

## Genomic Opportunities for Studying Sickle Cell Disease

Griffin P. Rodgers, M.D., M.A.C.P.

Director

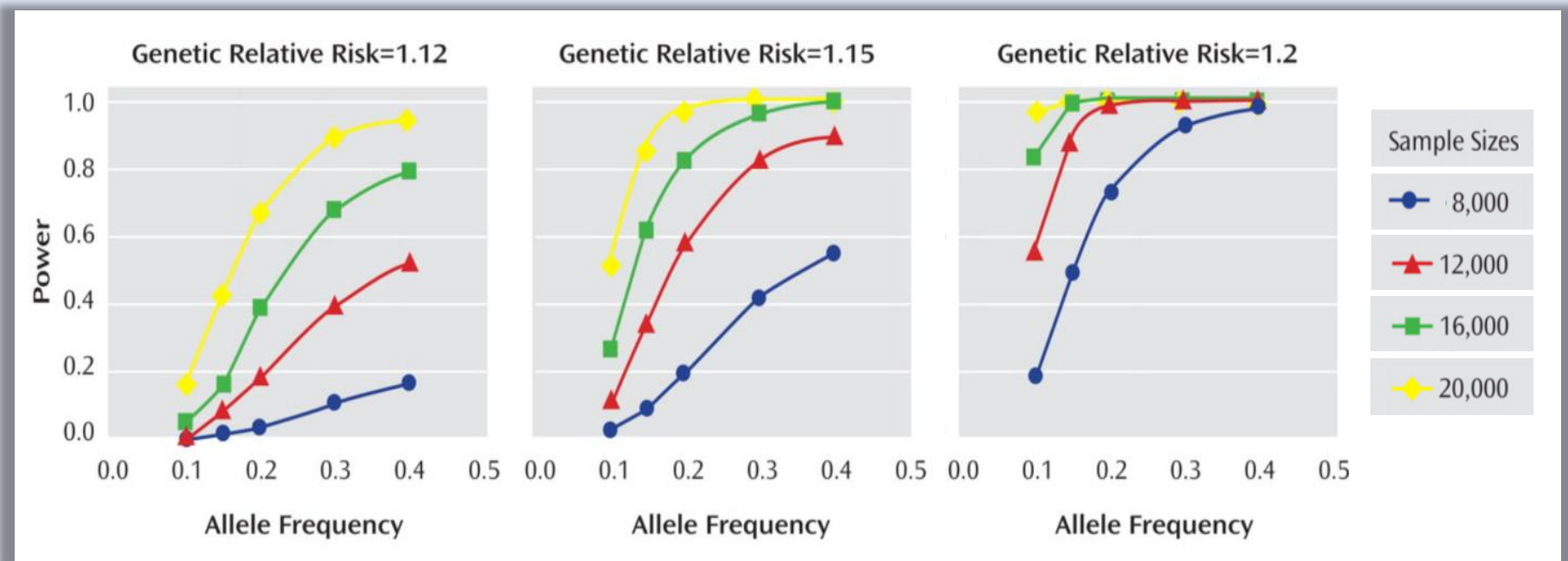
National Institute of Diabetes and Digestive and Kidney Diseases

December 8-9, 2011

# Finding Modifier Genes for Single Gene Traits May Require Large Cohorts

The study of modifier genes is crucial to understanding why patients with the same mutation often have widely varying clinical courses, as in SCD.

As with all complex traits, having a large sample size is crucial to find lower frequency alleles with modest effects.



# Large Cohorts in GWAS

There are several NIDDK consortia assembling large cohorts for important GWAS studies, including some with recent successes:

## Type 1 Diabetes Genetics Consortium (T1DGC)

- 7,514 cases and 9,045 controls

## Inflammatory Bowel Disease (IBD) Genetics Consortium

- 3,230 cases and 4,829 controls

## Chronic Kidney Disease (CKDGen)

- 67,093 individuals including 5,807 individuals with CKD

## Kidney disease in Type 1 Diabetics (GENIE)

- 1,726 cases

# Lessons Learned

Assembling large cohorts is difficult, especially if it involves aggregating existing collections. It is very important to consider:

## Phenotype(s)

- Finding the right phenotype is critical for gene discovery

## Logistical challenges

- Availability of samples and data in usable and sharable forms

## Ethical challenges

- Consents and IRB approvals may differ widely among cohorts

## Resource building

- How can the collection remain useful for subsequent analyses?

# NIDDK Support of Sickle Cell Disease Research

The importance of the initiative being planned is that it aims to identify new genes that play a role in the clinical manifestations of SCD.

The NIDDK looks forward to helping support the difficult follow-on work that will investigate each new gene of interest in the context of normal and SCD physiology.

NIDDK  
NIDDK

NATIONAL INSTITUTE OF  
DIABETES AND DIGESTIVE  
AND KIDNEY DISEASES

AND KIDNEY DISEASES  
DIABETES AND DIGESTIVE  
AND KIDNEY DISEASES

---

NIDDK

NATIONAL INSTITUTE OF  
DIABETES AND DIGESTIVE  
AND KIDNEY DISEASES

