

Guidance for Phase III of the Genomics Curriculum

www.GenomicMedicineInitiative.org

For your disease-gene association of choice, answer these questions:

1. Which of the three common Genome-Wide Association (GWA) study designs was used?
2. How were study participants selected and what measures were taken to avoid common problems? How many participants were included in the study?
3. Which technology platform was used for genotyping? How many SNPs were included and what was their origin (e.g. HapMap I, HapMap II and/or other)?
4. Were the results replicated? If so, how? Were any functional studies performed and, if so, what were they?

Required Reading

The NEW ENGLAND JOURNAL of MEDICINE

REVIEW ARTICLE

CURRENT CONCEPTS

Genomewide Association Studies and Human Disease

John Hardy, Ph.D., and Andrew Singleton, Ph.D.

Human genetic variation and its contribution to complex traits

Kelly A. Frazer, Sarah S. Murray, Nicholas J. Schork and Eric J. Topol

Abstract | The last few years have seen extensive efforts to catalogue human genetic variation and correlate it with phenotypic differences. Most common SNPs have now been assessed in genome-wide studies for statistical associations with many complex traits, including many important common diseases. Although these studies have provided new biological insights, only a limited amount of the heritable component of any complex trait has been identified and it remains a challenge to elucidate the functional link between associated variants and phenotypic traits. Technological advances, such as the ability to detect rare and structural variants, and a clear understanding of the challenges in linking different types of variation with phenotype, will be essential for future progress.

JAMA[®]

Online article and related content
current as of January 31, 2010.

How to Interpret a Genome-wide Association Study

Thomas A. Pearson; Teri A. Manolio

JAMA. 2008;299(11):1335-1344 (doi:10.1001/jama.299.11.1335)

<http://jama.ama-assn.org/cgi/content/full/299/11/1335>