

**Harvard Medical School
Curriculum Vitae**

Date Prepared: December 23, 2013
Name: Mark S. Boguski, M.D., Ph.D., Chief Medical Officer, Genome Health Solutions, Inc.
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Work FAX: 617-608-2598
Place of Birth: Cleveland, OHIO

Education

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| 1976 | B.A. (Natural Sciences) | The Johns Hopkins University, Baltimore, MD |
| 1986 | M.D., Ph.D. (Molecular Biology) | Washington University, St. Louis, MO |

Postdoctoral Training

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| 1987-1988 | Resident, Anatomic Pathology | Barnes, Jewish and Childrens Hospitals, St. Louis, MO |
| 1988-1989 | Medical Staff Fellow, Computational Biology | National Institutes of Health, Bethesda, MD |

Faculty Academic Appointments

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|--------------|---|---|
| 1989-1995 | Senior Staff Fellow | National Institutes of Health, Bethesda, MD |
| 1995-2000 | Senior Investigator (tenured) | National Institutes of Health, Bethesda, MD |
| 1995-2000 | Adjunct Professor of Molecular Biology and Genetics | The Johns Hopkins University School of Medicine, Baltimore, MD |
| 2002-2004 | Adjunct Professor of Medicine and Genetics | University of Washington School of Medicine Seattle, WA |
| 2008-present | Research Associate in Biomedical Informatics | Center for Biomedical Informatics, Harvard Medical School, MA |
| 2008-present | Associate Professor of Pathology | Harvard Medical School and Beth Israel Deaconess Medical Center, MA (on leave 2013) |

Other Professional Positions

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| 1995-1998 | Commissioned Officer, Rank O-5 (Commander) | U.S. Public Health Service, Bethesda, MD |
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| 2000-2001 | Senior Vice President of Research and Development | Rosetta Inpharmatics, Kirkland, WA (acquired by Merck & Co. 2001) |
| 2001-2002 | Visiting Investigator | Fred Hutchinson Cancer Research Center, Seattle, WA |
| 2002-2004 | Founding Director | Allen Institute for Brain Science, Seattle, WA |
| 2005-2007 | Vice President and Global Head, Genome and Proteome Sciences | Novartis Institutes for Biomedical Research, Cambridge, MA and Basel, Switzerland |
| 2013-present | Founder and Chief Medical Officer | Genome Health Solutions, Inc. |

Major Administrative Leadership Positions

National and International

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| 2000-2001 | Senior Vice President, Research and Development | Rosetta Inpharmatics, Kirkland, WA (acquired by Merck & Co., 2001) |
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- Managed a group of 90 employees consisting of experimental biologists, physicists, mathematicians, engineers, software and database developers
- One of four executive officers responsible for all aspects of the company, including the planning and execution of a \$114 million initial public offering in August, 2000 and Rosetta's acquisition by Merck & Co. in August 2001
- Worked closely with Business Development on planning, negotiation and execution of collaborative research transactions up to \$150 million

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| 2002-2004 | Founding Director | Allen Institute for Brain Science |
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- Conducted feasibility studies, detailed financial and scientific planning; identified and launched inaugural (Brain Atlas) project
- Formed a Scientific Advisory Board; identified, recruited and hired key staff in neuroanatomy, informatics, genomics, automation engineering, finance and operations
- Identified and transferred or in-licensed key enabling technologies
- Located, established and supervised initial build-out of research facility
- Secured IRS status for the Institute as a Medical Research Organization
- Lobbied on Capitol Hill and at NIH for Federal support
- Designed and executed the formal launch plan and represented the Institute to the popular press and scientific and medical communities

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| 2005-2007 | Vice President and Global Head, Genome and Proteome Sciences | Novartis Institutes for Biomedical Research, Cambridge, MA and Basel, Switzerland |
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- Managed a global research division with an annual budget of \$100.8 million and 314 employees in four countries
- Created a new research and technology division by merging two preexisting departments spanning automated, high-throughput biology, functional genomics, proteomics, research computing, statistics, knowledge management, data mining and competitive intelligence
- Managed numerous internal collaborations with Disease Areas, Translational Medicine and Early Clinical Development and external collaborations with biotechnology companies and academic centers

Committee Service

Local

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|-----------|---------------------------|------------------|-----------------------|
| 1998-1999 | Scientific Advisory Board | Founding Advisor | Ardais Corporation |
| 2008-2011 | Scientific Advisory Board | Advisor | Gene Network Sciences |
| 2009-2011 | Scientific Advisory Board | Advisor | Genome Quest |

Regional

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| 1998-2000 | Scientific Advisory Board | Advisor & Reviewer | Merck Genome Research Institute |
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National and International

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| 1993-1998 | Scientific Advisory Board | Advisor | Sequana Therapeutics |
| 1996-2001 | Genome Research Review Committee | Reviewer | National Human Genome Research Institute |
| 1996-1998 | Steering Committee, Cancer Genome Anatomy Project | Member | National Cancer Institute |
| 1997-1999 | Genetics Advisory Group | Advisor | The Wellcome Trust |
| 1998-2000 | Scientific Advisory Board | Advisor | Rosetta Inpharmatics, Inc. |
| 1998-2000 | Scientific Advisory Board | Advisor | Orion Genomics LLC |
| 2001-2004 | Genome Resources and Sequencing Priorities Panel | Advisor & Reviewer | National Human Genome Research Institute |
| 2002-2003 | Scientific Advisory Board | Advisor | Vialactia Biosciences |
| 2003-2008 | Council of Scientific Advisors | Advisor | Translational Genomics Institute |
| 2003-2008 | Scientific Advisory Board | Advisor | Keystone Symposia |
| 2010-2011 | Personalized Healthcare Committee | Member | College of American Pathologists |
| 2010-2011 | Emerging Technologies Committee | Member | College of American Pathologists |
| 2010-2011 | New Business Models Committee | Member | College of American Pathologists |
| 2011 | External Advisory Board | Member | M.D. Anderson Cancer Center |
| 2011 | Planning Committee, Interest Group 9 | Member | Institute of Medicine |
| 2013 | Diagnostic Intelligence and Health Information Technology Committee | Member | College of American Pathologists |

Professional Societies

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|--------------|---|----------------|
| 2001-present | Institute of Medicine of the National Academies | Elected member |
| 2001-present | American College of Medical Informatics | Elected member |

Editorial Activities

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| 1996 | Editor, special issue on Genomes & Evolution | Current Opinion in Genetics & Development |
| 1996-1999 | Editor | Genome Research |
| 2000-2001 | Board of Reviewing Editors | Science |
| 2003-2007 | Editor-in-Chief | GENOMICS |

Honors and Prizes

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| 1985 | Gerty T. Cori Predoctoral Fellowship and Prize |
| 1996 | Regents Award from the National Library of Medicine for “Scholarly and Technical Contributions to the Construction of the Human Transcript Map” |
| 1998 | NIH Director's Award for contributions to the "NCI Tumor Gene Index Project" |
| 2001 | One of the Inaugural “All-Stars” (in the fields of Bioinformatics and Gene Expression) elected by the readers of Genome Technology magazine |
| 2006 | " Pioneer of Proteomics " U.S. National Cancer Institute |
| 2006 | " Visionary and Influencer " The Personalized Medicine Coalition |
| 2006-2007 | Members of my Division of Genome and Proteome Sciences at Novartis received the following awards: <ul style="list-style-type: none">• VIVA (Vision, Innovation, Value, Achievement) awards (2006, 2007)• Team Innovation Awards (2006, 2007)• Global Information Technology Innovation Award (2007) |

Report of Funded and Unfunded Projects

Funding Information

Past

| | | | |
|-----------|---|--------------------------|--|
| 1988-2000 | NIH Intramural Program | PI | Computational biology |
| 2000-2001 | Venture capital, then public stock issued by Rosetta Inpharmatics | R&D Director | Microarray technology development & applications |
| 2001-2002 | Personal donation by Craig Mundie, CTO of Microsoft Corporation | PI | Proteomics and informatics development program |
| 2002-2004 | Paul G. Allen Foundation | Research Director | Allen Brain Atlas |
| 2005-2007 | Novartis Institutes for Biomedical Research | Vice President, Research | Genome and proteome sciences |
| 2009 | Harvard Catalyst Pilot Grant | PI | Drug Repurposing |

Report of Local Teaching and Training

Teaching of Students in Courses

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| 1990 | Course and Workshop in Computational Biology (Course Director and Lecturer) National Center for Biotechnology Information, NLM, NIH Postdoctoral fellows and professional staff, ~40 attendees |
| 1995-1999 | Bioinformatics section of annual course in Molecular Biology (Lecturer) The Johns Hopkins University School of Medicine, Department of Molecular Biology and Genetics. Graduate students, ~15 enrolled per year |
| 1996-1999 | Current Topics in Genome Analysis (Course co-Director and Lecturer) National Human Genome Research Institute, NIH. Postdoctoral and medical staff fellows, ~200 attendees per year |
| 2009-2011 | Genomic Medicine Training Initiative (Course Director and Lecturer) BIDMC Department of Pathology. Pathology Residents, 12-15 residents per year |

Formal Teaching of Residents, Clinical Fellows and Research Fellows (post-docs)

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| 1993-1994 | Todd Lowe, Ph.D. | <u>Assistant Professor, Department of Biomedical Engineering</u> , University of California, Santa Cruz |
| 1994-1997 | Douglas Bassett, Ph.D. | Executive Director and co-Site Head, Rosetta Inpharmatics/Merck Research Laboratories, Seattle, WA |
| 1995-2000 | Wohciech Makalowski, Ph.D | Professor & Head, Institute for Bioinformatics, University of Muenster, Germany |
| 1996-2000 | Oxana Pickeral, Ph.D. | Senior Associate, Booz Allen Hamilton, McLean, VA, USA |
| 1996-2000 | Jiong Zhang, Ph.D. | Atherosclerosis Research Unit, Children's Hospital Research Institute, Oakland, CA |
| 1996-2000 | Peter Kuehl, M.D., Ph.D. | Pulmonary and Critical Care Associates of Baltimore, Union Memorial Hospital |
| 1997-1998 | Olga Ermolaeva, Ph.D. | Staff Scientist, National Center for Biotechnology Information, NIH, Bethesda, MD |
| 1997-1998 | Kim Pruitt, Ph.D. | Staff Scientist and <u>RefSeq</u> Project Supervisor, National Center for Biotechnology Information, NIH, Bethesda, MD |
| 1998-2000 | Stephen Edwards, Ph.D. | Systems Biologist, National Health and Environmental Effects Research Laboratory, Research Triangle Park, NC |
| 1998-2000 | Hagit Shatkay, Ph.D. | <u>Assistant Professor of Computer Science</u> , Queen's University, Kingston, Ontario |
| 1999-2000 | Suzanne Szak, Ph.D. | Molecular Profiling Analysis Group, Biogen Idec, Cambridge, MA |

Report of Regional, National and International Invited Teaching and Presentations

Invited Presentations and Courses

Regional

- 1985 Structural Relations among the Mammalian Apolipoproteins. University of Chicago Pritzker School of Medicine, Department of Medicine, Chicago, IL October 4
- Molecular Systematics of the Apolipoproteins and their Genes. Baylor College of Medicine, Department of Medicine, Houston, TX February 21
- Evolution of the Apolipoproteins. University of California at Los Angeles, Molecular Biology Institute, Los Angeles, CA October 24
- 1986 On Computer-assisted Analysis of Biological Sequences. University of Texas Health Science Center, Department of Molecular Genetics, Dallas, TX August 1
- 1989 The Importance of Repeated Sequences in Protein Structure and Evolution. University of Michigan Medical Center, Department of Pathology and Howard Hughes Medical Institute, Ann Arbor, MI November 30
- 1990 Baroque Periodic Proteins and the Music of the Spheres. Washington University, Department of Biochemistry and Molecular Biophysics, St. Louis, MO May 3
- Biosequence Databases: Concepts, Contents and Applications. Institute for Defense Analysis, Supercomputer Research Center, Laurel, MD December 19
- 1991 Adventures in Information Space: Computers, Databases and the New Biology. Washington University, Department of Pharmacology and Molecular Biology, St. Louis, MO November 7
- The Importance of Repetitive Sequences in Protein Structure, Function and Evolution. Center for Bio/Molecular *Science* and Engineering, U.S. Naval Research Laboratory, Washington, D.C. February 7
- 1992 The Development and Uses of a New Database of Expressed Sequence Tags. Symposium on Sequence Analysis of Nucleic Acids and Proteins. University of Rochester School of Medicine. Rochester, NY October 29
- Sequence Homologies and Motifs Among Proteins that Regulate Ras-like GTPases. Department of Computer Science and the Institute for Molecular Evolutionary Genetics. Pennsylvania State University, University Park, PA. November 11
- 1993 Adventures in Information Space: Biomedical Discoveries in Sequence Databases. Countway Library of Medicine, Harvard University School of Medicine, Boston, MA April 2
- How to make discoveries in molecular sequence databases. Department of Biomathematics, University of Texas M.D. Anderson Cancer Center, Houston, TX, November 9
- GenBank and other computational resources at the U.S. National Center for Biotechnology Information. Department of Biomathematics, University of Texas M.D. Anderson Cancer Center, Houston, TX November 10
- 1994 Research and Resources at the National Center for Biotechnology Information. University of Colorado Health Sciences Center, Denver, CO February 4
- GenBank Select and a “Chromocentric” View of the Sequence Universe. Department of Molecular Biology and Genetics and Howard Hughes Medical Institute, Johns Hopkins University, Baltimore, MD April 14
- Gene Discovery in NCBI’s Database of Expressed Sequence Tags. Department of Molecular Biotechnology, University of Washington, Seattle, WA June 3
- 1995 The turning point in genome research. Zeneca Pharmaceuticals, Wilmington, DE October 30

The turning point in genome research. Wyeth-Ayerst Research Laboratories, Princeton, NJ
November 30

Adventures in Information Space: GenBank, Genomes, and a Turning Point in Biomedical
Research. University of Louisville School of Medicine, Department of Biochemistry, Louisville,
KY December 4

1996

Adventures in Information Space: GenBank, Genomes, and a Turning Point in Biomedical
Research. University of Minnesota Medical School, Institute of Human Genetics, Minneapolis, MN
February 22

Adventures in Information Space: GenBank, Genomes, and a Turning Point in Biomedical
Research. Department of Molecular Genetics, University of Texas Southwestern Medical Center,
Dallas, TX March 18

Adventures in Information Space: GenBank, Genomes, and a Turning Point in Biomedical
Research. Department of Anatomy and Cell Biology, Temple University School of Medicine,
Philadelphia, PA September 12

Bioinformatics. Discovery/Clinical Symposium on Molecular Medicine. Wyeth-Ayerst Research,
Princeton, NJ December 2

The Design of Bioinformatics Search Engines and Tools to Explore Genome Databases. 21st
Century Biology: Informatics in the Post Genomic Era. Fred Hutchinson Cancer Research Center,
Seattle, WA December 19

Adventures in Information Space: GenBank, Genomes and a Turning Point in Biomedical Research.
Rockefeller University, New York, NY. February 20

Narrowing the Gap between Sequence and Function. A symposium at the New York Academy of
Sciences on "Bioinformatics and the Discovery of Novel Therapeutics." New York, NY. February
25

21st Century Library of Medicine: The Books are Our Genes. A Symposium on "Genomics and
Gene Therapy: Meaning for the Future of Science and Medicine." Harvard Institute of Human
Genetics, Cambridge, MA March 26

Adventures in Information Space: GenBank, Genomes and a Turning Point in Biomedical Research.
Genome Lecture Series. Stanford University School of Medicine, Department of Genetics, Palo
Alto, CA May 30

Closing the Gap between Sequence and Function: Bioinformatics and High-throughput Biology.
University of California at San Francisco, San Francisco, CA May 31

Adventures in Information Space: GenBank, Genomes and a Turning Point in Biomedical Research.
Research Genetics, Huntsville, AL October 2

Adventures in Information Space: GenBank, Genomes and a Turning Point in Biomedical Research.
Affymetrix, Santa Clara, CA October 13

Adventures in Information Space: GenBank, Genomes and a Turning Point in Biomedical Research.
Catholic University, Washington, DC December 1

1998

Adventures in Information Space: GenBank, Genomes and a Turning Point in Biomedical Research.
Emory University Center for Molecular Medicine, Atlanta, GA. March 5

Adventures in Information Space: GenBank and Computational Biology. Boston Museum of
Science Symposium on Biotechnology Education. Boston, MA. March 9

Adventures in Information Space: GenBank, Genomes and a Turning Point in Biomedical Research.
University of Michigan, Department of Genetics. Ann Arbor, MI. March 16

Adventures in Information Space: GenBank, Genomes and a Turning Point in Biomedical Research. Vanderbilt University Careers Symposium. Nashville, TN. May 1

Adventures in Information Space: GenBank, Genomes and a Turning Point in Biomedical Research. Thomas Jefferson High School for Science and Technology. Fairfax, VA. June 4

Comparative Genomics of Rodents and Humans with Applications to QTL Mapping. NIAAA Workshop on QTL Mapping. Rockville, MD. August 21

Adventures in Information Space: GenBank, Genomes and a Turning Point in Biomedical Research. Distinguished Alumni Lecture, Washington University Medical Scientist Training Program, Annual Retreat. Potosi, MO. September 26

Large Scale Gene Expression Technologies: Closing the Gap Between Sequence and Function. Technology Seminar Series, Strategic Technologies Office, National Cancer Institute. Lipsett Auditorium, NIH. Bethesda, MD. October 22

Closing the Gap between Sequence and Function: The Frontier of Computational Biology and Functional Genomics. Medical University of South Carolina. Charleston, S.C. November 9

Adventures in Information Space: GenBank, Genomes and a Turning Point in Biomedical Research. Medical University of South Carolina. Charleston, S.C. November 9

Adventures in Information Space: GenBank, Genomes and a Turning Point in Biomedical Research. St. Jude Children's Research Hospital. Memphis, TN. December 4

Adventures in Information Space: GenBank, Genomes and a Turning Point in Biomedical Research. National Institute of General Medical Sciences, NIH. Bethesda, MD. December 10

1999

Overview and Discussion of the Emerging Field of Bioinformatics. Curriculum Development Workshop of the Quality Education for Minorities (QEM) Network. Spellman College. Atlanta, GA. January 22

Closing the Gap between Sequence and Function: The Frontier of "Functional Genomics." 193rd Meeting of the National Heart, Lung, and Blood Institute Advisory Council. Bethesda, MD. February 4

Closing the Gap between Sequence and Function: The Frontier of "Functional Genomics." Genentech, Inc. South San Francisco, CA. March 3

Closing the Gap between Sequence and Function: The Frontier of "Functional Genomics." Department of Biomedical Engineering, The Johns Hopkins University School of Medicine, Baltimore, MD. March 19

Closing the Gap between Sequence and Function: The Frontier of Computational Biology and Functional Genomics. Department of Genetics, University of Pennsylvania, Philadelphia, PA. April 12

Closing the Gap between Sequence and Function: The Frontier of Computational Biology and Functional Genomics. Symposia on Bioinformatics and Genomics in the 21st Century. University of Texas Medical Branch, Galveston, TX. April 15

"Reflections" on "Functional Genomics: Technology Development & Research Applications." The Banbury Center, Cold Spring Harbor Laboratory, New York. April 28

Bridging the Gap between Genomes and Function: The Frontier of Computational Biology and Functional Genomics. Yale University School of Medicine, New Haven, Connecticut. June 9

Bridging the Gap between Genomes and Function: The Frontier of Computational Biology and Functional Genomics. Advances in Genome Sciences Seminar Series, University of Michigan and Warner-Lambert/Parke-Davis Pharmaceutical Company, Ann Arbor, Michigan. October 11

Bridging the Gap between Genomes and Function: The Frontier of Computational Biology and Functional Genomics. 9th Annual Robert Steel Foundation International Symposium, "Genomics and Human Cancer," Memorial Sloan-Kettering Cancer Center Rockefeller Research Laboratories, New York, N.Y. October 13-15

In Vivo, In Vitro, In Silico: The Convergence of Biotechnology and Information Technology. NHLBI Intramural Staff Retreat, Airlie Center, Virginia. October 18

Higher Bandwidths through the Bottleneck: Semi-automated Approaches to Expression Array Interpretation. Merck Genome Research Institute, DNA Microarray Gene Expression Program, Dakin House, Pennsylvania. October 21

In Vivo, In Vitro, In Silico: The Frontier of Computational Biology and Functional Genomics. Pennsylvania State University Life Sciences Consortium Colloquium, University Park, PA. November 9

In Vivo, In Vitro, In Silico: The Frontier of Computational Biology and Functional Genomics. North Carolina State Genomic Science Seminar Series, Raleigh, NC. November 15

Bridging the Gap between Genomes and Function: The Frontier of Computational Biology and Functional Genomics. Advanced Topics in Molecular Genetics Lecture Series, Center for Drug Evaluation and Research and Development, Food and Drug Administration, Rockville, MD. December 7

2000 Higher Bandwidths through the Bottleneck: Semi-automated Approaches to Expression Array Interpretation. Yale University School of Medicine, New Haven, CT. January 17

In Vivo, In Vitro, In Silico: The Frontier of Computational Biology and Functional Genomics. Princeton University Bioinformatics Symposium, Princeton, NJ. January 19

In Vivo, In Vitro, In Silico: The Frontier of Computational Biology and Functional Genomics. "Biotechnology: the Future is Now," a course at the Smithsonian Institution, Washington, DC. February 9

In Vivo, In Vitro, In Silico: The Frontier of Computational Biology and Functional Genomics. University of Virginia, Charlottesville, VA. February 10

In Vivo, In Vitro, In Silico: The Frontier of Computational Biology and Functional Genomics. The College of Physicians of Philadelphia, Philadelphia, PA. February 17

Higher Bandwidths through the Bottleneck: Semi-automated Approaches to Expression Array Interpretation. Functional Genomics Lecture Series, Whitehead Institute/MIT, Cambridge, MA. February 24

Higher Bandwidths through the Bottleneck: Semi-automated Approaches to Expression Array Interpretation. Memorial Sloan Kettering Cancer Center, New York, NY. March 15

Higher Bandwidths through the Bottleneck: Semi-automated Approaches to Expression Array Interpretation. Chips, SNP and Functional Genomics Lecture Series, Department of Molecular Biology and Genetics, Johns Hopkins Medical Institutions, Baltimore, MD. April 28

Higher Bandwidths through the Bottleneck: Semi-automated Approaches to Expression Array Interpretation. Chemical/Life Sciences Division, Agilent Technologies, Palo Alto, CA. August 2

2001 Experimental annotation of the human genome using microarray technology. University of Washington, Seattle, WA. April 4

Research Challenges and Opportunities in Post-genome Biology. Vanderbilt University, Nashville, TN. October 1

Research Challenges and Opportunities in Post-genome Biology. The Genome Institute of the

- Novartis Foundation, La Jolla, CA. October 31
- Genomics: past, present and future. American Museum of Natural History. New York, New York. November 10
- 2002 Bioinformatics: past, present and future. Department of Genome Sciences, University of Washington. Seattle, WA. January 30
- Bioinformatics and genome sciences. Transcriptome 2002: From functional genomics to systems biology. Seattle, WA. March 10
- Bioinformatics: past, present and future. Institute for Systems Biology Inaugural Symposium. Seattle, WA. March 26
- Bioinformatics and Genome Sciences. University of Pennsylvania. Philadelphia, PA. April 4
- Bioinformatics and Genome Sciences. Indiana University. Indianapolis, IN. April 9
- Bioinformatics and Genome Sciences. Harvard-Partners Center for Genetics and Genomics. Boston, MA. May 9
- Bioinformatics and Genome Sciences. Zymogenetics, Inc. Seattle, WA. May 15
- Pharmacogenomic studies of the Pregnane X Receptor and Its Target Genes. Fred Hutchinson Cancer Research Center. Seattle, WA. June 6
- Bioinformatics: past, present and future. The Salk Institute. La Jolla, CA. September 19
- The Emergence of actual Human Disease as a Model for Discovery Research. Bay Area Clinical Genomics Symposium, San Francisco, CA. October 25
- Bioinformatics: past, present and future. Biomedical Computation Symposium (BCATS), Stanford University, Palo Alto, CA. October 26
- Bioinformatics: past, present and future. Montana State University, Bozeman, MT. November 8
- The Emergence of actual Human Disease as a Model for Discovery Research Washington Biotechnology & Biomedical Association, Seattle, WA. November 13
- 2003 Bioinformatics: past, present and future. Boston University Bioinformatics Graduate Program. Boston, MA. March 27
- 2004 Intersections of Genomics, Bioinformatics and Neuroscience. Stowers Institute, Kansas City, MO. April 23
- 2005 The end of the interlude? Reflections on bioinformatics, proteomics, systems biology and experimental medicine. BioSilico 2005, Cambridge, MA. October 25
- 2006 The end of the interlude? Reflections on bioinformatics, proteomics, systems biology and experimental medicine. MIT Enterprise Forum of Cambridge, Cambridge, MA. February 8
- The end of the interlude? Reflections on bioinformatics, proteomics, systems biology and experimental medicine. IBM Research, Westchester, NY. July 25
- The End of the Interlude: Reflections on Bioinformatics, Proteomics, Systems Biology and Experimental Medicine. Northeastern University, Boston, MA. September 28
- Proteomics, Systems Biology and Knowledge Mining in Drug and Biomarker Discovery. Microsoft Biotechnology Executives Forum. Cambridge, MA. October 19
- Genomics, Proteomics, Systems Biology and Knowledge Mining in Biomedical Research. University of Basel Biozentrum, Basel, Switzerland. October 26
- Proteomics, Systems Biology and Knowledge Mining in Biomedical Research. J.B. Little

- Symposium. Harvard School of Public Health. Boston, MA. November 3
- 2007 Proteomics, Systems Biology and Knowledge Mining in Biomedical Research. Broad Institute, Cambridge, MA. May 2
- Proteomics, Systems Biology and Knowledge Mining in Biomedical Research. Duke University, Institute for Genome Science and Policy, Durham, NC. June 27
- Proteomics, Systems Biology and Knowledge Mining in Drug and Biomarker Discovery. Countway Library of Medicine, Harvard Medical School, Boston, MA. September 11
- Proteomics, Systems Biology and Knowledge Mining in Drug and Biomarker Discovery. Vanderbilt University School of Medicine, Department of Biomedical Informatics, Nashville, TN. November 7
- Proteomics, Systems Biology and Knowledge Mining in Drug and Biomarker Discovery. Department of Pathology, Beth Israel Deaconess Medical Center, Boston, MA. November 9
- Proteomics, Systems Biology and Knowledge Mining in Drug and Biomarker Discovery. University of Pennsylvania School of Medicine, Philadelphia, PA. December 5
- 2008 Proteomics, Systems Biology and Knowledge Mining in Drug and Biomarker Discovery. Princeton University, Lewis-Silger Institute, Quantitative and Computational Biology Seminar Series, Princeton, NJ. February 18
- Proteomics, Systems Biology and Knowledge Mining in Drug and Biomarker Discovery. Stowers Institute for Medical Research. Kansas City, MO. February 21
- Is the World Ready for MyGenomeOnTheWeb.com? Panel Discussion, BioInnovations 2008, MIT Sloan School of Management, Cambridge, MA. April 18
- Online Health Information Retrieval by Consumers and the Challenge of Personal Genomics. Futures Conference on Personalized Medicine. The Gulf Coast Consortia. Houston, TX. June 14
- Clinical and Recreational Genomics: Personal Experiences with Direct-To -Consumer Genotyping. Massachusetts General Hospital, Psychiatric Genetics Program. Boston, MA. September 16
- Combined Pathology Grand Rounds. Brigham and Women's Hospital. Boston, MA. October 6
- 2009 Clinical and Educational Genomics: Personal Experiences with Direct-To -Consumer Genotyping. Roche 454, New Haven, CT. March 11
- Clinical and Educational Genomics: Personal Experiences with Direct-To -Consumer Genotyping. Vertex Pharmaceuticals, Cambridge, MA. July 15
- Customized Care 2020: How Medical Sequencing and Network Biology Will Enable Personalized Medicine. University of Utah School of Medicine. Salt Lake City, UT. November 5
- Personalized Genomic Medicine: A Three-Part Story. Intermountain Health Care, Clinical Genetics Institute. Salt Lake City, UT. November 6
- Clinical and Educational Genomics: Personal Experiences with Direct-To -Consumer Genotyping. Continuing Medical Education Workshop on Personalized Medicine: Are We There Yet? Intermountain Health Care and University of Utah School of Medicine. Salt Lake City, UT. November 7
- How Medical Sequencing and Network Biology will enable Personalized Medicine. Brown University, Center for Computational Molecular Biology Distinguished Lecture Series, Providence, RI. November 11

- 2010 Next Generation Genome Sequencing: Applications for Personalized Medicine and Public Health. Channing Laboratory, Harvard School of Public Health, Boston, MA. February 23
- Bringing Genomic Advances to the Clinic. Summer Scholars Program in Bioinformatics and Integrative Genomics. Harvard Medical School. Boston, MA July 28, 2010.
- Health Communication at the Nexus of Social Media and Popular Culture. Harvard School of Public Health. Boston, MA. February 10, 2011
- 2011 Personalized Genomic Medicine: Technologies, Training and Clinical Applications. Technology Assessment Group, Department of Health Policy and Management, Harvard School of Public Health. Boston, MA. February 24, 2011
- Cancer Genomics and the Impact of Next Generation Sequencing Symposia. University of Pittsburgh Cancer Institute. Pittsburg, PA. March 10, 2011
- Personalized Genomic Medicine: The Future is Now. Life Technologies. Carlsbad, CA. January 18, 2011.
- Participatory Medicine, Social Media & Their Roles in Clinical Trial Recruitment, Product Marketing & Post-Marketing Surveillance. New York Pharma Forum. New York, NY. January 26, 2011
- 2012 Cancer at the Crossroads: Will tumor genome analysis become the new standard of care? Harvard Medical School Longwood Seminars “Mini-Med School” on Cancer Genetics. Boston, MA. April 3, 2012.
- Cancer, Data and the Idolatry of the \$1000 Genome. Grand Rounds, Albert Einstein College of Medicine. Bronx, NY. September 20, 2012
- Pathologists and the Third Wave of Medical Genomics. Montefiore Medical Center. Bronx, NY. September 20, 2012.
- Big Data + Analytics = Precision Diagnosis and Better Outcome. Medical College of South Carolina. Charleston, SC. December 11, 2012.
- 2013 Case Studies in Cancer Genomics. Grand Rounds at Merrimack Valley Hospital, Steward Health Care System. Haverhill, MA. January 23, 2013
- Case Studies in Cancer Genomics: Big Data + Analytics = Better Outcomes. Blood Center of Wisconsin Annual Innovation Retreat. Lake Geneva, WI. May 29, 2013.
- Pathology and the Third Wave of Medical Genomics. 22nd Annual Beaumont Molecular Pathology and Medicine Conference. Oakland University William Beaumont School of Medicine. Troy, MI. September 25, 2013.
- Advances in Precision Diagnostics for Personalized Medicine. From Diagnosis to Delivery: Transformations in Cancer Care. Genentech Regional Symposium. Pittsburgh, PA. September 27, 2013
- Advances in Precision Diagnostics for Personalized Medicine. From Diagnosis to Delivery:

Transformations in Cancer Care. Genentech Regional Symposium. Jersey City, NJ. November 8, 2013

Pathologists and the Third Wave of Medical Genomics. Grand Rounds at Baystate Medical Center. Springfield, MA. November 25, 2013.

National

- 1984 Rat apolipoprotein A-IV nucleotide sequence contains 11 tandem repeat units for a 22-amino acid amphipathic segment. American Society of Biological Chemists 75th Annual Meeting, Minisymposium on Apolipoprotein Molecular Biology, St. Louis, MO June 5
- 1985 Comparative Analysis of Repeated Sequences in Rat HDL-associated Apolipoproteins A-I, A-IV and E. American Heart Association 57th Scientific Sessions, Minisymposium on Apolipoprotein Molecular Biology, Miami, FL November 12
- 1990 The Importance of Repetitive Protein Sequences in Biology and Medicine: Use of Medline as a Primary Tool for Basic Research in Molecular Biology. American Medical Informatics Association, Snowbird, UT June 23
- 1995 How to make discoveries in molecular sequence databases. Joint meeting of the American Society for Biochemistry and Molecular Biology and the American Chemical Society, San Francisco, CA May 22
- How to make discoveries in DNA and protein sequence databases. Summer Research Conference of the American Urological Association, Houston, TX August 5
- Integrated information retrieval tools for discovery in DNA and protein sequence databases. 45th Annual meeting of the American Society of Human Genetics, Minneapolis, MN October 25
- 1996 The Turning Point in Genome Research. The Genome Project: Commercial Implications, sponsored by the Cambridge Healthtech Institute, Fairmont Hotel, San Francisco, CA March 4
- Internet for Oncologists: Hunting for Genes on the World Wide Web. 32nd Annual meeting of the American Society of Clinical Oncology, Philadelphia, PA May 18-19
- Hunting for Genes on the World Wide Web. Health Care Track: Transforming Medicine. Harvard Conference on the Internet and Society, Harvard University, Cambridge, MA May 30
- Hunting for Genes in Computer Databases. Institute of Medicine Annual Meeting. National Academy of Sciences, Washington, DC October 14
- A gene map of the human genome. ASM conference on Yeast Genetics & Human Disease. Baltimore, MD November 16
- 1997 Bioinformatics Issues in Large-scale Studies of Gene Expression: Resources, LIMS and Query Systems. Meeting on Microarray Technologies and Applications. Tucson, AZ January 22
- cDNAs: Phylum-hopping, Transcript Mapping and Gene Expression Applications. Association of Biomolecular Resource Facilities '97: Techniques at

- the Genome/Proteome Interface. Baltimore, MD February 12
- Closing the Gap between Sequence and Function. "Functional Genomics: From Genes to Drugs." 5th Annual *Nature Genetics* Conference, Washington, DC April 17
- Adventures in Information Space: GenBank, Genomes and a Turning Point in Biomedical Research (Keynote Address) Association of American Medical Colleges, 4th Annual conference on Graduate Research, Education and Training. Leesburg, VA September 25
- 1998 Data Management and Analysis in Gene Expression Arrays. 2nd Workshop on Methods and Applications of DNA Microarray Technology. Tucson, AZ. January 11
- Adventures in Information Space: GenBank, Genomes and a Turning Point in Biomedical Research. Pharmacology: Session on Utilizing the Resources of the Genome Project, Experimental Biology (FASEB) '98. San Francisco, CA. April 19
- Closing the Gap between Sequence and Function: The Frontier of Computational Biology and Functional Genomics. *Glycobiology* '98. Baltimore, MD. November 12
- 1999 Adventures in Information Space: A Turning Point in Biomedical Research. CHI 6th Annual Conference on the Human Genome Project: Commercial Implications. San Francisco, CA. March 1
- 1999 Closing the Gap between Sequence and Function: The Frontier of Computational Biology and Functional Genomics. "Surfing the Genome," a short course of the American Society for Pharmacology and Experimental Therapeutics (ASPET). Experimental Biology '99. Washington, D.C. April 17
- Higher Bandwidths through the Bottleneck: Semi-automated Approaches to Expression Array Interpretation. The Nature Genetics Microarray Meeting, Scottsdale, AZ. September 23
- In Vivo, In Vitro, In Silico*: The Frontier of Computational Biology and Functional Genomics. National Research Council, Board of Science, Technology and Economic Policy conference on "Government-industry Partnerships in Biotechnology and Computing," National Academy of Sciences, Washington, D.C. October 25-26
- 2000 Bridging the Gap between Genomes and Function: The Frontier of Computational Biology and Functional Genomics. Annual meeting of the Society of Medical Administrators, Naples, Florida. January 10
- 2002 Bioinformatics and Genome Sciences. Radiation Research Society, 49th Annual Meeting. Reno, NV. April 24
- The Emergence of actual Human Disease as a Model for Discovery Research. Chips-to-Hits® Conference, Philadelphia, PA. October 28
- 2003 Bioinformatics: past, present and future. Canadian Bioinformatics Workshop. Vancouver, B.C. February 17
- 2004 Neurogenomics and the Allen Brain Atlas. Bio-IT World Conference, Boston, MA. April 31
- Neurogenomics and the Allen Brain Atlas. Conference on "A Decade of

- Neuroscience Informatics: Looking Ahead.” National Institutes of Health, Bethesda, MD. April 26
- 2006 Proteomics, Systems Biology and Knowledge Mining in Drug and Biomarker Discovery. Microsoft eScience Workshop. Johns Hopkins University, Baltimore, MD. October 13
- 2009 New Media for Increasing Health Awareness and Medical Knowledge. 12th Annual Meeting, National Coalition for Health Professional Education in Genetics. Bethesda, MD. September 22
- 2010 [The Impact of Personalized Medicine Today](#). Health Leaders Media Roundtable, PricewaterhouseCoopers, Boston, MA. March 19
- How will eHealth and Consumer Genetics Transform Translational Research and Medicine? Health 2.0 Panel, Network Biology 2.0: Connecting Genomes to Disease Progression and Drug Response. Broad Institute, Boston, MA. April 15
- Personalized Genomic Medicine: The Future is Now. Best of Futurescape at CAP’10 The Pathologists’ Meeting™. Chicago, IL. September 27, 2010
- Personalized Genomic Medicine: The Future is Now. Banbury Center Meeting on Genome-Era Pathology. Lloyd Harbor, NY. October 14, 2010.
- Personalized Genomic Medicine: The Future is Now. American Society for Human Genetics High School Workshop. Washington, D.C. November 1, 2010.
- 2011 Enabling Personalized Medicine through Health Information Technology. Brookings Institution. Washington, D.C. January 28, 2011.
- Personalized Genomic Medicine: A Pathologist’s Perspective. Institute of Medicine Board on Health Sciences Policy, Integrating Large-Scale Genomic Information into Clinical Practice, a public workshop in the IOM’s Roundtable on Translating Genomic-Based Research for Health. Washington, D.C. July 19, 2011.
- 2012 Cancer and the Fallacy of the \$1000 Genome. Health Foo. Cambridge, MA. May 19, 2012.
- Whole Genome Diagnostics and the Off-Label Use of Drugs. GEN Thompson Reuters Webinar. May 30, 2012.
- Pathologists and the Third Wave of Medical Genomics. The Clinical Genome Conference. San Francisco, CA. June 12, 2012.
- Genomic Data Analysis in Clinical Practice and Research. Association of Pathology Chairs Annual Meeting. Monterey, CA. July 10, 2012
- The \$1,000 Genome: Twilight of the Idol? Association for Pathology Informatics Annual Meeting. Chicago, IL. October 12, 2012
- The Idolatry of the \$1,000 Genome. HMS Leadership Summit and Board of Fellows Meeting. Boston, MA. October 25, 2012. <http://vimeo.com/58578986>

Big Data + Analytics = Precision Diagnosis and Better Outcomes. Whitney Symposium, GE Global Research Center. Niskayuna, NY. October 24, 2012.

Genomic Pathology: A Real-Time Demonstration. American Society for Clinical Pathology Annual Meeting. Boston, MA. November 1, 2012.

2013 Big Data for Genomic Health. I-Study™ Think Tank, Cambridge Healthtech Institute. Seaport World Trade Center. Boston, MA. April 12, 2013.

International

- 1985 Structure and Systematics of the Apo-A-IV Gene. Seventh International Atherosclerosis Symposium, Workshop on Genetic Regulation of Apoprotein Synthesis, Melbourne, Australia October 8
- 1991 Integrated Information Retrieval in Molecular Biology: Application to Repetitive Sequence Proteins. Institute of Biochemistry, University of Lausanne, Switzerland December 7
- 1992 A Public Resource for Expressed Sequence Tags. Genome Mapping and Sequencing Meeting. Cold Spring Harbor, New York May 8
A Public Resource for Expressed Sequence Tags. Genome Sequence and Analysis Conference IV. Hilton Head, SC September 29
- 1993 Linking Yeast Genetics to Mammalian Genomes using dbEST - database of Expressed Sequence Tags. European Community meeting on In silico analysis of yeast chromosomes, Orsay, France May 28
Update on dbEST - NCBI's database for Expressed Sequence Tags. Genome Sequence and Analysis Conference V. Hilton Head, SC October 25
- 1994 Adventures in Information Space: Biomedical Discovery in a Molecular Sequence Milieu. (Plenary Lecture) 9th Annual Conference of the North American Serials Interest Group. Vancouver, Canada June 5
- 1995 Integrated information retrieval for discovery in DNA and protein sequence databases. 5th Annual Molecular Biology Conference, Queenstown, New Zealand August 14
- 1999 Closing the Gap between Sequence and Function: The Frontier of Computational Biology and Functional Genomics. HUGO Human Genome Meeting, Brisbane, Australia. March 28
In Vivo, In Vitro, In Silico: The Frontier of Computational Biology and Functional Genomics. Federation of Asian and Oceanian Biochemists and Molecular Biologists, Conference on Genome Diversity and Bioinformatics, Dunedin, New Zealand. December 2
- 2000 *In Vivo, In Vitro, In Silico: The Frontier of Computational Biology and Functional Genomics. Keynote Address, 11th Nippon Telegraph and Telephone Science Forum on High Technology in the 21st Century, Tokyo, Japan. April 6*
- 2001 What is Bioinformatics? Session on the Business/IP Interface in Bioinformatics. Biotechnology Industrial Organization, International Convention and Exhibition,

- San Diego, CA. June 26
- 2003 Intersections of Genomics, Bioinformatics and Neuroscience. Neurogenomics Research Symposium, Society for Neuroscience 33rd Annual Meeting, New Orleans, LA. November 6
- 2004 Neurogenomics and the Allen Brain Atlas. Symposium on “The Biology of Genomes.” Cold Spring Harbor, NY. May 13
- 2008 Proteomics, Systems Biology and Knowledge Mining in Drug and Biomarker Discovery. Keystone Symposium on Biomarker Discovery, Validation and Applications. Granilbakken Conference Center, Lake Tahoe, CA. February 3-8
- Online Health Information Retrieval by Consumers and the Challenge of Personal Genomics. Drug Information Association, 44th Annual Meeting. Boston, MA. June 25
- 2009 Empowering the ePatient. Bio-IT World Conference, eHealth Solutions Track. Boston, MA. April 28
- 2009 Innovation Challenges for the Pharmaceutical Industry, Directorate General of the European Commission, Brussels, Belgium. December 8-9.
- 2010 Personalized Genomic Medicine: The Future is Now. Novartis Molecular Diagnostics Leadership Forum. New York, NY. November 15, 2010.
- 2011 Whole Genome Analysis as a Universal Diagnostic. American College of Medical Genetics Annual Meeting. Vancouver, BC, Canada. March 19, 2011
- Whole Genome Analysis as a Universal Diagnostic. 16th Annual Executive War College on Lab and Pathology Management. New Orleans, LA, May 3, 2011
- Whole Genome Analysis as a Universal Diagnostic. Biomarker World Congress. Cambridge Healthtech Institute. Philadelphia, PA. May 4, 2011
- 2012 Are clinical genomes already become semi-routine for patient care? Molecular Medicine Tri-Conference, Cambridge Healthtech Institute. San Francisco, CA. February 23, 2012.
- Molecular Genetic Testing and the Emergence of Computational Pathology. 17th Annual Executive War College Conference on Laboratory & Pathology Management. New Orleans, LA. May 3, 2012.
- Pathologists, Participatory Medicine and the Third Wave of Medical Genomics. Biomarker World Congress. Cambridge Healthtech Institute. Philadelphia, PA. May 22, 2012.
- 2013 Decision Support for Precision Genomic Medicine. Molecular Medicine Tri-Conference, Cambridge Healthtech Institute. San Francisco, CA. February 14, 2013.
- Essential Ways to Leverage Lab Informatics to Add Clinical Value to Your Lab-Developed Tests. 18th Annual Executive War College Conference on Laboratory & Pathology Management. New Orleans, LA. May 2, 2013.

Report of Clinical Activities and Innovations

Current Licensure and Certification

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| 1986-1988 | Missouri State Board of Healing Arts |
| 1987 | Diplomate of the National Board of Medical Examiners |
| 1988-2000 | Maryland State Board of Physician Quality Assurance |

Report of Scholarship

Publications

Peer reviewed publications in print or other media

1. Neuwelt, E. A., **Boguski, M. S.**, Frank, J. J., Appich, K. P. and Levy, C. C. (1978) Possible sites of origin of human plasma ribonucleases as evidence by isolation and partial characterization of ribonucleases from several human tissues. *Cancer Research* 38:88-93.
2. **Boguski, M. S.**, Hieter, P. A. and Levy, C. C. (1980) Identification of a cytidine-specific ribonuclease from chicken liver. *J. Biol. Chem.* 255:2160-2163.
3. **Boguski, M. S.**, Elshourbagy, N. A., Taylor, J. M. and Gordon, J. I. (1984) Rat apolipoprotein A-IV contains 13 tandem repetitions of a 22-amino acid segment with amphipathic helical potential. *Proc. Natl. Acad. Sci. USA* 81:5021-5025.
4. **Boguski, M. S.**, Elshourbagy, N. A., Taylor, J. M. and Gordon, J. I. (1985) Comparative analysis of repeated sequences in rat apolipoproteins A-I, A-IV and E. *Proc. Natl. Acad. Sci. USA* 82:992-996.
5. Lowe, J. B., **Boguski, M. S.**, Sweetser, D. A., Elshourbagy, N. A., Taylor, J. M. and Gordon, J. I. (1985) Human liver fatty acid binding protein: isolation of a full-length cDNA and comparative analysis of orthologous and paralogous proteins. *J. Biol. Chem.* 260:3413-3417.
6. Elshourbagy, N. A., **Boguski, M. S.**, Liao, W. S. L., Jefferson, L. S., Gordon, J. I. and Taylor, J. M. (1985) Expression of rat apolipoprotein A-IV and A-I genes: mRNA induction during development and in response to glucocorticoids and insulin. *Proc. Natl. Acad. Sci. USA* 82:8242-8246.
7. Elshourbagy, N. A., Walker, D. W., **Boguski, M. S.**, Gordon, J. I. and Taylor, J. M. (1986) The nucleotide and derived amino acid sequence of human apolipoprotein A-IV mRNA and the close linkage of its gene to the genes of apolipoproteins A-I and C-III. *J. Biol. Chem.* 261:1998-2002.
8. **Boguski, M. S.**, Birkenmeier, E. H., Elshourbagy, N. A., Taylor, J. M. and Gordon, J. I. (1986) Evolution of the apolipoproteins: structure of the rat apo-A-IV gene and its relationship to the human genes for apo-A-I, C-III and E. *J. Biol. Chem.* 261:6398-6407.
9. **Boguski, M. S.**, Freeman, M., Taylor, J. M., Elshourbagy, N. A. and Gordon, J. I. (1986) On computer-assisted analysis of biological sequences: proline punctuation, consensus sequences and apolipoprotein repeats (REVIEW). *J. Lipid Res.* 27:1011-1034.
10. Elshourbagy, N. A., Walker, D. W., Paik, Y. K., **Boguski, M. S.**, Freeman, M., Gordon, J. I. and Taylor, J. M. (1987) Structure and expression of the human apolipoprotein A-IV gene. *J. Biol. Chem.* 262:7973-

7981.

11. Cole, K. D., Fernando-Warnakulasuriya, G. J. P., **Boguski, M. S.**, Freeman, M., Gordon, J. I., Clark, W. A., Law, J. H. and Wells, M. A. (1987) Primary structure and comparative sequence analysis of an insect apolipoprotein: apolipoprotein-III from *Manduca sexta*. *J. Biol. Chem.* 262:11794-11800.
12. Kanost, M. R., **Boguski, M. S.**, Freeman, M., Gordon, J. I., Wyatt, G. R. and Wells, M. A. (1988) Primary structure of apolipoprotein-III from the migratory locust, *Locusta migratoria*: potential amphipathic structures and molecular evolution of an insect apolipoprotein. *J. Biol. Chem.* 263:10568-10573
13. Trainor, C.D., Evans, T., Felsenfeld, G. and **Boguski, M. S.** (1990) Structure and Evolution of a Human Erythroid Transcription Factor. *Nature* 343:92-6, 1990
14. Sikorski, R. S., **Boguski, M. S.**, Goebel, M. and Hieter, P. (1990) A Repeating Amino Acid Motif in *CDC23* Defines a Family of Proteins and a New Relationship Among Genes Required for Mitosis and RNA Synthesis. *Cell* 60:307-17, 1990.
15. Benson, D., **Boguski, M. S.**, Lipman, D. J. and Ostell, J. (1990) The National Center for Biotechnology Information. *Genomics* 6:389-391.
16. Peitsch, M. and **Boguski, M. S.** (1990) Is Apolipoprotein D a mammalian bilin binding protein? *The New Biologist* 2:197-206.
17. Opipari, A.W., **Boguski, M.S.** and Dixit, V.M. (1990) The A20 cDNA induced by Tumor Necrosis Factor encodes a novel type of Zinc-Finger Protein. *J. Biol. Chem.* 265:14705-14708.
18. Ballester, R., Marchuk, D., **Boguski, M.S.**, Saulino, A., Letcher, R., Wigler, M. and Collins, F. (1990) The *NF1* Locus Encodes a Protein Functionally Related to Mammalian GAP and Yeast IRA Proteins. *Cell* 63:851-9, 1990.
19. Wang, Y., **Boguski, M.S.**, Riggs, M. Rodgers, L. and Wigler, M. (1991) Sar1, a gene from *Schizosaccharomyces pombe* encoding a protein that regulates ras1. *Cell Regulation* 2(6):453-465.
20. Kinzler, K.W., Nilbert, M.C., Su, L.-K., Vogelstein, B., Bryan, T.M., Levy, D.B., Smith, K.J., Preisinger, A.C., Hedge, P., McKechnie, D., Finniear, R., Markham, A., Groffen, J., **Boguski, M.S.**, Altschul, S.F., Horii, A., Ando, H., Miyoshi, Y., Miki, Y. Nishisho, I., Nakamura, Y. (1991) Identification of FAP locus genes from chromosome 5q21. *Science* 253: 661-665.
21. Peitsch, M. C. and **Boguski, M.S.** (1991) The first lipocalin with enzymatic activity. *Trends Biochem. Sci.* 16(10):363.
22. Marchuk, D.A., Saulino, A.M., Tavakkol, R., Swaroop, M., Wallace, M.R., Andersen, L.B., Mitchell, A.L., Gutmann, D.H., **Boguski, M.S.** and Collins, F.S. (1991) cDNA cloning of the Type 1 Neurofibromatosis Gene: complete sequence of the NF1 gene product. *Genomics* 11:931-940.
23. Duronio, R., Gordon, J.I. and **Boguski, M.S.** (1992) Comparative analysis of the β transducin family with identification of several new members including *PWPI*, a nonessential gene of *Saccharomyces cerevisiae* that is divergently transcribed from *NMT1*. *Proteins Struct. Func. Genet.* 13:41-56.
24. **Boguski, M.S.**, Hardison, R.C., Schwartz, S. and Miller, Webb. (1992) Analysis of conserved domains and sequence motifs in cellular regulatory proteins and transcription control regions using new software tools for multiple alignment and visualization. *The New Biologist* 4:247-260.
25. **Boguski, M.S.**, Murary, A.W., and Powers, S. (1992) Novel repetitive sequence motifs in the α and β subunits of farnesyl-protein transferases and homology of the ~~MAD2 gene product~~ of yeast. *The New Biologist* 4:408-411.
26. Gutmann, D.H., **Boguski, M.**, Marchuk, D., Wigler, M., Collins, F.S., and Ballester, R. (1993) Analysis of the neurofibromatosis type 1 (NF1) GAP-related domain by site-directed mutagenesis. *Oncogene* 8:761-769.

27. Lai, C.-C., **Boguski, M.**, Broek, D. and Powers, S. (1993) Influence of guanine-nucleotides on complex formation between Ras and Cdc25 proteins. *Mol. Cell. Biol.* 13(3):1345-1352.
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29. **Boguski, M.S.**, Lowe, T.M.J. and Tolstoshev, C.M. (1993) dbEST - database for "expressed sequence tags." *Nature Genetics* 4(4):332-333.
30. Gregory, P.E., Gutmann, D.H., Mitchell A., Park, S., **Boguski, M.**, Jacks, T., Wood, D.L., Jove, R. and Collins, F.S. (1993) Neurofibromatosis type 1 gene product (neurofibromin) associates with microtubules. *Somat. Cell. Mol. Genet.* 19: 265-274.
31. Lawrence, C.E., Altschul, S.F., **Boguski, M.S.**, Liu, J.S., Neuwald, A. and Wootton, J.C. (1993) Detecting subtle sequence signals: a Gibbs sampling strategy for multiple alignment. *Science* 262:208-214.
32. Tugendreich, S., **Boguski, M.S.**, Seldin, M. and Hieter, P. (1993) Linking yeast genetics to mammalian genomes via the EST database: identification and mapping of the human homolog of *CDC27*. *Proc. Natl. Acad. Sci. USA* 90:10031-10035.
33. **Boguski, M.S.** and McCormick, F. (1993) Proteins regulating Ras and its relatives. *Nature* 366:643-654
34. Miller, W., **Boguski, M.S.**, Raghavachari, B., Zhang, Z. and Hardison, R.C. (1994) Constructing aligned sequence blocks. *J. Computational Biology* 1:51-64.
35. Tugendreich, S., Bassett, Jr., D.E., McKusick, V., **Boguski, M.S.** and Hieter, P. (1994) Genes conserved in yeast and humans. *Hum. Mol. Genet.* 3:1509-1517
36. Miller, W. and **Boguski, M.S.** (1994) A note about computing all local alignments. *Comp. Appl. Biosci.* 10:455-456.
37. Benson, D.A., **Boguski, M.S.**, Lipman, D.J., Ostell, J. (1994) GenBank. *Nucl. Acids Res.* 22:3441-3444
38. **Boguski, M.S.**, Tolstoshev, C.M., Bassett, D. (1994) Gene discovery in dbEST. *Science* 265:1993-1994
39. Hu, H.M., O'Rourke, K., **Boguski, M.S.** and Dixit, V.M. (1994) A novel RING finger protein interacts with the cytoplasmic domain of CD40. *J. Biol. Chem.* 269:30069-30072.
40. Jiang, X. C., C. Bruce, T. Cocke, S. Wang, **M. Boguski** and A. R. Tall (1995). Point mutagenesis of positively charged amino acids of cholesteryl ester transfer protein: conserved residues within the lipid transfer/lipopolysaccharide binding protein gene family essential for function. *Biochemistry* 34(21): 7258-63.
41. **Boguski, M.S.** and Schuler, G.D. (1995) ESTablishing a human transcript map. *Nature Genetics* 10:369-371.
42. **Boguski, M.S.** (1995) The turning point in genome research. *Trends Biochem. Sci.* 20:295-296.
43. **Boguski, M.S.** (1995) Hunting for genes in computer databases. *New Engl. J. Med.* 333(10):645-647.
44. Bassett, D.E., **M.S. Boguski**, F. Spencer, R. Reeves, M. Goebel and P. Hieter (1995). Comparative genomics, genome cross-referencing and XREFdb. *Trends Genet.* 11:372-373.
45. Madej, T., **M.S. Boguski** and S. Bryant (1995). Threading analysis suggests that the obese gene product may be a helical cytokine. *FEBS Lett.* 373:13-18.
46. Benson, D.A., **Boguski, M.S.**, Lipman, D.J., Ostell, J. (1996) GenBank. *Nucl. Acids Res.* 24: 1-5.
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48. Bassett, D.E., **M.S. Boguski**, Hieter, P. (1996) Yeast genes and human disease. *Nature* 379:589-590.
49. Makalowski, W., J. Zhang, and **M.S. Boguski** (1996) Comparative analysis of 1,196 mouse and human full-length mRNA and protein sequences. *Genome Research* 6:846-857.
50. G.D. Schuler, **M.S. Boguski**, E.A. Stewart, L.D. Stein, G. Gyapay, K. Rice, R.E. White, P. Rodriguez-Tome, A. Aggarwal, E. Bajorek, S. Bentolila, B.B. Birren, A. Butler, A.B. Castle, N. Chiannilkulchai, A. Chu, C. Clee, S. Cowles, P.J.R. Day, T. Dibling, N. Drouot, I. Dunham, S. Duprat, C. East, C. Edwards, J.-B. Fan, N. Fang, C. Fizames, C. Garrett, L. Green, D. Hadley, M. Harris, P. Harrison, S. Brady, A. Hicks, E. Holloway, L. Hui, S. Hussain, C. Louis-Dit-Sully, J. Ma, A. MacGilvery, C. Mader, A. Maratukulam, T.C. Matise, K.B. McKusick, J. Morissette, A. Mungall, D. Muselet, H.C. Nusbaum, D.C. Page, A. Peck, S. Perkins, M. Piercy, F. Qin, J. Quackenbush, S. Ranby, T. Reif, S. Rozen, C. Sanders, X. She, J. Silva, D.K. Slonim, C. Soderlund, W.-L. Sun, P. Tabar, T. Thangarajah, N. Vega-Czarny, D. Vollrath, S. Voyticky, T. Wilmer, X. Wu, M.D. Adams, C. Auffray, R. Berry, R. Brandon, A. Dehejia, P.N. Goodfellow, R. Houlgatte, J.R. Hudson Jr., S.E. Ide, K.R. Iorio, W.Y. Lee, N. Seki, T. Nagase, K. Ishikawa, N. Nomura, C. Phillips, M.H. Polymeropoulos, M. Sandusky, K. Schmitt, J.M. Sikela, K. Swanson, R. Torres, J.C. Venter, N.A.R. Walter, J. S. Beckmann, J. Weissenbach, R.M. Myers, D.R. Cox, M.R. James, D. Bentley, P. Deloukas, E.S. Lander, and T.J. Hudson (1996). A gene map of the human genome. *Science* 274: 540-546. (Also see <http://www.ncbi.nlm.nih.gov/SCIENCE96>)
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52. Bassett, D.E., **Boguski, M.S.**, Spencer, F., Reeves, R., Kim, S., Weaver, T., and Hieter, P. (1997) Genome Cross-referencing and XREFdb: Implications for the Identification and Analysis of Genes Mutated in Human Disease. *Nature Genetics* 15:339-344.
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genome: an analysis of 2,820 orthologous rodent and human sequences. *Proc. Natl. Acad. Sci. USA* 95:9407-9412.

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Thesis

Structure, Function and Evolution of the Mammalian Apolipoproteins and their Genes (1985) Washington University, Division of Biology and Biomedical Sciences, St. Louis, MO.

Narrative Report (limit to 500 words)

My research interests over the past [25 years](#) have been concentrated around several major and overlapping [themes](#):

- Bioinformatics and Computational Biology
- The relationships between genes and human disease
- Signaling and Structural Biology
- Genome and Proteome Research, including
 - Comparative Genomics and Evolution
 - Genome-wide transcript mapping
 - Functional Genomics
 - Pharmacogenomics
 - Neurogenomics
 - Proteomics for drug and biomarker discover
- Precision Medicine and Comparative Effectiveness Research

Many of these activities have involved not computational biology alone, or experimental biology alone, but a synergistic fusion of both approaches to scientific investigation.

Bioinformatics & Computational Biology

My interest in computational biology started in graduate school in the early 1980s (more than ten years before the term [bioinformatics](#) first appeared in the literature). I used and developed some of the primitive software tools of the time to study the structure, function and evolution of mammalian apolipoprotein genes. My subsequent work in computational biology has, over the years, involved algorithm development (e.g. Gibbs sampler, text mining), database design, development and implementation (dbEST, XREFdb, ArrayDB) and data mining, data analysis and data annotation (examples below). One database effort in particular, the database of Expressed Sequence Tags ([dbEST](#), 1993), has enjoyed a particularly long and influential life contributing first to gene discovery and subsequently to succeeding generations of genomics applications, namely transcript mapping, design and construction of microarrays, discovery *in silico* of single nucleotide polymorphisms and, ultimately, analysis and annotation of the human genome.

Genes & Disease

My group collaborated in the cloning and/or in-depth analysis of the products of a number of human disease genes including: *Neurofibromatosis Type 1*, *Familial Adenomatous Polyposis (APC)*, *Choroideremia*, *Multiple Endocrine Neoplasia Type 1* and *Cerebral Cavernous Malformation*. In addition, we pioneered the use of **comparative genomics** methods (see below) to gain broad insights into the relationships and conservation of

human disease genes in experimentally-tractable model organisms including yeast, nematodes, *Drosophila* and rodents.

Signaling and Structural Biology

Sequence analysis of the NF1 gene and its GTPase-activating (GAP) domain led us into the *ras* pathway where we worked for a number of years on identifying structural and functional motifs in *ras* signaling proteins. Notably, we elucidated the secondary structures of prenyltransferase subunits. Extensive studies were also performed on other proteins and protein families. We predicted that, unlike other apolipoproteins which possessed amphipathic α -helical structures, ApoD was a member of the lipocalin family composed of two orthogonal β -sheets. We also identified the first lipocalin with enzymatic activity, prostaglandin D synthase. Our [threading analysis](#) of the mouse *obese* gene product (leptin) predicted its unexpected structure and function as a helical cytokine, later confirmed by other groups using NMR, crystallography and cloning of the leptin receptor.

Genome and Proteome Research

Comparative Genomics & Evolution - Our group first coined the term *comparative genomics* in 1995 to describe our work on the large-scale sequence analysis of the homologs of human disease genes in model organisms and the first comparative genomics database, [XREFdb](#). Over the next six years we studied thousands of gene sets in humans, rats, mice, *Drosophila*, nematodes and yeast and established the basic [evolutionary parameters](#) for interpretation of conserved protein-encoding genes in the human genome.

Transcript Mapping - Clusters of human genes and ESTs (“[UniGenes](#)”) were utilized to construct the first comprehensive [transcript map of the human genome](#) (1996, 1998). Historically, this was the first instance of *Science* magazine using the World Wide Web to publish results, provide hyper-linked information resources and supplemental data sets. These maps facilitated and accelerated the positional cloning of hundreds of genes and this mapping approach was widely applied to other organisms.

Functional Genomics - We used human UniGenes to design and construct the first human cDNA microarray (representing 10,000 genes) and were first to provide a [rigorous definition of functional genomics](#) for the community. While on sabbatical at NHGRI, our group implemented the first relational database and analysis system, [ArrayDB](#), for microarray data. This design was copied by numerous academic and commercial groups. Our group was also first to apply methods of [statistical text-mining](#) to the interpretation of gene expression profiles. In the 2001 Genome Issue of *Nature*, we immediately followed the first publication of the human genome sequence with a paper showing how to use microarray technology to experimentally annotate and correct computational gene predictions.

Pharmacogenomics - We cloned and sequenced the [pregnane X receptor \(PXR\) gene](#) that encodes the key transcription factor regulating the expression of genes encoding drug and xenobiotic metabolizing enzymes. We also identified functional sequence polymorphisms in the promoters of these genes, cytochromes P450 3A (CYP3A), and studied the genotypes and corresponding molecular phenotypes in several populations differing in their drug-metabolizing abilities.

Neurogenomics - We pioneered the application of genome-scale approaches to neurobiology with the construction of a comprehensive, 3-dimensional transcript map of the mouse brain, the [Allen Brain Atlas](#).

Proteomics and Knowledge Mining. At Novartis, my division was responsible for the application of [proteomics technologies and computational knowledge-mining](#) for drug target and biomarker discovery.

Precision Medicine & Comparative Effectiveness Research

At Harvard Medical School and Beth Israel Deaconess Medical Center, my current research interests and activities include the applications of “next generation” genome sequencing technologies and network biology to the development of precision diagnostics and personalized medicine. I am also working on new approaches to drug “repurposing” involving “empowered patients,” social networking technologies, personal genotyping and health record data-mining to identify potential new uses for existing drugs. In 2009, I founded the [Genomic Medicine Initiative](#), the first training program of its kind, to prepare pathology residents to practice personalized medicine in the Genome Era.

