Name of journal: World Journal of Medical Genetics
ESPS Manuscript NO: 15311
Columns: Review

Clinical applications of high-throughput genetic diagnosis in inherited retinal dystrophies: Present challenges and future directions

Gemma Marfany, Roser Gonzàlez-Duarte

Abstract

The advent of Next Generation Sequencing (NGS) techniques has greatly simplified the molecular diagnosis and gene identification in very rare and highly heterogeneous Mendelian disorders. Over the last two years, these approaches, especially Whole Exome Sequencing (WES), alone or combined with homozygosity mapping and linkage analysis, have proved to be successful in the identification of more than 25 new causative retinal dystrophy genes. NGS-approaches have also identified a wealth of new mutations in previously reported genes and have provided more comprehensive information concerning the landscape of genotype-phenotype correlations and the genetic complexity/diversity of human control populations. Although Whole Genome Sequencing (WGS) is far more informative than WES, the functional meaning of the genetic variants identified by the latter can be more easily interpreted, and final diagnosis of inherited retinal dystrophies is extremely successful, reaching 80%, particularly for recessive cases. Even considering the present limitations of WES, the reductions in costs and time, the continual

Google

Clinical applications of high-throughput genetic diagnosis in inherited reti

网页 新闻 图片 视频 更多 搜索工具

找到约 43,000 条结果 （用时 1.10 秒）

Google 学术：Clinical applications of high-throughput genetic diagnosis in inherited retinal dystrophies: Present challenges and future directions

Perspective on genes and mutations causing retinitis ... - Daiger - 被引用次数：237

Molecular genetics of human retinal disease - Rattner - 被引用次数：185

Disease-targeted sequencing: a cornerstone in the ... - Rehm - 被引用次数：83

Genetic testing - Wiley Online Library

onlinelibrary.wiley.com › ... › Vol 42 Issue 1 › Abstract ▾ 翻译此页

作者：RL Gillespie - 2014 - 被引用次数：2 - 相关文章

2013年8月21日 - Genetic testing is of increasing clinical utility for diagnosing inherited eye disease. ... New high-throughput next-generation sequencing (NGS) methods, which permit the .... For example, inherited retinal dystrophies such as RP are .... Systems to transfer data between centres will also present challenges.

Get PDF (1190K) - Wiley Online Library

onlinelibrary.wiley.com/doi/10.1111/ceo.12159/pdf ▾ 翻译此页

作者：RL Gillespie - 2014 - 被引用次数：2 - 相关文章

Genetic testing is of increasing clinical utility for diag- ... of families, and in the future will direct gene-specific ... a family should also be present in other affected ..... inherited retinal dystrophies such as RP are charac- ... tive means of diagnosis but remains challenging,44–46 ... higher throughput diagnostic technologies.

Perspective on Genes and Mutations Causing Retinitis ...

www.ncbi.nlm.nih.gov › ... › PubMed Central (PMC) ▾ 翻译此页

作者：SP Daiger - 2007 - 被引用次数：234 - 相关文章

Success in identifying the causes of inherited retinal diseases has many implications ... Molecular diagnosis alone will therefore neither replace clinical testing nor fully .... other RP genes, and numerous inexpensive, high-throughput techniques exist for ... RELEVANCE TO CLINICAL PRACTICE AND FUTURE DIRECTIONS.

Current mutation discovery approaches in Retinitis

Google Clinical applications of high-throughput genetic diagnosis in inherited retin

网页 新闻 图片 视频 更多 搜索工具

找到约 44,900 条结果（用时 0.21 秒）

Google 学术：Clinical applications of high-throughput genetic diagnosis in inherited retinal dystrophies: Present challenges and future directions

Perspective on genes and mutations causing retinitis ... - Daiger - 被引用次数：239

Molecular genetics of human retinal disease - Rattner - 被引用次数：185

Disease-targeted sequencing: a cornerstone in the ... - Rehm - 被引用次数：83

Genetic testing - Wiley Online Library

onlinelibrary.wiley.com › ... › Vol 42 Issue 1 › Abstract ▾ 翻译此页

作者：RL Gillespie - 2014 - 被引用次数：2 - 相关文章

2013年8月21日 - Genetic testing is of increasing clinical utility for diagnosing inherited eye disease. ... New high-throughput next-generation sequencing (NGS) methods, which permit the .... For example, inherited retinal dystrophies such as RP are .... Systems to transfer data between centres will also present challenges.

Get PDF (1190K) - Wiley Online Library

onlinelibrary.wiley.com/doi/10.1111/ceo.12159/pdf ▾ 翻译此页

作者：RL Gillespie - 2014 - 被引用次数：2 - 相关文章

Genetic testing is of increasing clinical utility for diag- ... of families, and in the future will direct gene-specific ... a family should also be present in other affected ..... inherited retinal dystrophies such as RP are charac- ... tive means of diagnosis but remains