# Article information:

The illusion of polygenic disease risk prediction - ScienceDirect  
<https://www.sciencedirect.com/science/article/pii/S1098360021016233>

# Article summary:

1. Genetic associations of etiological significance are often interpreted as having predictive significance, leading to the expectation that polygenic risk scores can be used to predict disease and identify those at high risk for preventive intervention.

2. However, odds ratios or hazard ratios derived from polygenic risk scores do not directly indicate the discriminatory value of a screening test, and a strong risk factor may still be a poor screening test.

3. The detection rate for a given false-positive rate is necessary to assess the discriminatory value of a screening test, and the screening performance of current polygenic risk score studies is poor, with only about 15% of cases identified for a false-positive rate of 5%.

# Article rating:

Appears moderately imbalanced: The article provides some useful information, but is missing several important points or pieces of evidence that would be required to present the discussed topics in a balanced and reliable way. You are encouraged to seek a more balanced perspective on the presented issues by exploring the provided research topics and looking at different information sources.

# Article analysis:

该文章提出了一个问题，即基因组医学和医学筛查之间的界面问题。作者认为，基因关联通常被解释为具有预测意义，而实际上这种解释是错误的。作者指出，虽然基因组关联研究已经发现了许多与疾病和良性特征相关的常见DNA变异，但这些结果不能直接用于预测疾病风险。

文章批评了两篇最近广泛报道的论文，这些论文显示了多基因风险评分与一些常见疾病之间的关联。作者认为，这些论文中提出的多基因风险评分可以用于识别高风险人群，并进行预防干预。然而，作者认为这种做法是错误的，因为相对风险比如几率比或危害比并不能直接有效地评估多基因风险评分作为筛查测试的区分能力。

文章指出，在评估筛查测试的区分能力时需要考虑检测率（敏感性）和风险得分截断点等参数。然而，在实践中往往只给出几率比或危害比等相对风险指标，并没有给出具体参数值。此外，文章还指出，一个强风险因素并不一定是一个好的筛查测试，因为几率比或危害比通常只比较风险分布的两个尾部，而忽略了中间区域的人群。

文章提供了一个免费的在线工具（Risk Screening Converter），可以将几率比等相对风险指标转换为相关的筛查性能指标。作者使用这个工具评估了两篇论文中提出的多基因风险评分，并发现其筛查性能非常差。作者认为，在这种情况下，如果干预措施是有效、廉价和安全的，那么最好不要进行先前的测试，而是直接提供干预措施。

总之，该文章提醒人们在解释基因组关联研究结果时应该更加谨慎，并且需要考虑到筛查测试的实际效果。然而，该文章可能存在一些偏见和局限性。例如，作者没有考虑到某些疾病可能具有遗传异质性和环境交互作用等复杂因素。此外，作者也没有探讨如何在实践中平衡个体隐私和公共利益等问题。

# Topics for further research:

* Genetic heterogeneity and gene-environment interactions
* Balancing individual privacy and public interest in genomic screening
* Limitations of using relative risk measures in screening tests
* Importance of considering test performance parameters in screening tests
* Effectiveness
* cost
* and safety of interventions in screening programs
* Ethical and social implications of genomic screening and risk assessment

# Report location:

<https://www.fullpicture.app/item/706d9428ebf729eaf96f06e45deecab2>