

Band	GenU Score	General examples	Specific examples
A	1	<ul style="list-style-type: none"> ▪ All DNA extractions to include <ul style="list-style-type: none"> ○ extract > test locally ○ extract > DNA banking ▪ All RNA extraction 	
		<ul style="list-style-type: none"> ▪ Sample receipt, booking in, and processing of all sample types. Covers: <ul style="list-style-type: none"> ○ 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 	<ul style="list-style-type: none"> ▪ 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
A	1	<ul style="list-style-type: none"> ▪ DNA/cell culture sample export 	<ul style="list-style-type: none"> ▪ An additional A is counted for any exports only of DNA or cell cultures
		<ul style="list-style-type: none"> ▪ Cell freezing/storage – long term liquid nitrogen storage 	<ul style="list-style-type: none"> ▪ Freezing/storage – this is a one-off charge for potentially long-term storage
B	2	<ul style="list-style-type: none"> ▪ Single amplicon (genotyping or sequencing) 	<ul style="list-style-type: none"> ▪ 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
		<ul style="list-style-type: none"> ▪ 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 	<ul style="list-style-type: none"> ▪ 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
C	4	<ul style="list-style-type: none"> ▪ Genotyping 2-4 amplicons ▪ Sequencing: Very small gene with 2-4 exons/amplicons ▪ Sequencing: Predictive tests, confirmations and carrier tests 	<ul style="list-style-type: none"> ▪ CF-ARMS, CF-OLA, CF-HT ▪ AS/PWS ▪ FraX if Southern blotted ▪ DM, Friedreich's ataxia ▪ RT PCR BCR/ABL1

Band	GenU Score	General examples	Specific examples
		<ul style="list-style-type: none"> ▪ 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 	
		<ul style="list-style-type: none"> ▪ 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 and FISH (if 2-4 hybridisations) ▪ Kit based MLPA ▪ FISH only testing for constitutional or acquired samples with 2-4 FISH hybridisations 	<ul style="list-style-type: none"> ▪ Includes slide making/banding and FISH preparation for all probe types ▪ Parental follow up samples: any method NB. proband sample acts as a positive control ▪ E.g. CLL FISH panel ▪ Haematology monitoring samples included as follow up
D	7	<p>Postnatal constitutional whole genome screen by karyotyping or array analysis without a rapid aneuploidy pre-screen includes. This includes any additional conventional staining or FISH tests requested/required including confirmation of array findings, if required, for the proband</p>	<ul style="list-style-type: none"> ▪ Includes slide making and G-banding and processing steps post DNA extraction. ▪ Covers blood and solid tissue referrals ▪ G-band analysis appropriate to referral reason and if necessary other conventional staining (eg C band, NOR) to aid interpretation.
E	10	<ul style="list-style-type: none"> ▪ 5-19 amplicons (MLPA to count as 2 amplicons when part of full screen) ▪ All linkage tests including UPD 	<ul style="list-style-type: none"> ▪ Sequencing MECP2 by Sanger or NGS ▪ DMD linkage ▪ AS/PWS if linked markers used
		<ul style="list-style-type: none"> ▪ Prenatal constitutional whole genome screen by karyotyping or array analysis without a rapid aneuploidy pre-screen includes any additional conventional staining or FISH tests requested/required including array confirmation for the proband ▪ Postnatal constitutional whole genome screen by karyotyping or array analysis including a rapid 	<ul style="list-style-type: none"> ▪ 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. ▪ Postnatal covers blood and solid tissue referrals ▪ Includes long term culture, slide making and G- banding and processing steps post DNA extraction

Band	GenU Score	General examples	Specific examples
		<p>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</p> <ul style="list-style-type: none"> ▪ 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 	<ul style="list-style-type: none"> ▪ 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)	<ul style="list-style-type: none"> ▪ 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.	<ul style="list-style-type: none"> ▪ 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)	<ul style="list-style-type: none"> ▪ Sequencing FBN1 ▪ Sequencing BRCA1+BRCA2
		1-50 genes analysed by NGS	<ul style="list-style-type: none"> ▪ Sequencing 12 genes for Noonan Spectrum Disorders
H	40	Over 100 amplicons	<ul style="list-style-type: none"> ▪ Sequencing a group of genes in parallel that contribute to a single report
		51-500 genes analysed by NGS	<ul style="list-style-type: none"> ▪ 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