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Strategies to prevent hereditary disorders

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Abstract

Biomedical advances have led to a relaxation of natural selection in the human population of developed countries. In the absence of strong purifying selection, spontaneous and frequently deleterious mutations tend to accumulate in the human genome and gradually increase the genetic load; that is, the frequency of potentially lethal genes in the gene pool. This leads to unprecedented incidence increase of many complex diseases such as autism spectrum disorder (ASD), diabetes and all sorts of autoimmune compromises. We provide further evidence of negative impact of the genetic load on human health reported by state of the art techniques such as exome and whole genome sequencing. In particular, we dissect genetic compromises leading to severe cases of epilepsy and mental retardation. We further discuss strategies proposed to prevent significant accumulation of the genetic load and to reduce the incidence of hereditary disorders of Mendelian and complex etiology. We describe potential benefits of pre-conception carrier tests, pre-implantation genetic screening (PGS) along with recent advanced in prenatal aneuploidy tests. According to recent reports, prevention of the complex diseases based on genetic tests frequently is not feasible and presents a particular challenge. Strategies based on multi-generation cryoconservation of germinal material combined with artificial twinning promise significant future benefits to lower incidence of such conditions.

Biography

Tchourbanov Alexander has completed his Ph.D at the age of 29 years from University of Nebraska-Lincoln and postdoctoral studies from Loyola University Chicago Stritch School of Medicine and Children's hospital, New Orleans. As a director of bioinformatics at the New Mexico State University, Alexander has been actively involved in the building of the novel algorithms for the analysis of the next generation sequencing data. Recently, Alexander has been awarded a young international visiting scientist fellowship to conduct research at the Beijing Institute of Genomics. Currently, he works at the BIO5 institute developing CLIA-certified exome sequencing pipeline. He authored 19 papers in reputed peer-review journals.

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