	Chromosome analysis (Karyotype)	Single gene test*	Chromosome microarray (CMA)	NGS Panel Test	Whole Exome Seq (WES)	Whole Genome Seq (WGS)
Aneuploidy	+	--	+	--	--	+
Balanced chromosome rearrangements	+	--	--	--	--	+
Unbalanced chromosome rearrangements	+	--	+	--	--	+
Copy number variants (CNV) -Large >5Mb -Small: 0.1-1Mb	 + --	 -- --	 + +	 -- +	 -- #	 + +
Insertions/deletions (Indels) <100bp	--	+	--	+	+	+
Single nucleotide variants (SNV)	--	+	--	+	+	+
Trinucleotide repeat	--	+	--	--	--	--
Mitochondrial DNA variants (mtDNA)	--	+	--	+ (mtDNA panel)	--	+

* = For simplicity, a number of tests have been condensed into this single gene category. We're including Sanger sequencing; MLPA; PCR; Methylation studies; Southern blot etc.

+ can be detected / -- not recommended

- can sometimes be detected, but this test not recommended for CNVs.

WES isn't an ideal test to identify CNVs. This is often why a CMA is ordered prior to WES.

	Single Gene* (genetic test)	Chromosome microarray (CMA)	NGS Panel test	Whole Exome Sequence (WES)	Whole Genome Sequence (WGS)
Indication	When suspect conditions caused by variants in one or a small number of genes, e.g. Huntington disease.	Indicated for patients presenting with intellectual disability, developmental delay, multiple congenital anomalies, dysmorphic features.	When patients have a relatively well defined phenotype and you suspect a condition caused by a variant in one of several genes.	When suspect a monogenic disorder caused by a variant in one of a number of genes. Patients may show non-specific phenotype.	Currently, clinical use of WGS is limited due to cost/ availability of services. This is likely to change in the future.
Benefits	Very specific and accurate; less change of incidental or uncertain findings.	Medicare funding for certain indications; relatively cheap test compared to WGS/WES.	Can provide better coverage of genes of interest compared to exome; less chance of incidental findings compared to exome; often cheaper than exome.	Sequencing of the whole exome allows for future re-analysis as knowledge improves.	Covers non-coding regions of sequence; can identify some variants not detectable by exome e.g. mtDNA, triplet repeats.
Limitations	Limited utility when phenotype is non-specific or for conditions where multiple genes are associated.	Does not identify triplet repeats, balanced translocations, or point mutations.	Only sequences the genes covered on the panel; no capture of exome sequence so unable to re-analyse later.	Incomplete coverage of exome (false negatives); higher chance of incidental and secondary findings; time consuming variant interpretation.	Incomplete coverage; not all identified variants are interpretable; expensive & limited availability.

** This table is very high level and has been simplified for teaching purposes. In particular, we have oversimplified the single gene category (this includes methods such as Sanger sequencing, Southern Blot, PCR, MLP, methylation studies etc).*