The Yorkshire and North East Genomic Laboratory Hub

Tests Offered Within Y&NE GLH

KEY
End to End Testing completed at site
Shared Process With GLH

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Y&NE GLH Central Laboratory Leeds

CITT codes	Test Name	Test description	Technology	Overall Responsibilty	Shared Service
R210.2	Inherited MMR deficiency (Lynch syndrome)	Inherited MMR deficiency (Lynch syndrome) (503)	Small panel	Leeds	Y
R210.5	Inherited MMR deficiency (Lynch syndrome)	MLH1;MSH2;MSH6;PMS2	MLPA or equivalent	Leeds	Y
R209.1	Inherited colorectal cancer (with or without polyposis)	Inherited colorectal cancer (with or without polyposis) (54)	Small panel	Leeds	Y
R209.2	Inherited colorectal cancer (with or without polyposis)	MLH1;MSH2;MSH6;PMS2	MLPA or equivalent	Leeds	Y
R211.1	Inherited polyposis - germline test	Inherited colorectal cancer (with or without polyposis) (54)	Small panel	Leeds	Y
R211.2	Inherited polyposis - germline test	APC	MLPA or equivalent	Leeds	Y
	Myeloid Panel	HMDS		HMDS	Y
	Lymphoid Panel	HMDS		HMDS	N

Y&NE GLH Sheffield

CITT codes	Test Name	Test description	Technology	Previously done by Newcastle	Overall Responsibility	Shared Service
R124	Combined factor V and VIII deficiency	Panel	NGS	N	Sheffield	N
R90	Bleeding and platelet disorders	Panel	NGS	N	Sheffield	N
R96	Iron metabolism disorders - NOT common HFE mutations	Panel	NGS	N	Sheffield	N
R97	Thrombophilia	Panel	NGS	N (only did single SERPIN1 gene test)	Sheffield	N
R347	Inherited predisposition to acute myeloid leukaemia (AML)	Panel	NGS	N	Sheffield	N
R92	Rare anaemia	Panel	NGS	N	Sheffield	N
R229	Confirmed Fanconi anaemia or Bloom syndrome	Panel	NGS	N	Sheffield	N
R91	Cytopenia - NOT Fanconi anaemia	Panel	NGS	N	Sheffield	N
R90	Bernard Soulier	Panel	NGS	N	Sheffield	N
R90	Fibrinogen	Panel	NGS	N	Sheffield	N
R119	Factor X deficiency	Single gene	NGS	N	Sheffield	N
R120	Factor XI deficiency	Single gene	NGS	Y	Sheffield	N
R122	Factor XIII deficiency	Panel	NGS	N	Sheffield	N
R112	Factor II deficiency	Single gene	NGS	N	Sheffield	N
R115	Factor V deficiency	Single gene	NGS	N	Sheffield	N
R116	Factor VII deficiency	Single gene	NGS	Y	Sheffield	N
R117	Factor VIII deficiency	Single gene	NGS	Y	Sheffield	N
R118	Factor IX deficiency	Single gene	NGS	Y	Sheffield	N
R121	VWD	Single gene	NGS	Y	Sheffield	N
R123	Combined vitamin K-dependenc clotting factor deficiency	Panel	NGS	N	Sheffield	N
R124	Combined factor V and VIII deficiency	Panel	NGS	N	Sheffield	N
R338	Monitoring for G(M)CSF escape mutations	Single gene	NGS	N	Sheffield	N
R313	Neutropaenia consistent with ELANE mutations	Single gene	NGS	N	Sheffield	N
R90	Glanzmann thrombasthemia	Panel	NGS	N	Sheffield	N
R95	Iron Overload - hereditary haemochromatosis	Single gene	Taqman	Y	Sheffield	N
R260	Fanconi Anaemia/Bloomn syndrome	Breakage analysis	Chromosome scoring	?	Sheffield	N

Y&NE GLH Newcastle

CITT codes	Test Name	Test description	Technology	Overall Responsibilty	Shared Service
R47.1	Angelman syndrome	AS/PWS critical region Methylation testing	Methylation testing	Newcastle	N
R47.2	Angelman syndrome	AS/PWS critical region MLPA or equivalent	MLPA or equivalent	Newcastle	N
R48.1	Prader-Willi syndrome	AS/PWS critical region Methylation testing	Methylation testing	Newcastle	N
R48.2	Prader-Willi syndrome	AS/PWS critical region MLPA or equivalent	MLPA or equivalent	Newcastle	N
R53.1	Fragile X	FMR1 STR STR testing	STR testing	Newcastle	N
R53.1	Fragile X	FMR1 STR STR testing	STR testing	Newcastle	N
R404.2	Fragile X (Premature ovarian failure)	FMR1 STR STR testing	STR testing	Newcastle	N
R404.2	Fragile X (Premature ovarian failure)	FMR1 STR STR testing	STR testing	Newcastle	N
R53.1	Fragile X (FXTAS)	FMR1 STR STR testing	STR testing	Newcastle	N
R53.1	Fragile X (FXTAS)	FMR1 STR STR testing	STR testing	Newcastle	N
R69.1	Hypotonic infant with a likely central cause	SNRPN DMR	Methylation testing	Newcastle	N
R69.1	Hypotonic infant with a likely central cause	SNRPN DMR	MLPA or equivalent	Newcastle	N
R69.4	Hypotonic infant with a likely central cause	DMPK STR	STR testing	Newcastle	N
R70.1	Spinal muscular atrophy type 1 diagnostic test	SMN1	MLPA or equivalent	Newcastle	N
R72.1	Myotonic dystrophy type 1	DMPK STR	STR testing	Newcastle	N
R73.2	Duchenne or Becker muscular dystrophy	DMD	MLPA or equivalent	Newcastle	N
R77.1	Hereditary neuropathy - PMP22 copy number	PMP22	MLPA or equivalent	Newcastle	N
R402.2	Premature ovarian insufficiency	FMR1 STR STR testing	STR testing	Newcastle	N
R402.2	Premature ovarian insufficiency	FMR1 STR STR testing	STR testing	Newcastle	N
R377.1	SNP Array Service	Microarray only	Illumina 850 SNP array	Newcastle	Y