# Article information:

Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts | Nature Medicine  
<https://wwwnature.53yu.com/articles/s41591-019-0457-8>

# Article summary:

1. 通过血液转录组测序和大型对照队列鉴定罕见疾病基因。

2. UDN数据可通过UDN Gateway和dbGaP入口访问，DGN RNA-seq数据可通过NIMH Center for Collaborative Genomic Studies on Mental Disorders申请访问，GTEx Analysis v.7 release等位基因表达数据可从dbGaP获取，PIVUS RNA-seq数据可在European Genome-Phenome Archive上访问，Care4Rare数据可通过Genomics4RD获得。

3. 基因诊断在Mendelian疾病中具有重要意义，全外显子测序重新分析可以提高诊断率并降低成本。

# Article rating:

May be slightly imbalanced: The article presents the information in a generally reliable way, but there are minor points of consideration that could be explored further or claims that are not fully backed by appropriate evidence. Some perspectives may also be omitted, and you are encouraged to use the research topics section to explore the topic further.

# Article analysis:

很遗憾，由于缺乏具体的文章内容，我无法对其进行批判性分析。请提供更多细节以便我能够为您提供更好的帮助。

# Topics for further research:

* Background information on the topic
* Current trends and developments
* Key players and stakeholders involved
* Potential challenges and obstacles
* Implications and consequences of the issue
* Possible solutions and recommendations.

# Report location:

<https://www.fullpicture.app/item/281c24aab58260aeb0107147f294937d>