Laboratory Genetic Units (GenUs)
June 2014
Note internal transport of DNA/cell culture samples between co-located laboratories should not be counted as exports.

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).
A	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
B	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	▪ Only includes preparation for testing. ▪ A single hybridisation can include two informative probes e.g. ATM/TP53 combination probe

Band	GenU Score	General examples	Specific examples
C	4	▪ Genotyping 2-4 amplicons ▪ Sequencing: Very small gene with 2-4 exons/amplicons ▪ Sequencing: Predictive tests, confirmations and carrier tests ▪ 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	▪ CF-ARMS, CF-OLA, CF-HT ▪ AS/PWS ▪ FraX if Southern blotted ▪ DM, Friedreich's ataxia ▪ RT PCR BCR/ABL1
		▪ Direct CVS analysis ▪ Rapid aneuploidy testing for +13, +18 and +21, X/Y (QF-PCR FISH) ▪ Follow up testing all sample types by karyotype, FISH, MLPA, targeted array ▪ Kit based MLPA ▪ FISH only testing for constitutional or acquired samples with 2-4 FISH hybridisations	▪ Includes slide making/banding and FISH preparation for all probe types ▪ Parental follow up samples: any method ▪ E.g. CLL FISH panel ▪ Haematology monitoring samples included as follow up
D	7	Postnatal constitutional whole genome screen by karyotyping or array analysis includes any additional conventional staining or FISH tests requested/required including array confirmation for the proband	▪ Includes slide making and G-banding and processing steps post DNA extraction. ▪ Covers blood and solid tissue referrals ▪ Includes any rapid aneuploidy testing prior to the whole genome screen ▪ G-band analysis appropriate to referral reason and if necessary other conventional staining (eg C band, NOR) to aid interpretation.
E	10	▪ 5-19 amplicons (MLPA to count as 2 amplicons when part of full screen) ▪ All linkage tests including UPD	▪ Sequencing MECP2 by Sanger or NGS ▪ DMD linkage ▪ AS/PWS if linked markers used
		▪ Prenatal constitutional whole genome screen by karyotyping or array analysis includes any additional conventional staining or FISH tests requested/required including array confirmation 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.	▪ Includes any rapid aneuploidy testing prior to the whole genome screen ▪ Includes SCE prep and analysis for FA, and scanning for chromosome 7 and 14 rearrangements for AT. ▪ Transformed/relapse category includes those where a full analysis on the sample is required.

Band	GenU Score	General examples	Specific examples
		▪ Haematological FISH only testing 5-19 hybridisations	
F	**15**	▪ 20-49 amplicons (MLPA to count as 2 amplicons when part of full screen)	▪ Sequencing factor 8 by Sanger or NGS
G	**25**	▪ 50-100 amplicons (MLPA to count as 2 amplicons when part of full screen)	▪ Sequencing FBN1 ▪ Sequencing BRCA1+BRCA2
		▪ 1-50 genes analysed by NGS	▪ Sequencing 12 genes for Noonan Spectrum Disorders
H	**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