



Band	GenU Score	General examples	Specific examples
A	1	 All DNA extractions to include extract > test locally extract > DNA banking All RNA extraction 	
		 Sample receipt, booking in, and processing of all sample types. Covers: Sample preparation, setting up of culture(s) and processing of sample to provide a cell suspension for cytogenetic analyses, processing of PET samples for FISH, DNA extraction 	 Samples processed for both Cytogenetic and Molecular Genetic Studies are considered as separate. Interpretation/undertaking segregation of results from another laboratory. Re-issue of report for sample previously tested (repeat request for same test). Proband samples processed as a positive control for other family members
Α	1	 DNA/cell culture sample export 	 An additional A is counted for any exports only of DNA or cell cultures
		 Cell freezing/storage – long term liquid nitrogen storage 	 Freezing/storage – this is a one-off charge for potentially long-term storage
В	2	 Single amplicon (genotyping or sequencing) 	 FraX PCR Haemochromatosis Factor V Jak2 HD (diagnostic and predictive tests) Other triplet disorders where a single PCR is required (eg SBMA) Y deletions FLT3 NPM1
		 Embryo preparation of PGD analysis FISH only testing for constitutional or acquired samples with a single FISH hybridisation as the only test Follow up FISH testing for all sample types with a single FISH hybridisation as the only test 	 Only includes preparation for testing. A single hybridisation can include two informative probes e.g. ATM/TP53 combination probe Follow up of microarray findings using a single FISH probe
С	4	 Genotyping 2-4 amplicons Sequencing: Very small gene with 2-4 exons/amplicons Sequencing: Predictive tests, confirmations and carrier tests 	 CF-ARMS, CF-OLA, CF-HT AS/PWS FraX if Southern blotted DM, Friedreich's ataxia RT PCR BCR/ABL1





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		MS-PCR	
		 MLPA with no other test (including DMD) 	
		Prenatal tests to include the MCC	
		1 lane on Southern	
		 Triplet disorders that require two PCRs (allele specific and TP-PCR) 	
		Identity/paternity tests	
		Direct CVS analysis	Includes slide making/banding and FISH preparation for all probe types
		 Rapid aneuploidy testing for +13, +18 and +21, X/Y (QF-PCR FISH) 	Parental follow up samples: any method NB. proband sample acts as a positive control
		 Follow up testing all sample types by karyotype, FISH, MLPA, targeted array and FISH (if 2-4 hybridisations) 	 E.g. CLL FISH panel Haematology monitoring samples included as follow up
		Kit based MLPA	
		 FISH only testing for constitutional or acquired samples with 2-4 FISH hybridisations 	
D	7	Postnatal constitutional whole genome screen by karyotyping or array analysis without a rapid	 Includes slide making and G-banding and processing steps post DNA extraction.
		aneuploidy pre-screen includes. This includes any additional conventional staining or FISH tests	Covers blood and solid tissue referrals
		requested/required including confirmation of array findings, if required, for the proband	 G-band analysis appropriate to referral reason and if necessary other conventional staining (eg C band, NOR) to aid interpretation.
Е	10	5-19 amplicons (MLPA to count as 2 amplicons when	Sequencing MECP2 by Sanger or NGS
		part of full screen)	 DMD linkage
		 All linkage tests including UPD 	 AS/PWS if linked markers used
		 Prenatal constitutional whole genome screen by karyotyping or array analysis without a rapid 	 Includes SCE prep and analysis for FA, and scanning for chromosome 7 and 14 rearrangements for AT.
		aneuploidy pre-screen includes any additional conventional staining or FISH tests requested/required including array confirmation for the proband	 Transformed/relapse category includes those where a full analysis on the sample is required.
		Postnatal constitutional whole genome screen by	Postnatal covers blood and solid tissue referrals
		karyotyping or array analysis including a rapid	Includes long term culture, slide making and G- banding and processing steps post DNA extraction





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		 aneuploidy pre-screen test. This includes any additional conventional staining or FISH tests requested/required. Includes confirmation of array findings, if required, for the proband Chromosome breakage studies, eg FA, or AT Diagnostic, transformed or relapsed Haematological (marrow, blood, lymph node, effusion) or tumour whole genome screen by karyotyping or array analysis includes any additional conventional staining or FISH tests requested/required. Haematological FISH only testing 5-19 hybridisations 	 Rapid aneuploidy testing for +13, +18 and +21, X/Y (QF-PCR FISH)
F	15	20-49 amplicons (MLPA to count as 2 amplicons when part of full screen)	 Sequencing factor 8 by Sanger or NGS
		Prenatal constitutional whole genome screen by karyotyping or array analysis including a rapid aneuploidy pre-screen test. This includes any additional conventional staining or FISH tests requested/required. Includes confirmation of array findings, if required.	 Includes long term culture, slide making and G- banding and processing steps post DNA extraction Rapid aneuploidy testing for +13, +18 and +21, X/Y (QF-PCR FISH)
G	25	50-100 amplicons (MLPA to count as 2 amplicons when part of full screen)	Sequencing FBN1Sequencing BRCA1+BRCA2
		1-50 genes analysed by NGS	 Sequencing 12 genes for Noonan Spectrum Disorders
Η	40	Over 100 amplicons	 Sequencing a group of genes in parallel that contribute to a single report
		51-500 genes analysed by NGS	Sequencing 105 genes for Retinal Degeneration

Note internal transport of DNA/cell culture samples between co-located laboratories should not be counted as exports Shared activity within co-located laboratories only attracts the GenU (single band) for the shared activity