

NGS

Francesco Fiorentino, PhD

“GENOMA” Molecular Genetics Laboratories, Rome, Milan, Italy

Recent advances in next-generation sequencing (NGS) technologies have stimulated an increasing interest in its application in the field of reproductive medicine for Preimplantation Genetic Screening (PGS) purposes. NGS-based PGS has demonstrated reliable in detecting whole chromosome aneuploidies, mosaicism occurrences and segmental changes in embryos, with the potential to improve chromosomal diagnosis on embryos especially in terms of test performance, high-throughput, automation and sensitivity. In addition, NGS technique has the advantage of allowing the assessment of the mitochondrial genome from the same biopsy. The additional data obtained may provide unprecedented amounts of genetic information from human embryos which could be useful for diagnostic and research purposes, with the potential to revolutionize preimplantation diagnosis.