Stacey Bolk Gabriel, Ph.D.

[redacted]

Research and Management Experience

Broad Institute of MIT and Harvard (formerly Whitehead Institute Center for Genome Research)
11/98 - Present

Director, Genomics Platform, Broad Institute of Harvard and MIT, 5/2012 - Present

- Direct the Broad Institute's largest platform, and the largest US genome center, comprising 180 people dedicated to all sample handling, microarray, genotyping, and sequencing activities.
- Responsible for $90M annual budget for genomic activities.
- Oversee project management and data analysis activities, primarily in support of cancer and medical genetics.
- Oversee technology development and evaluation and implementation of new technology platforms.
- In addition, maintain all leadership activities listed below (as Program Co-Director)

Co-Director, Genome Sequence and Analysis Program and Medical and Population Genetics Program, Broad Institute of Harvard and MIT, 1/2009 – 5/2012

- Responsible for planning, execution and delivery of a portfolio of cancer and medical sequencing projects as part of the NHGRI Large-scale sequencing grant (PI, Lander) with a budget total of ~$40M annually.
- At most recent Large-scale sequencing grant renewal named Co-PI with Eric Lander.
- As Principle Investigator (PI), secured over $100M in other NIH awards over a period of 5 years aimed at large scale genotyping and sequencing.
- Direct the activity of cross-disciplinary teams, totaling 60 people, including project managers, analysts, computational biologists and software engineers in the analysis of next-gen sequence data as applied to an array of cancer genomics and medical genetics projects.
- Serve as co-chair of Data Production committee for the International 1000 Genome Project, as well as serving as a member of the Executive and Steering committee for The Cancer Genome Atlas.

Director, Genetic Analysis Platform, Broad Institute of Harvard and MIT, 5/2004 – 1/2009

- Responsible for creating, scaling and directing the Genetic Analysis Platform of the Broad Institute. The platform encompasses all production and data management activities related to nucleic acid analysis (gene expression, genotyping, re-sequencing).
- At peak period (2006-2008) operated the platform with yearly revenue of $45 million, overseeing a staff of 65 individuals (project managers, research scientists, software engineers and computational biologists).
- Key milestones included producing microarray data on over 1000,000 DNA samples over an 18 month period.
- Directed data production for over 50 publications describing Genome-wide Association findings.

Associate Director, High throughput biology, Medical and Population Genetics Program, Whitehead Institute Center for Genome Research, 5/2003 - 5/2004

- In this role spearheaded the expansion of SNP genotyping activity from targeted activity for Human HapMap project to a centralized technology platform with dedicated activity in technology development, large-scale production, data management and analysis.
- Oversaw successful completion of the Whitehead's contribution to the Human Hap Map project ($10 M project).

Scientific Director, SNP genotyping / HapMap Program, Whitehead Institute Center for Genome Research, 2/2002 – 5/2003

- Responsible for all aspects of the center's contribution to the International HapMap Project.
- Oversaw a team of 15 technicians, analysts and software engineers.
- Played active role in project design and quality control.
- Served on International HapMap project Steering committee.

Research Scientist, Program in Medical and Population Genetics, Whitehead Institute Center for Genome Research, 11/98 – 2/2002

- Investigated the haplotype structure of the human genome. Research resulting in first author Science publication, widely regarded as laying the foundation for the Human HapMap project.
- Technical development and implementation of the first platforms to be used for large high throughput SNP genotyping

Graduate Student, Department of Genetics, Case Western Reserve University, Cleveland, OH, 1/94– 11/98
Advisor: Aravinda Chakravarti, Ph.D.

- Dissertation "Genetic Dissection of Complex Human Disease"

Research Assistant, Department of Human Genetics, Chakravarti Lab, University of Pittsburgh, Pittsburgh, PA, 5/92-1/94

- Conducted lab work and phlebotomy in support of human genetic studies

Education and degrees

Doctor of Philosophy, Genetics. Case Western Reserve University, Department of Genetics, 1998.
Dissertation: Hirschsprung disease: Genetic dissection of a complex phenotype. Advisor: Aravinda Chakravarti.

Bachelor of Science, Molecular Biology. Carnegie Mellon University, Pittsburgh, PA, 1993.

Peer Review and other Extramural Activities

February 2007-present	External Advisory Committee, NHLBI Resequencing and Genotyping Service
March 2008-present	External Advisory Committee, USC Epigenome Center
November 2008-2009	Editorial Board, Human Genetics
February, 2009	Genomics, Computational Biology and Technology, Ad Hoc reviewer
March 2009	Editorial Board, Genome Research
July, 2009	NHLBI Special Emphasis Panel
July, 2009-present	Genomics, Computational Biology and Technology, Study section, Standing member
January 2010 - present	Genome Canada Science and Industry Advisory Committee, Member

August 2011 – present Scientific Advisory Board, PAAR4KIDS, St. Jude Research Hospital

Publications

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BOLK, S., M. ANGRIST, J. XIE, M. YANAGISAWA, J. M. SILVESTRI *et al.*, 1996 Endothelin-3 frameshift mutation in congenital central hypoventilation syndrome. Nat Genet **13:** 395-396.

BOLK, S., A. PELET, R. M. HOFSTRA, M. ANGRIST, R. SALOMON *et al.*, 2000 A human model for multigenic inheritance: phenotypic expression in Hirschsprung disease requires both the RET gene and a new 9q31 locus. Proc Natl Acad Sci U S A **97:** 268-273.

BOLK, S., E. G. PUFFENBERGER, J. HUDSON, D. H. MORTON and A. CHAKRAVARTI, 1999 Elevated frequency and allelic heterogeneity of congenital nephrotic syndrome, Finnish type, in the old order Mennonites. Am J Hum Genet **65:** 1785-1790.

BUSTAMANTE, M., F. ROGER, M. L. BOCHATON-PIALLAT, G. GABBIANI, P. Y. MARTIN *et al.*, 2003 Regulatory volume increase is associated with p38 kinase-dependent actin cytoskeleton remodeling in rat kidney MTAL. American journal of physiology. Renal physiology **285:** F336-347.

DE BAKKER, P. I., R. YELENSKY, I. PE'ER, S. B. GABRIEL, M. J. DALY *et al.*, 2005 Efficiency and power in genetic association studies. Nat Genet **37:** 1217-1223.

FREEDMAN, M. L., D. REICH, K. L. PENNEY, G. J. MCDONALD, A. A. MIGNAULT *et al.*, 2004 Assessing the impact of population stratification on genetic association studies. Nat Genet **36:** 388-393.

GABRIEL, S., and L. ZIAUGRA, 2004 SNP genotyping using Sequenom MassARRAY 7K platform. Current protocols in human genetics / editorial board, Jonathan L. Haines ... [et al.] **Chapter 2:** Unit 2 12.

GABRIEL, S. B., R. SALOMON, A. PELET, M. ANGRIST, J. AMIEL *et al.*, 2002 Segregation at three loci explains familial and population risk in Hirschsprung disease. Nat Genet **31:** 89-93.

GABRIEL, S. B., S. F. SCHAFFNER, H. NGUYEN, J. M. MOORE, J. ROY *et al.*, 2002 The structure of haplotype blocks in the human genome. Science **296:** 2225-2229.

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SCHAFFNER, S. F., C. FOO, S. GABRIEL, D. REICH, M. J. DALY *et al.*, 2005 Calibrating a coalescent simulation of human genome sequence variation. Genome Res **15:** 1576-1583.
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WEESE-MAYER, D. E., S. BOLK, J. M. SILVESTRI and A. CHAKRAVARTI, 2002 Idiopathic congenital central hypoventilation syndrome: evaluation of brain-derived neurotrophic factor genomic DNA sequence variation. Am J Med Genet **107:** 306-310.
WINCKLER, W., S. R. MYERS, D. J. RICHTER, R. C. ONOFRIO, G. J. MCDONALD *et al.*, 2005 Comparison of fine-scale recombination rates in humans and chimpanzees. Science **308:** 107-111.
FRAZER, K. A., D. G. BALLINGER, D. R. COX, D. A. HINDS, L. L. STUVE *et al.*, 2007 A second generation human haplotype map of over 3.1 million SNPs. Nature **449:** 851-861.
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GREENWAY, S. C., A. C. PEREIRA, J. C. LIN, S. R. DEPALMA, S. J. ISRAEL *et al.*, 2009 De novo copy number variants identify new genes and loci in isolated sporadic tetralogy of Fallot. Nat Genet **41:** 931-935.
HOSHIDA, Y., S. M. NIJMAN, M. KOBAYASHI, J. A. CHAN, J. P. BRUNET *et al.*, 2009 Integrative transcriptome analysis reveals common molecular subclasses of human hepatocellular carcinoma. Cancer research **69:** 7385-7392.
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MUSUNURU, K., J. P. PIRRUCCELLO, R. DO, G. M. PELOSO, C. GUIDUCCI *et al.*, 2010 Exome sequencing, ANGPTL3 mutations, and familial combined hypolipidemia. N Engl J Med **363:** 2220-2227.
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