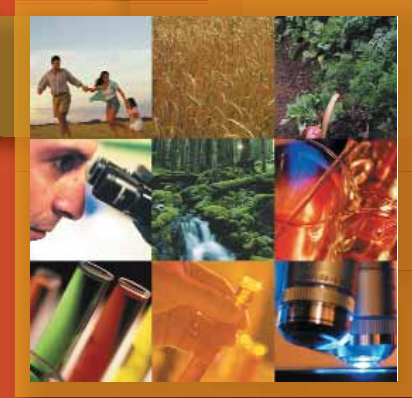


2020 Vision:
Adaptation to Environmental Change

Vision 2020 :
L'adaptation au changement environnemental



GenomeCanada International Conference

October 17-19 octobre 2007
Château Frontenac, Québec City
Québec, Canada

Agenda at a Glance

Aperçu de l'ordre du jour

Wednesday, October 17 ■ Mercredi 17 octobre

- 7:00 - 10:00 Registration ■ Inscription
- 8:00 - 9:15 Breakfast ■ Petit déjeuner
- 9:30 - 12:30 Plenary ■ Plénière
- 12:30 - 13:45 Lunch ■ Déjeuner
- 14:00 - 16:30 **Session 1 ■ Séance 1**
Systems Biology ■ Biologie des systèmes
- Session 2 ■ Séance 2**
Infectious Diseases
Maladies infectieuses
- 18:00 - 20:00 **Poster Session and Reception**
Séance d'affiches et vin d'honneur

Thursday, October 18 ■ Jeudi 18 octobre

- 7:30 - 8:45 Breakfast ■ Petit déjeuner
- 9:00 - 11:30 **Session 3 ■ Séance 3**
Behavioural Genetics
Génétique du comportement
- Session 4 ■ Séance 4**
Application of Genomics to Environmental
Problems ■ Application de la génomique
aux problèmes d'environnement
- 12:00 - 13:45 Lunch ■ Déjeuner
- 14:00 - 16:30 **Session 5 ■ Séance 5**
Genomics and Diversity
Génomique et diversité
- Session 6 ■ Séance 6**
Probing Biodiversity through Genomics
Explorer la biodiversité au moyen
de la génomique
- 18:00 - 21:00 **Reception and Banquet**
Vin d'honneur et banquet

Agenda at a Glance Aperçu de l'ordre du jour

Friday, October 19 ■ Vendredi 19 octobre

- 7:30 - 8:45 Breakfast ■ Petit déjeuner
9:00 - 12:00 Plenary ■ Plénière
12:15 - 13:30 Lunch ■ Déjeuner
13:30 Adjournment ■ Clôture

Program Committee ■ Comité du programme

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Consultant
Québec, Canada

William Bridger (Co-chair)

R.M. Spencer & Associates
Alberta, Canada

John Aitchison

Institute for Systems Biology
Washington, USA

Michel Bergeron

Université Laval
Québec, Canada

Nancy Press

Oregon Health and Science University
Oregon, USA

Steven Rothstein

University of Guelph
Ontario, Canada

Troy Duster

New York University
New York, USA

Paul Hebert

University of Guelph
Ontario, Canada

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Québec, Canada

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Genome Canada

Claudine Renaud

Genome Canada



Calvin R. Stiller
Chairman ■ Président du conseil d'administration

I am delighted to welcome you to Genome Canada's second International Conference. Last year we urged our speakers to gaze into the future and contemplate their work from the perspective of genomics and proteomics research in the year 2020 and beyond. It was so successful, that this year, we have sharpened the focus to address the role genomics plays in environmental issues. Not only is *2020 Vision: Adaptation to Environmental Change* a timely and controversial subject for scientists and society as a whole, it is an area where genomics and proteomics research have much to contribute.

Images of melting polar ice and disastrous weather reflect changing environmental conditions around the world. Scientists, sociologists and politicians are acutely aware of the impact these and other warning signs have on our future worldwide. By hosting this conference, Genome Canada is hoping to identify new research directions for genomics and inspire the search for more and better answers to how and why our environment is changing and what we can do about it.

Internationally acclaimed speakers from around the world will be sharing their thoughts and visions about where *Adaptation to Environmental Change* will lead. Congratulations on joining us for what will be an exciting and illuminating few days.

Je suis heureux de vous accueillir à la deuxième Conférence internationale de Génome Canada. L'an dernier, nous avons demandé à nos conférenciers de penser à ce que pourraient être l'avenir et leurs travaux, du point de vue de la recherche en génomique et en protéomique, en 2020 et par la suite. Les résultats ont été si concluants que cette année, nous avons choisi de nous concentrer sur le rôle que joue la génomique en environnement. Non seulement *Vision 2020 : Adaptation au changement environnemental* est-il un sujet opportun et controversé pour les chercheurs et la société en général, mais il est également un domaine où la recherche en génomique et en protéomique a beaucoup à offrir.

Des images de la fonte de la glace polaire et de conditions météorologiques désastreuses témoignent des changements de l'environnement partout dans le monde. Les chercheurs, les sociologues et les politiciens sont très conscients des répercussions de ces manifestations et d'autres signaux d'avertissement sur notre avenir, partout sur la planète. Par cette conférence, Génome Canada espère cibler de nouvelles orientations de recherche en génomique et inspirer la quête de réponses plus nombreuses et plus pertinentes sur les changements qui marquent l'environnement, les raisons de ces changements, et les moyens d'action possibles.

Des conférenciers de renommée internationale vous feront part de leurs réflexions sur ce à quoi nous mènera l'*adaptation au changement environnemental* et de leurs visions à cet égard. Je vous félicite d'être des nôtres pendant ces quelques jours fort intéressants et instructifs.

Martin Godbout
President and CEO ■ Président et chef de la direction



Welcome to Genome Canada's second International Conference which focuses its *2020 Vision* on the environment. Issues such as global warming and climate change have dominated the media over the past year, but few articles have examined the impact of our changing environment at the genomic level. This year's conference, addresses the theme of *Adaptation to Environmental Change* from a world-wide genomics perspective.

Speakers from Canada, the United States, Great Britain, Europe, New Zealand and Asia will present exciting research on systems biology, infectious diseases, behavioural genetics, the application of genomics to environmental problems, genomics and diversity, and biodiversity. The conference Steering Committee challenged each speaker to envision not only what their research reveals about adaptation to environmental change today, but also where it might lead in the next decade. They will be looking at developments in genomics and proteomics as well as the environmental, ethical, economic, legal and social (GE³LS) implications of this adaptation.

I am sure you will enjoy the 2007 edition of Genome Canada's *2020 Vision: Adaptation to Environmental Change*.

A handwritten signature in black ink, appearing to read 'Martin Godbout'.

Je vous souhaite la bienvenue à la deuxième Conférence internationale, *Vision 2020*, qui se concentre cette année sur l'environnement. Des questions comme le réchauffement de la planète et les changements climatiques ont retenu l'attention des médias au cours de la dernière année, mais peu d'articles se sont attardés aux répercussions de notre environnement en changement à l'échelle de la génomique. La conférence de cette année a pour thème l'*Adaptation au changement environnemental* du point de vue de la génomique, partout dans le monde.

Des conférenciers du Canada, des États-Unis, de la Grande-Bretagne, de l'Europe, de la Nouvelle-Zélande et de l'Asie présenteront d'intéressantes recherches sur la biologie des systèmes, les maladies infectieuses, la génétique du comportement, l'application de la génomique à la résolution de problèmes environnementaux, la génomique et la diversité, de même que la biodiversité. Le Comité directeur de la conférence a mis au défi chacun des conférenciers d'envisager non seulement ce que révèle aujourd'hui leur recherche sur l'adaptation au changement environnemental, mais également où leurs travaux pourraient mener au cours de la prochaine décennie. Les conférenciers examineront les progrès de la génomique et de la protéomique, de même que les répercussions de cette adaptation sur l'environnement, l'éthique, l'économie, le droit et la société (GE³DS).

Je suis certain que cette nouvelle édition de *Vision 2020 : Adaptation au changement environnemental* saura vous plaire.

A handwritten signature in black ink, appearing to read 'Martin Godbout'.

Plenary 9:30 - 12:30 Plénière

Salle de Bal

Chair ■ Président : William Bridger

9:30 - 10:15



David Cox
Perlegen Sciences Inc.
California, USA

Human Genetic Variation and Healthcare

11:00 - 11:45



Claire M. Fraser-Liggett
University of Maryland
Maryland, USA

Understanding our Microbial Selves through the
Science of Metagenomics

10:15 - 11:00



John D. Aitchison
Institute for Systems Biology
Washington, USA

The Promise and Practice of Systems Biology

11:45 - 12:30



Bartha Maria Knoppers
Université de Montréal
Québec, Canada

Across the Genomes: Whither "Identifiability"?

Session 1 14:00 - 16:30 Séance 1

Place d'Armes

Chair ■ Président : John D. Aitchison

14:00 - 14:30



Richard Rachubinski
University of Alberta
Alberta, Canada

Systems Biology Approaches to an Understanding of
Peroxisome Function

15:30 - 16:00



Simon Letarte
Institute for Systems Biology
Washington, USA

Mass Spectrometry Proteomics: New Enabling
Technologies for Biomarker Discovery and P4 Medicine

14:30 - 15:00



Ulf Nehrass
Institut Pasteur Korea
Seoul, Korea

From Drugs to Genes: Visualizing Chemical Genomics

16:00 - 16:30



Carl L. Hansen
University of British Columbia
British Columbia, Canada

Highly Integrated Microfluidic Systems for Systems Biology

Session 2 14:00 - 16:30 Séance 2

Jacques Cartier

Chair ■ Président : Michel G. Bergeron

14:00 - 14:30



David Relman
Stanford University
California, USA

Exploration of the Human Indigenous Microbial
Communities

15:30 - 16:00



Antonello Covacci
Novartis
Siena, Italy

The Future of Bacterial Genomics from the Host's
Point of View

14:30 - 15:00



Lorne Tyrrell
University of Alberta
Alberta, Canada

Genomic Applications to Infectious Diseases

16:00 - 16:30



Michel G. Bergeron
Université Laval
Québec, Canada

Rapid Molecular Diagnostic at Point-of-Care: A Daily
Reality by 2020

Session 3 9:00 - 11:30 Séance 3

Frontenac

Chair ■ Présidente : Nancy Press

9:00 - 9:30



Wendy Johnson
University of Edinburgh
Edinburgh, UK

Genetic and Environmental Influences on the Wrong
Models of Cognitive Ability and Personality

10:30 - 11:00



Eric Turkheimer
University of Virginia
Virginia, USA

Behavior Genetics Finally Delivers as a Tool for
Understanding the Environment

9:30 - 10:00



James R. Flynn
University of Otago
Dunedin, New Zealand

Unpacking Intelligence

11:00 - 11:30



Nancy Press
Oregon Health and Science University
Oregon, USA

Phenotypes, Social Constructionism, and the Spurious
Finding: Behavioral Genetics in Social Context

12:00 - 13:45 Lunch ■ Déjeuner

Session 4 9:00 - 11:30 Séance 4

Jacques Cartier

Chair ■ Président : Steven Rothstein

9:00 - 9:30



David Layzell
Queen's University
Ontario, Canada

Towards a Transformative and Sustainable Bioeconomy:
Opportunities for Genomics Research

10:30 - 11:00



Stefan Jansson
Umeå University
Umeå, Sweden

Natural Variation in *Populus*

9:30 - 10:00



Steven Rothstein
University of Guelph
Ontario, Canada

Using Genomic Knowledge to Improve Crop Plants during
a Time of Environmental Challenges

11:00 - 11:30



Ben F. Koop
University of Victoria
Victoria, British Columbia

Applications of Genome Research in Salmonids (cGRASP)
to Environmental Change

Session 5 14:00 - 16:30 Séance 5

Frontenac

Chair ■ Président : Troy Duster

14:00 - 14:30



Troy Duster
New York University
New York, USA

A Futuristic Projection to 2020 of New and Emergent Social Issues Generated by Research in Human Molecular Genetics

15:30 - 16:00



Deborah Bolnick
University of Texas
Texas, USA

Racing to Conclusions: Understanding Human Genomic Diversity and Population Structure in the 21st Century

14:30 - 15:00



Duana Fullwiley
Harvard University
Massachusetts, USA

Environments of Human Practice on the Future of Race Thinking

16:00 - 16:30



Stephen L. Minger
King's College London
London, UK

Therapeutic and Research Potential of Human Stem Cells

Session 6 14:00 - 16:30 Séance 6

Jacques Cartier

Chair ■ Président : Paul Hebert

14:00 - 14:30



Paul Hebert
University of Guelph
Ontario, Canada

Fractals, Genomes and a Barcoded World

15:30 - 16:00



Mostafa Ronaghi
Stanford Genome Technology Centre
California, USA

Advanced Technologies for Determination of Biodiversity

14:30 - 15:00



Hendrik Poinar
McMaster University
Ontario, Canada

Our Frozen Heritage: Metagenomics to Paleogenomics

16:00 - 16:30



Dan Janzen
University of Pennsylvania
Pennsylvania, USA

Wild Biodiversity through a New Microscope - the Barcode: Some ethical, environmental and economic implications of a new way of seeing what you are looking at

Jean Lemire

Biologist, film producer and director ■ Biologiste, producteur et réalisateur

The Antarctic Mission ■ La mission Antarctique



Banquet - 18:00-21:00 - Salle de Bal

Educated as a biologist, Jean Lemire has over 20 years of experience in marine biology under his belt. As a scientist affiliated with the University of Hawaii, and at the Mingan Island Cetacean Study Research Station, his knowledge of whales led to his participation in a great many research projects in the international arena.

In 1987, he began a parallel career in filmmaking and founded Les Productions Ciné-Bio, a firm specializing in scientific consulting, research and script development for documentary productions.

In 2001, he founded Les Productions Glacialis inc., a new production company specializing in natural sciences, marine science, arts and culture.

In 2003–2004, he produced the following films: *Short Infinity* and *Arctic Mission*, a collection of five documentary and feature-length films to be shown in movie theatres, on the effects of climate change in the Arctic. *Arctic Mission* met with seldom-seen popular acclaim. It reached hundreds of millions of television viewers around the world and received the prestigious Earth Watch award, presented by the National Geographic Society.

In 2006, he co-produced and co-directed *The White Planet*, a feature-length environmental film to be shown in movie theatres. Sold in nearly 60 countries, this film is a powerful visual testimonial on the threatened world of the Arctic, the first and hardest-hit casualty of global climate change.

In 2006, he was voted personality of the year by La Presse/Radio-Canada, in the category Soft Science, Pure Science and Technology. The readers of the popular magazine, *Sélection du Readers Digest*, named him Hero of the Year 2006 in the environmental area.

In Autumn of 2005, Jean Lemire embarked on his greatest adventure to date. On board the *Sedna IV*, accompanied by a crew of sailors, filmmakers and scientists, he went off in conquest of Antarctica, the last continent. *Antarctic Mission* would become one of the great expeditions of modern times: 430 days of sailing, isolation and extreme adventures. A feature-length film to be shown in movie theatres, a documentary series and a series on the adventure are now being put together.

Biologiste de formation, Jean Lemire a plus de 20 ans d'expérience en biologie marine. Scientifique associé à l'Université d'Hawaï et à la Station de recherche des îles Mingan, ses connaissances sur les baleines l'amènent à participer à de nombreux projets de recherche sur la scène internationale.

En 1987, il entame une carrière parallèle en cinéma et fonde Les Productions Ciné-Bio, une entreprise spécialisée en consultation scientifique, en recherche et en scénarisation pour les productions documentaires.

En 2001, il fonde les productions GLACIALIS inc., une toute nouvelle maison de production spécialisée dans le domaine des sciences naturelles, de la mer, des arts et de la culture. Il produit en 2003 - 2004 : *7 km2 d'infini* et *Mission Arctique*, une collection de 5 films documentaires et un long-métrage pour les salles de cinéma sur les effets des changements climatiques en Arctique.

Succès populaire rarement égalé, *Mission Arctique* rayonne sur la scène internationale, avec des centaines de millions de téléspectateurs et est récipiendaire du prestigieux « Earth Watch Award », remis par la National Geographic Society.

En 2006, il coproduit et coréalise *La Planète Blanche*, un long-métrage environnemental pour les salles de cinéma. Vendu dans près de 60 pays, le film offre un puissant témoignage visuel sur le monde menacé de l'Arctique, première victime des changements climatiques.

La même année, il est élu personnalité de l'année La Presse/Radio-Canada, dans la catégorie Sciences humaines, sciences pures et technologie. Les lecteurs du populaire magazine *Sélection – Reader's Digest* lui décerne le titre de Héros de l'année 2006 dans le secteur environnement.

À l'automne 2005, Jean Lemire embarque dans sa plus grande aventure à ce jour. À bord du *Sedna IV*, entouré d'une équipe de marins, cinéastes et scientifiques, il part à la conquête du dernier continent, l'Antarctique. « *Mission Antarctique* » devient l'une des plus grandes expéditions des temps modernes : 430 jours de navigation, d'isolement et d'aventures extrêmes. Un long-métrage pour les salles de cinéma, une série documentaire et une série sur l'aventure sont présentement en montage.

(Photo : Martin Leclerc)

Plenary 9:00 - 12:00 Plénière

Salle de Bal

Chair ■ Présidente : Cindy Bell

9:00 - 9:50



Eddy Rubin
University of California
California, USA

Energy Genomics

11:10 - 12:00



David Bentley
Illumina
California, USA

New DNA Sequencing Technology and
Changing Horizons

9:50 - 10:40



Ron M. Fournay
Royal Canadian Mounted Police/GRC
Ottawa, Canada

Forensic DNA Analysis - Past and Future Challenges

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John D. Aitchison

Dr. Aitchison studied biochemistry, specializing in biotechnology and genetic engineering, at McMaster University in Ontario, Canada. There, in the laboratory of Dr. Richard Rachubinski, he investigated the molecular mechanisms responsible for sorting proteins into peroxisomes. After receiving his Ph.D. in 1992, Dr. Aitchison performed his postdoctoral work in the laboratory of Nobel Laureate Dr. Günter Blobel at Rockefeller University. In Dr. Blobel's lab, Dr. Aitchison applied classic cell biology techniques and yeast genetics to the study of protein import into the nucleus. During this time, he began to apply large-scale proteomics to the problem, which he continued as an Assistant Professor in the Faculty of Medicine and Dentistry at the University of Alberta from 1997-2000. In September 2000, Dr. Aitchison joined the Institute for Systems Biology (ISB) with the goal of applying systems approaches to the study of fundamental cell biology. He is currently a Professor at the ISB, and holds Adjunct Professorships at the University of Alberta and the University of British Columbia, and Affiliate Faculty status at the University of Washington.



David Bentley is Chief Scientist at Illumina Inc (Sequencing), developing new DNA sequencing technology for fast, accurate sequencing of complex genomes. His major research interest is the study of human sequence variation. He was previously Head of Human Genetics and founding Member of the Board of Management at the Wellcome Trust Sanger Institute, where he played leading roles in the Institute's contribution to the human genome reference sequence, The SNP Consortium and the International HapMap Project.



Michel G. Bergeron

Dr. Michel G. Bergeron is Professor and Chairman of the Division of Microbiology and of the Centre de recherche en infectiologie of Université Laval in Québec City. After receiving his MD in 1968 from Université Laval, he trained in Internal Medicine at McGill University and in Infectious Diseases and Microbiology at Tufts University and MIT. In 1974, Dr. Bergeron founded the Centre de recherche en infectiologie, one of the largest infectious diseases research centres in North America. His latest innovative DNA-based tests identify pathogens and their antibiotic resistance genes from clinical samples in under an hour instead of 2 to 3 days. He and his team are now developing compact discs (CDs) to quickly read DNA and detect microbes in less than 15 minutes, in the doctor's office. Dr. Bergeron has written or co-authored more than 380 scientific publications, given more than 550 presentations at universities and international meetings and trained more than 80 graduate and postgraduate students and fellows. He is on the editorial board of several international journals and a fellow of the Infectious Diseases Society of America and the Canadian Academy of Health Sciences. He is a member of the Scientific Advisory Committee of the Council of Canadian Academies and of the Canadian Space Agency. Dr. Bergeron has received the Prix du Québec Wilder-Penfield, the province's highest scientific distinction in bio-medical research and was recently awarded Genome Québec's "Biotechnology Award of Tomorrow" at the 2007 Genesis Awards Gala. Dr. Bergeron's current project is an international consortium of 116 researchers from 16 countries called Diagnostics for Life dedicated to developing point-of-care testing (POCT) devices.

Rapid Molecular Diagnostic at Point-of-Care: A Daily Reality by 2020

Today, as in the time of Pasteur, more than 140 years ago, it still takes two days to identify most microbes and detect their resistance to antibiotics. The Centre de recherche en infectiologie (CRI), an internationally recognized research group in infectious diseases (IDs) with 250 highly qualified personnel (HQP) from Université Laval in Québec City, Canada has created, in collaboration with several research institutions and industrial partners from Canada and abroad, an integrated collaborative research team focused on rapid (<1h) nucleic acid-based diagnostic technologies for better control of IDs. Over the years, this multidisciplinary team has developed state-of-the-art technologies and know-how on biomarkers, polymeric biosensors, microfluidic technologies, ultra-sensitive optical detection technology, rapid nucleic acid extraction methods, microbial genomics database of evolutionary-conserved microbial chromosomal genes, antibiotic resistance genes and toxin genes, bioinformatics strategies for optimal primer/probe design and data analysis, multiplex PCR and real-time PCR, capture probe design and microarray hybridization. The integration of these technologies has resulted in the development of the first two real-time PCR assays ever approved by the FDA (ID1-StrepBTM, Bergeron et al., *N Engl J Med* 2000; 343:175-9 and ID1-MRSATM, Huletsky et al. *J. Clin Microbiol.* 2004, 42: 1875-84) and of the first compact disk (CD) based device able to detect DNA at room temperature in 15 minutes (Peytavi et al., *Clin Chem.* 2005, 51:1836-44). We recently discovered FCR (Fluorescence Chain Reaction), an ultra-sensitive technology that amplifies the detection signal and which could well replace PCR (Ho et al., *J Am. Chem. Soc.* 2005, 127:12673-6). Diagnostics is a neglected area of research worldwide. For example, only 3.5% of all public and

private research funds are devoted to diagnosis. Moreover, technologies for affordable molecular diagnostics of IDs are considered by world health experts as a priority for improving life in developing countries (Daar et al., *Nat Genet.* 2002, 32:229-32). The Bill & Melinda Gates Foundation also considers molecular diagnostics as one of their priorities (Burgess et al., *Nature.* 2006, 23:444 Suppl 1:1-2). As diagnostics is upstream and guides treatment, well planned rapid nucleic acid-based point-of-care tests (POCTs) will become powerful management and preventive tools not only for IDs, the leading cause of mortality worldwide, but also for many other aspects of human, animal and environmental health, including defence preparedness, forensics, agriculture, or food and cosmetics industries. Genomically-based POCT will aim at developing on-site testing in hospitals (delivery room, emergency room, intensive care unit, etc.), primary care facilities (doctor's office, dispensaries, etc.), pharmacies, remote settings (space, battlefields, Africa, etc.), or even, after adequate validation, for home-based self-care testing such as required for theranostic monitoring of chronic diseases.

We are now building a Canada-based international research consortium on POCT called Diagnostics for Life supported by an extraordinary interactive infrastructure located at Université Laval in Québec City to provide affordable and comprehensive rapid (<15min) point-of-care diagnostic tests and devices to meet public health and environmental challenges. We believe genomic POCT will, over the next 20 years, revolutionize global health in both developed and developing worlds.



Deborah Bolnick is an Assistant Professor in the Department of Anthropology at the University of Texas at Austin. She studied anthropology at Yale University, and received her Ph.D. in anthropology in 2005 from the University of California, Davis. Dr. Bolnick's research focuses on the patterns of human genetic variation and how they are shaped by culture, language, history, and geography. She uses both ancient and modern DNA to reconstruct Native American prehistory and to investigate prehistoric population movements. Dr. Bolnick also investigates contemporary understandings of the relationship between race, ethnicity, and genomics. Her research has been published in a variety of journals, including *Molecular Biology and Evolution*, *American Journal of Physical Anthropology*, *American Journal of Human Genetics*, and *Proceedings of the National Academy of the Sciences USA*.

Racing to Conclusions: Understanding Human Genomic Diversity and Population Structure in the 21st Century

During the last decades of the 20th century, a new scientific consensus seemed to emerge regarding racial groups and the pattern of human genetic diversity. This consensus reflected a large body of evidence showing that human races are not genetically distinct, and it suggested that racial groupings provide a poor representation of the genetic diversity in our species. However, a number of recent studies seem to conflict with this consensus. These new genomic studies have used some novel technologies to identify statistically significant differences between continental groupings, and they argue that continental or racial groupings must be considered in studies of human disease and human population structure. In this presentation, I will evaluate why the conclusions of these recent studies differ from the earlier consensus, and I will discuss the implications for our understanding of human genomic diversity and population structure.



Antonello Covacci

Antonello Covacci is head of the Cellular Microbiology and Bioinformatics Unit at Novartis Vaccines and Diagnostics in Siena, Italy. After a degree in internal medicine at the University of Florence he was a postdoctoral fellow at the Hormone Research Institute, UCSF and a visiting scholar at Stanford University. He has been involved in the identification of the Ptl genes on pertussis toxin secretion during a pre-doctoral stage at Sclavo Research Center – Siena, working with Rino Rappuoli. A large fraction of his scientific career has been dedicated to the molecular characterization of the Cag pathogenicity island of *Helicobacter pylori* and the effector molecule CagA. This is the first example of a bacterial oncogene that alters the terminal differentiation process of epithelial cells by interacting with a tumor suppressor molecule. He is supporting the development of a prophylactic vaccine against *Helicobacter pylori* and a research effort for defining a new proteic vaccine against *Streptococcus pneumoniae*. In collaboration with members of the Novartis Vaccines Research Center he is involved in exploring the genetic structure of bacterial populations using advanced computational methods. He was elected a member of the European Molecular Biology Organization in 2001.

The Future of Bacterial Genomics from the Host's Point of View

The advent of whole-genome sequencing of bacteria and advances in bioinformatics have revolutionized the study of bacterial pathogenesis, enabling the targeting of possible vaccine candidates starting from genomic information. Nowadays, the availability of hundreds of bacterial genomes allows the identification of the genetic differences across several genomes from the same species. The unexpected degree of intra-species diversity suggests that a single genome sequence is not entirely representative and does not offer a complete picture of the genetic variability of a species. The practical consequence is that, in many cases, a universal vaccine is possible only by including a combination of antigens and this combination must take into account the pathogen population structure. Comparisons of multiple genomes to provide insights into conserved or unique families of proteins or functional domains are needed to continuously improve the precision of annotation and to identify the basic building blocks of proteins, trace the evolution of virulence mechanisms, potentially reconstruct complex structures, and identify and design novel immunogens. These increasing needs are helping to drive the beginning of the next phase based on a Grid of supercomputers connecting major scientific institutions, with decentralized databases

containing a repository of any available data. All these operations will be performed in real time to any scientist able to formulate fundamental questions using indexing and data retrieval "à la Google™". The sharing of genomic information would represent a major effort to identify host factors controlling virulence or susceptibility loci for a particular infection widening the universe of genomic information and favoring the power of tomorrow's science.



Dr. Cox is internationally recognized for his research on the molecular basis of human genetic disease. After receiving his B.A. and M.S. degrees from Brown University in Rhode Island, Dr. Cox obtained his M.D. and Ph.D. degrees from the University of Washington, Seattle. He then completed his Pediatric Residency at the Yale-New Haven Hospital in New Haven, Connecticut and was a Fellow in both genetics and pediatrics at the University of California San Francisco. From 1980 to 1993, Dr. Cox held faculty positions in the Departments of Pediatrics, Biochemistry and Psychiatry at the University of California San Francisco. In 1993, he accepted a position as a Professor of Genetics and Pediatrics at the Stanford University School of Medicine as well as the Co-director of the Stanford Genome Center. In October of 2000, Dr. Cox left his position at Stanford University to become the Chief Scientific Officer of Perlegen Sciences, Inc. Dr. Cox is certified by both the American Board of Pediatrics and the American Board of Medical Genetics. He has served on several international and national councils and commissions including the Council of the Human Genome Organization (HUGO) and the National Bioethics Advisory Commission (NBAC) and the Health Sciences Policy Board of the Institute of Medicine. Dr. Cox's honors include election to the Institute of Medicine of the National Academy of Sciences.



Troy Duster

Troy Duster is Silver Professor of Sociology and Director of the Institute for the History of the Production of Knowledge at New York University – and he also holds an appointment as Chancellor's Professor at the University of California, Berkeley. From 1996-98, he served as member and then chair of the joint NIH/DOE advisory committee on Ethical, Legal and Social Issues in the Human Genome Project (The ELSI Working Group). He is past-president of the American Sociological Association (2004-2005), a member of the Board of Advisors of the Social Science Research Council, and in 2003-2004 served as chair of the Board of Directors of the Association of American Colleges and Universities. He is the former Director of the American Cultures Center and the Institute for the Study of Social Change, both at the University of California, Berkeley.

A Futuristic Projection to 2020 of New and Emergent Social Issues Generated by Research in Human Molecular Genetics

This presentation will explore several social and political implications of emerging developments in human genetic research and databasing. The first will address the new conundrums of attempts by governments and agencies to control chimeric research in an international and global context. The second concerns the increasing movement towards universal DNA databases, for both forensic and health purposes. And finally, the third will address how genetic discoveries will be targeted to specific and "diverse" human populations, raising the specter of the molecular reinscription of social categories of ethnicity and race.



James R. Flynn is Professor Emeritus at the University of Otago and recipient of the University's Gold Medal for Distinguished Career Research. His name has been given to the 'Flynn effect', the documentation of massive IQ gains from one generation to another, and he has been profiled in Scientific American. The American Psychological Association has devoted a symposium and a book to his research. Along with Prof. R J. Sternberg of Yale, he is a Distinguished Associate of the Psychometrics Centre at Cambridge. The International Society for Intelligence Research (ISIR) has named him "Distinguished Scientist of the Year" (2007). His talk is based on his current book, *What Is Intelligence?* (www.cambridge.org). It has been described by Prof. S. J. Ceci of Cornell as "calling into question fundamental assumptions about the nature of intelligence that have driven the field for the past century."

Unpacking Intelligence

There is a line of analysis that runs: there is a general intelligence factor (g); it is based in brain physiology and can be enhanced only by eugenic trends; since trends are dysgenic, pessimism is in order. I will attempt to show:

- (1) That social trends fragment g into a number of cognitive abilities
- (2) That these are functionally independent of one another
- (3) That the brain is more like our muscles than we had thought
- (4) That various cognitive abilities are susceptible to huge gains over time - heritability estimates notwithstanding
- (5) However, that the future may belong to enhanced critical acumen.



Ron M. Fourney

Dr. Fourney received his Ph.D. in Biochemistry and conducted post-doctoral studies in molecular basis of cancer predisposition as a National Cancer Institute of Canada and Alberta Cancer Board Research Fellow. He joined the RCMP as a civilian member and molecular genetics specialist in 1988. Dr. Fourney is a founding member of the RCMP DNA program and has been instrumental in the development and implementation of forensic DNA typing for Canada. He represents the RCMP on numerous national and international committees tasked with the development of DNA identification methods for forensic human identification. He has also played key roles in numerous investigations including organization and management of the SR111 DNA Typing task force for the DNA identification of the victims of the Swissair Flight 111 aircraft disaster. He has continued his interest in enhancing forensic DNA technology and has specialized in fluorescent Short Tandem Repeat detection analysis, robotic automation and comprehensive strategic planning for DNA data banks and high throughput DNA analysis. Dr. Fourney is closely involved with the privacy and security issues of DNA human identification and was a key content expert in the design of the Canadian DNA Data Bank Legislation. Dr. Fourney is currently the Director, National Services and Research Branch which falls under the Forensic Science and Identification Services (FS&IS) of the RCMP National Police Services. His new role includes the research and development of science and technologies to aid in national criminal investigations and the advancement of forensic applications within the National Police Services, as well as internationally, through cooperative partnerships. He is a member of the editorial boards for the Journal of Bio-Techniques and the Journal of Forensic Sciences. He has an academic cross appointment as adjunct professor in the Department of Biology, Carleton University (Ottawa-Carleton Institute of Biology). In April 2006, her Excellency the Governor General of Canada appointed Dr. Fourney Officer of the Order of Merit of the Police Forces in recognition for his contributions to Canada in the field of forensic DNA science.



Claire M. Fraser-Liggett, Ph.D. is Director of the Institute of Genome Sciences at the University of Maryland School of Medicine in Baltimore, MD. Until 2007, she was President and Director of The Institute for Genomic Research (TIGR) in Rockville, MD, which has been at the forefront of the genomics revolution since it was founded in 1992. In a landmark publication in 1995, a group of TIGR investigators reported on the first complete genome sequence of a free-living organism, *Haemophilus influenzae*. With the benefit of 12 years of hindsight, it is clear that this new approach changed the way that microbiology has been studied and has, to date, produced DNA sequence data from nearly 1000 different species across the phylogenetic tree. TIGR quickly became the world's leader in the new field of microbial genomics, attracting significant funding from the DOE, NIH, and NSF to build this program. Her group's success with microbial genomics expanded to include work on other organisms, and TIGR can take credit for playing a key role in the sequencing and analysis of the first plant species, *Arabidopsis thaliana*, and the first human pathogenic parasite, *Plasmodium falciparum*. Dr. Fraser-Liggett's productivity is reflected in her more than 200 publications, and the fact that for the past 10 years she has been the most highly cited investigator in the field of microbiology. But just as important as her large number of highly cited papers is the impact that her body of work has had on our understanding of microbial diversity and evolution. Her contributions to the fields of genomics and microbiology have been acknowledged by many agencies and professional societies and her list of awards include the E.O. Lawrence Award, the highest honor bestowed on research scientists by the Department of Energy, the Promega Biotechnology Award from the American Society of Microbiology, and the Charles Thom Award from the Society for Industrial Microbiology. She has served on many advisory panels for all of the major Federal funding agencies, the National Research Council, the Department of Defense, and the intelligence community. She has served on the Editorial Boards of a number of high impact journals. In addition, she has contributed her time as a Board member for universities, research institutes, and other non-profit groups because of her commitment to the education of our next generation of scientists.



Duana Fullwiley

Duana Fullwiley is an anthropologist of science and medicine concerned with how personal identity, health status, and molecular genetics findings increasingly intersect. She is an assistant professor of Anthropology and African and African American studies at Harvard University. She is currently completing a book manuscript on cultural, genetic, pharmacological and historical bearings on sickle cell disease (and trait) expressions in Senegal, West Africa.

Environments of Human Practice on the Future of Race Thinking

We are entering an era where the first genome transplantation has just been achieved, which has been pronounced as a possible mechanism to resolve humans' great environmental resource and medical dilemmas. We are also living in an era of continued genocide and ethnic divisions in places such as Darfur that have environmental resource scarcity as well as older tensions of human difference, and who matters as 'human,' at their source. How simple organisms can be harnessed and manipulated to provide technological remedies for alternative fuel sources with genome transfers will most likely never fully solve our humanistic challenges, such as racial thinking, that often structure the problems of ethnic violence in regions where feuds over fuel, water, land, etc., are taking place on the planet. Logics of race difference — as fundamentally human, biological difference — seem to re-emerge alongside some of the most stunning advancements and innovations of each era. In this talk I will focus on the genome era and present an ethnographic instance of how scientific work environments, as settings of human practice, help to shape technologies that both deconstruct and reconstruct race thinking in the United States. In so doing, I will reassess the role of the environment, as well as human cultural and economic practices, in geneticists' presentation and re-

presentation of human diversity. For these purposes, human diversity is not merely rooted in forces often talked about as 'natural' (evolutionary, adaptive) biological difference. Rather, it is also linked to processes that are 'unnatural,' meaning that human difference becomes seized upon and commodified as race (or other divisive identifiers) through political and economic stakes that could have happened otherwise. In thinking about the future, we can highlight 'sustainable' ways of representing human diversity in all of its complexity, detail and, to come full circle, similarity.



Dr. Carl Hansen is a new Assistant Professor in the Department of Physics and Astronomy at the University of British Columbia who joins the recently established UBC systems biology initiative. He is a Michael Smith Scholar, an associate faculty member at the Institute for Systems Biology (Seattle) and an associate member of the Michael Smith Labs (UBC). Dr. Hansen directs a research program aimed at exploiting highly integrated microfluidic technologies to address fundamental bottlenecks in biological and medical research. Dr. Hansen helped develop Multilayer Soft Lithography (MSL), a breakthrough fabrication technology which allows for the robust integration of thousands of active microvalves within a compact biochip. He is a co-inventor on 29 US and international patent applications (8 issued) related to microfluidic technologies for biology. Dr. Hansen's previous work in the area of structural biology has resulted in the successful commercialization of the Topaz Crystallization System (Fluidigm Corp.), the first product based on MSL technology. The Hansen lab conducts interdisciplinary research on the development and validation of new technologies for biological research. Current projects included the use of microfluidic devices for high-throughput live-cell imaging, single cell analysis, proteomics and genomics.

Highly Integrated Microfluidic Systems for Systems Biology

Microfluidic devices have the potential to accelerate discovery in the biological and medical sciences by allowing for the complex processing of biological samples on a compact chip format. Through impressive economy of scale and parallelization these devices can increase throughput while dramatically reducing sample consumption and cost. Moreover, the unique properties of the microenvironment can be exploited to enable assays that are difficult or impossible to achieve in macroscopic devices. A recent breakthrough in fabrication techniques called Multilayer Soft Lithography provides a robust and flexible technology platform upon which these devices can be realized. This technique allows for the dense integration of active valves, pumps and logic gates in a soft polymer device. I will discuss our recent work in the development and application of MSL technologies for a variety of applications including single cell genetic analysis, dynamic studies of protein signaling, and structural biology.



Paul Hebert

A native of Kingston, Ontario, Dr. Hebert completed his undergraduate work in biology at Queen's University, his PhD in genetics at Cambridge University and postdoctoral fellowships at the University of Sydney and at the Natural History Museum in London. He took up a faculty position at the University of Windsor in 1976 where he remained until repositioning to the University of Guelph in 1990 where he holds a Canada Research Chair in Molecular Biodiversity. Over his career, Dr. Hebert has served as Director of the Great Lakes Institute at Windsor, as Chair of the Department of Zoology at Guelph, and as Chairman of the Huntsman Marine Science Centre in St. Andrews. He is currently Director of the Biodiversity Institute of Ontario and of the Canadian Barcode of Life Network. Dr. Hebert has authored 275 papers, most employing molecular markers to probe issues related to biological diversity. He has supervised more than 70 graduate students and postdoctoral fellows over his career, has received varied national and international scientific awards and is a Fellow of the Academy of Sciences of Canada.

Fractals, Genomes and a Barcoded World

In contrast to whole genome assembly projects, DNA barcoding represents the acquisition of sequence information for a single gene region across all species. Because of the fractal nature of genomes, barcode data yield insights which are both taxonomically comprehensive and genomically representative. One particularly important barcode application lies in the development of automated systems for species identification. Results from the first 30K animal species reveal that resolution is high and that rule sets are remarkably robust across groups with strident differences in biology. Further, early investigations suggest these conclusions are extensible to other domains of eukaryotic life. Aside from assembling a reference library for life, the next decade promises devices which integrate PCR and sequencing to allow point-of-contact identifications. Furthermore, massively parallelized sequencing platforms will employ barcode libraries to revolutionize our capacity to monitor environmental conditions using biota. Given these prospects, barcoding the world seems irresistible and the International Barcode of Life Project plans to deliver coverage for 500K species within 5 years.



Stefan Jansson is professor in Plant Cell and Molecular Biology at Umeå Plant Science Centre, Department of Plant Physiology, Umeå University, Sweden. He got his PhD at the same University in 1992, studying the gene coding for photosynthetic light-harvesting proteins in Scots pine. During his postdoc in KVL, Copenhagen, Denmark he initiated his project "Structure and function of the chlorophyll a/b-binding proteins" studying the role of the light-harvesting antenna in photosynthesis and photoprotection using *Arabidopsis* as the main model system. During almost 10 years, he has worked with tree genomics (*Populus*). He has there worked with EST sequencing, DNA microarray production, bioinformatics, genetical genomics and also in the *Populus* genome sequencing consortium. His group has, among other things, studied gene expression in trees grown in the field. Lately, the project has been directed towards natural variation, i.e. developing the techniques and strategies to identify the genes behind important traits in trees, using for example association genetics, microarrays and metabolomics.

Natural Variation in *Populus*

Access to the full genome sequence of *Populus* gives new opportunities to study natural variation. *Populus* species are typically dioecious and wind-pollinated, and have therefore in general a higher genetic variation than most other plants typically studied, with high nucleotide diversity, limited linkage disequilibrium and population structure. Their long generation times make also traditional tools like QTL mapping less useful for genetic analysis. We are studying natural variation in the most widespread of the European *Populus* species, aspen (*Populus tremula*). We study several traits related to biotic interactions, for example herbivory. We also seek to study natural variation in leaf metabolite composition, and its relation to fungal endophytes. Our model trait, however, is phenology, in particular autumn senescence that is under tight genetic control and where strong latitudinal clines are present. We are also exploiting association mapping as a tool for connecting phenotypic and genotypic variation, and study variation in gene expression levels.



Dan Janzen

While originally focused on tropical animal-plant relationships (1963-1985), Janzen is more recently conducting an inventory of tropical caterpillars, their parasites, and their food plants, and does research on the conservation of tropical biodiversity through its non-damaging development (see <http://janzen.sas.upenn.edu>). His 430 publications encapsulate much of this. He and his biologist wife, Winnie Hallwachs, are architects of Area de Conservación Guanacaste (ACG) in northwestern Costa Rica (<http://www.acguanacaste.ac.cr>), decreed a UNESCO World Heritage Site in 1999. Janzen received the first Crafoord Prize in biology from the Swedish Royal Academy of Science (1984), and the Kyoto Prize in Basic Biology (1997). A member of the US National Academy of Sciences (1992) and honorary member of the Costa Rican National Park Service, he promotes the relevance and potential of conservation of tropical wildland biodiversity for global understanding, national sustainable development, and individual quality of life. His current focus is finding the funds to endow the entire national park system of Costa Rica, and facilitating global bioliteracy through the emergence of the ability by all people to be able to identify any organism anywhere anytime through DNA barcoding.

Wild Biodiversity through a New Microscope - the Barcode: Some ethical, environmental and economic implications of a new way of seeing what you are looking at

The world is on the verge of losing its human-brain-based ability to know which species is where, when and doing what, at a given moment in real time. Fortunately, through global technological advances we have regained the ability to apply what we have learned over the past three centuries, and construct a whole new knowledge capacity with and on top of these building blocks. This technological, and to some degree philosophical, "rescue" of the taxonomic process has arrived just in the nick of time. As our taxonomic skills were diminishing, societal pressure was gradually reducing the number of surviving species to classify. It seems that whatever is not explicitly protected, disappears. How ironic that we now have the technology to enable people everywhere to actually "read" nature, a bioliteracy once possessed by only a very few fanatics. Introducing bioliteracy to the world at large means that people will have the opportunity to be vastly better informed about their environment, for better or for worse. Will it have the same impact, for example, as teaching first-graders to read or the invention of the printing press? How dramatically will this new literacy affect humanity's ethical, environmental and economic relationship to wild biodiversity?



Wendy Johnson grew up in Tacoma, Washington, the eldest of three daughters of a civil engineer and psychologist/special education teacher. She graduated from Occidental College in Los Angeles, California, with a degree in mathematics. She spent several years as a commercial insurance underwriter and consulting pension actuary in San Francisco and Los Angeles, and then settled down as a consulting casualty actuary for Coopers and Lybrand in San Francisco. She married Glenn Evans in 1989, and they have a daughter and son. With Glenn, she founded Pacific Actuarial Consultants in 1991, and they maintained this actuarial practice together until 2001. In 1995, however, Wendy began to study psychology at San Francisco State University, and obtained a Master's Degree in Developmental Psychology there in 1999. She entered the doctoral program in the Psychology Department at the University of Minnesota in 2000, and completed her degree there in 2005. She pursues research involving the structure and nature of general intelligence and special mental abilities, personality structure and development, antecedents of individual and sex differences in academic achievement, antecedents of later-life health and psychological well-being, and contributions of cognitive ability to later-life outcomes, with particular emphasis on understanding the underlying transactions between genetic and environmental influences. She is currently a Research Council of the United Kingdom Fellow in the Department of Psychology at the University of Edinburgh.

Genetic and Environmental Influences on the Wrong Models of Cognitive Ability and Personality

Investigators interested in the structures of cognitive ability and personality often point to the presence of stable or substantial genetic and environmental influences and correlations as evidence for the accuracy or relevance of their phenotypic models. In this talk I will show that even a demonstrably inaccurate structural model of cognitive ability and even random sets of personality items show evidence of genetic and environmental influences and correlations comparable to those from carefully compiled and validated models of cognitive ability and personality scales. This calls into question the accuracy of the inference that the presence of genetic influences indicates that traits have been accurately measured. It also suggests that genetic correlations will tend to parallel phenotypic correlations, and that the underlying reasons for genetic correlations are the same as those that underlie phenotypic correlations. That is, genetic influences on one trait may also affect the other or vice versa, or genetic influences on some third trait may also affect the two traits of primary interest, and we cannot distinguish among these possibilities with the genetic correlation alone. I will also provide data comparing differences in cognitive ability and personality between

members of monozygotic and dizygotic twin pairs and differences between randomly matched pairs. The data indicate that, though twins are more similar in these traits than random pairs, the extent to which this is true is surprisingly small. This suggests that genetic influences on behavior are more subtle than many people expect and helps to explain the difficulty we have had in identifying the genes for cognitive ability and personality.



Bartha Maria Knoppers

Bartha Maria Knoppers, PhD, holds the Canada Research Chair in Law and Medicine and the Chaire d'excellence Pierre Fermat (France). She is Professor at the Faculté de droit, Université de Montréal and Senior Researcher at the Centre de recherche en droit public (CRDP). She is a graduate of McMaster University (B.A.), University of Alberta (M.A.), McGill University (LL.B., B.C.L.), Cambridge University, U.K., (D.L.S.), Sorbonne Paris I) (Phd.) and was admitted to the Bar of Québec in 1985. She has authored numerous articles and books. Professor Knoppers is former Chair of the International Ethics committee of the Human Genome Organization (HUGO), (1996-2004). She was a member of the International Bioethics Committee of the United Nations, Educational, Scientific and Cultural Organization (UNESCO) which drafted the Universal Declaration on the Human Genome and Human Rights (1993-1997). She is also Co-Founder of the International Institute of Research in Ethics and Biomedicine (IIREB). From 2000-2006 she served on the Board of Genome Canada, and in 2003, became Chair of the Ethics Working Party of the International Stem Cell Forum. Professor Knoppers received a Doctor of Laws Honoris Causa from the University of Waterloo (2001), a Doctor of Medicine Honoris Causa from Université de Paris V (René Descartes) (2002) and a Doctor of Laws Honoris Causa from McMaster University, Ontario (2007). In 2002, she was elected Fellow of the American Association for the Advancement of Science, was selected as one of the 50 nation builders in Canada by the Globe and Mail, and named Officer of the Order of Canada. She founded the international Public Population Project in Genomics (P3G) in 2003. In that same year, she was elected Fellow of The Hastings Center (Bioethics), New York, member of the International Ethics Committee of the World Anti-Doping Agency and, in April 2005, Fellow of the Canadian Academy of Health Sciences (CAHS).

Across the Genomes: Whither "Identifiability"?

While both the complexity and yet convergence of genome research are constantly reaffirmed, co-adaptation and co-evolution are not without consequence for policy debates. Examining some of the social constructs used to describe the ethical issues raised, will set the stage for determining the feasibility of concepts such as common heritage, global public goods and open-access databases. These latter approaches assume certain principles based on communitarian values. Even if affirmed and proven to be feasible, is the realization of these values and approaches threatened by the increasing ability to "re-identify" research participants, as made possible by the resulting international, large-scale genomic projects?



Ben Koop is Professor in biology in the Department of Biology and Medical Science at the University of Victoria. Dr. Koop obtained his Ph.D. from the University of Wayne State Medical School in 1988. Dr. Koop is originally from Fort St. John, B.C. Dr. Koop's research interests lie in the fields of Molecular Biology, Genetics and Evolution. Dr. Koop's research looks at broad molecular evolutionary problems using a very highly multidisciplinary approach. In 2001, Dr. Koop received the Natural Sciences and Engineering Research Council Steacie Fellowship – one of Canada's premier science and engineering prizes and is currently a Canada Research Chair.

Applications of Genome Research in Salmonids (cGRASP) to Environmental Change

The cGRASP project is designed to provide a foundation for understanding the genome of salmon with a focus on Atlantic salmon. The genomic information gained from one species has been shown to be applicable to other salmonid species and will benefit conservation and enhancement of wild stocks, aquaculture and environmental assessments. Genomic resources also enable us to address fundamental scientific questions concerning the evolution of salmonid genomes, and will facilitate monitoring the expression of genes and proteins in a wide variety of natural and altered environments. Towards these goals, a physical map from 250,000 fingerprinted BAC clones has provided 4200 contigs. Complete characterization of the duplicated MHC class I loci and the TCRA locus provide information on the impact of genome duplication in the salmon. In addition, more than 70 cDNA libraries have been constructed from a wide variety of tissues and different developmental stages. From these and other libraries more than 430,000 DNA sequence reads from both the 5' and 3' ends of these expressed sequence tags (ESTs) have been completed. An analysis of contigs, gene IDs, gene families and polymorphisms is underway. These sequences have been combined into over 81,000 unique transcripts in Atlantic salmon and are being examined

together with over 51,000 unique contigs/genes in Rainbow trout. With the EST sequences, a microarray of 16,000 cDNAs has been constructed and initial expression studies have indicated that the array works very well for all salmonids and even some more distant species. Preliminary studies have also shown the array to provide a wealth of new data in the study of cellular and tissue responses to pollutants, diseases and stress, as well as in the study of reproduction and development. Initial results show a great sensitivity of gene expression patterns to small environmental, disease challenges and physiological changes. Microarrays provide a powerful new tool in environmental, conservation, physiological and fish health studies.



David Layzell

Dr. Layzell is a world-renowned plant physiologist whose recent work explores how biological systems can address the challenges of climate change and clean energy. His work has led to the invention of a number of novel scientific instruments and technologies, resulting in seven U.S. patents and the launching of Qubit Systems Inc., a successful Queen's University spin-off company. By coupling elegant empirical measurements of plant metabolism with mathematical models of relevant biophysical processes, Dr. Layzell has identified and described aspects of plant metabolic control that were previously unknown, especially in the area of biological nitrogen fixation and assimilate partitioning in plants. A recipient of the C.D. Nelson Award from the Canadian Society of Plant Physiologists, and an NSERC Steacie Fellowship, Dr. Layzell's research accomplishments have also been recognized with his induction to the Royal Society of Canada. He has also served as Chair of several NSERC Committees and on the Management Boards of a number of national research networks. Dr. Layzell is Founder, President and Chief Executive Officer of the BIOCAP Canada Foundation, a national, not-for-profit organization that coordinates funds and communicates bioeconomy research and commercialization focused on addressing priorities for climate change and energy security. Since 2002 and with the support of industry, provincial and federal governments, BIOCAP has invested about \$8M to leverage over \$50M (cash) in research to support the work of 270 faculty and 300+ graduate students at 35 universities across Canada.

Towards a Transformative and Sustainable Bioeconomy: Opportunities for Genomics Research

Climate change and the supply of clean energy are among the greatest challenges facing human society in the next century. Canada's vast biological resources can be managed and used to provide solutions in the form of carbon sinks, greenhouse gas emission reductions and biomass energy and fuels. This presentation will explore the magnitude of both the challenge and opportunity in Canada, and then identify some of research areas that would benefit from work in the field of genomics.



Educated as a biologist, Jean Lemire has over 20 years of experience in marine biology under his belt. As a scientist affiliated with the University of Hawaii, and at the Mingan Island Cetacean Study Research Station, his knowledge of whales led to his participation in a great many research projects in the international arena. In 1987, he began a parallel career in filmmaking and founded Les Productions Ciné-Bio, a firm specializing in scientific consulting, research and script development for documentary productions. In 2001, he founded Les Productions Glacialis inc., a new production company specializing in natural sciences, marine science, arts and culture. In 2003–2004, he produced the following films: *Short Infinity* and *Arctic Mission*, a collection of five documentary and feature-length films to be shown in movie theatres, on the effects of climate change in the Arctic. *Arctic Mission* met with seldom-seen popular acclaim. It reached hundreds of millions of television viewers around the world and received the prestigious Earth Watch award, presented by the National Geographic Society. In 2006, he co-produced and co-directed *The White Planet*, a feature-length environmental film to be shown in movie theatres. Sold in nearly 60 countries, this film is a powerful visual testimonial on the threatened world of the Arctic, the first and hardest-hit casualty of global climate change. In 2006, he was voted personality of the year by La Presse/Radio-Canada, in the category Soft Science, Pure Science and Technology. The readers of the popular magazine, *Sélection du Readers Digest*, named him Hero of the Year 2006 in the environmental area. In Autumn of 2005, Jean Lemire embarked on his greatest adventure to date. On board the *Sedna IV*, accompanied by a crew of sailors, filmmakers and scientists, he went off in conquest of Antarctica, the last continent. Antarctic Mission would become one of the great expeditions of modern times: 430 days of sailing, isolation and extreme adventures. A feature-length film to be shown in movie theatres, a documentary series and a series on the adventure are now being put together.



Simon Letarte

Simon Letarte is a Senior Research Scientist at the Institute for Systems Biology (ISB) in Ruedi Aebersold's group. He obtained his B. Sc. in chemistry at Laval University and his Ph.D. under the supervision of Michel Bertrand at University of Montreal, where he applied mass spectrometry for rapid bacteria identification. He got involved in proteomics during his postdoc at the Montreal Proteomics Network with Michel Desjardins, where he studied changes in protein expression during phagosome maturation. His interests at the ISB are quantitative mass spectrometry-based proteomics, metabolomics and biomarker discovery. More specifically, he has been developing methods for large-scale proteome comparisons, including label-free quantification and targeted peptide sequencing. Simon also uses a Multiple Reaction Monitoring (MRM) platform for absolute protein quantification, where preselected proteotypic peptides are produced with heavy isotopes, spiked into serum and used to measure endogenous protein concentrations

Mass Spectrometry Proteomics: New Enabling Technologies for Biomarker Discovery and P4 Medicine

Traditional mass spectrometry-based proteomics has plateaued in recent years. The decade-old model of automatically selecting the few most intense peptides and fragmenting them to infer amino acid sequences is inefficient for biomarker discovery: abundant proteins are identified over and over again, casting a shadow over the low abundance, biologically relevant proteins that go unnoticed by the mass spectrometer. Comparative proteomics of multiple disease states is better achieved by label-free, MS1 proteome comparisons followed by targeted analysis of statistically significant, differentially abundant peptides. New software such as the Corra platform has emerged over the last years to make those kind of analyses possible for large sample populations. Multiple Reaction Monitoring (MRM) is also a new paradigm, where triple quadrupole mass spectrometers are used to rapidly jump from precursor-fragments pairs (transitions) as a confirmation of the amino acid sequence. Monitoring specific transitions is achieved faster and with more sensitivity than scanning the entire mass range for every peptide. The MRM approach requires careful preparation beforehand to select which proteins to look for. However, it lends itself to absolute quantification by spiking known amounts of heavy labeled peptides and comparing the ratio between the

two isotopologues to infer absolute quantification of endogenous peptides. MRM is useful for biomarker discovery and validation, but the potential of measuring absolute amounts of proteins over time is really what opens the door to proteomics in P4 medicine. One can envision a future where, once a panel of proteomic and metabolomic biomarkers have been identified for most common diseases patients can assess their health status on an individual basis by using medical devices derived from mass spectrometry technology. Immersive data visualization and pattern recognition will help highlight the changes and cross-reference it to patient history, genetic background and public databases.



Dr. Stephen Minger is the Director of the Stem Cell Biology Laboratory and a Senior Lecturer in the Wolfson Centre for Age Related Diseases at King's College London. Dr. Minger received his PhD in Pathology (Neurosciences) in 1992 from the Albert Einstein College of Medicine. From 1992-1994, he was a post-doctoral fellow at the University of California, San Diego, where he first began to pursue research in neural stem cell biology. In 1995, Dr. Minger was appointed an Assistant Professor in Neurology at The University of Kentucky Medical School. He moved his stem cell research programme to Guy's Hospital in 1996 and was appointed a Lecturer in Biomolecular Sciences at King's College London in 1998. Over the last 15 years, his research group has worked with a wide range of somatic stem cell populations, as well as mouse and human embryonic stem (ES) cells. In 2002, together with Dr. Susan Pickering and Professor Peter Braude, Dr. Minger was awarded one of the first two licenses granted by the UK Human Fertilisation and Embryology Authority for the derivation of human ES cells. His group subsequently generated the first human embryonic stem cell line in the UK and was one of the first groups to deposit this into the UK Stem Cell Bank. They have gone on to generate five new human ES cell lines, including one that encodes the most common genetic mutation resulting in Cystic Fibrosis and another one that contains the Huntington's disease mutation. In addition to the derivation of human ES cell lines, the Stem Cell Biology Laboratory is focused on the generation of a number of therapeutically relevant human somatic stem cell populations from embryonic stem cells. These include cardiac, vascular, retinal, mesenchymal and neural stem/progenitor cell populations, as well as pancreatic β -cells and oligodendrocyte progenitors. The Stem Cell Biology research team has established a number of significant collaborations with biological and clinical scientists throughout the UK specifically related to clinical translation of stem cells for a wide variety of human disorders. Dr. Minger is also one of the co-organisers, together with Dr. Chris Mason of UCL, of the London Regenerative Medicine Network, a grass-roots, research-led organisation designed to stimulate clinical translation of cell- and gene-based therapies within London. He is also the Senior Editor of Regenerative Medicine, a new journal launched in Jan 2006 by Future Medicines, which recently won the 2006 ALPSP/Charlesworth Award for Best New Journal. Stephen is also the Stem Cell Expert and Member of the UK Gene Therapy Advisory Committee (GTAC) at the Department of Health. Research of the Stem Cell Biology Laboratory is supported by the UK Medical Research Council, The European Union, GlaxoSmithKline PLC, The Novartis Institute, The Oliver Bird Foundation, The UK Department of Trade and Industry, The Huntington's Disease Association, The HighQ Foundation, The Alzheimer's Research Trust, The Charitable Foundation of Guy's and St Thomas' Hospitals, the BBSRC, and the EPSRC amongst others.

Therapeutic and Research Potential of Human Stem Cells

Stem cells offer great therapeutic potential for human disease conditions where the loss of specific cell types is the major pathophysiological feature of individual disorders. Embryonic stem (ES) cells derived from 6-8 day old human embryos offer the most therapeutic potential as these cells are capable of generating every cell and tissue type in the human body. If we can control the differentiation of ES cells, then cell replacement for profound human disorders such as Parkinson's disease, insulin-dependent diabetes, heart disease, stroke, multiple sclerosis, rheumatoid arthritis, spinal cord damage, and macular degeneration could become standard new therapies. This presentation will examine the state of the art in the

development of stem cell therapies and outline some of the technical and ethical considerations as we progress toward clinical and research application of human ES cells.



Ulf Nehrass

After studying Biochemistry at Cambridge University (UK), Ulf Nehrass moved to EMBL (Heidelberg, Germany) and obtained his PhD from Heidelberg University. His graduate work under Prof E.C. Hurt led to the functional determination of the first nucleoporin. This study was thematically followed in his postdoctoral work with Nobel Prize Laureate Günter Blobel at Rockefeller University (New York, USA), where Dr. Nehrass focused on the molecular mechanisms of nuclear transport, as well as the structural organization of nuclear envelope components. In 1998, Dr. Nehrass started an independent group at the Institut Pasteur in Paris, where he began working on nuclear structure function relationship. In a number of publications featuring advanced microscopy and imaging approaches, his group zoomed in on nuclear structure function relations, showing that the spatial organization of the nucleus constitutes an integrated level of gene regulation. Employing molecular genetics and live tracking of genes during activation stages, the Nehrass group eventually provided formal proof of Blobel's gene-gating hypothesis, which he first proposed in 1985. After becoming the Chef d'Unité and Directeur de Recherche in the Paris Institute, in 2004 Dr. Nehrass became Director and CEO of the newly founded Institut Pasteur Korea located in Seoul. Addressing infectious and chronic disease models at the level of the cell, Institut Pasteur Korea exploits advanced visualization and imaging tools for therapy development and systems biology. Featuring a chemical genetics platform, screening facilities and a medicinal chemistry department, Institut Pasteur Korea has developed into a world leading translational research center discovering both the molecular underpinnings of disease and new therapeutic interventions.

From Drugs to Genes: Visualizing Chemical Genomics



Anthropologist Hendrik Poinar has received international acclaim and media attention for his research on two fronts: the discoveries he's made about ancient humans from their fossilized remains; and the work he's done determining the timing and origin of the human immunodeficiency virus (HIV) from some of the oldest samples of archival HIV, collected between 1959 and 1980. An assistant professor in the departments of Anthropology, and Pathology and Molecular Medicine at McMaster University in Hamilton, Ontario, Poinar moved from the Max Planck Institute for Evolutionary Anthropology in Germany, where he was a post-doctoral fellow from 2000-2003. He also completed a post-doctoral fellowship at Oregon State University from 1999-2000, after obtaining his Ph.D. from the Ludwig Maximilians Universität München. As head of a world-class molecular anthropology lab that will devise novel techniques to extract information from ancient DNA, Poinar's own specialty is the chemical and molecular analysis of fossilized feces, or coprolites. His findings are used to address evolutionary and anthropological questions that range from the diet and gender differences of hunter-gather populations to the language abilities of Neanderthals to whether or not early humans interbred with Neanderthals. Recently Poinar and colleagues were the first to use a metagenomics approach to sequence portions of the woolly mammoth genome. Using these high-throughput parallel sequencing machines, it will be possible to sequence entire genomes of extinct organisms, and measure evolution in action. The author of more than 40 publications, including book chapters and articles in leading journals such as Nature and Science, Poinar's research has attracted significant grants from the provincial and federal governments and he has also been named the winner of the 2004 Petro-Canada Young Innovator Award.

Our Frozen Heritage: Metagenomics to Paleogenomics

The permafrost (perennially frozen soil) regions of Eastern Beringia, unglaciated Yukon Alaska and Siberia, preserve some of the most remarkable archives of geologically and biologically relevant information in the northern hemisphere. Among these archives are sub-fossil forest beds, soft tissues and bones of extinct and extant mammals, birds, exceptionally preserved plant and insect fossils, and — especially remarkable — biological trace fossils in the form of isolated DNA fragments, proteins, and other metabolic byproducts. New parallel sequencing machines designed for metagenomic analyses can be used to mine the permafrost of all frozen "in-time" DNA and RNA molecules, which can shed light on past climates (insect DNA, plant DNA), migration patterns of large megafauna (i.e. mammoths) and the time lines of their extinction from our planet. In addition with well preserved remains of plants and animals stored in permafrost over hundreds of thousands of years, it is theoretically possible to reconstruct the genomes of these extinct organisms and address fundamental questions on adaptation, selection and evolution over geologically meaningful time spans.



Nancy Press

Nancy Press received her B.A. degree from Sarah Lawrence College with an emphasis on social psychology. After working in publishing for several years, she entered a graduate program in cultural anthropology, receiving her Ph.D. from Duke University in 1986. This was followed by an NIMH postdoctoral fellowship in medical anthropology, awarded at the Mental Retardation Research Center of UCLA's Neuropsychiatric Institute where she achieved the rank of Associate Professor before accepting an appointment at the Oregon Health & Science University in Portland, Oregon. She is currently a professor in the Schools of Nursing and Medicine at that institution. Dr. Press has been a major contributor to the development of the field of the ethical, legal and social implications of genetics (ELSI). She served on the NIH-DOE Task Force on Genetic Testing, the ELSI Research and Program Evaluation Group (ERPEG), and was co-chair of the Implementation Workshop of the Genetic Testing for Cystic Fibrosis Consensus Development Conference. She is senior author on JAMA and Journal of Clinical Oncology papers produced by Principal Investigators of the 15-study, Cancer Genetics Studies Consortium. Working in the fields of reproductive genetics, behavioral genetics, and cancer genetics, the thread which ties Press' work together is the question of how cultural factors shape the choices and understandings of scientists, clinicians, and the general public in developing, using, and reacting to genetic information.

Phenotypes, Social Constructionism, and the Spurious Finding: Behavioral Genetics in Social Context

A primary challenge for behavioral genetics is the selection and definition of phenotypes for which genotype correlations are sought. The concepts of *social constructionism* and *medicalization* can help explain how certain behaviors become defined and thus become available for a search for their genetic underpinnings and why those searches have had only limited success. This talk will examine these issues and attempt to delineate what the social consequences, for scientists and for the broader society, are of the failures as well as the continued attempts.



Richard Rachubinski is Professor in the Department of Cell Biology at the University of Alberta. He received his Ph.D. from McGill University and did postdoctoral work both at McGill and at the Rockefeller University. He returned to Canada to join the Department of Biochemistry at McMaster University and assumed his current position in 1993. He is a leader in cell biology in the areas of protein targeting and organelle biogenesis. He has redefined our views of the formation of cell membranes and the movement of proteins across them. His studies on the peroxisome, an organelle essential for lipid homeostasis, have led to the identification of numerous mutations causing lethal genetic disorders, collectively called the peroxisome biogenesis disorders. He has assembled within the Department of Cell Biology a group of outstanding young biologists investigating the assembly of the varied membrane compartments making up the cell. He has been the recipient of numerous awards for scientific excellence and is currently Canada Research Chair in Cell Biology and an International Research Scholar of the Howard Hughes Medical Institute. He was made a Fellow of the Royal Society of Canada in 2002.

Systems Biology Approaches to an Understanding of Peroxisome Function

The peroxisome is an ubiquitous organelle that compartmentalizes a variety of important biochemical functions, notably the oxidation of fats and the inactivation of reactive oxygen species. The requirement for functional peroxisomes is underscored by the lethality of a group of inherited peroxisomal disorders, collectively called the peroxisome biogenesis disorders, in which peroxisomes fail to assemble. One of the exceptional properties of peroxisomes is their highly dynamic nature with respect to changing environmental conditions.

We have recently undertaken systems biology approaches using baker's yeast *Saccharomyces cerevisiae* to try to achieve a global and predictive understanding of peroxisomal assembly, function and dynamics. In our practice of systems biology, we aim to enumerate and quantify all relevant molecular constituents and their interactions involved in the global response governing peroxisome induction; computationally and mathematically integrate different data types; mathematically model our biological system; and test predictions arising from our systems analysis of peroxisomes. Our systems biology approaches have centered on 1) transcriptional networks, with a focus on parallel combinatorial control to control timing of the transcriptional response with respect to changes in

the environment; 2) quantitative proteomics using isotope-coded affinity tags to evaluate the enrichment of proteins in peroxisomes and define bona fide time- and condition-specific constituents of peroxisomes (proteins move); and 3) a comprehensive screen of the yeast gene deletion library expressing a fluorescently labeled peroxisomal protein to reveal the complexity of the peroxisomal biogenic response through quantitative imaging and to identify novel aspects of, and players in, peroxisome biology. Our ultimate goal is a reliable and predictive model of the peroxisome with regard to its biogenesis, function and response in an ever changing environment.

This work was supported by grants from the Canadian Institutes of Health Research and the Howard Hughes Medical Institute to R.R. and the National Institutes of Health to J.A.



David Relman

David Relman, M.D., is associate professor of medicine, and of microbiology and immunology at Stanford University. He is also chief, infectious diseases section, at the VA Palo Alto Health Care System in Palo Alto, California. His research is directed towards the characterization of the human indigenous microbial communities, with emphasis on understanding variation in diversity, succession, the effects of disturbance, and the role of these communities in health and disease. This work brings together approaches from ecology, population biology, environmental microbiology, genomics and clinical medicine. In addition, his research explores the classification structure of humans and non-human primates with systemic infectious diseases, based on patterns of genome-wide gene transcript abundance in blood and other tissues. The goals of this work are to recognize classes of pathogen and predict clinical outcome at early time points in the disease process, as well as to gain further insights into virulence. Past scientific achievements include the description of a novel approach for identifying previously-unknown pathogens, the identification of a number of new human microbial pathogens, including the agent of Whipple's disease, and some of the most extensive analyses to date of the human indigenous microbial ecosystem. (see <http://relman.stanford.edu>). Among his other activities, Dr. Relman currently serves as Chair of the Board of Scientific Counselors of the National Institute of Dental and Craniofacial Research (NIH), Chair of the Institute of Medicine's Forum on Microbial Threats (U.S. National Academies of Science), member of the National Science Advisory Board for Biosecurity, and advises several U.S. Government departments and agencies on matters related to pathogen diversity, the future life sciences landscape, and the nature of present and future biological threats. He is a member of the American Academy of Microbiology. Dr. Relman received the Squibb Award of the IDSA in 2001, and was the recipient of both the NIH Director's Pioneer Award, and the Distinguished Clinical Scientist Award from the Doris Duke Charitable Foundation, in 2006.

Exploration of the Human Indigenous Microbial Communities

Complex microbial ecosystems occupy the cutaneous and mucosal surfaces of humans. Recent advances have highlighted both the tremendous diversity of these communities and their importance to host physiology, yet, we have only scratched the surface. Questions remain about the ecological processes that establish and maintain the human microbiota throughout life. Furthermore, basic features of the human microbial ecosystem remain poorly described, including variability in diversity, in space and time. Host individuality imposes a strong signature on patterns of diversity. In turn, our indigenous microbial ecosystem defines who we are as individuals. Assembly of the oral and the gut microbiota may also involve both stochastic historical events and contemporary environmental factors. Approaches that combine community ecology, molecular microbial ecology, and metagenomics may improve our understanding of health and disease within the communal human organism. By understanding the patterns of diversity associated with human health, we may be able to preserve and restore health more effectively. By recognizing the early signs of impending disturbance, we may be able to predict and avoid disease."



Mostafa Ronaghi is a principal investigator at Stanford Genome Technology Center. He has developed several molecular tools, which are widely being used. He has co-founded four companies and helped several companies in the start-up process. In June 1997 he co-founded Pyrosequencing AB (Changed the name to Biotage in 2003), a genomics company that develops and markets Pyrosequencing machine and its chemicals. Pyrosequencing AB had an IPO in June 2000 in Stockholm Stock Exchange after three rounds of financing and since then has acquired three other companies. Dr. Ronaghi co-founded ParAllele BioScience in November 2001. ParAllele BioScience developed and marketed a highly multiplexed technology for genic testing. ParAllele BioScience was acquired by Affymetrix in May 2005 after two rounds of financing. Dr. Ronaghi co-founded NextBio in 2004. NextBio is a specialized search engine for life science data. NextBio is a venture-backed company with thousands of users shortly after product release. Dr. Ronaghi started Avantome, a DNA sequencing company. Avantome will develop product enabling DNA sequencing at any lab performing biological research. He earned his Ph.D. from The Royal Institute of Technology. Dr. Ronaghi holds more than 20 pending and issued patents and has written more than 50 peer-reviewed publications.

Advanced Technologies for Determination of Biodiversity

At Stanford Genome Technology Center, we are developing new technologies for high-throughput biological applications. In this presentation, we discussed development of two portable devices for studies of unknown and known flora. One portable device is using a CMOS-based image sensor for detection of light generated from Pyrosequencing reaction which is emerging as a standard technique for rapid determination of the content of an unknown flora. This technology is very powerful for massive sequencing of hundreds of thousand of targets in a single run, however, it may be expensive for frequent monitoring of a flora. We are also developing inexpensive assays based on hybridization. Development, advantages and disadvantages of these technologies will be discussed.



Steven Rothstein

Dr. Steven Rothstein has done research in plant molecular genetics including work in plant nutrition and reproductive biology. Dr. Rothstein received a bachelor's degree in chemistry from Swarthmore College, Swarthmore, Pennsylvania, in 1975; a PhD from the University of Wisconsin, Department of Biochemistry, in 1980; and did his post-doctoral training at the Plant Breeding Institute in Trumpington, Cambridge, England, from 1980 to 1983. From 1984 to 1988 he was a scientist for Ciba-Geigy Corporation (Novartis) and then became an associate Professor in the Department of Molecular Biology and Genetics at the University of Guelph. Steven was also the NSERC Industrial Research Chair. In 1995, Dr. Rothstein became a full Professor and Department Chair and held this position until 1998. From 1998 to 2001 he served as the Trait and Technology Research Director of Agronomic Traits at Pioneer Hi-Bred. In this position, he was responsible for research in using genomics to enhance breeding selection and in the development of transgenic corn with enhanced agronomic traits. Currently, Dr. Rothstein is a Senior University Research Chair and Professor in the Department of Molecular and Cellular Biology, University of Guelph. Nitrogen fertilizer is required for high crop yields but is also the single largest economic cost for farmers and its wide-scale use leads to an array of environmental problems. Much of Dr. Rothstein's current research is focused on using a variety of approaches including functional genomics on both model systems like Arabidopsis and on corn to try to develop lines with improved nitrogen use efficiency. This would allow the maintenance of high yields while lowering the economic and environmental costs.

Using Genomic Knowledge to Improve Crop Plants during a Time of Environmental Challenges

Crop production world-wide has increased based on a combination of enhanced crop genetics and the use of fertilizers and other agricultural chemicals. This augmentation of production was sufficient to allow for a large increase in human population and living standards. However, over the last decade it has become increasingly difficult to meet the continually increased usage of the important grain and oilseed crops in an economically and environmentally sustainable fashion. Crop production has a profound effect on many environmental parameters from land use to water and air pollution to the production of greenhouse gases. At the same time, environmental changes can have a profound effect on crop production, particularly changes that affect the availability of water and nutrients. To help deal with this issue, we are interested in using gene knowledge to improve complex multi-gene agronomic traits in important crop plants. There are two basic approaches: first one can develop a greater understanding of the extant genetics using a variety of genomic tools along with detailed phenotypic analyses; second, one can generate new genotypic variation via transformation. Different approaches for these will be described for maize that can be used for any complex trait of interest. The efficient use of nitrogen fertilizer is one such trait and is the

focus of much of our work and will be used as an example to describe what needs to be done to accomplish trait improvement. Its importance is due to the fact that the purchase of nitrogen fertilizer is the single largest input cost for farmers for many crops and its use leads to significant environmental problems. In addition, in many developing countries, the cost of nitrogen fertilizer is prohibitive making it extremely important to use what is available in the most efficient way possible.



Eddy Rubin is Director of the Department of Energy's Joint Genome Institute and Director of the Genomics Division at the Lawrence Berkeley National Lab. He received his B.A. in physics from UC San Diego, his M.D. from the University of Rochester Medical Center, and his Ph.D. in biophysics from the University of Rochester. In 1988 he joined the Berkeley Lab to develop computational and biological approaches to the analysis of DNA sequence data. From sequence interpreter he became a sequence generator, leading the Joint Genome Institute's contribution of a significant fraction of the human genome's finished sequence. Under his direction JGI sequences organisms from extreme-philés and primitive plants and animals to his own work, which includes relatives of humans such as extinct *Homoneanderthalensis*.



Eric Turkheimer

Eric Turkheimer received his B. A. from Haverford College in 1976 and studied clinical psychology and behavior genetics under Lee Willerman and John Loehlin at the University of Texas at Austin. After completing his Ph.D. at the University of Texas in 1985 and a clinical internship at the University of California, San Francisco in 1986, he accepted a faculty position in the Department of Psychology at the University of Virginia, where he is currently Professor and Director of Clinical Training. Turkheimer is an associate editor for Psychological Assessment, and has served on the editorial boards of Journal of Abnormal Psychology and Developmental Psychology. His overarching research goal is to explore the possibilities and limitations of behavior genetics as a means of expanding the scope and rigor of human behavioral science. His research has encompassed many of the substantive and methodological themes common to behavioral genetic researchers: data from adoptees, twins, siblings, parents and children to investigate intelligence, personality, psychopathology and family dynamics; experimental and quasi-experimental research designs, statistical modeling, synthesis of empirical results, and, perhaps most characteristically, philosophy of science. His current research includes detection of G by E interactions in twin studies of intelligence, development of statistical methods for analyses of children of twins, and the use of twins to establish quasi-experimental control in studies of developmental associations between parenting behavior and offspring outcomes in adolescence.

Behavior Genetics Finally Delivers as a Tool for Understanding the Environment

From the very beginning, behavior geneticists have argued that the genetics of behavior was applicable not only to genetics but also to studies of environmental influences on behavior. If we could show that some variation in behavior is attributable to genetics, the argument went, the rest would have to be attributable to the environment, and environmental scientists could study that free of the distracting confounds of genetic influence. Somehow it never happened: behavior genetics was half a century's worth of everything turning out to be more genetic and less environmental than anyone thought. I propose that a new model of behavior genetics, directed at purported causal relationships between risk factors and outcomes in development, has finally delivered on the promise. The appropriate goal of human behavior genetics has never been to identify genetic influence; rather it has been to isolate causation in a naturalistic field where experimentation is usually impossible.



Dr. Lorne Tyrrell holds the CIHR/GSK Chair in Virology in the Department of Medical Microbiology and Immunology at the University of Alberta. He has focused his research since 1986 on viral hepatitis. His work on the development of antiviral therapy was supported by CIHR and Glaxo Canada. It resulted in the licensing of the first oral antiviral agent to treat chronic hepatitis B infection – lamivudine – in 1998. Today, lamivudine is licensed in over 170 countries worldwide for the treatment of HBV. In 1998, Dr. Tyrrell collaborated with Drs. N. Kneteman and David Mercer to develop the first non-primate animal to support hepatitis C replication. This animal model is mice with human/mouse chimeric livers. This animal model supports HCV, HBV, and malaria replication. Dr. Tyrrell is now focusing on the viral and host genetic factors that lead to chronic HBV or HCV infection. The mouse model is proving very useful in these studies. From these studies we should develop a better understanding of why some people clear HCV infection and others become chronic carriers. These results may also provide accurate predictions of the patient's response to current antiviral therapy with pegylated interferon and ribavirin.

For his studies on viral hepatitis, Dr. Tyrrell has received numerous prestigious awards including the Gold Medal of the Canadian Liver Foundation (2000), the Alberta Order of Excellence (2000), Officer of the Order of Canada (2002), Fellow of the Royal Society (2004), FNG Starr Award of the Canadian Medical Association (2004), and the Principal Award of the Manning Foundation (2005).

Genomic Applications to Infectious Diseases

Infectious diseases remain the major cause of morbidity and mortality worldwide despite tremendous progress which was made using vaccines, antibiotics, antivirals and public health measures. Over the last 30 years there have been over 30 new organisms identified that cause new diseases. These include some challenging diseases we currently face such as HIV and AIDS, hepatitis C virus as a major cause of cirrhosis and liver cancer, SARS and avian influenza. Genomic approaches have been critical in the rapid identification of new pathogens (eg. SARS), vitally important to our knowledge of disease pathogenesis (eg. avian influenza), understanding the unique mechanism that the infecting organs use to evade the host immune response (HCV) and identifying new targets for therapeutic approaches. We have just begun to explore the variable presentation of diseases caused by a common organism which should make the study of the variable host responses a rich area for future genomic studies in infectious diseases.

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NorCOMM: High Throughput Mammalian Functional Analysis for the Discovery of Novel Determinants of Human Disease

NorCOMM (North American Conditional Mouse Mutagenesis project) is funded by a major grant from Genome Canada. NorCOMM is a large-scale research initiative to develop and distribute a resource of mouse embryonic stem (ES) cell lines carrying single conditional knockout mutations across the mouse genome. We are creating a publicly accessible library of ES cells suitable for drug discovery, target discovery and validation, and investigating mouse models of human diseases. We are working closely with international mouse knock-out partner projects funded by the European Commission (EuCOMM) and the NIH (KOMP: Knockout Mouse Project). Together, the projects will accomplish complete coverage of the mouse genome using a combination of high throughput random gene trap mutagenesis and systematic high-throughput targeting of remaining untrapped genes. Gene trapping in ES cells is an insertional mutagenesis approach to functional genomics. The gene trap vector is designed to generate sequence, expression and functional information for each clone. Since the project began on April 1, 2006 NorCOMM's three major gene trap centres have contributed more than 50,000 mutant ES cell lines to the Canadian Mouse Mutant Repository in Toronto. This large and growing archive of ES cells is publicly available on a cost-recovery basis and access to clones is unrestricted and nonexclusive. Complementing the trapping approach, over the next two years NorCOMM will create at least 500 targeted cell lines by specifically targeting genes not mutated in the gene trap resource. To ensure maximum coverage of the mouse genome and to minimize redundancy of effort, NorCOMM is working closely with

EuCOMM and KOMP to develop the Canadian list of genes for targeting. NorCOMM is inviting input from the Canadian scientific community on which genes would have the greatest impact if a knockout mouse ES cell line were available. We have established an online Gene Submission Form on our website www.NorCOMM.org. Researchers can request genes of interest and the information provided becomes part of the NorCOMM gene targeting prioritization process. Submissions through this form are confidential and secure.

Poster Abstracts Résumés d'affiches

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Public Health Genomics (PHG) and Public Participation

The intermingling of genomics with population health raises ethical and policy challenges that have the potential to affect the lives of many citizens. Experts and health practitioners are now recognizing the importance of involving the public in the design of public health policies. We investigated four PHG related projects, CARTaGENE, the British Columbia Biobank project, Generation Scotland and the UK Biobank projects that used public participation to foster dialogue and found that there are several challenges to effective public participation. Each project was examined for i) the goals of public participation; ii) definition of the public participation process; iii) the targeted public; iv) public representatives; v) mechanisms of public participation; vi) timing of public participation; vii) level of public influence and involvement; viii) cultural and practical barriers; and ix) post-evaluation of participation results. The main obstacle that emerged from this research is that of terminology and definition of public participation, which remains unclear. Other emerging issues include defining the public considering there is a range of publics and stakeholders that could be influential; establishing who the representatives are; selecting appropriate mechanisms of public participation from the variety of different techniques that are sometimes not well understood; timing and the lack of evidence regarding the impact of the interventions on the development of health policies. Inspired by these limitations rooted in the conceptual as well as operational stages of public participation, we developed a list of criteria for effective public participation in PHG. This list involves the evaluation of cost-effectiveness in

public participation, consideration for the levels of involvement, independence, influence, public representativeness, resource accessibility, decision-making structure, orientation of goals and public transparency.

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La participation du public dans les plans nationaux de lutte à une pandémie d'influenza

Contexte : Face à la menace d'une pandémie d'influenza, plusieurs pays ont décidé de mettre en place des mesures rassemblées dans des plans nationaux. Vu l'importance des pouvoirs et des responsabilités assumés par les États en cas de pandémie, la lecture des différents plans de lutte à une pandémie d'influenza fait apparaître des enjeux éthiques de la plus grande importance. Pourtant, malgré la récente émergence d'une littérature qui s'intéresse aux aspects éthiques des pandémies, encore trop peu de travaux se sont penchés sur la participation du public en tant que facteur de valorisation éthique des plans. Ce travail entend donc présenter l'analyse des différents plans nationaux et définir la place réservée à la participation du public à l'intérieur de ces plans.

Méthode : Au total, 22 plans ont été examinés au cours de l'année 2006.

Seuls les plans émanant de la plus haute autorité compétente dans chacun des pays ont été retenus. La grille d'analyse utilisée s'est constituée principalement autour de trois thèmes: l'utilisation de la génomique, les questions éthiques et la participation du public.

Résultats : Aucun plan ne fait la promotion de l'utilisation de la génomique humaine pour lutter contre les pandémies. Aussi, la majorité des plans examinés ne contiennent à peu près aucune considération sur les enjeux éthiques. Trois plans font exception à cette règle (Canada, Suisse et Nouvelle-Zélande). Ces plans proposent des cadres éthiques dont l'application relève uniquement des experts. Finalement, l'immense majorité des plans prévoient des mécanismes de communication unidirectionnels dans lesquels le citoyen est perçu avant tout comme un exécutant, ou encore comme un simple vecteur de la maladie.

Conclusion : La participation du public peut contribuer à la valorisation éthique des plans en liant leur application à des impératifs de bonne gouvernance, de décentralisation des pouvoirs décisionnels et d'émancipation des communautés.

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Developing Genome Resources for *Clostridium difficile*

The emergence of a virulent strain of *Clostridium difficile* (PFGE NAP1/ribotype 027/toxinotype III) has led to recent outbreaks across North America and Europe associated with increased morbidity and mortality. We are using complete genome sequencing and bioinformatics approaches to study clinical isolates representing a range of strain types, geographic locations, and year of isolation. Our aims are (i) to provide detailed knowledge of genomic diversity within *C. difficile*; (ii) to identify polymorphisms valuable for the development of DNA-based diagnostic and typing strategies; and (iii) to identify candidate mutations and genes associated with disease pathogenesis or showing potential value in the development of therapeutic and immunization strategies. Using a Roche/454 GS-FLX massively parallel DNA sequencing system, whole-genome shotgun sequencing and assembly has been performed for 5 of 9 *C. difficile* samples originally isolated between 1985 and 2007. Based on preliminary assemblies and gene annotations, *C. difficile* genome size appears larger (4.29 Mb to 4.42 Mb) in non-NAP1 and pre-epidemic NAP1 than in NAP1 isolates from the 2004-2007 epidemic (4.05 Mb to 4.11 Mb). The changes in genome size are largely due to differential levels of mobile DNA element activities in the isolates. Otherwise, genome-level alignments display a high level of collinearity, suggesting that the *C. difficile* genome has not undergone major rearrangements. Our preliminary analyses indicate that 83% of the *C. difficile* genome is occupied by ~3800 protein-coding genes, and the entire gene set is almost entirely shared between NAP1 and non-NAP1 isolates.

While continuing to perform whole genome sequencing on the remaining isolates, we are developing and implementing an analysis pipeline to characterize gene presence/absence and allelic diversity across all sequenced isolates. Our computational prediction of lost or gained genes and gene variants will be correlated with isolate characteristics (strain type, year of isolation) and predicted gene function information to identify and prioritize genomic regions and candidate genes with increased potential for clinical applications including diagnostics, molecular epidemiology, immunization strategies and therapeutic treatments.

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Human Genomic Databases as Global Public Goods

The variation between the genomes of individual human beings is small. There are also variations between different populations around the world; all these variations give insight into human biology. However, because they are small, in order to study these variations, researchers need a large amount of data. Population-based human genetic databases provide a powerful resource for these human research studies, but one database alone is not sufficient to fully understand the biology of individual human beings or to compare the variation between populations. We need to link the information from many of these databases together.

As population-based human genetic databases are located in different parts of the world, linking their data necessitates that data cross borders, thus introducing the complexity of international law and international coordination. In addition, the data takes on a 'personal' quality when primary DNA sequences are linked to "personal data" from different records (i.e. genealogical, medical, socio-demographical data). This extends the need for privacy protections over and above existing personal data protection laws.

We propose to study the interests that come into play from the perspective of both the individual State where the database are funded and housed and that of the health needs of populations, using the notion of Global Public Goods as a strategic means of sharing genomic research resources among humanity.

Poster Abstracts Résumés d'affiches

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Comparative genomics of promoter sequences in *Lentinula edodes* and *Coprinus cinereus* by *in silico* analysis of Serial Analysis of Gene Expression (SAGE) tags

Lentinula edodes, or Shiitake mushroom, is a widely cultivated edible mushroom. *Coprinus cinereus* is a multicellular basidiomycete with a completed 108,616,200 genome sequence assembly of the monokaryotic strain Okayama-7 in 2003 and ongoing genome annotation. Our long term goal is to understand the mechanisms of fruiting body initiation and development, which remain largely unknown. One approach to achieve this goal is to analyze the diversity (or conservation) of promoter sequences in these fungi. Serial Analysis of Gene Expression (SAGE) was used: we constructed longSAGE libraries from 4 developmental stages of *L. edodes*, and two 5'-SAGE libraries from dikaryotic mycelial and primordial stages to comparatively analyze the transcriptomes of *C. cinereus* and *L. edodes*. The 5'-SAGE was based on template switching during cDNA synthesis, which introduced restriction site to the 5' end of mRNA transcripts. The first 17-bp of each transcript was extracted by a type II restriction enzyme, MmeI. Two 17-bp tags were ligated to form ditags, which were sequenced directly by the sequencing-by-synthesis method (Roche Genome Sequencer). Individual SAGE tags obtained from the ditags represented transcription start sites (TSS), and 1500 bp upstream of the TSS were extracted as the input sequences for transcription factor binding site (TFBS) analysis by MEME (<http://meme.sdsc.edu/meme/intro.html>). Results generated by MEME were further analyzed by comparing putative TFBSs against the JASPAR database (http://mordor.cgb.ki.se/cgi-bin/jaspar2005/jaspar_db.pl). Individual motifs identified were subjected to P-Match algorithm ([\[bin/pub/programs/pmatch/bin/p-match.cgi\]\(http://www.gene-regulation.com/cgi-bin/pub/programs/pmatch/bin/p-match.cgi\)\) to search for similar known fungal motifs in the TRANSFAC database. Comparison of TFBS motifs found in *L. edodes* and *C. cinereus* were discussed. We compared promoter sequences putatively important for fruiting body initiation and development in *L. edodes* and *C. cinereus*, indicating the potential of using *C. cinereus* as a model platform for studying fruiting processes of edible mushrooms.](http://www.gene-regulation.com/cgi-</p></div><div data-bbox=)

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Genetic susceptibility and potentiality: Cases studies of direct-to-consumer advertising for genetic testing

The burgeoning genetic knowledge has expanded the knowledge of the possible role of genetics in what have been traditionally considered behavioral disorders, including attention deficit hyperactivity disorder (ADHD), depression, and substance abuse. New genes have also been associated, but not linked with potential to succeed at various lifestyle choices, including athletic sports. The push to commercialize early genetic associations has manifested in the direct-to-consumer advertising of tests that claim to determine individual susceptibility to alcoholism. A marked example of a well-marketed 'genetic potentiality test' is the Human Sports Performance test for the ACTN3 gene, purported to measure a candidate's suitability for endurance sports. This study looks at detailed case studies of companies engaging in direct-to-consumer advertising of genetic tests for susceptibility to alcoholism and Human Sports Performance. These companies constitute a subset of a larger sample from an earlier study examining direct-to-consumer advertising for genetic testing. Promotion and risk communication strategies employed by the companies promoting alcohol-related genetic testing are coded to develop themes. Genetic counseling arrangements, and information provision, representation of benefits and risks and use of credibility markers are examined. Tests can be ordered directly by consumers without the requirement for mediation by an independent physician or genetic counselor. Companies websites provide anti-reductionist disclaimers that

may be at odds with messages conveyed in advertising text. Provision of consumer testimonials, emphasis on employee credentials and use of logos of accrediting bodies are used on company websites as makers of credibility. The results are discussed in the context of public health and personal health and lifestyle management.

Poster Abstracts Résumés d'affiches

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Towards More Effective GE³LS Integration

In its most recent funding competition in 2005, Genome Canada included a requirement that "funded researchers must consider the GE³LS (i.e. ethical, economic, environmental, legal, and social issues related to genomics and proteomics research) aspects of their research and, where appropriate, seek advice from GE³LS experts to develop a plan to address those GE³LS issues directly raised by the research." Integrating genomic science and GE³LS research within a single project represents an innovation in the structure of research funding, one that endeavours to produce more socially viable technologies by addressing social interests directly within the research and development process.

The objective of this poster is to examine early "on-the-ground" experiences with the Genome Canada model of integration. Genome Canada held a workshop in July 2007 that brought together genomic science researchers, GE³LS researchers, and others with experience in GE³LS integration (including representatives from Genome Canada and the regional Genome Centres). The workshop addressed approaches, accomplishments, opportunities and challenges of GE³LS integration by providing: a national forum for information sharing and discussion of lessons learned through experience with Genome Canada-funded integrated research; a 'snapshot' of GE³LS integration; i.e., GE³LS integration in its present state of practise; and, an opportunity for GE³LS and genomics and proteomics researchers and others to jointly explore and to better understand the factors that facilitate, enable and leverage successful integration. This poster identifies and discusses major themes that emerged from the workshop, including the following:

- Building relationships is essential to integration of GE³LS research;
- Successful integration of GE³LS research takes time;
- The purpose and function of integrated GE³LS research needs to be clearly defined;
- "Embedded" GE³LS research is not the same as "integrated" GE³LS research;
- Scientific and GE³LS research may follow different timelines; and,
- Effective knowledge translation from GE³LS research needs attention.

The discussion of these themes is aimed at providing a useful starting point for a more sophisticated understanding of the integrated model in practice and for developing future approaches to funding integrated research.

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Development of Congenic Mouse Lines for use in the Analysis of Genetic Complexity in Caffeine Metabolism

Caffeine is predominantly metabolized in both mice and humans, through 3-demethylation via the cytochrome P450 isoform CYP1A2, an important enzyme in the phase I metabolism of many drugs and other xenobiotics. The regulation of basal expression of this gene is not well understood. The APN mouse strain was derived from outbred Swiss Webster mice through selective inbreeding for low caffeine 3-demethylation. A genome-wide scan was conducted for quantitative trait loci contributing to this phenotype in the F2 generation of an intercross between APN and C3H/HeJ, a strain with comparatively high caffeine 3-demethylation activity. Three QTLs were identified, on chromosomes 1, 4 and 9. Here, we describe the derivation of congenic strains containing C3H/HeJ intervals corresponding to each of the QTLs, on an APN background. A combination of phenotypic and genetic marker-assisted selection was used. Caffeine metabolism was measured by HPLC analysis of serum from mice dosed with caffeine p.o. Genomic DNA was screened with a panel of short tandem repeat markers spanning the genome. Mice were selected for preservation of the C3H/HeJ chromosome segment in the QTL interval as well as maximum decrease in residual donor strain genome. Breeders were selected based on the narrowing of the interval area with each successive mating. From this process we have been able to produce congenic mice for each of these QTL intervals, identified as C1 : APN.C3H-(D1Mit495-D1Mit292), C4 : APN.C3H-(D4Mit196-D4Mit232), and C9 : APN.C3H-(D9Mit347-D9Mit2). Each of these congenic lines retains a caffeine metabolism phenotype, while only the C4 and C9 lines show altered Cyp1a2 gene expression, as determined by RT-PCR. The C1 line

does show altered expression of other phase I and phase II drug metabolizing enzymes, as determined by DNA microarray analysis. Candidate loci for 2 of 3 QTLs involved in caffeine 3-demethylation have been identified, and the presence of a modifier locus affecting Cyp1a2 expression has been confirmed. Hence, the congenic lines described herein are proving to be a valuable model for the study of drug metabolism involving CYP1A2.

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Surveying the full landscape of human alternative splicing using custom-designed microarrays and data analysis tools

We have recently described the development and application of a microarray platform for the global quantitative analysis of alternative splicing (AS) in mammalian cells and tissues. As part of a Genome Canada funded project, we are expanding this system to provide a comprehensive-as-possible survey of AS in normal human cells and tissues, and also in specific disease contexts. As an initial step, we have designed microarrays containing exon body and exon junction probes to detect virtually all possible AS patterns for a set of 3,620 human genes, which represent ~5000 AS events that are conserved between human and mouse. Data has been collected from hybridizing cDNA from over 50 normal human tissues. An efficient preprocessing pipeline has been implemented, and a previously developed algorithm (GenASAP) has been used to confirm the reliability of the data. In parallel, a novel machine learning algorithm has been developed to measure in a robust manner the relative abundance of individual exons across transcripts using the microarray data. These approaches are allowing the discovery of new splice variants and splicing regulatory patterns in humans. We will next scale these procedures to survey splicing patterns for all protein-coding human genes. In addition, we will profile splicing patterns in the context of human aging and different disease states, including certain cancers. This work should significantly advance our understanding of the role of AS in human biology.

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Access to Data and Materials in the Life Sciences - A Study of the *Caenorhabditis elegans* Knockout Project as a Comparative Model for Knockout Mouse Repositories and Databases

Shortly after the release of the *C.elegans* genome sequence in 1998, the *C.elegans* Gene Knockout Consortium was formed with the goal of knocking out every gene in the *C.elegans* genome. We are investigating the effects of policies regarding access to data and materials generated by this project as a comparative model for knockout mouse databases and repositories.

The mission of the *C.elegans* Knockout project is to facilitate genetic research of this model system. To do this, several unique policies have been adopted by the project. 1) Ease of access to mutants: all mutants are stored in one central repository: the *Caenorhabditis* Genetics Centre at the University of Minnesota and the available mutants are listed on the WormBase database. 2) No withholding of mutants: Researchers are able to request the knockout of a specific gene but once a mutant is created it is available to all researchers without delay. 3) Low Cost: All mutants are distributed for a flat fee of \$7/strain to academic users. 4) No Restrictions on Use: Investigators are only requested to acknowledge the source of mutants in any publications; no material transfer agreements are used. The North American Conditional Mouse Mutagenesis (NorCOMM) project, in collaboration with the American (KOMP) and European (EUCOMM) knockout projects, aims to provide universal access to the mutant mice and ES cells to researchers for a minimal cost. Our study of the *C.elegans* project through interviews with key players in the *C.elegans* and the broader research community will give us insight into the impact of the

policies adopted by this project. A comparison with knockout mouse consortia policies will provide a better understanding of the impacts of these databases/repositories and their policies on research and innovation.

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Gene Expression of DNA in *Salmonella* Using Different Clustering Techniques

The Genome of more than 300 species of bacteria has been sequenced. The genome of bacteria is fluid and can be inserted mediated by Lateral Gene transfer Process (LGP), genetic recombination mechanism, which plays a major role in bacterial evolution (Boucher, et al 2003).

The theme is to study a small group of pathogen, *salmonella* sp. causing epidemic illness like diarrhea and intestinal disease mostly in children. Severity of salmonella infection is likely to increase due to HIV epidemics in under developing countries.

Salmonella sp. is food borne pathogens causing gastro intestinal infection and illness. Our bioinformatics tools and techniques are novel, safe and rapid research solution. This poster reveals the intelligent dataset and filtration of genes causing the disease, indicates the way to check the growth of this bacteria.

METHOD:

Hybridization of the array with two labeled different fluors for parallel analysis. Comparing the gene expression patterns by measuring the mRNA level in normal and pathological cells. The cDNA are generated with manually depositing small part of genes of interest from the known location on the glass surface. The out put of the depositions are groups of cDNA amplified by Polymerase Chain Reaction (PCR). CDNA probes hybridized to the array and loose probe washed off. The probes are tagged with fluoroscense reporter molecules with detectable light stimulated by laser. Each spot of array scanned by microscope with computerized scanner. Then various softwares are used to change them into numerical data.

RESULT :Analysis of microarray data through clustering techniques (Fuzzy c-means) we found most useful. There was tremendous decrease in values from 5134 genes to 154 genes remaining. Hence 3% genes are as the genes of interest.

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Canada's Health Biotechnology Collaboration with Developing Countries: A Preliminary Analysis of Entrepreneurial Linkages

Health biotechnology is a potent tool that can contribute towards improving nations' ability to treat debilitating diseases and increase their economic competitiveness. But research shows that biotechnology knowledge agglomerates in industrialized nations. Private firms are crucial for the functioning of the health biotechnology sector. Innovative firms are at the core of nations' health biotechnology innovation systems; they integrate various types of knowledge to develop new health products and processes. Collaboration with Canadian health biotechnology firms can help developing countries to build capacity in this field.

In this poster we report the preliminary results of a survey conducted of 181 Canadian health biotechnology firms on their linkages with developing countries' partners. Such an exploratory survey exercise is a good tool to map and analyze current patterns and the extent of collaboration between the Canadian health biotechnology private sector and the developing world. We examine the proportion of Canadian health biotechnology firms involved in linkages with developing countries. We map the locations of the countries Canadian firms have partners in. We look at the types of activities and technologies involved in these collaboration initiatives. We examine the output of partnerships between the Canadian health biotechnology private sector and their developing countries' partners and finally, we look at the reasons Canadian firms cite for working with developing countries.

Preliminary analysis of the survey data shows that around a quarter of the

Canadian health biotechnology firms are involved in partnerships with developing countries. Their strongest linkages are with China and India, but the firms have linkages with various smaller developing countries as well. The linkages are also seen to have potential in resulting in joint publications, patents and products. Accessing markets in emerging economies was a major reason cited by Canadian health biotechnology firms for collaborating with developing countries. Bi-directional knowledge flows are also important reasons for collaboration. Such results point to need for further research into this phenomenon to inform policy options that encourage successful North-South collaboration in health biotechnology.

Poster Abstracts Résumés d'affiches

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The Atlantic Medical Genetics and Genomics Initiative (AMGGI)

The human phenome comprises the set of all phenotypes attributable to sequence variation in the human genome. Clinical geneticists define that component of the phenome resulting from rare and deleterious sequence variants with relatively high penetrance (i.e. mutations). Defined in this way, monogenic or "Mendelian" mutations represent a tiny subset of all sequence variation present in humans worldwide, but in the complete phenome they have the most interpretable genotype-phenotype correlation and functional consequences. As of July 15, 2007, OMIM had 2060 independent gene entries with described allelic variants, out of an estimated 22,000 independent protein-coding transcription units. Thus the great majority of genes remain to be understood in terms of genetically defined function.

The Atlantic Medical Genetics and Genomics Initiative (AMGGI) is a four-year, \$9.1 million Genome Canada project to ascertain, collect and molecularly characterize 25-30 new monogenic disorders in Atlantic Canada. AMGGI is unique, collecting families across multiple medical disciplines (ophthalmology, neurology, pediatrics, nephrology, oncology, etc), and utilizing centralized technology platforms to bring an industrial scale approach to clinical genetics. AMGGI is also breaking new ground in bridging the traditional gap between research and clinical diagnostics, and includes a research component to study the impact of genetic information on patients, health care workers, and the Canadian health care system.

AMGGI currently has 31 disorders of likely genetic etiology under molecular genetic investigation. To date, we have defined the causal genes for two

novel disorders, in the fields of ocular and cardiovascular medicine. One of these, a new gene for sudden cardiac death, is of both scientific and profound diagnostic significance for the communities at risk. The second, a gene for Schnyder crystalline corneal dystrophy, is of potential relevance to general lipid metabolism, with implications for cardiovascular disease and cancer. AMGGI has also verified mutations segregating in the Atlantic population in known genes for multiple developmental disorders. Genome-wide mapping in families segregating novel phenotypes has identified 5 chromosomal linkages for which the causal mutations are actively being sought. We will present a selection of results from these projects.

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Nutrigenomics and Behavioural Change: A Public Health Perspective

The science of nutrigenomics—the integration of functional genomics, nutrition and health—is viewed as one of the next waves in health interventions utilizing the knowledge of the human genome. Although in its infancy, nutrigenomics holds the promise of improving therapeutic outcomes and preventing disease by allowing individuals to personalize nutritional information to their genome thereby utilizing the benefits and minimizing the risks of specific diets or dietary components. Despite the potential of nutrigenomics, there are reasons to be skeptical about this emerging science. This presentation will assess the claims of nutrigenomics in light of a public health understanding of behavioural change. Commentators have noted that the science of nutrigenomics is not at a point where it can support large number of claims of health benefits to individuals. Despite optimism, genetic advances are not close to bringing us to the 'brink of a nutritional revolution.' Moreover, there is considerable ignorance regarding the clinical impact of nutrigenomics, as the technology is too new to have been subjected to any systematic or epidemiological studies. Although it is unknown what impact nutritional advice based on genotype will have, behavioural change is exceptionally difficult to modify. From a public health perspective, behavioural change is often impeded by environmental, ecological and socio-political factors. Claims of behavioural changes in response to information gained from nutrigenomics, therefore, may be spurious and should be viewed, at a minimum, with scepticism.

This presentation can be divided into three parts. The first part will summarize the claims of the public health benefits of nutrigenomics made

in academic literature, industry, research groups and the popular media. The second part will briefly review some relevant theories of behavioural change. The third part will identify some of the difficulties associated with the use of genetic information to modifying behaviour. This presentation will conclude that while nutrigenomics is often portrayed as resulting in behavioural change and harnessing the potential to overhaul nutritional science, it remains an open question whether nutrigenomics will have a significant impact on behaviour at a population level.

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Genomic Applications in screening assessment of *Bacillus cereus* group biotechnology micro-organisms

The Canadian Environmental Protection Act (CEPA 1999) includes a Domestic Substance list of micro-organisms (MOs) that were present in Canadian commerce between 1984 and 1986. Many of these MOs have not undergone screening assessments for ability to elicit pathogenicity and/or toxicity to humans and mammalian surrogates. We are developing in vitro genomic-based methods and data to establish a baseline for toxicity/pathogenicity testing of *Bacillus* biotech-organisms regulated under CEPA and the Pest Control Products Act. In Canada, subspecies of *Bacillus thuringiensis* (Bt) have been permitted for commercial use as biopesticides (e.g., israelensis (Bti) and kurstaki (Btk)) and other applications (e.g. Bt 13367 and Bc 14579). Bt is generally considered a close genetic member of a group containing *B. cereus* (Bc) and *B. anthracis* (Ba). This presentation provides an overview of genomic applications carried out by our group aimed towards screening assessment of Bc group biotechnology MOs.

Genome content of various Bc group MOs were assessed by hybridization of genomic DNAs to oligonucleotide arrays designed from the Bc 14579 genome. Hybridizations were scored for different classes of virulence factors such as hemolysins, lipases and toxins and validation of observations has been made by isolating and cloning hemolysins and sequencing of discrete regions of the genomes. Effects of microbes on eukaryotic macrophage transcription were assessed using two macrophage-like cell lines: J774A.1 (ATCC TIB-67) and monocyte/macrophage-like cell line RAW264.7 (ATCC TIB-71). Transcripts of cells exposed to Bc spores were profiled by microarray and PCR.

Compared to controls, RAW264.7 cells exposed to Bc spores show significant differences in transcript abundance of pro-inflammatory cytokines and immune response genes in early time points. In contrast, J774A.1 showed very few genes with > 2-fold difference in transcript levels in early time points. This data set shows that the two immune-cell lines differ in response to Bc spores at the transcript level and appear to recognize and phagocytize spores and modulate inflammation related signalling differently.

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Comparative transcriptomics using full-genome tiling arrays

Recent studies have indicated that the genome is far more widely transcribed than previously assumed. Whether this "hidden transcriptome" is functional remains a topic of active debate as many of the expressed sequences evolve at a neutral rate, strongly suggesting that they are neither protein-coding nor structural RNAs. Even if the primary sequence is not conserved, however, these RNAs may still serve a functional role, for example as lineage-specific regulatory elements or markers of chromatin state. Determining the extent to which the new transcripts are functional is important because if they are functional then they are also candidates to search for the ~50% of mapped disorders for which no causative mutation can be identified. To assess the role of non-coding transcripts in the genome, we are profiling expression in four different human and mouse tissues (brain, heart, liver and testis), using high-density Affymetrix arrays that cover the complete non-repetitive genome at a 35 bp resolution. Both total- and polyA+ RNA fractions were assayed for each tissue, to enable a more readily distinction of coding and non-coding transcripts. Our experimental approach allows for assessment of non-coding RNA function by looking at the degree of evolutionary conservation of expression, as well as tissue-specific regulation. Differential regulation across tissues provides evidence of biological function even in the absence of evolutionary constraint. Moreover, while the vast majority of coding sequences have already been identified, we still expect to identify novel exons for known protein-coding transcripts using gene-finding algorithms such as GenRate. Here we will present preliminary results of our studies of a genomic region that is syntenic between human and mouse.

Poster Abstracts Résumés d'affiches

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Policymaking, harmonization and population biobanks

Policymaking in an international context is often delegated to recognized organizations (e.g. WHO, UNESCO, OECD). Similarly, international societies of concerned individuals and experts contribute to the elaboration of guidance, while being respectful of cultural and national differences. In the field of population genomics, are the GE3LS issues simply multiplied by the sheer number of participants? At the Centre de recherche en droit public (CRDP), Université de Montréal, a Policymaking Core has been established to inform the international community of the possibility and importance of prospective approaches to traditional issues such as: consent, access, governance and commercialization. If these issues are not discussed and some level of harmonization achieved, the population health goal of these ambitious large projects (biobanks) cannot be achieved, due to the absence of policy interoperability.

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An integrated genetic/physical map for the old world monkey, *Cercopithecus aethiops*

The vervet monkey (African green monkey; *Cercopithecus aethiops*, *Chlorocebus aethiops sabaeus*) is an important model for studying human diseases and complex traits. The vervet monkey's higher chromosome number ($2n=60$ for vervet vs. $2n=46$ for human) also makes it a useful model for the characterization and study of evolutionary recent (<5 MYA) genome rearrangements. We have embarked on a Genome Canada/Génome Québec Competition III project to sequence the ends of >200,000 vervet monkey bacterial artificial chromosomes and compute a genomic framework map of the vervet genome. Using alignment information to infer orthology to the human, chimpanzee and rhesus genome sequences, we have integrated gene and genetic marker information onto the framework map, and use it to identify regions of genome colinearity as well as regions of rearrangements. This resource will aid in genetic mapping and fine-mapping studies, as well as clarifying the repositioning of vervet chromosome ends, centromeres, and other types of genomic reorganizations.

As of September 2007 we have deposited >213,500 BAC end sequences into NCBI/Genbank, representing 52.7% of our goal and ~5-fold coverage of the genome. 76% of all end-sequenced BACs display genome colinearity. In aggregate this clone set spans 84% of the human genome, with the remaining 503.2 Mb distributed across 1622 gaps, including the 229.6 Mb corresponding to the 24 human centromere gaps. Using a combination of gap information (position, size, and sequence content) and end sequence information indicating non-orthology (no counterpart in the human genome) or non-colinearity (change in genome structure)

we are currently characterizing genome rearrangements including translocations, inversions, duplications, and centromere repositioning. Current progress in BAC end sequencing, weekly updated quality assessments, and comparative mapping tools and results are all available on our website at http://www.genomequebec.mcgill.ca/compgen/submit_db/vervet_project.

Poster Abstracts

Résumés d'affiches

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High resolution DNA-microarray-based genotyping of copy number variants in the human genome

Recent studies have demonstrated that human genetic variation includes widespread differences in total genomic content in the form of copy number variations (CNVs). Although our current understanding of the CNV map is still incomplete, rapid progress is being made to more comprehensively discover and characterize CNVs in the human population. In order to accurately determine how CNVs may contribute to the etiology and pathogenesis of complex diseases or for CNVs to act as useful markers for such diseases, a detailed analysis of their structures and genotypic frequencies in the general population is required. To achieve these goals, we have employed high-resolution custom oligonucleotide DNA microarrays to characterize copy number variant regions (CNVRs) in samples from the HapMap collection.

We have developed a database of ~16 million oligonucleotide probes that cover the non-repeat masked regions of the genome at an average spacing of 100 bp, selected according to stringent thermodynamic and sequence characteristics. In addition, we generated statistically robust approaches for calling CNV intervals in individual samples, methods for grouping variants from multiple samples into CNVRs, as well as processes to genotype the copy number state of individual samples in designated CNVRs. When we analyzed 27 CNVRs that were identified by fosmid-end sequencing and genotyped for copy number states in eight individuals from the HapMap collection, we found that concordance of our genotyping calls were 98% across methodologies and platforms. Using an

automated algorithm to call the variant intervals, we found a 99% concordance rate with intervals defined by fosmid-end sequencing in regions without homozygous deletions in both the reference and the sample DNA. In another study, we profiled thirty individual DNA samples from the HapMap collection across previously identified CNVRs and found that many regions were smaller and more variable than previously reported. Such refinement of CNVRs will help enable the development of accurate microarray-based CNV genotyping assays for the determination of genotypic frequencies at CNVRs. Combined, these results demonstrate that highly reproducible, sensitive, and specific microarray measurements can determine how CNVs vary across populations, paving the way to increasing the utility of CNVs in disease-related research.

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