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遗传变异分类标准与指南 免责声明

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分段编辑按钮



These ACMG Standards and Guidelines were developed primarily as an educational resource for clinical laboratory geneticists to help them provide quality clinical laboratory services. Adherence to these standards and guidelines is voluntary and does not necessarily assure a successful medical outcome. These Standards and Guidelines should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, the clinical laboratory geneticist should apply his or her own professional judgment to the specific circumstances presented by the individual patient or specimen. Clinical laboratory geneticists are encouraged to document in the patient's record the rationale for the use of a particular procedure or test, whether or not it is in conformance with these Standards and Guidelines. They also are advised to take notice of the date any particular guideline was adopted and to consider other relevant medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures.

ACMG的这些标准与指南主要是为帮助临床实验室遗传学家提供优质的临床检验服务,开发基础教育资 源。遵守这些标准和指南是自愿的且不一定确保成功的医疗效果。这些标准和指南并不包含所有合适的程 序和检测,或者是否定了其他通过合理方法获得相同结果的程序和检测。在判断任何具体的程序或测试的 合理性时,临床实验室遗传学家应根据个体病人或物种的具体情况应用自己的专业进行判断。不管这些数 据是否标准和指南一致,我们鼓励临床实验室遗传学家将病人的记录进行归档,以用于特定的程序或测 试。我们还建议他们关注任何特定指南被采纳使用的日期,并考虑该日期之后出现的其他可用的相关医疗 和科学信息。还需审慎考虑到知识产权可能会限制某些测试和其他程序的性能。

摘要

The American College of Medical Genetics and Genomics (ACMG) previously developed guidance for the interpretation of sequence variants.1 In the past decade, sequencing technology has evolved rapidly with the advent of high-throughput next-generation sequencing. By adopting and leveraging next-generation sequencing, clinical laboratories are now performing an ever-increasing catalogue of

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整个指南编辑。

農传变异分类标准与指南

- Key Words 关键词
- * 1.引言
- + 3.总论
- + 3.2 命名
- 3.3 文献及数据库使用 反向链接
- 3.4 生物信息学计算预
- ◆ 4.1 PVS1 无效变异
- ◆ 4.2 PS1 相同氨基酸改变
- 4.3 PS2 PM6 新生变异
- ◆ 4.4 PS3 BS3 功能研究
- + 4.5 PS4 PM2 BA1 BS1 BS2 变异频率及对照人群的使用
- 4.6 PM1 热点突变和/或关键 的、得到确认的功能域
- 4.7 PM3 BP2 顺式/反式检测
- 4.8 PM4 BP3 由于框内缺失/ 插入和末端缺失导致的蛋白长 度改变
- ◆ 4.9 PM5 同一位置新的错义变
- ◆ 4.10 PP1 BS4 共分离分析
- ◆ 4.11 PP2 BP1 变异谱
- 4.12 PP3 BP4 生物信息分析
- ◆ 4.13 PP4 表型支持
- ◆ 4.14 PP5 BP6 有信誉的来源
- ◆ 4.15 BP5 可替代座位观察
- ◆ 4.16 BP7 同义变异
- 5. 序列变异报导
- ◆ 5.1 结果









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