WESSEX REGIONAL GENETICS LABORATORY – RARE DISEASE TESTS – SERVICE LIST – from 1st October 2020

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 £35.

DISEASE / DISORDER		**Routine TAT (Calendar days)**	**Gene(s) or locus**	**Description of test** *(including reference intervals/ clinical decision values where applicable)*	**Category**
CYTOGENETIC TESTS					
<u>Prenatal Diagnosis:</u> Common aneuploidy testing **R22.1, R401.1** Array CGH **R22.2** Karyotype **R297.1** FISH		 3 14 14 14	 Chr 13, 18, 21, X, Y Genome-wide CNVs Targeted probe	 QF PCR Array CGH G banding FISH	
<u>Fetal loss referrals:</u> Common aneuploidy testing **R22.1, R318.1** Array CGH **R22.2, R318.2** Karyotype **R297.1** FISH DNA storage		 42 42 42 42 N/A	 Chr 13, 18, 21, X, Y Genome-wide CNVs Targeted probe	 QF PCR Array CGH G banding FISH	
<u>Postnatal Cytogenetic referrals:</u> Common aneuploidy testing **R26.1** Array CGH (multiple indications) Targeted follow up testing **R375.1** Karyotype **R297.1** FISH		 3[1]/42 42 42 42 42 [1]urgent neonates		 QF PCR Array CGH Array or FISH G banding FISH	
MOLECULAR GENETIC TESTS					
Albinism or congenital nystagmus **R39** *TYR* and *OCA2* dosage analysis*		84 42	23-gene panel *TYR, OCA2*	Next-generation sequencing MLPA	NGS service Targeted mutation test

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Albright hereditary osteodystrophy, pseudohypoparathyroidism and pseudopseudohypoparathyroidism Methylation testing **R293.2**		42	*GNAS*	MS-MLPA	Imprinting analysis
UPD(20) follow-up **R263**		42	No specific gene	Microsatellite analysis	
GNAS sequencing **R293.1**		42	*GNAS*	NGS + Sanger sequencing	Single gene screen
Alpha 1 - antitrypsin deficiency **R191**		42	*SERPINA1*	2 common mutations (S & Z)	Targeted mutation test
Angelman syndrome **R47** Chromosome 15 abnormalities		42	*SNURF*:TSS-DMR	MS-MLPA	Imprinting analysis
UPD(15) follow-up **R263**		42	No specific gene	Microsatellite analysis	
Aniridia, *PAX6*-related*		42	*PAX6*	Next-generation sequencing + MLPA	NGS service
Aortopathy panel	*See* Thoracic aortic aneurysm or dissection				
Beckwith-Wiedemann syndrome **R49** Chromosome 11 abnormalities		42	ICR1 and ICR2	MS-MLPA	Imprinting analysis
UPD(11) follow-up **R263**		42	11p15.5	Microsatellite analysis	
CDKN1C sequencing **R49.3**		42	*CDKN1C*	Sanger sequencing	Single gene screen
Blepharophimosis, ptosis and epicanthus inversus (BPES) **R43**		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* separate Oncology service list (on Oncology genetics page of our website)				
Chronic lymphoblastic leukaemia (CLL)	*See* separate Oncology service list (on Oncology genetics page of our website)				

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Cowden syndrome	*See PTEN* hamartoma tumour syndrome				
Cystic fibrosis Routine diagnostic or carrier testing **R184.1, R185.1** Urgent carrier testing **R185.1** Newborn screen from blood spots **R253**		42 14 4 working days	*CFTR* *CFTR* *CFTR*	50 most common UK mutations 50 most common UK mutations 4 most common mutations in the UK	Targeted mutation test
Factor V Leiden and Prothrombin mutations*		42	*F5, F2*	NGS genotyping	Targeted mutation test
Familial testing for known variants Predictive testing **R242** Carrier testing **R244** Parental/segregation testing **R375**	*Please send to appropriate specialist laboratory for the familial condition*				Targeted mutation test
Fragile X syndrome (FRAXA) **R29.3, R53** or Premature ovarian insufficiency (POI) **R402.2** Standard fluorescent PCR Amplidex™ PCR		42 42	*FMR1* *FMR1*	Expand™ long-template PCR kit Amplidex™ *FMR1* PCR kit *(Normal: up to 45 repeats; Intermediate: 46-58 repeats; Premutation: 59-200 repeats; Full mutation: >200 repeats)*	Repeat expansion test
Haematological malignancies	*See* separate Oncology service list (on Oncology genetics page of our website)				
Haemochromatosis	*See* Iron overload – hereditary haemochromatosis testing				
Hypogonadotropic hypogonadism **R148** 14-gene panel		84	14-gene panel	Next-generation sequencing	NGS service
Infertility	*See* Cystic fibrosis; Y microdeletions				

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Inherited breast cancer and ovarian cancer **R208**		42	*BRCA1, BRCA2, PALB2*	Next-generation sequencing + MLPA for *BRCA1* and *BRCA2*	NGS service
Inherited ovarian cancer (without breast cancer) **R207**		42	8-gene panel	Next-generation sequencing + MLPA for *BRCA1* and *BRCA2*	NGS service
Iron overload – hereditary haemochromatosis testing **R95**		42	*HFE*	Two common mutations	Targeted mutation test
Kagami-Ogata syndrome **R268** Methylation abnormalities Paternal UPD(14) follow-up **R263**		 42 42	 *MEG3:*TSS-DMR No specific gene	 Methylation-sensitive MLPA Microsatellite analysis	Imprinting analysis
Kallmann syndrome	*See* Hypogonadotropic hypogonadism				
Léri–Weill dyschondrosteosis (LWD)	*See* Short stature – SHOX deficiency				
Marfan syndrome	*See* Thoracic aortic aneurysm or dissection				
Multi-locus imprinting disorder*		42	No specific gene	Methylation-sensitive MLPA for chromosomes 6, 7, 11, 14, 15 and 20	Imprinting analysis
Multiple exostoses **R390**		42	*EXT1, EXT2*	Next-generation sequencing + MLPA	NGS service
Myeloproliferative neoplasia (MPN)	*See* separate Oncology service list (on Oncology genetics page of our website)				
Neurofibromatosis type 1 (NF1) **R222** including Legius syndrome*		42	*NF1, SPRED1*	Next-generation sequencing + MLPA for *NF1*	NGS service
Noonan syndrome* Full screen Data analysis only		 84 84	 14-gene panel 14-gene panel	 Next-generation sequencing Next-generation sequencing (data)	NGS service
Oculopharyngeal muscular dystrophy (OPMD) **R75**		42	*PABPN1*	Fluorescent PCR *(Normal: 10 repeats; Pathogenic: 11-17 repeats)*	Repeat expansion test
Prader-Willi syndrome **R48** Chromosome 15 abnormalities UPD(15) follow-up **R263**		 42 42	 *SNURF*:TSS-DMR No specific gene	 MS-MLPA Microsatellite analysis	Imprinting analysis

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Premature ovarian failure (POF) / Primary ovarian insufficiency (POI), Fragile X-related	*See* Fragile X syndrome				
Prenatal testing for known pathogenic variants*	*Please send to appropriate specialist laboratory for the familial condition*				Targeted mutation test
Primary ciliary dyskinesia (PCD)	*See* Respiratory ciliopathies				
Pseudohypoparathyroidism (PHP)	*See* Albright hereditary osteodystrophy				
PTEN hamartoma tumour syndrome **R213**		42	*PTEN*	Mutation screening by NGS + MLPA	NGS service
Respiratory ciliopathies including non-CF bronchiectasis **R189**		84	46-gene panel	Next-generation sequencing	NGS service
RNA studies (investigating the effect of sequence variants on splicing) **R296**		42	No specific gene	Analysis of DNA variants for splicing abnormalities	Specialised testing
Rubinstein-Taybi syndrome*		42	*CREBBP, EP300*	Next-generation sequencing + MLPA	NGS service
Russell-Silver syndrome	*See* Silver-Russell syndrome				
Sequencing of known variants (confirmations, family or predictive testing)	*Please send to appropriate specialist laboratory for the familial condition*				Targeted mutation test
Short stature – SHOX deficiency **R52** including Léri–Weill dyschondrosteosis (LWD)		42	*SHOX*	Sanger sequencing and MLPA	Single gene screen
Silver-Russell syndrome (Growth failure in early childhood) **R147.2**					Imprinting analysis
Chromosome 11 abnormalities		42	ICR1 only	MS-MLPA	
UPD(7) by MS-MLPA		42	No specific gene	MS-MLPA	
UPD(11) follow-up **R263**		42	No specific gene	Microsatellite analysis	
Thoracic aortic aneurysm or dissection (TAAD) **R125**		84	32-gene panel	Next-generation sequencing + MLPA for *FBN1* only	NGS service
Temple syndrome **R267**					Imprinting analysis
Methylation abnormalities		42	*MEG3*:TSS-DMR	MS-MLPA	
Maternal UPD(14) follow-up **R263**		42	No specific gene	Microsatellite analysis	

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Transient neonatal diabetes mellitus (TNDM) (6q24-related neonatal diabetes, **R143.3**) UPD(6) follow-up **R263**		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 has been carried out by WRGL)		42	No specific gene	Microsatellite analysis	UPD confirmation
X-inactivation studies **R111**		42	No specific gene	Methylation analysis by restriction enzyme digestion *(Random = <80:20 ratio; skewed = >91:9 ratio).*	Specialised testing
Y microdeletions (AZFa, b, and c) **R411**		42	No specific gene	MLPA	Targeted mutation test