# “ACMG 遗传变异分类标准与指南”中文版

# 网站使用说明

## 1、打开网页

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## 3、进入指南中文版全文


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目录栏超链接，点击可快速定位
整个指南编辑，不分段
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分段编辑按钮
遗传变异分类标准与指南
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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。
编辑
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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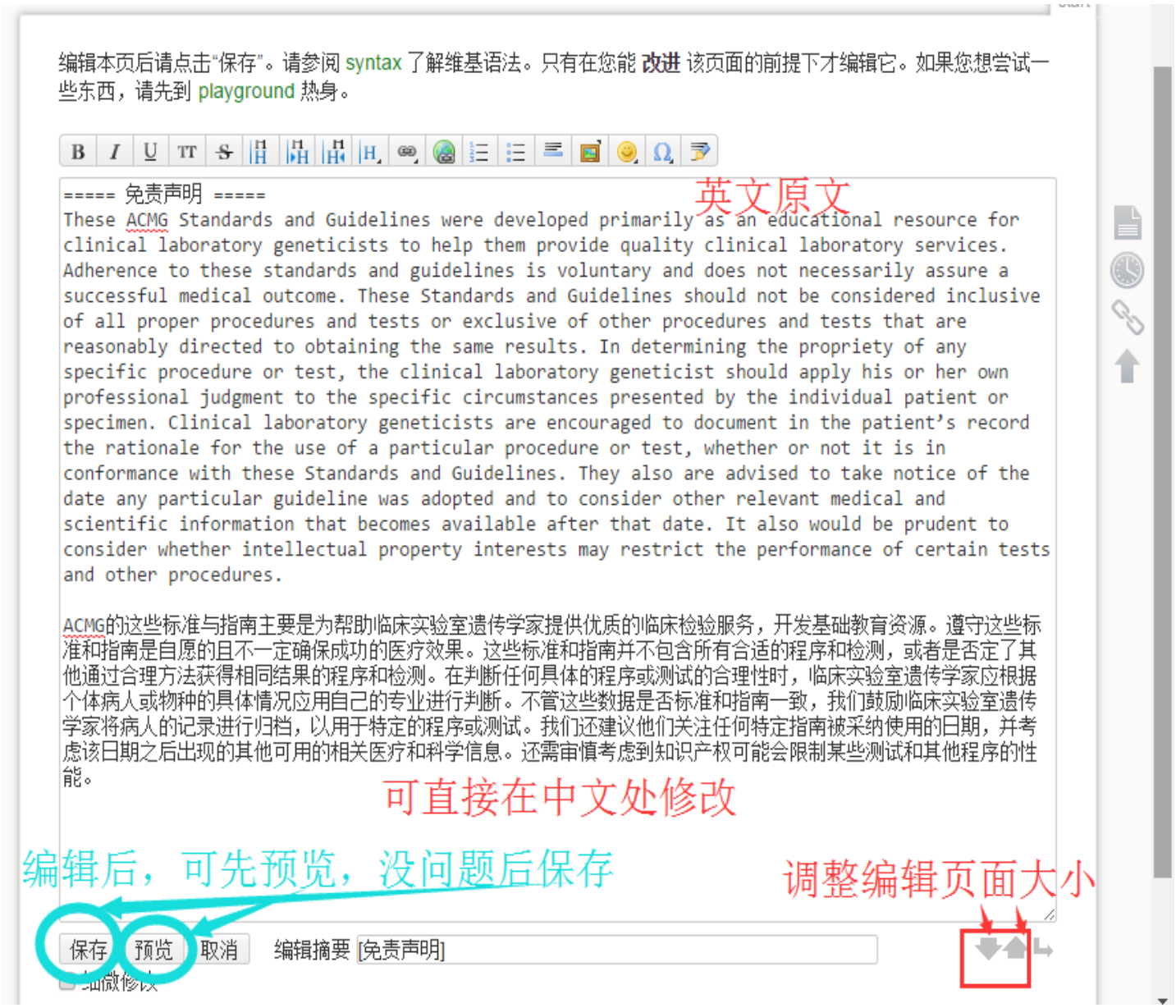


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In November 2013 the workgroup held a workshop at the AMP meeting with more than 50 attendees, presenting the revised classification criteria and two potential scoring systems. One system is consistent with the approach presented here and the other is a point system whereby each criterion is given a number of points, assigning positive points for pathogenic criteria and negative points for benign criteria, with the total defining the variant class. With an audience-response system, the participants were asked how they would weight each criterion (as strong, moderate or supporting, or not used) during evaluation of variant evidence. Again, the responses were incorporated into the classification system presented here. It should be noted that while the majority of respondents did favor a point system, the workgroup felt that the assignment of specific points for each criterion implied a quantitative level of understanding of each criterion that is currently not supported scientifically and does not take into account the complexity of interpreting genetic evidence.

2013年11月，工作组在AMP会议期间举行了超过50人参加的研讨会，提出了修订后的分类标准和两个评分系统。一个系统与这里介绍的方法是一致的，另一个系统是一个点系统，每一项标准都有一个点数，正点为致病标准，负点为良性标准，根据总点数进行变异分类。参与者使用此系统并进行反馈，回答在评估变异证据过程中他们如何权衡各个标准（如强，中度或支持，或不使用）。参与者的反馈结果经分析后会再次综合到这里介绍的分类系统中。应该但要指出的是，虽然大多数调查对象更倾向于点数评分系统，但本工作组认为，每个标准中具体点数的设置量化了对每个标准的理解，但是这一方法量化指标目前缺乏科学依据，并且没有考虑解读遗传证据解读时的复杂性。

The workgroup also evaluated the literature for recommendations from other professional societies and working groups that have developed variant classification guidelines for wellstudied genes in breast cancer, colon cancer, and cystic fibrosis and statistical analysis programs for quantitative evaluation of variants in select diseases.While those variant analysis guidelines