Nussbaum

• Phenotyping. It is unclear to me what is meant by Adult and Pediatric sites pursuing entirely separate “paths for phenotyping”. What is meant by “path” here?

• The Informed consent slide does not say what the parents/child are being consented for? Does it differ between studies? What is the plan if actionable findings occur for adult onset disease in a child? Removal of pediatric data once subjects become adults?
Pediatric Reactor Slide 2

• Fact the adult and pediatric phenotypes are different is described as obstacle. Why an obstacle? Why not a strength?
• TPMT genotype imputation – has it been linked to patient response? How many have been exposed to 6TG?
Pediatric Reactor Slide 3

• Slide 17 - DGV is a valuable resource. How about ISCA?
• Slide 23 - Is the 83% concordance between sequencing and imputed haplotypes what is to be expected? Are there ethnic differences in success of imputation?