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Title:

Molecular Genetics of Rare Diseases: From Research to Prevention and Treatment.

Abstract:
Inbred communities, such as the Negev Bedouins and various Jewish cohorts, are susceptible to autosomal recessive diseases. Through human molecular genetics studies and generation of novel bioinformatics tools, followed by molecular biochemistry and generation and analysis of in-vitro and in-vivo disease models, we deciphered the molecular basis of nearly 50 human diseases. Many of those diseases are common throughout the Arab world, and others - in various Jewish diasporas. Initiating and continuously implementing massive carrier testing programs, our work eliminates many severe hereditary diseases, with major impact on infant mortality rates and public health. Moreover, in-depth studies of monogenic cases of common diseases, such as ADHD, atrial fibrillation and gout, enable elucidation of novel molecular pathways of those diseases. Finally, our work paves the way for novel treatment modalities of both rare and common human diseases.