GSP Meeting
4.13.2016

Genomes

No/low power for noncoding regions of the genome; Need larger numbers for power to detect coding variants or more $, less cost, "severe" concentration eg 2 studies

Resources:

- analysis methods development
- common controls
- imputation reference (particularly for non-European Ancestry).

Drive cost decreases?

What would help make this work?
By how much?
When might it be practical?
eg,

- extremely well phenotyped or multiply-phenotyped samples (eg w/EMR);
- endophenotypes;
- complementary studies;
- imputation (#of samples w/GWAS);
- functional imputation; prospect for disruptive analysis methods (pathways, networks other func. data); tech change (reduced coverage),

How many diseases could we explore?

Exomes

Larger sample size for $ leads to higher power for coding variants. Expect to have power for multiple examples of (coding) variants for important diseases

Heavy focus on discovery for individual diseases. Is this goal new? Seems unambitious.

What more *positive* can we say towards program goals?

Does this assume a somewhat monotonic design?

What effect on CCDG centers?
X10 ok for exomes?

What effect on AC's? CMGs?

What effect on SV detection?

Do we have enough samples available?

What happens to common controls?

How many diseases could we explore?

High-level:

How should we weight the ability to demonstrate the program can find disease variants, vs other points of the program, eg., stimulating the next generation of work on WGS (analysis, costs, technology), and developing resources (ie common controls). Is there a path that does both?

Can we imagine an extreme example— WGS but limited to two phenotypes (50-75K samples each over 4 years).