single locus	12CEN	Aneusomy 12 detection
D13S25 single locus	13q14	Monosomy or long arm deletion detection
D13S319 single locus	13q14	Monosomy or long arm deletion detection
D20S108 single locus	20q12	Monosomy or long arm deletion detection
DDIT3 breakapart	12q13	All variants Formerly CHOP
DEK/NUP214 dual fusion	t(6;9)(p22;q34)	
Dermatofibrosarcoma protruberans panel chromosomes COL1A1 and PDGFB breakapart	17q22 and 22q13	Detection of t(17;22)(q22;q13) and amplification in supernumerary ring
DXZ1/DYZ3 single locus	XCEN/YCEN	Sex mismatched monitoring
EGR1/D5S23,D5S721	5q31/5p15.2	Monosomy or long arm deletion detection
EML4 breakapart	2p21	All variants
ETV6 breakapart	12p13	All variants
ETV6/RUNX1 dual fusion	t(12;21)(p13;q22)	Formerly TEL/AML1
EWSR1 breakapart	22q12	All variants
EWSR1/FLI1 dual fusion	t(11;22)(q24;q12)	
FGFR1/D8Z2 breakapart + single locus	8p11/8CEN	All variants and FGFR1 amplification
FOXO1 breakapart	13q14	All variants Formerly FKHR See also PAX3
FUS breakapart	16p11	All variants
GLIOMA PANEL single locus Mix 1 – EGFL3,TP73/ANGPTL1,ABL2 Mix 2 - ZNF44,ZK1,MAN2B1/GLTSCR1+2,CRX	1p36/1q25 19p13/19q13	Loss of 1p relative to 1q and loss of 19q relative to 19p
HER2/D17Z1 single locus	17q11.2-q12	HER2 amplification detection

HER2/TOP2A/D17Z1 single locus	17q11.2-q12/ 17q21-22/17CEN	HER2 amplification detection and TOP2A deletion
IGH breakapart	14q32	All variants
IGH/BCL2 dual fusion	t(14;18)(q32;q21)	
IGH/CCND1,MYEOV dual fusion	t(11;14)(q13;q32)	
IGH/FGFR3 dual fusion	t(4;14)(p16;q32)	
IGH/MAF dual fusion	t(14;16)(q32;q23)	
IGH/MAFB dual fusion	t(14;20)(q32.33;q11.1-q13.1)	MM
IGH/MYC/D8Z2 dual fusion	t(8;14)(q24;q32)	Aneusomy 8 also detected
IGK breakapart	2p12	All variants
IGL breakapart	22q11	All variants
IRF4/DUSP22 breakapart	6p25.3	All variants
JAZF1 breakapart	7p15.1-2	All variants
KMT2A breakapart	11q23	All variants
MALT breakapart	18q21	All variants
MAML2 breakapart	11q21	All variants
MDM2/D12Z1 single locus	12q14.3-q15/12CEN	MDM2 amplification detection
MECOM	3q26.2	Tricolour All variants
MELANOMA PANEL single locus D6Z1/RREB1/MYB/CCND1	6CEN/6p25/6q23/11q13	
MYB single locus	6q23	Long arm deletion detection
MYC breakapart	8q24	All variants
MYC/D8Z2 single locus	8q24/8CEN	Detection of gain of 8q24 relative to 8CEN
MYCN/D2Z1 single locus	2p24/2CEN	N-MYC amplification detection
NUP98 breakapart	11p15	AML, ALL, CML-bc
PAX3 breakapart	2q35	All variants See also FOXO1
PBX1/HLF/TCF3 dual fusion	t(1;19)(q23;p13.3)/t(17;19)(q22;p13.3)	ALL
PDGFRA /LNX/ SCFD2 breakapart	4q12 FIP1L1/CHIC2/PDGFRA rearrangement	Tricolour All variants
PDGFRB breakapart	5q32	All variants
PIK3CA single locus	3q26.32	PIK3CA amplification detection
PML/RARA dual fusion	t(15;17)(q22;q21)	
PROSTATE PANEL Mix 1 – TMPRSS2/ERG breakapart Mix 2 - PTEN/CEP10 single locus	21q22 10q23/10CEN	Mix 1 – Tricolour, all variants Mix 2 - Long arm deletion detection
PTEN/CEP10 single locus	10q23/10CEN	Long arm deletion detection
PZRY8	Xp22.33 and Yp11.32	
RARA breakapart	17q21	All variants
RB1	13q14	Monosomy or long arm deletion detection
RELN/TES/7CEN	7q22.1/7q31.2/7CEN	Monosomy or long arm deletion detection
ROS1 breakapart	6q22	All variants
RUNX1/RUNX1T1 dual fusion	t(8;21)(q22;q22)	Formerly AML1/ETO
SEC63/D6Z1	6q21/6CEN	Long arm deletion detection
SS18 breakapart	18q11.2	All variants Formerly SYT
TCF3 breakapart	19p13.3	All variants Formerly EA2
TCR breakapart	14q11.2	All variants
TCRb breakapart	7q34	All variants.
TP53/D17Z1 single locus	17p13.1/17CEN	Short arm deletion detection
TP53/MPO single locus	17p13.1/17q22	i(17q) detection
TRB breakapart	7q34	All variants
Uveal Melanoma single locus D3Z1/D8Z2	3 centromere and 8 centromere	Aneuploidy detection
YWHAE breakapart	17p13.3	All variants
ZNF217	20q13.2	ZNF217 amplification

				detection
Test	**Specimen Type**	**Volume**	**Turnaround Time**	**Notes/Comments**
FLT3 ITD, FLT3 TKDand NPM1 ex 12 testing for patients with AML	Blood / bone marrow	0.5-5ml EDTA or Li Heparin	3 - 7 days	
Follicular lymphoma/DLBCL – FISH only	PETS 2x4µm sections on slides		21 days	
Fragile X syndrome	Blood	0.5-5ml EDTA	2-8 weeks	
Friedreich Ataxia	Blood	0.5-5ml EDTA	4 weeks	
Fumarate Hydratase Deficiency (*FH* sequencing and MLPA)	Blood or Fibroblasts	0.5-5ml EDTA	2-6 weeks	
Gilbert syndrome	Blood	0.5-5ml EDTA	2-6 weeks	
Glanzmann thrombasthenia	Blood	0.5-5ml EDTA	2-6 weeks	
Glioma –FISH only	PETS 4x4µm sections on slides		7 days	
Glucose Transporter 1 (GLUT1) deficiency syndrome (SLC2A1 sequencing and MLPA)	Blood	0.5-5ml EDTA	2-6 weeks	
Glutaric Acidaemia Type 1(GA1)	Blood or Fibroblasts	0.5-5ml EDTA	2-6 weeks	
Glycogen Storage Disease Next Generation Sequencing Panels: **Liver, Muscle, Heart, Generalised Panel**	Blood	0.5-5ml EDTA	12 weeks	* see website for panel content Further sub-panels available
Glycogen Storage Disease Type 0 (GYS2 – liver, GYS1 - muscle)	Blood	0.5-5ml EDTA	2-6 weeks	
Glycogen Storage Disease Type II (Pompe disease) (GAA)	Blood	0.5-5ml EDTA	2-6 weeks	
Glycogen Storage Disease Type III (AGL)	Blood	0.5-5ml EDTA	2-6 weeks	
Glycogen Storage Disease Type IV (Andersen disease) (GBE1)	Blood	0.5-5ml EDTA	2-6 weeks	
Glycogen Storage Disease Type V (McArdle disease) (PYGM)	Blood	0.5-5ml EDTA	2-6 weeks	
Glycogen Storage Disease Type VII (Tarui disease)(PFKM)	Blood	0.5-5ml EDTA	2-6 weeks	
Haemochromatosis	Blood	0.5-5ml EDTA	4 weeks	
Haemophilia A/Factor VIII deficiency-Next Generation Sequencing	Blood	0.5-5ml EDTA	8 Weeks	
Haemophilia B/Factor IX deficiency	Blood	0.5-5ml EDTA	2-6 weeks	
Hemiplegic Migraine Next Generation Sequencing Panel*	Blood	0.5-5ml EDTA	8 weeks	* see website for panel content
Her2 –FISH only	Paraffin Embedded Tissue Biopsy		7 days	Contact Lab prior to referral
Hereditary Ataxia Next Generation Sequencing Panel*	Blood	0.5-5ml EDTA	12 weeks	* see website for panel content
Hereditary Leiomyomatosis with renal cell carcinoma (HLRCC/MCUL) (*FH* sequencing and MLPA)	Blood	0.5-5ml EDTA	2-6 weeks	
Hereditary Non Polyposis Colorectal Cancer (HNPCC) Tumour Microsatellite Instability Analysis (MSI)	Blood and PETS 8x10µm sections in universal	0.5-5ml EDTA	8 weeks	
Hereditary Spastic Paraparesis (HSP) Next Generation Sequencing Panel*	Blood	0.5-5ml EDTA	12 weeks	* see website for panel content
Huntington disease	Blood	0.5-5ml EDTA	4 weeks	
Hyperammonaemia Next Generation	Blood	0.5-5ml EDTA	12 weeks	* see website for

Sequencing Panel*				panel content Sub-panels available
Hypophosphatasia (ALPL)	Blood	0.5-5ml EDTA	2-6 weeks	
Isovaleric acidaemia (IVA) - full sequencing	Blood	0.5-5ml EDTA	2-6 weeks	
Isovaleric acidaemia (IVA) newborn screening – c.941C>T common 'benign' mutation	Blood	0.5-5ml EDTA	3 days	
JAK2 (V617F mutation)	Blood / Bone Marrow	0.5-5ml EDTA	14 days	
JAK2 Exon 12 mutation screen (polycythaemia rubra vera/PRV)	Blood / Bone Marrow	0.5-5ml EDTA	14 days	
Karyotype	See Chromosome			
KRAS (v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog) for CRC and NSCLC and other	PETS 8x10µm sections in universal		7 days	
Leucine Proline-Enriched Proteoglycan (*LEPRE1*) – autosomal recessive OI.	Blood	0.5-5ml EDTA	2-8 weeks	
Li Fraumeni (TP53 gene sequencing and MLPA)	Blood	0.5-5ml EDTA	2-8 weeks	
Long Chain 3-Hydroxyacyl CoA-dehydrogenase (LCHAD) Deficiency – c.1528G>C common mutation	Blood or Guthrie spots	0.5 -5ml EDTA	2 weeks	
Lynch syndrome / Hereditary Non Polyposis Colorectal Cancer (HNPCC) Gene panel (including MLPA)*	Blood	0.5-5ml EDTA	6 weeks full screen, 2 weeks predictive	* see website for panel content
Malt Lymphoma – FISH only	PETS 2x4µm sections on slides		21 days	
Mantle cell lymphoma - FISH only	PETS 2x4µm sections on slides		21 days	
Medium Chain Acyl CoA-dehydrogenase (MCAD)Deficiency - c.985A>G common mutation - Full sequencing	Blood or Guthrie spots	0.5-5ml EDTA	2-6 weeks	
Medium Chain Acyl CoA-dehydrogenase (MCAD) Deficiency newborn screening - c.985A>G common mutation - Full sequencing	Blood or Guthrie spots	0.5-5ml EDTA	3 days - 2 weeks	
Medullary cystic and glomerulocystic kidney disease (autosomal dominant tubulointerstitial kidney disease) Next Generation Sequencing panel	Blood	1-5ml EDTA	6 weeks full screen, 2 weeks predictive	*see website for panel content
Microarray	Blood	2-3ml EDTA	28 days	
Microarray (skin)	Skin biopsy	1-2mm cubed Sterile tissue culture medium pots	28 days	
Microarray (fetal loss)	Placental biopsy at cord insertion site, fetal membrane, villi, cord biopsy, skin biopsy	1cm cubed Sterile tissue culture medium pots	28 days	
Microarray (prenatal)	Chorionic villus samples and Amniotic Fluid samples	3-4 fronds sterile universal in CVS culture medium	14 days	

MPL Exon 10 mutation screen	Blood / bone marrow	0.5-5ml EDTA	14 days	
Mucoepidermoid carcinoma – FISH only	PETS 2-4x4µm sections on slides		14 days	
Multiple Endocrine Neoplasia Type 1 (*MEN1* gene sequencing & MLPA)	Blood	0.5-5ml EDTA	2-6 weeks	
Multiple Endocrine Neoplasia Type 2 (MEN2) and Hirschsprung disease (*RET* gene)	Blood	0.5-5ml EDTA	2-6 weeks	
Myelodysplastic syndromes (MDS)	Bone Marrow	0.25-1ml BM in 5-10ml of transport medium **OR** 1ml BM/VB in Li Hep	21 days	
Myeloma – FISH only	Bone marrow	0.5-5ml EDTA	21 days	
Myeloproliferative disease (MPD)	Bone marrow	0.25-1ml BM in 5-10ml of transport medium **OR** 1ml BM/VB in Li Hep	21 days	
Myeloproliferative disorder/essential thrombocythaemia(ET)/polycythaemia rubra vera (PRV)/ myelofibrosis (MF) – see JAK2	-	-	-	
MYH9-related disorders	Blood	0.5-5ml EDTA	2-6 weeks	
Neonatal alloimmune thrombocytopenia (NAIT)	Blood	0.5-3ml EDTA	2-6 weeks	
Neuroblastoma –FISH only	PETS 2x4µm sections on slides		14 days	
Osteogenesis Imperfecta – autosomal dominant Next Generation Sequencing Panel*	Blood	0.5-5ml EDTA	6 weeks full screen, 2 weeks predictive	*see website for all CTD NGS panels & content
Osteogenesis Imperfecta-autosomal recessive Next Generation Sequencing Panel*	Blood	0.5-5ml EDTA	6 weeks full screen, 2 weeks predictive	*see website for all CTD NGS panels & content
Osteogenesis Imperfecta Type V (IFITM5)	Blood	0.5-5ml EDTA	2-6 weeks	
Osteoporosis and Osteoporosis – pseudoglioma syndrome (LRP5)	Blood	0.5-5ml EDTA	2-6 weeks	
Parkinsonism Next Generation Sequencing panel (please see Dystonia and Parkinsonism Next Generation Sequencing panel)	-	-	-	
Peptidylprolyl Isomerase B (cyclophilin B) PPIB	Blood	0.5-5ml EDTA	2-8 weeks	
Peroxisomal Biogenesis Disorders – Zellweger Syndrome Spectrum Next Generation Sequencing Panel	Blood	0.5-5ml EDTA	12 weeks	*see website for panel content Sub-panels available
Pharmacogenetics NGS panel for non-small cell lung cancer, colorectal cancer, GIST, glioma & melanoma	PETS 8x10µm sections in universal		7 days	*see website for panel content
Platelet-type pseudo von Willebrand Disease (*GP1BA* gene)	Blood	0.5-5ml EDTA	2-6 weeks	
Polycystic Kidney Disease (autosomal dominant) *PKD1 & PKD2* Full-gene sequencing and MLPA	Blood	1-5ml EDTA	2-6 weeks	

Polycystic kidney disease (autosomal recessive) PKHD1 gene	Blood	1-5ml EDTA	2-6 weeks	
Polycystic kidney disease modifier Next Generation Sequencing panel*	Blood	1-5ml EDTA	6 weeks full screen, 2 weeks predictive	*see website for panel content
Polycystic liver disease Next Generation Sequencing panel*	Blood	1-5ml EDTA	6 weeks full screen, 2 weeks predictive	*see website for panel content
Protein C Deficiency	Blood	0.5-5ml EDTA	2-6 weeks	
Protein S Deficiency	Blood	0.5-5ml EDTA	2-6 weeks	
Prothrombin (3' non 20210G>A prothrombin variants)	Blood	0.5-5ml EDTA	2-6 weeks	
Pseudoxanthoma Elasticum	Blood	0.5-5ml EDTA	2-6 weeks	
Quantitative BCR-ABL (MRD)	Blood/Bone marrow	0.5-5ml EDTA	2 weeks	Send to Lab immediately
Rhabdomyolysis/metabolic myopathies Next Generation Sequencing Panel*	Blood	0.5-5ml EDTA	12 weeks	*see website for panel content
Renal cell cancer Next Generation Sequencing panel*	Blood	1-5ml EDTA		*see website for panel content
Retinoblastoma	Blood	2-3ml Li Hep	28 days	
Sickle Cell Disease	Blood	0.5-5ml EDTA	2 weeks	
Sickle Cell Disease (newborn screening – transfused babies)	Dried Bloodspot	-	2 weeks	Referrals via Newborn Screening only
SNP Array – see Microarray				
Spinal Muscular Atrophy, 5q-linked	Blood	0.5-5ml EDTA	2-8 weeks	
Spinal Muscular Atrophy Next Generation Sequencing Panel*	Blood	0.5-5ml EDTA	12 weeks	* see website for panel content
Spinal and Bulbar Muscular Atrophy (SBMA) (Kennedy disease), X-linked	Blood	0.5-5ml EDTA	4 weeks	
Spinocerebellar Ataxia, types 1-3, 6, 7, 12 and 17	Blood	0.5-5ml EDTA	4 weeks	
Stickler syndrome sequencing panel	Blood	0.5-5ml EDTA	2-6 weeks	
SYNGAP1-associated intellectual disability	Blood	0.5-5ml EDTA	2-8 weeks	
Synovial sarcoma –FISH only	PETS 2x4µm sections on slides		21 days	
TPMT genotyping	Blood	3-5mL EDTA	7 days	
Trimethylaminuria/Fish Odour Syndrome (FMO3)	Blood	0.5-5ml EDTA	2-6 weeks	
MRD analysis for paediatric ALL by Ig/TCR gene rearrangements including UKALL2011 clinical trial	Bone marrow	2.5-10ml ACD. OR EDTA.	Dependant on time point	Peripheral blood in ACD is acceptable at diagnosis if WCC is > 20x10^9/l.
Urea Cycle Disorders (OTC, CPS1, NAGS, ASL, ASS1, ARG1)	Blood	0.5-5ml EDTA	2-6 weeks	
Very-long-chain-acyl-CoA dehydrogenase (VLCAD) Deficiency	Blood or fibroblasts	0.5-5ml EDTA	2-6 weeks	
von Willebrand Disease type 1-3r	Blood	0.5-5ml EDTA	2-6 weeks	
Warfarin resistance (*VKORC1* gene) and combined vitamin K clotting factor deficiency type 2	Blood	0.5-5ml EDTA	2-6 weeks	

Wilms tumour, aniridia, genitourinary anomalies and mental retardation (WAGR syndrome) by MLPA of WT1 gene	Blood	1-5ml EDTA	2-6 weeks	
Wilms Tumour, Frasier syndrome, Denys Drash syndrome, Steroid-resistant nephrotic syndrome, Meacham syndrome (Wilms Tumour Suppressor) WT1 gene sequencing and MLPA	Blood	0.5-5ml EDTA	2-6 weeks	
Wilson disease (ATP7B) - Full sequencing - MLPA and promoter sequencing	Blood	0.5-5ml EDTA	2-6 weeks	