Standard list of laboratory preparations for testing

Genetics is a fast moving area with continual developments in processes to improve test sensitivity and specificity to the benefit of patients. Production of diagnostic reports for both cytogenetic and molecular genetics involves a wide variety of practical and analytical techniques. Laboratories utilise both commercially available kits, in house assays and also develop bespoke tests to suit the individual nature of genetics changes. The steps required to produce diagnostic reports can broadly be divided into five main areas and include a variety of techniques including but not limited to the following.

Sample preparation: extraction of nucleic acids, cell culture, chromosome staining

Analytical techniques: chromosome analysis by karyotyping, in situ hybridisation and microarray. DNA analysis by PCR in combination with a variety of techniques such as fragment analysis, Sanger sequencing or next generation sequencing

Interpretation of results: Comparison of results to standards/controls to identify chromosomal imbalances, DNA sequence variation or size variation using a combination of manual and computational methodologies.

Reporting: Reports are primarily generated from tests performed in house but may also include validation and reporting of results generated from research studies. Reports include analytical results and their interpretation as well as advice on follow up and implications for family members where appropriate.

Add on tests: Clarification of results may also sometimes require additional specific tests e.g. determination of zygosity or identity, exclusion of maternal cell contamination of prenatal samples or X-inactivation studies.

Laboratories also provide storage and export services.