

Embryology and In vitro Fertilization

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Clinical exome sequencing screening of subfertile individuals participating in in vitro fertilization program: A pilot study

Anastasios Argyriou¹, Danny Dafnis¹, Ioannis Giakoumakis¹ and Pantelis Constantoulakis²

¹Mediterranean Fertility Institute, Greece

²Science Labs Medical Institute, Athens, Greece


Recent advances in genome analysis using next generation sequencing (NGS) allows simultaneously analyzing hundreds and thousands of genes for mutations that either cause or predispose to diseases and/or pathologic phenotypes. We have chosen to apply an advanced clinical exome sequencing panel (powered by Sophia Genetics DDM) on an Illumina NextSeq-500 platform that analyses in detail at least 11 Mb of human expressed DNA that contain more than 4500 genes with disease-causing mutations, according to the Human Gene Mutation Database (HGMD). We selected and targeted on 103 genes involved in human infertility, according to the Human Phenotype Ontology (HPO) database, which uses updated information from validated sources (Decipher, Orphanet, OMIM). The population we studied in this preliminary effort is 12 unrelated volunteers that were participating in IVF programs and exhibited unexplained subfertility and 2 control fertile subjects. Next generation sequencing and detailed bioinformatics analysis of the infertility related genes revealed pathogenic and/or likely pathogenic mutations in most of the

sub-fertile individuals, whereas no mutations were found in the 2 fertile controls in any of the genes studied. The number of samples analyzed is very small to draw conclusive results, but this pilot study suggests that genetic factors that predispose various fertility related functions may play significant role in cases of repeated IVF failures. Given the significant cost and health burden of repeated efforts for fertilization this new genomic era offers a novel approach in selecting the couples that must think of alternative reproduction options.

Speaker Biography

Anastasios Argyriou has 31 years of professional and research experience in Biology of Reproduction and Andrology. (Bachelor in Biology, University of Athens, Greece, DEA in Endocrinology and Development, University of Caen, France, Diploma in Andrology, University Paris XI, France, PhD in Physiology of Human Reproduction, University of Paris VI, France). As a Senior Clinical Embryologist in IVF Laboratories, he has attended multiple seminars in advanced IVF laboratory methods, teaching both undergraduate and Post-graduate students and published scientific papers and articles. He delivered lectures to academic and scientific audiences as a member of Greek and International Scientific Associations and a regular member of the National Committee of Medically Assisted Reproduction in Greece.

e: a.argyriou@yahoo.gr

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