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:**
http://www.sheffieldchildrens.nhs.uk/our-services/sheffield-diagnostic-genetics-service/next-generation-sequencing.htm

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.

Test	Specimen Type	Volume	Turnaround Time	Notes/Comments
Acute Lymphoblastic Leukaemia (ALL)/BCR-ABL	Blood/Bone marrow	0.5-5ml EDTA	2-4 weeks	Send to Lab immediately
Acute Lymphoblastic Leukaemia (ALL)/BCR ABL	Bone marrow/ leukaemic blood	0.25-1ml BM in 5-10ml of transport medium **OR** 1ml BM/VB in Li Hep	10 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	10 days	
Acute Myeloid Leukaemia /AML/ AML-M2/AML-17	Blood/Bone marrow	0.5-5ml EDTA	2-4 weeks	Send to Lab immediately
Acute Promyelocytic Leukaemia (APL)/AML M3/AML-17	Blood/Bone marrow	0.5-5ml EDTA	2-4 weeks	Send to Lab immediately
ADAMTS13 deficiency (thrombotic thrombocytopenic purpura)	Blood	0.5-5ml EDTA	8 weeks	*see website for panel content
Adrenoleukodystrophy (ALD) (X-linked)	Blood	0.5-5ml EDTA	2-8 weeks	
ALK Breakapart FISH (2p23)	PET Biopsy		1-2 weeks	Contact lab prior to referral
Alpha thalassaemia	Blood	0.5-5ml EDTA	2-8 weeks	
Alveolar rhabdomyosarcoma	PETS 2x4µm sections on slides		14 days	
Amylotrophic Lateral Sclerosis and Dementia Next Generation Sequencing Panel*	Blood	0.5-5ml EDTA	16 weeks	*see website for panel content
Anaplastic large cell lymphoma, ALK positive	PETS 2x4µm sections on slides		14 days	
Androgen Insensitivity Syndrome (Testicular Feminisation)	Blood	0.5-5ml EDTA	2-8 weeks	
Antithrombin Deficiency	Blood	0.5-5ml EDTA	2-8 weeks	
Aneuploidy FISH test	Amniotic fluid sample	2-5ml in sterile universal	2-3 days	
Apolipoprotein E (APOE)	Blood	0.5-5ml EDTA	2-8 weeks	
Array CGH (see CGH / microarray)	-	-	-	
Ataxia Next Generation Sequencing panel (Please see Hereditary Ataxia and Migraine panel)	-	-	-	
Bernard-Soulier syndrome (*GP1BA, GB1BB, GP9*)	Blood	0.5-5ml EDTA	2-8 weeks	
Beta thalassaemia	Blood	0.5-5ml EDTA	2-8 weeks	
Bladder cancer	PETS 2x4µm sections on slides		14 days	

Blooms syndrome	Blood	3-4ml Li Hep	28 days	Inform lab prior to sample dispatch
BRAF (V-raf murine sarcoma viral oncogenes homolog B1) p.Val600Glu mutation	PETS 8x10µm sections in universal		1-2 weeks	
Hereditary Breast & Ovarian Cancer *(BRCA1 & BRCA2)*	Blood	0.5-5ml EDTA	8 weeks full screen, 2 weeks predictive	Performed by Next Generation Sequencing* & MLPA
Breast Cancer (Her2) FISH	PETS 2x4µm sections on slides		1-2 weeks	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	2-8 weeks	
Burkitt lymphoma	PETS 2x4µm sections on slides		14 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	2-8 weeks	
Carnitine Palmitoyl Transferase Type2 (CPT2) Deficency	Blood or Fibroblasts	0.5-5ml EDTA	2-8 weeks	
Cartilage-associated protein (*CRTAP*) -autosomal recessive OI.	Blood	0.5-5ml EDTA	2-8 weeks	
CBCL/CTCL / Skin lymphoma/ mycosis fungoides (IgH or T cell gene rearrangements)	Paraffin embedded tissue biopsy	5 □m unmounted sections	2-8 weeks	
Cerebral AD Arteriopathy with Subcortical Infarcts & Leukoencephalopathy (CADASIL)	Blood	0.5-5ml EDTA	2-8 weeks	
CGH/microarray	Blood	2-3ml EDTA AND 2-3mls Li Hep	4 weeks	Please note blood in EDTA and Li Hep are required
Chimerism/pre or post bone marrow/stem cell transplant (BMT/SCT)/donor for BMT/SCT/matched unrelated donor (MUD) – Sex matched (Powerplex)	Blood/Bone marrow	0.5-5ml EDTA	2 weeks	
Chimerism/pre or post bone marrow/stem cell transplant (BMT/SCT)/donor for BMT/SCT/matched unrelated donor (MUD) – Sex mis-matched (FISH)	Blood/Bone marrow	2-3ml Li Hep	2 weeks	
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	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	2-3 weeks	
Chronic Lymphoproliferative Leukaemia (CLL) –FISH	Bone marrow	Universal with 5-10ml of transport medium /Li Hep	28 days	
Chronic Myeloid Leukaemia (CML) Karyotyping & BCR-ABL1 FISH	Bone marrow /leukaemic blood	0.25-1ml BM in 5-10ml of transport medium **OR** 1ml BM/VB in Li Hep	28 days	Urgent samples have a TAT of 10 days
Chronic Myeloid Leukaemia (CML) BCR-ABL1 Quantitation	Blood / Bone marrow	0.5-5ml EDTA	2 weeks	Send to Lab immediately
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 (see website for list of sub-panels and gene content)	Blood	0.5-5ml EDTA	12 weeks	*see website for panel content
Congenital Bilateral Absence of Vas Deferens (CBAVD)	Blood	0.5-5ml EDTA	2-8 weeks	
Congenital thrombotic thrombocytopenic purpura (ADAMTS13 deficiency)	Blood	0.5-5ml EDTA	2-8 weeks	
Crigler-Najjar Syndrome types I and II	Blood	0.5-5ml EDTA	2-8 weeks	
CYP2C19	Blood	1-3ml EDTA	8 weeks	
CYP3A4	Blood	1-3ml EDTA	8 weeks	
Cystic fibrosis	Blood or Dried bloodspots	0.5-5ml EDTA	2-8 weeks	
Dentatorubral-pallidoluysian atrophy (DRPLA)	Blood	0.5-5ml EDTA	2-6 weeks	
Dermatofibrosarcoma protuberans	PETS 4x4µm sections on slides		14 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	
Dystonia and Parkinsonism Next Generation Sequencing Panel	Blood	0.5-5ml EDTA	16 weeks	*see website for panel content
Dystonia 1 (DYT1) or Idiopathic Torsion Dystonia, dominant	Blood	0.5-5ml EDTA	2-6 weeks	
Dystrophia myotonica (DM)	Blood	0.5-5ml EDTA	2 weeks	
EGFR (exons 18-21)	PETS 8x10µm sections in universal		5 days	EGFR testing form required please contact laboratory
Ehlers Danlos Next Generation Sequencing Panels (Vascular, Classic and Kyphoscoliotic)	Blood	0.5-5ml EDTA	12 weeks	*see website for panel content
Ehlers-Danlos Syndrome Classical (*COL5A1* and *COL5A2*)	Blood	0.5-5ml EDTA	2-8 weeks	
Ehlers-Danlos Syndrome Classical (*COL5A1*) - Null allele	Skin biopsy/cultured fibroblasts		2-8 weeks	
Ehlers-Danlos Syndrome - Hypermobile (*TNXB*)	Blood	0.5-5ml EDTA	2-8 weeks	
Ehlers-Danlos Syndrome - KMH (*FKBP14*)	Blood	0.5-5ml EDTA	2-8 weeks	
Ehlers-Danlos Syndrome - Kyphoscoliotic (*PLOD1*)	Blood	0.5-5ml EDTA	2-8 weeks	

Ehlers-Danlos Syndrome - Musculocontractural (*CHST14*)	Blood	0.5-5ml EDTA	2-8 weeks	
Ehlers-Danlos Syndrome - vascular (*COL3A1*)	Blood	0.5-5ml EDTA	2-8 weeks	
Ehlers-Danlos Syndrome - arthrochalasic (*COL1A1* and *COL1A2*)	Blood	0.5-5ml EDTA	2-8 weeks	
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	PETS 2x4µm sections on slides		14 days	
Factor V deficiency (F5)	Blood	0.5-5ml EDTA	8 weeks	*see website for panel content
Factor XIII Deficiency	Blood	0.5-5ml EDTA	2-8 weeks	
Factor XI Deficiency (Haemophilia C)	Blood	0.5-5ml EDTA	2-8 weeks	
Familial Adenomatous Polyposis Coli (FAP) & MUTYH Gene Panel (*APC & MUTYH* sequencing and MLPA)*	Blood	0.5-5ml EDTA	2-8 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 (LDLR sequencing and MLPA; ApoB p.(Arg3527Gln) mutation analysis; PCSK9 p.(Asp374Tyr) mutation analysis).	Blood	0.5-5ml EDTA	2-8 weeks	
Familial motor neurone disease / amyotrophic lateral sclerosis with or without frontotemporal dementia (ALS/FTD) *C9orf72* gene	Blood	0.5-5ml EDTA	2 weeks	
Familial motor neurone disease / amyotrophic lateral sclerosis (ALS) *SOD1* gene	Blood	0.5-5ml EDTA	2-8 weeks	
Familial motor neurone disease / amyotrophic lateral sclerosis (ALS) *TARDBP* gene	Blood	0.5-5ml EDTA	2-8 weeks	
Familial Porencephaly (*COL4A1 & COL4A2*) by Next Generation Sequencing	Blood	0.5-5ml EDTA	12 weeks	*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
Fanconi Anaemia by Next Generation Sequencing (individual genes)	Blood	0.5-5ml EDTA	8 weeks	*see website for panel content
Fibrinogen disorders (FGA, FGB, FGG)	Blood	0.5-5ml EDTA	8 weeks	*see website for panel content
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	FLJ25076 (ICS)			

syndrome (ICS) and 5p15.2 Cri Du Chat microdeletion syndrome (CDC)	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)		KAL1	
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**
AFF1(MLLT2)/MLL 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/ASS Dual Fusion		t(9;22)(q34;q11)	Tricolour –Complex deletion rearrangement pattern monitoring possible
BLADDER PANEL - Single locus probes		3CEN/7CEN/9p21/17CEN	3, 7 and 17 aneuploidy

D3Z1/D7Z1/p16/D17Z1		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 probes	11q13/11CEN	CCND1 amplification detection
CDKN2C(p18)/CKS1B amplification in MM single locus probes	1p32.3/1q21	CDKN2C deletion / CKS1B
CERVICAL PANEL Single locus probes hTERC/MYC/D7Z1	3q26.2/8q24/7CEN	Detection of hTERC and/or MYC gain
CHD5/1qter single locus probes	1p36	Short arm deletion detection
CLL PANEL Single locus probes 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
D1Z2 Single locus probe	1p36	Short arm deletion detection
D8Z2 Single locus probe	8CEN	Aneusomy 8 detection
D12Z3 Single locus probe	12CEN	Aneusomy 12 detection
D7S522/D7Z1 Single locus probes	7q31/7CEN	Monosomy or long arm deletion detection
D13S25 Single locus probe	13q14	Monosomy or long arm deletion detection
D13S319 Single locus probe	13q14	Monosomy or long arm deletion detection
D20S108 Single locus probe	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 probes	17q22 and 22q13	Detection of t(17;22)(q22;q13) and amplification in supernumerary ring
DXZ1/DYZ3 Single locus probes	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
EVI1 (D3S1243/hTERC/RH123089) Breakapart	3q26.2	Tricolour All variants
EWSR1 Breakapart	22q12	All variants
EWSR1/FLI1 Dual Fusion	t(11;22)(q24;q12)	
FGFR1/D8Z2 Breakapart + single locus probe	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 probes 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 probes	17q11.2-q12	HER2 amplification detection
HER2/TOP2A/D17Z1 Single locus probes	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 probe	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
MALT Breakapart	18q21	All variants
MDM2/D12Z1 single locus probes	12q14.3-q15/12CEN	MDM2 amplification detection

MELANOMA PANEL Single locus probes D6Z1/RREB1/MYB/CCND1	6CEN/6p25/6q23/11q13	
MLL Breakapart	11q23	All variants
MYB Single locus probe	6q23	Long arm deletion detection
MYC Breakapart	8q24	All variants
MYC/D8Z2 single locus probes	8q24/8CEN	Detection of gain of 8q24 relative to 8CEN
N-MYC/D2Z1 Single locus probes	2p24/2CEN	N-MYC amplification detection
NUP98 breakapart	11p15	AML, ALL, CML-bc
p16(D9S1749-D9S1752)/CEP9 Single locus probes	9p21/9CEN	Short arm deletion detection
PAX3 Breakapart	2q35	All variants See also FOXO1
PBX1/TCF3 dual fusion probe	t(1;19)(q23;p13.3)	ALL
PDGFRA /LNX/ SCFD2 Breakapart	4q12 FIP1L1/CHIC2/PDGFRA rearrangement	Tricolour All variants
PDGFRB Breakapart	5q32	All variants
PIK3CA Single locus probe	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 probes	21q22 10q23/10CEN	Mix 1 – Tricolour, all variants Mix 2 - Long arm deletion detection
PTEN/CEP10 Single locus probes	10q23/10CEN	Long arm deletion detection
RARA Breakapart	17q21	All variants
RB1	13q14	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 a/d Breakapart	14q11.2	All variants
TP53/D17Z1 Single locus probes	17p13.1/17CEN	Short arm deletion detection
TP53/MPO Single locus probes	17p13.1/17q22	i(17q) detection
Uveal Melanoma Single Locus Probes D3Z1/D8Z2	3 centromere and 8 centromere	Aneuploidy detection
ZNF217	20q13.2	ZNF217 amplification detection

Test	**Specimen Type**	**Volume**	**Turnaround Time**	**Notes/Comments**
Follicular lymphoma/DLBCL	PETS 2x4µm sections on slides		14 days	
Fragile X syndrome	Blood	0.5-5ml EDTA	2-8 weeks	
Friedreich Ataxia	Blood	0.5-5ml EDTA	2-6 weeks	
Fructose-1,6-bisphosphatase deficiency	Blood	0.5-5ml EDTA	2-8 weeks	
Fumarate Hydratase Deficiency (*FH* sequencing and MLPA)	Blood or Fibroblasts	0.5-5ml EDTA	2-8 weeks	
GATA2	Blood / Bone Marrow	0.5-5ml EDTA	8 weeks	*see website for panel content
Gilbert syndrome	Blood	0.5-5ml EDTA	2-8 weeks	
Glanzmann thrombasthenia	Blood	0.5-5ml EDTA	2-8 weeks	
Glioma	PETS 4x4µm sections on slides		14 days	
Glucose Transporter 1 (GLUT1) deficiency syndrome (SLC2A1 sequencing and MLPA)	Blood	0.5-5ml EDTA	2-8 weeks	
Glutaric Acidaemia Type 1(GA1)	Blood or Fibroblasts	0.5-5ml EDTA	2-8 weeks	
Glycogen Storage Disease Next Generation Sequencing Panels:	Blood	0.5-5ml EDTA	8 weeks	*content for each panel is listed on

Liver, Muscle, Heart, Generalised Panel*				website
Glycogen Storage Disease Type 0 (GYS2 – liver, GYS1 - muscle)	Blood	0.5-5ml EDTA	2-8 weeks	
Glycogen Storage Disease Type 1a (von Gierke disease) (G6PC)	Blood	0.5-5ml EDTA	2-8 weeks	
Glycogen Storage Disease Type 1 non-a (SLC37A4)	Blood	0.5-5ml EDTA	2-8 weeks	
Glycogen Storage Disease Type II (Pompe disease) (GAA)	Blood	0.5-5ml EDTA	2-8 weeks	
Glycogen Storage Disease Type III (AGL)	Blood	0.5-5ml EDTA	2-8 weeks	
Glycogen Storage Disease Type IV (Andersen disease) (GBE1)	Blood	0.5-5ml EDTA	2-8 weeks	
Glycogen Storage Disease Type V (McArdle disease) (PYGM)	Blood	0.5-5ml EDTA	2-8 weeks	
Glycogen Storage Disease Type VI (Hers Disease)(PYGL)	Blood	0.5-5ml EDTA	2-8 weeks	
Glycogen Storage Disease Type VII (Tarui disease)(PFKM)	Blood	0.5-5ml EDTA	2-8 weeks	
Glycogen Storage Disease Type IX (X-linked) (PHKA2 – liver, PHKA1 - muscle)	Blood	0.5-5ml EDTA	2-8 weeks	
Glycogen Storage Disease Type IX (autosomal) (PHKB, PHKG2)	Blood	0.5-5ml EDTA	2-8 weeks	
Glycogen Storage Disease Type X (PGAM2)	Blood	0.5-5ml EDTA	2-8 weeks	
Haemochromatosis	Blood	0.5-5ml EDTA	2-8 weeks	
Haemophilia A/Factor VIII deficiency	Blood	0.5-5ml EDTA	2-8 weeks	
Haemophilia B/Factor IX deficiency	Blood	0.5-5ml EDTA	2-8 weeks	
Haemophilia C/Factor XI deficiency	Blood	0.5-5ml EDTA	2-8 weeks	
Her2	Paraffin Embedded Tissue Biopsy		1-2 weeks	Contact Lab prior to referral
Hereditary Ataxia and Migraine Next Generation Sequencing Panel*	Blood	0.5-5ml EDTA	16 weeks	* see website for panel content
Hereditary Leiomyomatosis with renal cell carcinoma (HLRCC/MCUL) (*FH* sequencing and MLPA)	Blood	0.5-5ml EDTA	2-8 weeks	
Hereditary Non Polyposis Colorectal Cancer (HNPCC) Gene panel (including MLPA)*	Blood	0.5-5ml EDTA	2-8 weeks	* see website for panel content
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	16 weeks	* see website for panel content
Hereditary Spastic Paraparesis (dominant, pure)-*SPAST* gene	Blood	0.5-5ml EDTA	2-8 weeks	Blood in EDTA required for MLPA
Hereditary Spastic Paraparesis (dominant, pure)-*ATL1* gene	Blood	0.5-5ml EDTA	2-8 weeks	
Hereditary Spastic Paraparesis (dominant, pure)-*REEP1* gene	Blood	0.5-5ml EDTA	2-8 weeks	
Huntington disease	Blood	0.5-5ml EDTA	2 weeks	
Hypophosphatasia (ALPL)	Blood	0.5-5ml EDTA	2-8 weeks	
JAK2 (V617F mutation)	Blood / Bone Marrow	0.5-5ml EDTA	2 weeks	
JAK2 Exon 12 mutation screen (polycythaemia rubra vera/PRV)	Blood / Bone Marrow	0.5-5ml EDTA	2 weeks	
Karyotype	See Chromosome			
KIT- D816V	Blood / bone marrow	1-3ml EDTA	10 days	
KRAS (v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog) for CRC	PETS 8x10µm sections in universal		5 days	

and NSCLC and other				
Leber Hereditary Optic Neuropathy (LHON)	Blood	0.5-5ml EDTA	2-8 weeks	
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	Blood or Guthrie spots	0.5 -5ml EDTA	2 weeks	
Malt Lymphoma	PETS 2x4µm sections on slides		14 days	
Mantle cell lymphoma	PETS 2x4µm sections on slides		14 days	
Medium Chain Acyl CoA-dehydrogenase (MCAD)Deficiency	Blood or Guthrie spots	0.5-5ml EDTA	2-8 weeks	
Microarray (see CGH)	-	-	-	
Microarray	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	
Mitochondrial Disorder, mitochondrial cytopathy, MELAS, MERRF, NARP	Blood	0.5-5ml EDTA	2-8 weeks	Contact Lab prior to referral
MPL Exon 10 mutation screen	Blood / bone marrow	0.5-5ml EDTA	2 weeks	
Multiple Endocrine Neoplasia Type 1 (*MEN1* gene sequencing & MLPA)	Blood	0.5-5ml EDTA	2-8 weeks	
Multiple Endocrine Neoplasia Type 2 (MEN2) and Hirschsprung disease (*RET* gene)	Blood	0.5-5ml EDTA	2-8 weeks	
MutYH-associated polyposis (MAP) (*MUTYH* gene) UK common mutation screen/carrier testing	Blood	0.5-5ml EDTA	2-4 weeks	
Myelodysplastic syndromes (MDS)	Bone Marrow	0.25-1ml BM in 5-10ml of transport medium **OR** 1ml BM/VB in Li Hep	28 days	
Myeloma - FISH	Blood / bone marrow	0.25-1ml BM in 5-10ml of transport medium **OR** 1ml BM/VB in Li Hep	28 days	
Myeloproliferative disease (MPD)	Bone marrow	0.25-1ml BM in 5-10ml of transport medium **OR** 1ml BM/VB in Li Hep	28 days	
Myeloproliferative disorder/essential thrombocythaemia(ET)/polycythaemia rubra vera (PRV)/ myelofibrosis (MF) – JAK2	Blood / Bone marrow	0.5-5ml EDTA	2 weeks	
MYH9-related disorders	Blood	0.5-5ml EDTA	8 weeks	*see website for panel content
Neonatal alloimmune thrombocytopenia (NAIT)	Blood	0.5-3ml EDTA	2-8 weeks	
Neuroblastoma	PETS 2x4µm sections on slides		14 days	
Osteogenesis Imperfecta – autosomal dominanr Next Generation Sequencing Panel	Blood	0.5-5ml EDTA	2-8 weeks	*see website for all CTD NGS panels & content
Osteogenesis Imperfecta-autosomal recessive Next Generation	Blood	0.5-5ml EDTA	2-8 weeks	*see website for all CTD NGS panels &

Sequencing Panel				content
Osteogenesis Imperfecta Type V (IFITM5)	Blood	0.5-5ml EDTA	2-8 weeks	
Osteoporosis and Osteoporosis-pseudoglioma syndrome (LRP5)	Blood	0.5-5ml EDTA	2-8 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 (PEX1, PEX6, PEX10, PEX12, PEX26)	Blood	0.5-5ml EDTA	2-8 weeks	
Phosphoglycerate Mutase (muscle, deficiency of)	Blood	0.5-5ml EDTA	2-8 weeks	
PIK3CA (Phosphatidylinositol 3-kinase, catalytic, alpha polypeptide)	PETS 8x10µm sections in universal		1-2 weeks	
Platelet-type pseudo von Willebrand Disease (*GP1BA* gene)	Blood	0.5-5ml EDTA	2-8 weeks	
Polycystic Kidney Disease (autosomal dominant) *PKD1 & PKD2* Full-gene sequencing and MLPA	Blood	1-5ml EDTA	2-8 weeks	
Polycystic Liver Disease (autosomal dominant) PRKCSH & SEC63 gene sequencing	Blood	1-5ml EDTA	2-8 weeks	
Protein C Deficiency	Blood	0.5-5ml EDTA	2-8 weeks	
Protein S Deficiency	Blood	0.5-5ml EDTA	2-8 weeks	
Prothrombin (3' non 20210G>A prothrombin variants)	Blood	0.5-5ml EDTA	2-8 weeks	
Pseudoxanthoma Elasticum	Blood	0.5-5ml EDTA	2-8 weeks	
Quantitative BCR-ABL (MRD)	Blood/Bone marrow	0.5-5ml EDTA	2 weeks	Send to Lab immediately
Retinoblastoma	Blood	2-3ml Li Hep	28 days	
RUNX1	Blood / Bone Marrow	0.5-5ml EDTA	8 weeks	*see website for panel content
SBDS	Blood / Bone Marrow	0.5-5ml EDTA	8 weeks	*see website for panel content
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
Spinal Muscular Atrophy, 5q-linked	Blood	0.5-5ml EDTA	2-8 weeks	
Spinal and Bulbar Muscular Atrophy (SBMA) (Kennedy disease), X-linked	Blood	0.5-5ml EDTA	2-6 weeks	
Spinocerebellar Ataxia, types 1-3, 6, 7, 12 and 17	Blood	0.5-5ml EDTA	2-6 weeks	
Synovial sarcoma	PETS 2x4µm sections on slides		14 days	
TPMT Pyrosequencing	Blood/buccal swabs	0.5- EDTA 5ml	4 weeks	
Trimethylaminuria/Fish Odour Syndrome (FMO3)	Blood	0.5- EDTA 5ml	2-8 weeks	
Tyrosine hydroxylase deficient dopa-responsive dystonia (Segawa)	Blood	0.5-5ml EDTA	2-8 weeks	
UKALL2003/UKALLR3 MRD TRIALS Research trial	Bone marrow	2.5-10ml	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	2-8 weeks	
Very-long-chain-acyl-CoA	Blood or fibroblasts	0.5-5ml	2-8 weeks	

dehydrogenase (VLCAD) Deficiency		EDTA		
von Willebrand Disease type 1-3r	Blood	0.5-5ml EDTA	2-8 weeks	
von Willebrand Disease platelet type pseudo (*GP1BA* gene)	Blood	0.5-5ml EDTA	2-8 weeks	
Warfarin resistance (*VKORC1* gene) and combined vitamin K clotting factor deficiency type 2	Blood	0.5-5ml EDTA	2-8 weeks	
Wilms Tumour, Frasier syndrome, Denys Drash syndrome, (Wilms Tumour Suppressor) WT1 gene sequencing and MLPA	Blood	0.5-5ml EDTA	2-8 weeks	
Wilson disease (ATP7B)	Blood	0.5-5ml EDTA	2-8 weeks	