OVERVIEW

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Clinically evaluating NGS and its extra information

There is growing use of next generation sequencing (NGS) in diverse fields of scientific research and clinical diagnostics. In the context of preimplantation genetic diagnosis (PGD), NGS has quickly transitioned from a research technique to a frontline clinical methodology, employed for the detection of aneuploidy in human preimplantation embryos. The rapid speed of adoption has been driven by a desire to lower the costs of comprehensive chromosome screening and by the perception that NGS offers the best chance of facilitating this of any method currently available. Additionally, NGS has the potential to provide a number of extra benefits, such as revealing the DNA sequence of individual genes and quantifying levels of mitochondrial DNA in embryo biopsy specimens. At present several NGS methods are available for the genetic assessment of human embryos, utilizing different sequencing platforms and/or alternative DNA amplification strategies. Depending on the technique, there may be differences in cost per sample, throughput, speed, quantity and quality of the DNA sequence data produced. There is little doubt that NGS represents an exciting opportunity for PGD laboratories to improve patient care. However, future developments, such as routine whole genome sequencing, will raise significant ethical questions and should be debated as a matter of urgency.