Genetic Science Spotlight

A comparison of multiple molecular genetic testing methods

Progressing landscape of genetic testing technology generates improved accuracy and streamlined processes at a reduced cost. Here's some handy information of the more popular genetic testing methods for your reference:

1. SANGER SEQUENCING for single base sequencing of smaller target genomic region

In Sanger sequencing, targeted DNA is copied numerous time, producing fragments of different length with fluorescent tags at the end of each fragment which is then used to produce readings. This method the gold standard (99.9997% accuracy) in genetic testing and is especially great for single sample and short genomic region analysis as its cost per run is low. However, it requires more DNA sample to start with and it is not cost effective in sequencing large DNA data with multiple target genes.

2. NEXT GENERATION SEQUENCING (NGS) for analysis for large genomic region such as in WES, WGS and multiple targeted genes panel

NGS anneals cleaved DNA to fragments of about 100-150bp onto a slide which is then amplified and separated before introducing fluorescent tagged nucleotides that would ligate to the fragments to produce readings. NGS can be ran in parallel, offering fast and has high throughput analysis. This makes NGS the best candidate for WES and WGS. It has a 99.3% accuracy and accuracy can be further enhanced by increasing the depth of the sequencing by running the test, for example 30x. Because of the large amount of data generated, data interpretation lengthy and tedious. NGS is used widely in cases where diagnosis is inconclusive and rare. Recent years, NGS has witnessed growth in cancer screening and non-invasive prenatal testing sector.

3. Multiplex Ligation-dependent Probe Amplification (MLPA) for detection of duplication and deletion variants

Instead of amplifying target DNA, MLPA amplifies the probe DNA which is then used to ligate with target DNA to generate signals. It is primary used to detect methylation (imprinting), del/dup and also repeats expansion that could not be detected by both Sanger and NGS. It is low in cost and has a fast turn-around time. However, MLPA could not be used to detect unknown point mutation. Genetic conditions caused by del/del variants such as Duchenne Muscular Dystrophy and Fragile X, and by methylation such as Prader Willi Syndrome and Angelman Syndrome are prime examples that has great diagnosis success by MLPA.

4. KARYOTYPING for analysis of chromosomal structures, translocation, aneuploidy and polyploidy

Karyotyping is a study of the chromosome. It is performed by staining cells that is grown in culture dish and examine under the microscope. It is simple, affordable and readily available. It is a basic study of the structure of chromosomes. Although it has limited resolution, it has been proved to be valuable in detecting structural changes, translocation, aneuploidy and polyploidy.

5. qPCR for small sample genotyping with simple and quick workflow

qPCR utilizes a thermostable DNA polymerase to amplify target DNA with fluorescent tagged probe. The amount of fluorescent released would be proportional to the quantity of the amplified DNA. This 30 years old technology has simple workflow and is highly customable could be used in genotyping and quantifying target region. To date, qPCR is used more frequently in real time gene expression and in microbiological studies. It is important to note that clinical and forensic uses for real-time PCR may be affected by inhibitors found in certain body fluids such as hemoglobin or urea.

This is a part of vast molecular genetic testing methods available in the market. Different genetic condition has different types of variants that need to be studied using specific methods for best outcome. In this constant evolving world of genetic, it is best to consult your clinical geneticist or genetic counsellor to ascertain the best test that would give then most valuable information in patient care and management.

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