“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

网页地址栏输入 acmg.cbgc.org.cn

2、登录

输入用户名、密码
3、进入指南中文版全文
4. 进入分段编辑页面

编辑完成后请先预览，没问题后保存。
5、点击“修订记录”，选择两个版本进行比较。
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 weigh 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.

The workgroup also evaluated the literature for recommendations from other professional societies and working groups that have developed variant classification guidelines for well-studied 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...