Quality Subcommittee

Association for Clinical Genetic Science
Part of the British Society for Genetic Medicine

Genetic Laboratory Reporting Time Targets

Proposed revised Medical Genetics Dashboard reference	Category	Examples of tests to be included in category
GEN04i	Proportion of prenatal and urgent postnatal rapid aneuploidy QF-PCR/FISH tests and PCR based tests where result is required urgently for prenatal diagnosis completed within 3 working days.	Prenatal test on DNA from CVS, AF, fetal blood (including rapid aneuploidy) by PCR, Sanger sequencing, FISH or chromosome analysis from direct CVS culture. Neonates with query Trisomy (FISH).
GEN04ii	Proportion of urgent haemato-oncology rapid PCR/FISH tests completed within 3 working days.	Urgent diagnostic haemato-oncology and molecular monitoring of acute leukaemias.
GEN04iii	Proportion of urgent postnatal blood karyotype tests completed within 10 calendar days.	Urgent postnatal blood karyotype and urgent chromosome breakage.
GEN04iv	Proportion of prenatal cytogenetics tests & urgent postnatal array CGH tests, PCR based tests for predictive testing and confirmation of neonatal results, and Southern blot analysis where the result is urgently needed for prenatal diagnosis completed within 14 calendar days.	Prenatal chromosome analysis, prenatal microarray, prenatal FISH and prenatal Southern blots (FSHD, Fragile X syndrome). Clinically urgent PCR, MLPA, Sanger sequencing, microarray and FISH tests (not prenatal); neonatal diagnosis or patient/partner pregnant. BRCA predictive tests. Predictive testing PCR, MLPA or Sanger sequencing where familial pathogenic mutation is known.
GEN04v	Proportion of urgent haemato-oncology tests completed within 14 calendar days.	Karyotyping for ? acutes, ? CML, ? transformation and ? relapse.
GEN04vi	Proportion of routine haemato-oncology tests completed within 21 calendar days.	All non-urgent referrals.
GEN04vii	Proportion of routine postnatal/solid tissue/bloods cytogenetic tests including array CGH tests, non-urgent PCR based tests where the familial mutation is known (excluding predictive tests) and specific mutation tests or gene tracking by microsatellite analysis completed within 28 calendar days.	Routine postnatal karyotype. Routine and solid tissue microarrays. Non-urgent microarray follow up tests. Chromosome breakage testing. Diagnostic and cascade testing by PCR, MLPA, Real time PCR. Methylation pyrosequencing analysis for FSHD2. Fragile X Assuragen test. Cascade Sanger sequencing of known variants if not urgent. Cascade MLPA or Sanger sequencing for familial segregation analysis of an unclassified variant.
GEN04viii	Proportion of mutation screening or tests that require Southern blot analysis and Next Generation Sequencing panels (NGS) of ≤10 genes completed within 56 calendar days.	Full (or partial) gene screen by Sanger sequencing. Small NGS panels of ≤10 genes. Southern blotting (e.g. FSHD and Fragile X syndrome).
GEN04ix	Proportion of Next Generation Sequencing (NGS) panels of >10 genes and other large scale sequencing work e.g. WES or WGS completed within 112 calendar days.	Large targeted NGS panels of >10 genes. Clinical exome panels. Whole exome and whole genome sequencing.