Appendix A: Turnaround Times

Clinical Urgency	Category (mapping to test directory)	Sub- categories	Calendar Days	Examples
URGENT	Ultra Rapid	N/A	3 days	QF-PCR for rapid trisomy detection Urgent haemato-oncology FISH/RT-PCR PCR-based tests where the result is needed urgently for prenatal diagnosis
URGENT	Ultra rapid	NA	7 days	NIPT
URGENT	Rapid	Rapid	14 days	Microarray for prenatal / urgent postnatal (e.g. neonatal referrals) Urgent Haemato-oncology karyotyping Mutation specific molecular pathology tests Southern blot tests where the result is needed urgently for prenatal diagnosis PCR-based tests for predictive testing and confirmation of neonatal results
		Complex rapid	21 days	Urgent panels and exomes for relevant indications NIPD
NON- URGENT	Standard	Somatic Cancer	21 days	Standard HO karyotyping (e.g. MDS) NGS panels for HO referrals NGS panels for molecular pathology referrals
		Rare Disease	42 days (6 weeks)	Standard paediatric microarray Standard single gene and small gene panel (<10 gene) sequencing Known familial mutation testing Standard STR based analysis Postnatal karyotyping (e.g. fertility or familial microarray follow-up)
NON- URGENT	Complex Standard	Rare Disease	84 days (12 weeks)	Large gene-panels (>10 genes) or WES for standard referral indications
			Part a) 42 days (6 weeks)	Expectation for delivery of centralised WGS (from DNA sample receipt to return of vcf and/or filtered variants to GLH)
			Part b) 42 days (6 weeks)	Validation/reporting of centralised WGS results after receipt at GLH