Perspective
Volume 13:02, 2025

Journal of Clinical & Medical Genomics

ISSN: 2472-128X

Open Access

Polygenic Risk Scores and their Relationship to Trait Heritability in Population Health

Agosto Schneider*

Department of Pediatrics and Neurology, University of Southern California, Los Angeles, Canada

Introduction

Polygenic Risk Scores (PRS) have become a key tool in the field of genomics and epidemiology, offering a way to predict an individual's genetic predisposition to complex diseases and traits. By aggregating the effects of multiple genetic variants across the genome, PRS provide a measure of an individual's genetic risk, based on the cumulative influence of thousands or even millions of Single Nucleotide Polymorphisms (SNPs). The potential applications of PRS are vast, ranging from personalized medicine and targeted interventions to improving population health strategies. However, a critical aspect of their utility lies in understanding their relationship to the heritability of traits in populations. Heritability refers to the proportion of the variation in a particular trait or disease that can be attributed to genetic differences within a population. Examining how PRS correlate with heritability can provide valuable insights into the genetic architecture of diseases, the extent to which genetic factors influence individual health, and the ways in which PRS can be used in clinical and public health settings [1].

Description

Heritability is a concept that has been central to genetics since the early 20th century. It represents the degree to which genetic variation contributes to phenotypic variation in a population. For complex traits, such as height, blood pressure, and susceptibility to diseases like cardiovascular conditions, diabetes, and psychiatric disorders, heritability estimates are typically derived from twin, family, and adoption studies. These studies compare the phenotypic similarity between individuals with varying degrees of genetic relatedness, helping to quantify the genetic contribution to the trait. However, traditional heritability studies often have limitations, particularly for traits influenced by many small genetic effects. In the past, such studies were less effective at uncovering the role of the vast number of genetic variants that contribute to complex diseases [2].

The advent of Genome-Wide Association Studies (GWAS) marked a significant turning point in our understanding of the genetic basis of complex traits. By scanning the entire genome for associations between specific genetic variants and traits, GWAS have identified thousands of loci associated with a wide range of diseases. However, while these studies have been instrumental in pinpointing genetic risk factors, they have also revealed that most genetic variants associated with complex diseases have only small individual effects. This means that while genetic factors contribute to disease risk, environmental and lifestyle factors also play a significant role in determining disease outcomes. Moreover, GWAS have typically focused on identifying specific SNPs associated with a trait, without providing a comprehensive measure of an individual's overall genetic risk. This is where PRS come into play [3]. Polygenic risk scores aggregate the contributions of numerous genetic variants, each contributing a small effect, to generate a single, composite score that can predict an individual's genetic predisposition to a trait or disease. The development of PRS is based on the results of GWAS, with each genetic variant included in the score being weighted by its effect size, as determined by the GWAS. For example, in the case of cardiovascular disease, a PRS might include thousands of genetic variants that have been associated with factors like cholesterol levels, blood pressure, and other markers of heart disease. By summing the effects of these variants, the PRS can provide an estimate of an individual's genetic risk for developing cardiovascular conditions. This approach has proven to be useful in research settings, particularly for understanding the genetic contributions to population-level variation in disease risk [4].

The relationship between polygenic risk scores and heritability is a complex one. On the one hand, PRS provide a way to quantify the genetic risk for a particular trait, helping to clarify the extent to which genetic factors contribute to the overall heritability of that trait. Since heritability is a population-level measure, PRS allow for the estimation of an individual's genetic risk relative to the broader population. However, the relationship between PRS and heritability is not always straightforward. One key challenge in using PRS to predict heritability is that the scores typically explain only a portion of the genetic variance for a trait. While PRS can capture the effects of common genetic variants, they do not account for rare variants or the complex interactions between genes and the environment. As a result, the heritability explained by PRS may be lower than the total heritability of the trait, which is derived from twin studies or family studies [5].

Conclusion

In conclusion, polygenic risk scores represent a powerful tool for understanding the genetic basis of complex traits and diseases, offering insights into the heritability of these traits at both the individual and population levels. While PRS have the potential to revolutionize personalized medicine and population health strategies, their relationship to heritability is not without challenges. The effectiveness of PRS in predicting disease risk depends on factors such as the heritability of the trait, the genetic architecture of the population, and the diversity of the population studied. Nonetheless, as genomic research continues to advance and as more diverse populations are included in genetic studies, the potential for PRS to improve public health outcomes becomes increasingly clear. By providing a more comprehensive understanding of genetic risk, PRS can help guide personalized prevention and treatment strategies, leading to better health outcomes for individuals and communities worldwide.

Acknowledgment

None.

Conflict of Interest

None.

***Address for Correspondence:** Agosto Schneider, Department of Pediatrics and Neurology, University of Southern California, Los Angeles, Canada; E-mail: schne.agosto@gmail.com*

Received: 01 April, 2025, Manuscript No. JCMG-25-165765; **Editor assigned:** 03 April, 2025, Pre QC No. P-165765; **Reviewed:** 17 April, 2025, QC No. Q-165765; **Revised:** 22 April, 2025, Manuscript No. R-165765; **Published:** 29 April, 2025, DOI: 10.37421/2472-128X.2025.13.335

References

1. Pi-Sunyer, Xavier. "The medical risks of obesity." *Postgrad Med* 121 (2009): 21-33.
2. Malik, Vasanti S., Walter C. Willet and Frank B. Hu. "Nearly a decade on—trends, risk factors and policy implications in global obesity." *Nat Rev Endocrinol* 16 (2020): 615-616.
3. Wen, Wanqing, Yoon-Shin Cho, Wei Zheng and Rajkumar Dorajoo, et al. "Meta-analysis identifies common variants associated with body mass index in east Asians." *Nat Genet* 44 (2012): 307-311.
4. Monda, Keri L., Gary K. Chen, Kira C. Taylor and Cameron Palmer, et al. "A meta-analysis identifies new loci associated with body mass index in individuals of African ancestry." *Nat Genet* 45 (2013): 690-696.
5. Phulka, Jobanjit S., Mishal Ashraf, Beenu K. Bajwa and Guillaume Pare, et al. "Current state and future of polygenic risk scores in cardiometabolic disease: A scoping review." Circulation: Genomic and *Precision Medicine* 16 (2023): 286-313.

How to cite this article: Schneider, Agosto. "Polygenic Risk Scores and their Relationship to Trait Heritability in Population Health." *J Clin Med Genomics* 13 (2025): 335.