WESSEX REGIONAL GENETICS LABORATORY – RARE DISEASE TESTS – SERVICE LIST – from 1st April 2022

Note: NHS referrals in England are funded directly by NHS England and are subject to the National Genomic Test Directory; referral criteria must satisfy the NHSE Rare and Inherited Disease eligibility criteria. Test Directory indications are in **bold.** Tests not currently covered by the Test Directory are marked with an asterisk*. Non-NHSE, private and overseas referrals will be invoiced. Samples received in error or with inadequate material for testing will be charged at £45.

DISEASE / DISORDER	Price (£)	Routine TAT (Calendar days)	Gene(s) or locus	Description of test *(including reference intervals/ clinical decision values where applicable)*	Category
CYTOGENETIC TESTS					
Fetal loss referrals:					
Microarray analysis **R22.2, R318.2**	435	42	Genome-wide CNVs	Analysis, interpreting and reporting	
DNA storage	45	N/A			
Postnatal cytogenetic referrals:					
Common aneuploidy testing **R26.1**	205	3[1]/42		QF PCR	
Microarray (multiple indications)	435	42		Analysis, interpreting and reporting	
Targeted follow up testing	205	42		Microarray analysis or FISH	
Karyotype **R297.1**	295	42		G banding	
FISH	295	42 [1]urgent neonates		FISH	
MOLECULAR GENETIC TESTS					
Albinism or congenital nystagmus **R39**	885	84	23-gene panel	Next-generation sequencing	NGS service
TYR and *OCA2* dosage analysis*	130	42	*TYR, OCA2*	MLPA	Targeted mutation test
Albright hereditary osteodystrophy, pseudohypoparathyroidism, pseudopseudohypoparathyroidism, acrodysostosis and osteoma cutis					Imprinting analysis
Methylation testing **R293.2**	245	42	*GNAS*	MS-MLPA	
UPD(20) follow-up **R263**	365	42	No specific gene	Microsatellite analysis	
GNAS sequencing **R293.1**	470	42	*GNAS*	NGS + Sanger sequencing	NGS service

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Alpha 1 - antitrypsin deficiency **R191**	245	42	*SERPINA1*	Two common mutations (S & Z)	Targeted mutation test
Angelman syndrome **R47** Chromosome 15 abnormalities UPD(15) follow-up **R263**	 245 365	 42 42	 *SNURF*:TSS-DMR No specific gene	 MS-MLPA Microsatellite analysis	Imprinting analysis
Aortopathy panel	*See* Thoracic aortic aneurysm or dissection				
Beckwith-Wiedemann syndrome **R49**; isolated hemihypertrophy or macroglossia **R50** Chromosome 11 abnormalities (ICR1 and ICR2) UPD(11) follow-up **R263** *CDKN1C* sequencing **R49.3**	 245 365 370	 42 42 42	 *H19-IGF2*:IG-DMR, *KCNQ1OT1*:TSS-DMR 11p15.5 *CDKN1C*	 MS-MLPA Microsatellite analysis Sanger sequencing	 Imprinting analysis Single gene screen
Blepharophimosis, ptosis and epicanthus inversus (BPES) **R43**	485	42	*FOXL2*	Full mutation screen + MLPA + fragment analysis	Single gene screen
Breast/ovarian cancer	*See* Inherited breast cancer and ovarian cancer				
Ovarian cancer	*See* Inherited ovarian cancer (without breast cancer)				
Chronic myeloid leukaemia (CML)	*See* Oncology genetics page on our website				
Chronic lymphoblastic leukaemia (CLL)	*See* Oncology genetics page on our website				
Confirmation of variants (e.g. from research studies or non-accredited sources)	130	42	*Please note: these may be sent to other specialist laboratories for testing as appropriate or where specified by the National Genomic Test Directory; prices for outsourced tests may differ from those quoted here.*		Targeted mutation test
Cowden syndrome	*See PTEN* hamartoma tumour syndrome				
Cystic fibrosis Routine diagnostic or carrier testing **R184.1, R185.1** Urgent carrier testing **R185.1**	 245 245	 42 14	 *CFTR* *CFTR*	 50 most common UK mutations 50 most common UK mutations	Targeted mutation test

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Newborn screen from blood spots **R253**	130	4 working days	*CFTR*	4 most common UK mutations	
Factor V Leiden and Prothrombin mutations*	245	42	*F5, F2*	NGS genotyping	Targeted mutation test
Familial testing for known variants Predictive testing **R242** Carrier testing **R244** Parental/segregation testing **R375**	245	 14 42 42	*Please note: these may be sent to other specialist laboratories for testing as appropriate or where specified by the National Genomic Test Directory; prices for outsourced tests may differ from those quoted here.*		Targeted mutation test
Haematological malignancies	*See* Oncology genetics page on our website				
Haemochromatosis	*See* Iron overload – hereditary haemochromatosis testing				
Hypogonadotropic hypogonadism **R148** 14-gene panel	 885	 84	 14-gene panel	 Next-generation sequencing	 NGS service
Inherited breast cancer and ovarian cancer **R208** Routine Urgent (including mainstream referrals)	775	 42 21	*BRCA1, BRCA2, PALB2*	Next-generation sequencing + MLPA for *BRCA1* and *BRCA2*	NGS service
Inherited ovarian cancer (without breast cancer) **R207**	885	42	8-gene panel	Next-generation sequencing + MLPA for *BRCA1* and *BRCA2*	NGS service
Iron overload – hereditary haemochromatosis testing **R95**	245	42	*HFE*	Two common mutations	Targeted mutation test
Kagami-Ogata syndrome **R268** Methylation abnormalities Paternal UPD(14) follow-up **R263**	 245 365	 42 42	 *MEG3:*TSS-DMR No specific gene	 Methylation-sensitive MLPA Microsatellite analysis	Imprinting analysis
Kallmann syndrome	*See* Hypogonadotropic hypogonadism				
Marfan syndrome	*See* Thoracic aortic aneurysm or dissection				
Multi-locus imprinting disorder **R417.1**	245	42	No specific gene	Methylation-sensitive MLPA for chromosomes 6, 7, 11, 14, 15, 19 and 20	Imprinting analysis
Myeloproliferative neoplasia (MPN)	*See* Oncology genetics page of our website				

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Neurofibromatosis type 1 (NF1) **R222** (including Legius syndrome*)	775	42	*NF1, SPRED1*	Next-generation sequencing + MLPA for *NF1*	NGS service
Oculopharyngeal muscular dystrophy (OPMD) **R75**	130	42	*PABPN1*	Fluorescent PCR *(Normal: 10 repeats; Pathogenic: 11-17 repeats)*	Repeat expansion test
Prader-Willi syndrome **R48**					Imprinting analysis
Chromosome 15 abnormalities	245	42	*SNURF*:TSS-DMR	MS-MLPA	
UPD(15) follow-up **R263**	365	42	No specific gene	Microsatellite analysis	
Prenatal testing for known pathogenic variants* (to include maternal cell contamination testing **R321** where a maternal sample is available)	375	3	*Please note: these may be sent to other specialist laboratories for testing as appropriate or where specified by the National Genomic Test Directory; prices for outsourced tests may differ from those quoted here.*		Targeted mutation test
Primary ciliary dyskinesia (PCD)	*See* Respiratory ciliopathies				
Pseudohypoparathyroidism (PHP)	*See* Albright hereditary osteodystrophy				
PTEN hamartoma tumour syndrome **R213**	485	42	*PTEN*	Mutation screening by NGS + MLPA	NGS service
Respiratory ciliopathies including non-CF bronchiectasis **R189**	1050	84	46-gene panel	Next-generation sequencing	NGS service
RNA studies (investigating the effect of sequence variants on splicing) **R296**	615	42	No specific gene	Analysis of DNA variants for splicing abnormalities	Specialised testing
Russell-Silver syndrome	*See* Silver-Russell syndrome				
Silver-Russell syndrome (Growth failure in early childhood) **R147.2**					Imprinting analysis
Chromosome 11 abnormalities (ICR1 only) and UPD(7) analysis	245	42	*H19-IGF2*:IG-DMR, *GRB10*:alt-TSS-DMR, *MEST*:alt-TSS-DMR	MS-MLPA	
UPD(11) follow-up **R263**	365	42	No specific gene	Microsatellite analysis	
Thoracic aortic aneurysm or dissection (TAAD) **R125**	1050	84	32-gene panel	Next-generation sequencing + MLPA for *FBN1* only	NGS service

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Temple syndrome **R267** Methylation abnormalities Maternal UPD(14) follow-up **R263**	245 365	42 42	*MEG3*:TSS-DMR No specific gene	MS-MLPA Microsatellite analysis	Imprinting analysis
Transient neonatal diabetes mellitus (TNDM) (6q24-related neonatal diabetes, **R143.3**) UPD(6) follow-up **R263**	245 365	42 42	*PLAGL1*:alt-TSS-DMR No specific gene	6q24 methylation, UPD and dosage analysis by MLPA Microsatellite analysis	Imprinting analysis
Uniparental disomy confirmation **R263** (where no prior imprinting analysis at WRGL)	600	42	No specific gene	Microsatellite analysis	UPD confirmation
X-inactivation studies **R111**	245	42	No specific gene	Methylation analysis by restriction enzyme digestion *(Random = <80:20 ratio; skewed = >91:9 ratio).*	Specialised testing