Rare Disease service list - Version: 1.7. Index: WGLS DOC 033467. Printed: 18-Aug-2026 13:24

WESSEX GENOMICS LABORATORY SERVICE (Salisbury) – RARE DISEASE TESTS – SERVICE LIST – from 1st April 2026

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. For current prices, please contact our Duty Scientist: shc-tr.WRGLdutyscientist@nhs.net .

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** DNA storage		42 N/A	Genome-wide CNVs	Analysis, interpreting and reporting	
Postnatal cytogenetic referrals: Common aneuploidy testing **R26.1** Microarray (multiple indications) Targeted follow up testing Karyotype **R297.1** FISH		3[1]/42 42 42 42 42 [1]urgent neonates		QF PCR Analysis, interpreting and reporting Microarray analysis or FISH G banding FISH	
MOLECULAR GENETIC TESTS					
Albinism or congenital nystagmus **R39** *TYR* and *OCA2* dosage analysis*		42 42	28-gene panel *TYR, OCA2*	Next-generation sequencing MLPA	NGS service Targeted mutation test
Albright hereditary osteodystrophy, pseudohypoparathyroidism, pseudopseudohypoparathyroidism, acrodysostosis and osteoma cutis Methylation testing **R293.2**		42	*GNAS*	MS-MLPA	Imprinting analysis

UPD(20) follow-up **R263** 3-gene panel **R293.1**		42 42	No specific gene *GNAS, PRKAR1A, PDE4D*	Microsatellite analysis NGS + Sanger sequencing	 NGS service
Alpha 1 - antitrypsin deficiency **R191**		42	*SERPINA1*	Two common mutations (S & Z)	Targeted mutation test
Angelman syndrome **R47** Chromosome 15 abnormalities UPD(15) follow-up **R263**		 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**		 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**		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 **R370;** or where a second technique required **R443**)		42	Sanger sequencing, RC-PCR (targeted NGS), MLPA, as appropriate *Please note: these may be sent to other specialist laboratories for testing as appropriate or where specified by the National Genomic Test Directory*		Targeted mutation test

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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 UK mutations	Targeted mutation test
Factor V Leiden and Prothrombin common variants*		42	*F5, F2*	NGS genotyping	Targeted mutation test
Familial testing for known variants Predictive testing **R242** Carrier testing **R244** Parental/segregation testing **R375**		14 42 42	Sanger sequencing, RC-PCR (targeted NGS), MLPA, as appropriate *Please note: these may be sent to other specialist laboratories for testing as appropriate or where specified by the National Genomic Test Directory*		Targeted mutation test
Haematological malignancies	*See* Oncology genetics page on our website				
Haemochromatosis	*See* Iron overload – hereditary haemochromatosis testing				
Hypogonadotropic hypogonadism **R148** 22-gene panel		42	22-gene panel	Next-generation sequencing	NGS service
Inherited breast cancer and ovarian cancer **R208**		42	7-gene panel	Next-generation sequencing	NGS service
Inherited MMR deficiency (Lynch syndrome) **R210**		42	4-gene panel	Next-generation sequencing + MLPA (includes copy number analysis for *EPCAM*)	NGS service

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Inherited ovarian cancer (without breast cancer) **R207**		42	9-gene panel	Next-generation sequencing	NGS service
Iron overload – hereditary haemochromatosis **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				
Marfan syndrome	*See* Thoracic aortic aneurysm or dissection				
Multi-locus imprinting disorder Methylation abnormalities **R417.1** 5-gene panel **R417.2**		 42 42	 No specific gene 5-gene panel	 Methylation-sensitive MLPA for chromosomes 6, 7, 11, 14, 15, 19 and 20 Next-generation sequencing	Imprinting analysis NGS service
Myeloproliferative neoplasia (MPN)	*See* Oncology genetics page of our website				
Neurofibromatosis type 1 (NF1) **R222** (including Legius syndrome)		42	*NF1, SPRED1*	Next-generation sequencing	NGS service
NICE approved PARP inhibitor treatment **R444** Breast cancer **R444.1** Prostate cancer **R444.3**		42	*BRCA1, BRCA2*	Next-generation sequencing	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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Prenatal testing for known pathogenic variants* (to include maternal cell contamination testing **R321** where a maternal sample is available)		3	Sanger sequencing, RC-PCR (targeted NGS), MLPA, as appropriate *Please note: these may be sent to other specialist laboratories for testing as appropriate or where specified by the National Genomic Test Directory*		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**		42	48-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
Russell-Silver syndrome	*See* Silver-Russell syndrome				
Silver-Russell syndrome (Growth failure in early childhood) **R147.2** Chromosome 11 abnormalities (ICR1 only) and UPD(7) analysis UPD(11) follow-up **R263**		42 42	*H19-IGF2*:IG-DMR, *GRB10*:alt-TSS-DMR, *MEST*:alt-TSS-DMR No specific gene	MS-MLPA Microsatellite analysis	Imprinting analysis
Thoracic aortic aneurysm or dissection (TAAD) **R125**		42	33-gene panel	Next-generation sequencing	NGS service
Temple syndrome **R267** Methylation abnormalities Maternal UPD(14) follow-up **R263**		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**		42 42	*PLAGL1*:alt-TSS-DMR No specific gene	6q24 methylation, UPD and dosage analysis by MLPA Microsatellite analysis	Imprinting analysis

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Uniparental disomy confirmation **R263** (where no prior imprinting analysis at 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

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