عنوان مقاله:

Exome sequencing reveals novel genes in ARID associated with ataxia families

محل انتشار:

اولین کنگره بین المللی و سیزدهمین کنگره ژنتیک ایران (سال: 1393)

تعداد صفحات اصل مقاله: 1

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خلاصه مقاله:

Intellectual disability (ID) afflicts about 1-3% of the general population. ARID is clinically and genetically heterogeneous often occurring within consanguineous families. There is a critical need to find underlying genetic causes for improving diagnosis, recurrent risk calculation, prognosis and providing prenatal diagnosis services. We investigated the potential ofwhole exome sequencing to delineate the genetic defects in six families with intellectual disability, ataxia and some othersymptoms like cataract and nystagmus. We identified six candidate variants, one known (new mutation) and five novelcandidate genes which haven't previously been implicated in ARID phenotype. Our data exhibit impressiveness of exomesequencing for rapid and also cheaper molecular diagnosis of clinically and genetically heterogeneous diseases in populations with high rate of consanguinity like Iran

کلمات کلیدی:

Intellectual Disability, ataxia, autosomal recessive, exome sequencing, gene