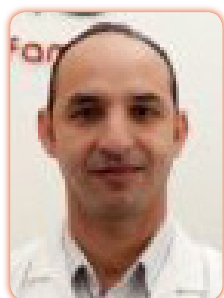


HUMAN GENETICS & GENETIC DISEASES

MOLECULAR MEDICINE & DIAGNOSTICS

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Reducing the burden of genetic diseases via IVF and preimplantation genetic diagnosis

It has been estimated that 5.3% of newborns worldwide will develop a genetic disorder. Certain inherited diseases seem to cluster in particular ethnic groups where consanguinity is common. For example, Canavan, Gaucher and Maple syrup urine diseases are more common in the Ashkenazi population, while individuals of Middle Eastern descent have higher incidence of beta-thalassemia and sickle cell anemia. Living with a genetic condition comes at a considerable financial cost to patients and dramatically impacts the healthcare system. Cystic fibrosis, for instance, is a life-threatening disease that is estimated to cost patients over \$300,000 in medical expenses during their lifetime. Mitochondrial complex I deficiency is an example of a hereditary disease that has no promising treatments and is typically fatal in early childhood. Mitochondrial replacement through three-parent IVF has recently been used to minimize chance of passing mitochondrial disease to the offspring. Although this technique has yet to be legally approved in most countries, a breakthrough genome-editing technology has the potential to cure mitochondria disease. Progenesis and collaborators are currently exploring CRISPR technology to correct mitochondrial mutations in humans. Standard practice for preventing genetic disorders in IVF involves parental carrier screening to identify disease-causing mutations, followed by preimplantation genetic diagnosis (PGD) to test embryos for the mutation before implantation. Industry standards for carrier screening typically include a few hundred genes linked to Mendelian disorders. PGD is used after carrier screening to test embryos for one specific genetic condition, but has the potential to screen for hundreds of human diseases simultaneously. The future of inherited disease control with IVF may eliminate the need for carrier screening by replacing it with comprehensive PGD testing. These tests can significantly reduce the risks of inheriting a genetic disease, alleviate the economic burden on patients and healthcare system and improve overall quality of life.

Biography

Nabil Arrach has 20 years of research experience in molecular genetics, both in preclinical and clinical settings. He has worked at several prestigious research centers, including the University of California, Berkeley, Sanford-Burnham Medical Research Institute and the University of California, Irvine. He was the first Scientist to optimize and validate next generation sequencing for PGS and PGD in 2013. He continues to focus on technology innovation to understand and solve clinical challenges in IVF. He holds a PhD in Molecular and Cellular Biology. He was speaker in numerous scientific conferences and the Founder of Progenesis Inc., a leader in next generation sequencing technology for preimplantation genetic screening and diagnosis.

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