



HUGO Symposium on

# Genomics and Ethics, Law and Society

Sequencing of Individual Genomics: Impact on Society and Ethics

1 – 3 November 2009, Geneva, Switzerland

**PROGRAMME AND ABSTRACT BOOK**



UNIVERSITÉ  
DE GENÈVE





HUMAN GENOME ORGANISATION

Symposium on Genomics & Ethics, Law & Society  
Sequencing of Individual Genomes: Impact of Society & Ethics

1 – 3 November 2009  
Geneva, Switzerland



## CONTENTS

Human Genome Organisation .....	1 - 4
University of Geneva .....	5
World Health Organization (WHO) .....	6 - 8
Welcome to Geneva .....	9
General Information .....	9 - 13
President's Message .....	14
Conference Chairman's Message .....	15
Programme (Daily Schedule) .....	16 - 21
Acknowledgements .....	22
International Scientific Committee .....	23 - 33
Panel of International Speakers .....	34 - 47
Abstracts – International Speakers .....	48 - 70
Abstracts – Posters .....	71 - 92
Abstracts Index .....	93 - 94

## ORGANISING COMMITTEE

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## **HUMAN GENOME ORGANISATION**

Human Genome Organisation (HUGO) is the international organisation of scientists involved in human genetics. HUGO was conceived in late April 1988, at the first meeting on genome mapping and sequencing at Cold Spring Harbor. The idea of HUGO was particularly Sydney Brenner's. He also suggested the name of the organisation and its acronym. At a rump session called to discuss the proposal at Cold Spring Harbor on April 30, 1988, Victor McKusick (Baltimore) was asked to serve as founding president. A 'Founding Council', was assembled from among those at the Cold Spring Harbor meeting, supplemented by others, to a total of 42 scientists from 17 countries. The initial membership included many of the individuals who went on to lead the human genome project through to its completion.

HUGO has, over the years, played an essential role behind the scenes of the human genome project. With its mission to promote international collaborative effort to study the human genome and the myriad issues raised by knowledge of the genome, HUGO has had noteworthy successes in some of the less glamorous, but nonetheless vital, aspects of the human genome project.

A truly international organisation, HUGO has been instrumental in reaching out to groups and nations that have not actively participated in the human genome project. Following a call to the members, HUGO has succeeded in recruiting scientists from 24 countries to act as ambassadors for their genome communities. These individuals conduct work in conveying the knowledge and implications of the human genome project to teachers, social workers and educational ministries - the 'first line' of society who must be informed and ready for the changes that knowledge of the genome will bring.

Maximising the benefits of the human genome project for all humanity, and not just the wealthy nations, requires international collaboration, resource sharing and continued dialogue. HUGO is constantly looking for new means of enabling international co-operation and building on the unique infrastructure that it already has in place through its truly global membership.

That collaboration is actively, and prominently, fostered by HUGO at the annual, highly successful, HGM meetings. HGM meetings have been held in locations as diverse as Shanghai and Cancun, Kyoto and Helsinki. They are the only occasions when the international human genome community draws together and provides platforms for interaction between scientists and bioethicists, between the people at the bench and the greater community. Twelve years of annual meetings have brought together over 10,000 attendees from 48 different countries – and public

discussion sessions at each meeting have disseminated information to the lay public about this rapidly increasing knowledge.

Under the Presidency of Edison Liu, who has taken office in May 2007, HUGO is entering a new and exciting phase of its development. Since inception, the administrative function of HUGO has been based in the Western hemisphere but is now moving, not only, with the times, but also with the directional emphasis of the science, to Singapore by early 2008. This reflects HUGO's awareness of the growth and increasing drive in all aspects of genomics that is being demonstrated throughout the region.

HUGO is entering the 20th year of its history by making an inflection in its direction. Now that the human genome has been sequenced, HUGO is seeking the biological meaning of its information content. To this end, it is focusing on the medical implications of genomic knowledge. Moving forward, HUGO is also working to enhance the genomic capabilities in the emerging countries of the world. The excitement and interest in genomic sciences in Asia, Middle East, South America, and Africa are palpable and the hope is that these technologies will help in national development and health.

### **Mission Statement**

- to investigate the nature, structure, function and interaction of the genes, genomic elements and genomes of humans and relevant pathogenic and model organisms;
- to characterise the nature, distribution and evolution of genetic variation in humans and other relevant organisms;
- to study the relationship between genetic variation and the environment in the origins and characteristics of human populations and the causes, diagnoses, treatments and prevention of disease;
- to foster the interaction, coordination, and dissemination of information and technology between investigators and the global society in genomics, proteomics, bioinformatics, systems biology, and the clinical sciences by promoting quality education, comprehensive communication, and accurate, comprehensive, and accessible knowledge resources for genes, genomes and disease;
- to sponsor factually-grounded dialogues on the social, legal, and ethical issues related to genetic and genomic information and championing the regionally-appropriate, ethical utilisation of this information for the good of the individual and the society.

## **HUGO Membership**

To become a HUGO member, please visit our website at **[www.hugo-international.org](http://www.hugo-international.org)** and register online. Annual HUGO membership subscription is fixed at £35 for students and post docs and £50 for non-students.

How do I join HUGO?

Simply register online and upload the following documents to complete your application.

- Full contact details
- 1 page CV
- List of no more than 5 recent publications
- Brief statement about your research interests

Please note: Members' contact details might be distributed electronically in the form of a membership list and a membership directory will be posted online in the member's only area in this website to other members, however this information will not be given to outside sources, other than for very specific and relevant purposes.

## **Membership Benefits**

- Reduced registration fees for HUGO events, conferences and training courses
- Annual voting to elect members to HUGO Council
- Access to announcements and information on this website
- Free subscription to The HUGO Journal
- Reduced subscription to The Annals of Human Genetics, Clinical Genetics, Science and AAAS
- Young scientists can apply for the HUGO Travel Award for educational visits
- Rights to nominate and vote at HUGO Council Member Election

## Upcoming Events

- HUGO Symposium on Genomics and Bioeconomy, end January 2010 Mexico City, Mexico
- HUGO 14<sup>th</sup> Human Genome Meeting 2010, 18th – 21st May 2010, Montpellier, France
- HUGO Symposium on Genomics and Hereditary Disorders, September/October 2010, Dubai, UAE
- HUGO-International Association for Breast Cancer Research 27<sup>th</sup> Meeting, early October 2010, Singapore
- HUGO 15<sup>th</sup> Human Genome Meeting 201, mid March 2011, Dubai, UAE
- HUGO 16<sup>th</sup> Human Genome Meeting 2012, Sydney, Australia
- HUGO 17<sup>th</sup> Human Genome Meeting 2013 cum International Congress of Genetics, Singapore

Visit [http://www.hugo-international.org/events\\_humangenomemtn.php](http://www.hugo-international.org/events_humangenomemtn.php) for more details and announcements!



## **UNIVERSITY OF GENEVA (UNIGE)**

The University of Geneva is an institution devoted to research, teaching and dialogue. It is the site of academic creativity and the transmission of knowledge.

Since its foundation in 1559 the UNIGE has developed ever higher ethical standards and steadily increased in quality and the will to innovation. Today it is one of Europe's leading universities.

The UNIGE also shares the international calling of its host city, Geneva, a centre of international and multicultural activities with a venerable cosmopolitan tradition. Its desire to expand its collaboration with partner institutions and broaden its appeal to researchers and students from around the world has made the UNIGE a "globalised university", a meeting place for academic disciplines and various cultures, and a forum for ideas.

We are pleased to have you visit this website, intended for a general international public, and look forward to welcoming you to Geneva.

### **About Faculty of Medicine**

#### **From DNA to bedside manner**

In its effort to continuously adapt its instruction to developments in biomedicine as well as to a changing society, the Faculty of Medicine is a leader in teaching. It was the Swiss pioneer of the new problem-based method, and the Faculty is today the first and only one of its kind in Switzerland to be officially accredited.

It collaborates closely with Geneva University Hospitals in the development of clinical medicine and houses the only School of Dental Medicine in French-speaking Switzerland, and the quality of its research has been broadly recognised. Diabetes, infectious diseases, genetics, neurosciences and transplants are some of the fields in which Faculty scholars enjoy international renown.

Website:

[http://www.unige.ch/international/etudageneve/acadstruct/medicine\\_en.html](http://www.unige.ch/international/etudageneve/acadstruct/medicine_en.html)

### **University of Geneva 450th Anniversary**

To celebrate the 450 years of University of Geneva, the UNIGE prepared programmes such as expositions, workshops, open doors, festive moments, lectures and open debate to all and to all during the period of February to December 2009.

Visit the official website of The University of Geneva 450th Anniversary <http://www.unige.ch/450/accueil.html#top> for more information.

## **WORLD HEALTH ORGANIZATION (WHO)**

WHO is the directing and coordinating authority for health within the United Nations system. It is responsible for providing leadership on global health matters, shaping the health research agenda, setting norms and standards, articulating evidence-based policy options, providing technical support to countries and monitoring and assessing health trends. In the 21st century, health is a shared responsibility, involving equitable access to essential care and collective defence against transnational threats.

### **The WHO agenda**

WHO operates in an increasingly complex and rapidly changing landscape. The boundaries of public health action have become blurred, extending into other sectors that influence health opportunities and outcomes. WHO responds to these challenges using a six-point agenda. The six points address two health objectives, two strategic needs, and two operational approaches. The overall performance of WHO will be measured by the impact of its work on women's health and health in Africa.

#### **1. Promoting development**

During the past decade, health has achieved unprecedented prominence as a key driver of socioeconomic progress, and more resources than ever are being invested in health. Yet poverty continues to contribute to poor health, and poor health anchors large populations in poverty. Health development is directed by the ethical principle of equity: Access to life-saving or health-promoting interventions should not be denied for unfair reasons, including those with economic or social roots. Commitment to this principle ensures that WHO activities aimed at health development give priority to health outcomes in poor, disadvantaged or vulnerable groups. Attainment of the health-related Millennium Development Goals, preventing and treating chronic diseases and addressing the neglected tropical diseases are the cornerstones of the health and development agenda.

#### **2. Fostering health security**

Shared vulnerability to health security threats demands collective action. One of the greatest threats to international health security arises from outbreaks of emerging and epidemic-prone diseases. Such outbreaks are occurring in increasing numbers, fuelled by such factors as rapid urbanisation, environmental mismanagement, the way food is produced and traded, and the way antibiotics are used and misused. The world's ability to defend itself collectively against outbreaks has been strengthened since June 2007, when the revised International Health Regulations came into force.

### **3. Strengthening health systems**

For health improvement to operate as a poverty-reduction strategy, health services must reach poor and underserved populations. Health systems in many parts of the world are unable to do so, making the strengthening of health systems a high priority for WHO. Areas being addressed include the provision of adequate numbers of appropriately trained staff, sufficient financing, suitable systems for collecting vital statistics, and access to appropriate technology including essential drugs.

### **4. Harnessing research, information and evidence**

Evidence provides the foundation for setting priorities, defining strategies, and measuring results. WHO generates authoritative health information, in consultation with leading experts, to set norms and standards, articulate evidence-based policy options and monitor the evolving global health situation.

### **5. Enhancing partnerships**

WHO carries out its work with the support and collaboration of many partners, including UN agencies and other international organisations, donors, civil society and the private sector. WHO uses the strategic power of evidence to encourage partners implementing programmes within countries to align their activities with best technical guidelines and practices, as well as with the priorities established by countries.

### **6. Improving performance**

WHO participates in ongoing reforms aimed at improving its efficiency and effectiveness, both at the international level and within countries. WHO aims to ensure that its strongest asset - its staff - works in an environment that is motivating and rewarding. WHO plans its budget and activities through results-based management, with clear expected results to measure performance at country, regional and international levels.

### **The role of WHO in public health**

WHO fulfils its objectives through its core functions:

- providing leadership on matters critical to health and engaging in partnerships where joint action is needed;
- shaping the research agenda and stimulating the generation, translation and dissemination of valuable knowledge;
- setting norms and standards and promoting and monitoring their implementation;
- articulating ethical and evidence-based policy options;
- providing technical support, catalysing change, and building sustainable institutional capacity; and
- monitoring the health situation and assessing health trends.

These core functions are set out in the 11th General Programme of Work, which provides the framework for organisation-wide programme of work, budget, resources and results. Entitled "Engaging for health", it covers the 10-year period from 2006 to 2015.



## **BIENVENUE À GENÈVE! (WELCOME TO GENEVA!)**

On behalf of the organisers, University of Geneva, WHO (co-sponsor) and HUGO, we would like to thank you for attending the second episode of the HUGO-WHO Symposia series on Genomic and Global Health – Genomics and Ethics, Law and Society (GELS 2009)!

We do hope you will take this opportunity to interact with our international panel of renowned speakers and contribute to the discussion forum in the afternoon of 2 November 2009.

### **GENERAL INFORMATION**

HUGO Information Desk is set up at the entrance to the plenary talk venue – Hall 1 (Level 1) at CIGG. Do drop by anytime for more information on HUGO, our activities as well as to get to know more about our new initiatives.

Speakers can preview and upload their presentation at the HUGO Information Desk. WIFI is free for all HUGO GELS 2009 participants, login detail as follows:

WIFI code : GELS2009  
Password : GELS2009

Registration is located at the entrance to the plenary talk venue – Hall 1 (Level 1) at CIGG where you can register and collect your delegate badge.

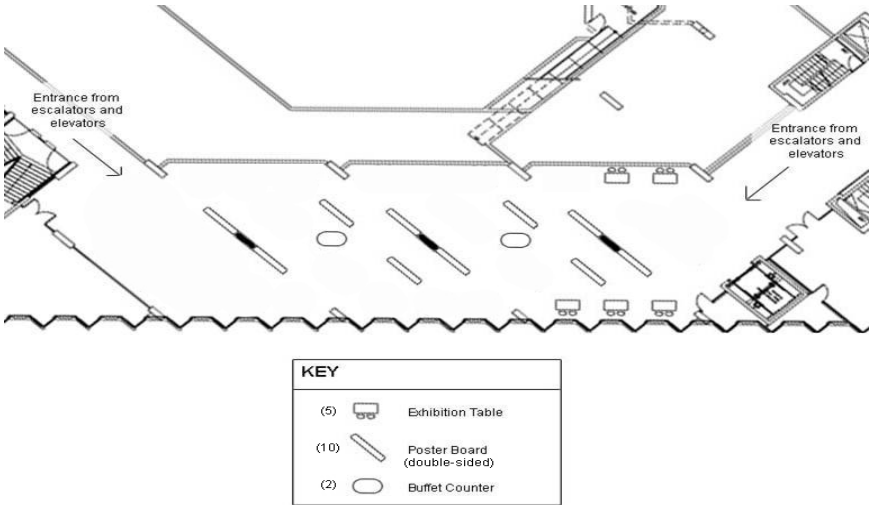
Registration is open on:

Sunday, 1 November 2009	8.00 am – 5.30 pm
Monday, 2 November 2009	8.00 am – 5.30 pm

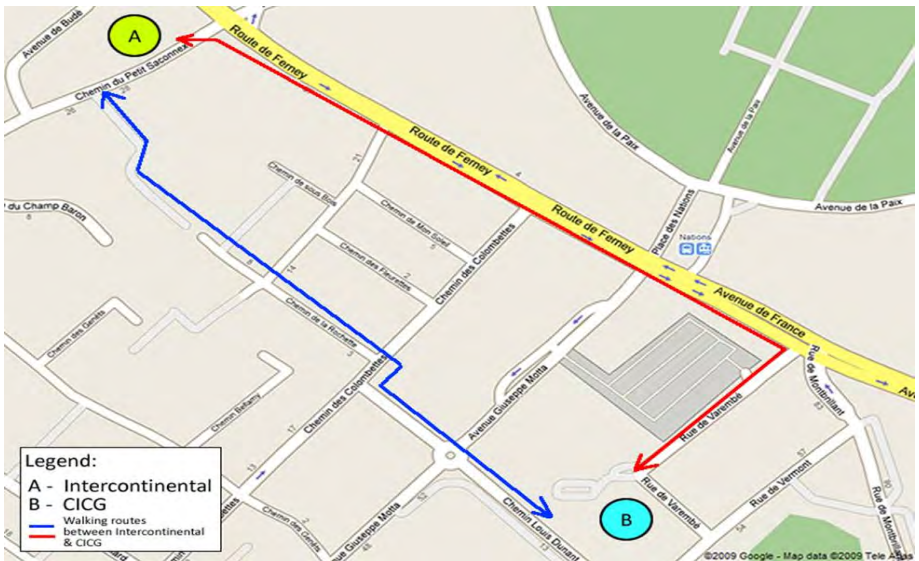
Badges are issued at registration and all are required to wear the badge at all times at CIGG. You may be refused entrance if you are unable to show your badge. An administration fee of CHF10 will be imposed on any replacements.

Tea breaks will be served at the exhibition/poster area (Espace Motta, Level 2 at CIGG) twice a day. Lunch will be served at CIGG Restaurant (Level 1) at CIGG (next to GELS 2009 plenary talk venue – Hall 1). You are required to present your lunch coupon to redeem your lunch at the CIGG Restaurant. Lunch is inclusive of one appetiser or dessert, one hot dish and mineral water or beer or soda or wine (tap water is available for free).

All poster sessions, located at the exhibition/poster area (Espace Motta, Level 2 at CIGG) are scheduled after lunch, do find some time to browse through and support your fellow delegates and to share your experiences with each other!



Conference Dinner is scheduled on 1 November 2009 (Sunday) at Intercontinental Geneva Hotel, a 15-minute walk from CICG. Do let us know if you need any assistance.





## DO YOU KNOW?

- Geneva is the second-most-populous city in Switzerland after Zurich. Situated where the Rhone River exits Lake Geneva (Lac Léman), it is the capital of the Republic and Canton of Geneva.
- Geneva is a worldwide centre for diplomacy and international cooperation, and is widely regarded as a global city, mainly because of the presence of numerous international organisations, including the headquarters of many of the agencies of the United Nations, the World Trade Organization (WTO), the World Economic Forum (WEF), the International Federation of Red Cross and Red Crescent Societies (IFRC), and the International Committee of the Red Cross (ICRC). It is also the place where the Geneva Conventions were signed, which chiefly concern the treatment of wartime non-combatants and prisoners of war. The city has been referred to as the world's most compact metropolis and the "Peace Capital".
- Museums and art galleries are everywhere in the city. Some are related to the many international organisations as the International Red Cross and Red Crescent Museum or the Microcosm in the CERN area. The Palace of Nations, home of the United Nations headquarters can also be visited.

- **Transport from Geneva International Airport**  
The airport is located at nearly 4 km from Geneva city centre. It is easily reachable by train or by bus using the united network of public transport Unireso. Only one single ticket is required to travel by train, tram, bus or boat in the area France-Vaud-Geneva. It takes only 6 minutes from/to Geneva city centre by train (every 12 minutes at rush hours). The airport railway station has direct access to the airport Check-in and Arrival levels. All trains stop at Geneva-Cornavin station (city centre).

The following urban buses stop every 8-15 minutes at rush hours at the airport (bus stops at the Check-in level, in front of or beside the train station) from/to:

Geneva city centre - Onex: bus 10;

**International organisations - Geneva city centre: bus 5 and 8; tram 13 and 15**

International organisations - lake: bus 28;

Meyrin: bus 28;

Le Lignon - Lancy - Plan-les-Ouates: bus 23.

- Tourist information and tickets can be obtained at the Unireso information counter in the Arrivals hall of the airport, on leaving customs control. Tickets can also be purchased from the machines located at bus stops (CHF or Euro change required) and at the Swiss railway station.
- **Travel free** on public transport during your stay in Geneva  
With effect from January 2008, you can pick up a free ticket for public transport from the machine in the baggage collection area at the Arrival level. This Unireso ticket, offered by Geneva International Airport, allows you to use public transport in Geneva free for a period of 80 minutes.

If you are staying at a hotel, a youth hostel or a campsite, the establishment will offer you the "Geneva Transport Card" that allows you to use public transport in

Geneva free of charge during your stay. Get more information from Genève Tourisme or the establishment concerned.

- **Public transport night service**  
You can use a collective taxi service (Taxibus) to get to the airport in the early morning, before public transport services start.  
On Friday and Saturday evenings, the N5 route serves the airport between 00:00 and 00:30 (Noctambus network).
- **Geneva is home to the University of Geneva, founded by John Calvin in 1559. The Graduate Institute of International and Development Studies is also in Geneva. The Graduate Institute was among the first academic institutions to teach international relations in the world. Also, the oldest international school in the world is located in Geneva, the International School of Geneva, founded in 1924 along with the League of Nations. Webster University, an accredited American university, also has a campus in Geneva. Moreover, the city is home to the Institut International de Lancy (founded in 1903). The Geneva School of Diplomacy and International Relations is a private university on the grounds of the Château de Penthes, an old manor with a park and view of Lake Geneva. The Canton of Geneva's public school system has écoles primaires (ages 4–12) and cycles d'orientation (ages 12–15). The obligation to attend school ends at age 16, but secondary education is provided by collèges (ages 15–19), the oldest of which is the Collège Calvin, which could be considered one of the oldest public schools in the world.**
- **Places of Interest**  
If you are passing through Geneva, do not miss visiting the following places:
  - Geneva's lakefront
  - Jet d'eau (water fountain)
  - Flower Clock
  - Saint Peter's Cathedral
  - Reformation Wall
  - Place Neuve
  - United Nations Building
  - Place du Bourg-de-Four
  - Carouge

For more information on Geneva's attractions, restaurant/dining guide, local customs, nightlife, touring information on the region, visit the Geneva Tourism and Convention Bureau website at <http://www.geneva-tourism.ch>

Sources: [www.wikipedia.org](http://www.wikipedia.org); [www.geneva-tourism.ch](http://www.geneva-tourism.ch)



## President's Message

The confluence of new and powerful genomic sequencing technologies, computational capabilities, and the ubiquity of the internet are converging to dramatically change our view of "self", of our heritage, and of disease. The scale of the speed and precision mirrors that of computer advances which, in the long run, gave us a synthetic advancement based on connectivity called the internet. Similarly, our ability to catalogue and ultimately to harness genetic complexity will provide solutions to human problems in ways we cannot currently even envisage. Our symposium on Genomics and Ethics, Law, and Society seeks to explore that inflection point we are experiencing in medical discovery and genetic awareness. The challenges are no longer technical but are social and legal. How should the consumer interpret the ramifications of her genetic signature now easily and directly available? How should such information be regulated and should it be regulated? How much risk is in an odds ratio of 1.2? How deterministic is genetics? Is there such a thing as a patent for a gene that is in all of us? We hope to explore these and other questions. As part of this conference, we invite all delegates to participate in an open discussion and to make your voices heard as we compose an analytical paper for HUGO on this topic of the new sequencing technology and society, ethics, and law.

A handwritten signature in black ink, appearing to read "Edison T. Liu". The signature is fluid and cursive, with a large, stylized "L" at the end.

Edison T. Liu  
President, Human Genome Organisation



## Conference Chairman's Message

Dear Participants and Guests

Welcome to Geneva, the international city of Science, International Relationships, International Health, and currently the city of the Genome!

The ability to read individual genomes and document the extensive genomic variability among individuals, coupled with the ability to score the equally extensive phenotypic variability of each person, provide a tremendous challenge: how to link genomic individuality with disease phenotypes and traits. This matchmaking effort is intimately related to a number of ethical, legal, and social issues. The Human Genome Organisation organises this timely conference on “Genomics, Ethics, Law, and Society” at the dawn of the personal genomics era; your participation in the debates would be extremely valuable and will certainly lead to a more mature and sophisticated confrontation with the plethora of these issues.

The University of Geneva celebrates his 450<sup>th</sup> anniversary this year, and the genome challenges are a crucial component of the celebrations, and a priority goal for the years to come. In the very centre of Geneva the University has currently organised a unique exhibition under the GeneDome entitled “Genome: au coeur du vivant” in which the whole haploid human reference genome will be read for a period of three months, and the visitors will enjoy a fascinating educational tour into the mysteries, history, and importance of genomes of people. I cannot imagine a better time and place of the HUGO conference!

Please also take a moment to enjoy Geneva and its friendly people. Rich in history, institutions, culture, and services, the city remains to be discovered, and could provide you fond memories.

Welcome again!

Stylianos E. Antonarakis, MD., DSc.  
Conference Chairman  
Professor and Chairman  
University of Geneva Medical School  
Department of Genetic Medicine and Development

# PROGRAMME (DAILY SCHEDULE)

## Day One (1 November 2009)

- 8.00 AM Registration
- 9.00 AM Opening Address by **Edison T. Liu**
- 9.05 AM Welcome Address by **Stylianos E. Antonarakis**
- 9.10 AM – 9.25 AM Welcome Lecture by **Ala Alwan**
- Session 1 – Science and its Capabilities**  
**Chair: Edison T. Liu**
- 9.25 AM – 10.10 AM **Olufunmilayo Olopade**  
*Advances In Breast Cancer: Pathways To Personalised Medicine*
- 10.10 AM – 10.30 AM *Tea Break*
- 10.30 AM – 11.15 AM **Edison T. Liu**  
*Genomic Sequencing: The Potential Impact Of The Technologies On Science And Society*
- 11.15 AM – 12.00 PM **Klaus Lindpaintner**  
*Future Of Health Care Industry – Strategies For Personalised Medicine*
- 12.00 PM – 12.45 PM **Mark McCarthy**  
*What will the new sequencing technologies deliver for science and society?*
- 12.45 PM – 2.15 PM **Lunch @ CIG Restaurant (Level 1)**  
**HUGO Council Meeting @ Room 15 (Level -1, 1.15 PM –2.15 PM)**  
**Poster session 1 (Level 2, 1.30 PM – 2.15 PM)**

**Session 2 – Personal Genomics: Redefining Privacy, Choice and the Internet**  
**Chair: Ruth Chadwick**

2.15 PM – 3.00 PM

**Ruth Chadwick**

*Redefining Privacy, Choice and the Internet*

3.00 PM – 3.45 PM

**Helen Nissenbaum**

*Privacy, Technology, Policy, And The Integrity Of Social Life*

3.45 PM – 4.05 PM

*Tea Break*

4.05 PM – 4.50 PM

**Linda Avey**

*Personal Genetics - Lessons Learned and the Opportunities Ahead*

4.50 PM – 5.35 PM

**Jeantine Lunshof**

*Redefining Privacy: Public Genomes and Open Consent*

5.35 PM – 6.20 PM

**Bartha Maria Knoppers**

*Personal Genomics and Privacy*

7.30 PM – 9.30 PM

**Conference Dinner** at InterContinental Geneva Hotel

**Day Two – (2 November 2009)**

**Session 3 – Genetic Determinism,  
Discrimination, Exceptionalism and  
Selection**

**Chair: Stylianos E. Antonarakis**

9.00 AM – 9.45 AM

**Stylianos E. Antonarakis**

*The Medical Genome*

9.45 AM – 10.30 AM

**Alastair V. Campbell**

*What – if Anything – is Special about ‘Genetic  
Privacy’?*

10.30 AM – 10.50 AM

*Tea Break*

10.50 AM – 11.35 AM

**Thomas H. Murray**

*Genetic Exceptionalism: A Reassessment*

11.35 AM – 12.20 PM

**David Cox**

*Genetic Determinism and Real Life: The  
Application and Misapplication of Human DNA  
Sequence Variation*

12.20 PM – 2.20 PM

**Lunch @ CIG Restaurant (Level 1)**

**HUGO Ethics Committee Meeting @ Room 16**  
(Level -1, 1.00 PM – 2.20 PM)

**The HUGO Journal Editorial Board Meeting**  
@ Room 15 (Level -1, 1.00 PM - 2.20 PM)

**Brocher Foundation Short Presentation @**  
Hall 1 (1.00 PM – 1.45 PM)

**Poster Session 2 (Level 2, 1.20 PM – 2.20 PM)**

2.20 PM – 5.20PM

**Session 4- Discussion Forum**

**Chair: Edison T. Liu, Ruth Chadwick, Bartha Maria Knoppers**

3.40 PM – 4.00 PM

*Tea Break*

6.30 PM – 8.45 PM

**Public Lecture\*** at University of Geneva (CMU Hall B400)

**Stylianos E. Antonarakis and Bartha Maria Knoppers**

*\*Buses depart from CICG to CMU at 5.35 PM sharp (30 minutes journey). Return buses depart from CMU at 9.00 PM and stop at Railway Station Cornavin (10 minutes journey) and InterContinental Geneve Hotel (10 minutes journey).*

**Day Three – (3 November 2009)**

**Session 5 – Equity and Justice : Access and Participation in the Developing World**  
**Chair: Abdallah Daar**

- 9.00 AM – 9.45 AM      **Abdallah Daar**  
*Genomics Initiatives in Developing Countries*
- 9.45 AM – 10.30 AM    **Raj S. Ramesar**  
*Parent of Origin Effect: Will Africa be Relegated to the Role of a Neglected Parent Watching its Successful Children Bicker Over Their Inheritance*
- 10.30 AM – 10.50 AM    *Tea break*
- 10.50 AM – 11.35 AM    **Partha Majumdar**  
*Ethical Dilemmas in the Conduct of Genomic Research: When Technological Developments Encounter Economic and Educational Underdevelopment*
- 11.35 AM – 12.20 PM    **Gerardo Jimenez-Sanchez**  
*Genomic Medicine in Mexico: Progress and challenges related to equity and justice.*
- 12.20 PM – 1.50 PM    **Lunch @ CIG Restaurant (Level 1)**  
**Poster Session 3 (Level 2, 1.05 PM – 1.50 PM)**

**Session 6 – Open Access, Open Markets:  
Intellectual Property?**

**Chair: Bartha Maria Knoppers**

1.50 PM – 2.35 PM

**Charles Auffray**

*Redefining intellectual property in the transition  
from genomics to systems medicine*

2.35 PM – 3 20 PM

**Timothy Caulfield**

*Are Gene Patents the Problem?: A Look at  
Emerging Evidence*

3.20 PM – 3.40 PM

*Tea break*

3.40 PM – 4.25 M

**Philippe Ducor**

*« Open access » aspects of DNA patenting*

4.25 PM – 5.10 PM

**James Toupin**

*The Development of the Law of Gene Patenting  
in the United States*

5.10 PM – 5.15 PM

**Closing**



## **ACKNOWLEDGEMENTS**

This symposium has received financial support from the following organisations:

**Wellcome Trust (Institutional Sponsor)**

**Genome Canada (Diamond Sponsor)**

**Springer (Gold Sponsor)**

Brocher Foundation (Silver Sponsor)  
Illumina (Silver Sponsor)

We would like to extend our utmost appreciation to all of their support.

## INTERNATIONAL SCIENTIFIC COMMITTEE



### **Ala ALWAN**

Assistant Director-General  
Noncommunicable Diseases and Mental Health  
World Health Organization

Dr Ala Alwan is Assistant Director-General for Noncommunicable Diseases and Mental Health since 1 February 2008. Dr Alwan graduated in Medicine from the University of Alexandria. He practiced medicine in Scotland and obtained his postgraduate training and qualifications in the United Kingdom. Following his return to Iraq, his home country, he held several positions in clinical and academic medicine and public health. He was Professor and Dean of the Faculty of Medicine, Mustansiriya University, Baghdad.

In 1992, he joined WHO as Regional Adviser for Noncommunicable Diseases in the Regional Office for the Eastern Mediterranean. He then served as WHO Representative in Oman, and Director, Division of Health Systems Development in the Eastern Mediterranean Region. In 1998, Dr Alwan was reassigned to WHO headquarters as Director for Noncommunicable Diseases Prevention and then Director of the Department of Noncommunicable Diseases Management. In 2001, he became WHO Representative in Jordan. From 2003 to 2005, he was Minister of Education and Minister of Health in the Government of Iraq. From 2005 to January 2008, he was Representative of the Director-General and Assistant Director-General for Health Action in Crises.



## **Stylianos E. ANTONARAKIS**

Professor and Chairman  
Department of Genetic Medicine and Development  
University of Geneva Medical School and University Hospitals of Geneva

Stylianos Antonarakis is the Director of the Division of Medical Genetics at the University of Geneva in Switzerland. Before moving to Geneva he was a professor at Johns Hopkins University in the USA. His lab participates in many projects involving the genetics of monogenic and polygenic disorders as well as the annotation of the human genome and particularly of human chromosome 21. He is on the editorial board of many high profile journals such as *Genome Research* and *Genomics* and has recently served as president of the European Society for Human Genetics. He has published more than 470 papers in peer-reviewed journals since 1982.



## **Alastair V. CAMPBELL**

Director

Centre for Biomedical Ethics, Yong Loo Lin School of Medicine, NUS,  
Singapore

Chen Su Lan Centennial Professor of Medical Ethics, NUS, Singapore

Professor Alastair V. Campbell is the Director of CBmE. Prior to this, he was Professor of Ethics in Medicine in the Medical School of the University of Bristol and Director of its Centre for Ethics in Medicine. He is a former President of the International Association of Bioethics. With keen interest in classical music, reading and skiing, Professor Campbell is also a prolific writer with more than 30 books and book chapters as well as dictionary entries to his name; and many more articles in refereed journals. Recent books include *The Body in Bioethics* (Routledge-Cavendish, 2009), *Health as Liberation* (Pilgrim Press, 1995) and *Medical Ethics*, 4th Edition, co-authored with Grant Gillett and Gareth Jones (Oxford University Press, 2005). He was until recently a member of the Medical Ethics Committee of the British Medical Association and Chairman of the Ethics and Governance Council of UK Biobank. In 1999, Professor Campbell was given the prestigious Henry Knowles Beecher award, which recognises his lifetime of contribution to ethics and the life sciences, and his excellence in scholarship, research and ethical inquiry. He is also Honorary Vice-President of the Institute of Medical Ethics, UK and an elected Fellow of The Hastings Center, New York.

Professor Campbell is a member of the Bioethics Advisory Committee to the Singapore Government, of the National Medical Ethics Committee of the Ministry of Health and he is a Board Member of the Health Sciences Authority of Singapore and of the National Medical Research Council.



## **Ruth CHADWICK**

Director

ESRC Centre for Economic and Social Aspects of Genomics (Cesagen)  
Cardiff University  
Chairperson, HUGO Ethics Committee

Ruth Chadwick is Director of the ESRC (Economic and Social Sciences Research Council) Centre for Economic and Social Aspects of Genomics (Cesagen), Cardiff University, UK. She also holds a Link Chair between Cardiff Law School and the School of English, Communication and Philosophy (ENCAP). She has co-ordinated a number of projects funded by the European Commission, including the EUROSCREEN projects (1994-6; 1996-9) and co-edits the journal *Bioethics* and the online journal *Genomics, Society and Policy*. She is Chair of the Human Genome Organisation Ethics Committee and has served as a member of several policy-making and advisory bodies, including the Panel of Eminent Ethical Experts of the Food and Agriculture Organisation of the United Nations (FAO), and the UK Advisory Committee on Novel Foods and Processes (ACNFP). She was editor-in-chief of the award winning *Encyclopedia of Applied Ethics* (1998), of which a second edition is now being prepared. She is an Academician of the Academy of Social Sciences and a Fellow of the Hastings Center, New York; of the Royal Society of Arts; and of the Royal Society of Medicine. In 2005 she was the winner of the World Technology Network Award for Ethics for her work on the relationship between scientific developments and ethical frameworks.



## **Abdallah S. DAAR**

Professor of Public Health Science and of Surgery , University of Toronto  
Director of Ethics and Commercialization, McLaughlin-Rotman Centre for  
Global Health

Member, Ethics Committee, HUGO

Chair, Board of Advisors, UN University International Institute for Global  
Health

Chair, Global Alliance for Chronic Diseases

Professor Abdallah S Daar is Professor of Public Health Sciences and of Surgery at the University of Toronto. He is also Senior Scientist and Director of the Program on Ethics and Commercialization, McLaughlin-Rotman Centre for Global Health, University Health Network and University of Toronto.

His major research focus is on the use of life sciences to ameliorate global health inequities, with a particular focus on building scientific capacity and increasing innovation in developing countries, in addition to studying how technologies can be rapidly taken from “lab to village”.

His work has spanned biomedical sciences, surgery, organ transplantation, bioethics and global health. His major international awards include the Hunterian Professorship of the Royal College of Surgeons of England and the UNESCO Avicenna Prize for Ethics of Science. He is a fellow of the Royal Society of Canada, the New York Academy of Sciences, the Canadian Academy of Health Sciences and of the Academy of Sciences of the Developing World (TWAS). He has published 5 books, and over 300 research articles and chapters in books. He has trained hundreds of graduate students.

Daar is a member of UNESCO's International Bioethics Committee and the Ethics Committee of the Human Genome Organisation. He was a member of the Africa Union High Level Panel on Modern Biotechnology, which published its seminal report, “Freedom to Innovate”, in 2007. His recent accomplishments include work on the Grand Challenges in Global Health, and he led a global study of Grand Challenges in Chronic Non-Communicable Diseases. He has advised the UN Secretary General's Office, UNESCO, WHO, and the Government of Canada.

He studied medicine at Makerere University, Uganda and University of London, before going to Oxford for clinical residency and fellowship training and a doctorate in immunology. He was on the faculty of Oxford University before moving to the Middle East to help start two new medical schools. He moved to the University of Toronto in 2001. Professor Daar is Chair of the Board of Advisors of the United Nations University International Institute of Global Health, and of the Board of the recently created Global Alliance for Chronic Diseases.



## **Bartha-Maria KNOPPERS**

Professor

Director, Centre of Genomics and Policy Faculty of Medicine

Department of Human Genetics, McGill University

Former Chairperson, HUGO Ethics Committee

Dr. Knoppers (Phd) holds the Canada Research Chair in Law and Medicine (Tier 1: 2001 - ). She is Professor at the Faculté de droit, Université de Montréal and Senior Researcher at the Centre for Public Law. Dr. Knoppers is former Chair of the International Ethics Committee of the Human Genome Organisation (1996-2004). She is Co-Founder of the International Institute of Research in Ethics and Biomedicine and Chair of the Ethics Working Party of the International Stem Cell Forum (U.K.). In 2003, she founded the international Public Population Project in Genomics (P3G) and became the Principal Investigator of CARTaGENE. CARTaGENE is a resource of samples and data on the genetic diversity of the population of Quebec.



## **Edison T. LIU**

Executive Director, Genome Institute of Singapore (Biomedical Sciences Institutes)

Professor of Medicine, National University of Singapore

President, Human Genome Organisation

Dr. Edison Liu received his residency training in internal medicine at Washington University, St. Louis, and clinical cancer fellowships at Stanford University (Oncology), and at the University of California at San Francisco (Hematology). He then pursued post-doctoral studies as a Damon-Runyan Cancer Research Fellow at the University of California at San Francisco in the laboratory of Dr. J. Michael Bishop. In 1987, he joined the faculty of Medicine at the University of North Carolina at Chapel Hill where he was director of UNC's Specialized Program of Research Excellence (SPORE) in Breast Cancer. In 1996, he joined the NCI as the Director of the Division of Clinical Sciences. In 2001, Dr. Liu assumed the position of Executive Director, Genome Institute of Singapore. His current scientific research investigates the dynamics of gene regulation on a genome scale that can explain biological states in cancer. Dr. Liu has contributed over 245 articles, reviews, and book chapters to the scientific literature. Dr. Liu also is the Executive Director of the Singapore Tissue Network, the national tissue repository in Singapore. Dr. Liu's awards include the Leukemia Society Scholar (1991-1996), the Brinker International Award for basic science research in Breast Cancer (1996), the Rosenthal Award from the American Association for Cancer Research (2000), the President's Public Service Medal for his work in helping Singapore resolve the SARS crisis, and a Doctor of Medicine Sciences honoris causa (2007). In 2007, Dr. Liu was elected the President of the Human Genome Organisation (HUGO).



## **Alex MAURON**

Professor of Bioethics, Medical Faculty  
University of Geneva

Alex Mauron studied biology at the University of Lausanne and in 1978 received his doctorate in molecular biology. He completed his post-doctoral studies at Stanford University in the USA and at the Institute for Biochemistry at the University of Geneva. Since 1987 Alex Mauron has worked in bioethics, first as a scientific advisor at the Fondation Louis-Jeantet de Médecine in Geneva and from 1995 as an associate professor for bioethics at the medical faculty of the University of Geneva (full professor since 2001). In addition to his research and teaching in bioethics, Alex Mauron has been presenting bioethical and policy issues in the public media on a regular basis. He has been a member of the Swiss Science and Technology Council since 2004. Alex Mauron was born on 26 July 1951 in Pasadena, California. He has dual Swiss and French nationality.



## **Thomas H. MURRAY**

President and CEO, The Hastings Center  
Member, HUGO Ethics Committee  
Board of Directors, Charity Navigator  
Physicians and Lawyers for National Drug Policy  
Consulting Editor in Bioethics, Johns Hopkins University Press

Thomas H. Murray is President and CEO of The Hastings Center. Dr. Murray was formerly the Director of the Center for Biomedical Ethics in the School of Medicine at Case Western Reserve University, where he was also the Susan E. Watson Professor of Bioethics. He serves on many editorial boards, has been president of the Society for Health and Human Values and of the American Society for Bioethics and Humanities, and among other current posts serves as Chair of the Ethical Issues Review Panel for the World Anti-Doping Agency and as International Expert Advisor to Singapore's Bioethics Advisory Committee. Dr. Murray has testified before many Congressional committees and is the author of more than 200 publications including *The Worth of a Child*, *The Cultures of Caregiving: Conflict and Common Ground among Families, Health Professionals and Policy Makers*, edited with Carol Levine, and *Genetic Ties and the Family: The Impact of Paternity Testing on Parents and Children*, edited with Mark A. Rothstein, Gregory E. Kaebnick and Mary Anderlik Majumder. He is also editor, with Maxwell J. Mehlman, of the *Encyclopedia of Ethical, Legal and Policy Issues in Biotechnology*. In January 2004 he received an honorary Doctor of Medicine degree from Uppsala University.



## **Tikki PANG**

Director, Research Policy and Cooperation (RPC/IER),  
World Health Organization

Dr Tikki PANG is presently Director, Research Policy and Cooperation, World Health Organization, Geneva, Switzerland. Prior to joining WHO, he was Professor of Biomedical Sciences, Institute of Postgraduate Studies and Research, University of Malaya, Kuala Lumpur, Malaysia. He holds a PhD in Immunology-Microbiology from the Australian National University, Canberra, Australia. He is a Fellow of the Royal College of Pathologists (UK), Institute of Biology (UK), American Academy of Microbiology (USA), Academy of Medicine of Malaysia, and Academy of Sciences for the Developing World (TWAS); and a Member of the International Molecular Biology Network (IMBN). Also, he is currently Secretary, WHO Research Ethics Review Committee, and Secretary, WHO Advisory Committee on Health Research. Research interests are in epidemiology, pathogenesis, laboratory diagnosis and prevention of infectious diseases, and in health research policy, health research systems, best practices in research, development of research capabilities in developing countries, and linkages between research and policy. He has published more than 200 scientific articles and 6 books on a variety of topics.



## **Jean-Dominique VASSALLI**

Rector of the University of Geneva

VASSALLI, Jean-Dominique - Rector of the University of Geneva Born in Geneva, Jean-Dominique Vassali obtained his Swiss Federal diploma of medicine at Geneva University in 1972, his Doctorate in Sciences (PhD) at Rockefeller University in 1977 and his Doctorate in Medicine at Geneva University in 1984. In 1994, he was appointed Vice Dean of the Faculty of medicine until 1999. Then he was appointed Vice Rector of Geneva University, and held this position until 2003. He was notably head of cases about the collaboration between Vaud and Geneva, and the valorization of the scientific discoveries. Since 2004, he presides the fundamental medicine Section of the Faculty of medicine. In July 2007, he was appointed Rector of Geneva University.

## PANEL OF INTERNATIONAL SPEAKERS



### **Charles AUFFRAY**

Genexpress team - Systemoscope Consortium  
Functional Genomics and Systems Biology for Health  
CNRS Institute of Biological Sciences - Villejuif – France  
Chairperson, HUGO Intellectual Property Committee

Dr. Charles Auffray, Research Director at CNRS, heads the Genexpress team in Functional Genomics and Systems Biology for Health, located in Villejuif, France, affiliated with the CNRS Institute of Biological Sciences.

Dr. Charles Auffray obtained the Agrégation in Physiology and Biochemistry at Ecole Normale Supérieure de Cachan in 1975, his PhD in Molecular Immunology and Genetic Engineering at Pierre and Marie Curie University of Paris VI and Pasteur Institute, Paris in 1981. From 1981-1983 he was a Post-doctoral Fellow and Junior Faculty at the Department of Biochemistry and Molecular Biology of Harvard University in Cambridge, USA; from 1983-1991 a Group Leader at the Institute of Embryology, CNRS and Collège de France, Nogent s/Marne; from 1991-1995 the Scientific Director of the Genexpress Program at Généthon in Evry, concurrently Head of the CNRS Research Unit in Molecular Genetics and Developmental Biology, then Functional Genomics and Systems Biology for Health in Villejuif (1991-2003).

Dr. Charles Auffray develops a systems approach to cancer and the physio-pathology of the immune, neuro-muscular and respiratory systems by integrating functional genomics tools with mathematical, physical and computational approaches. He is actively involved in the development of open-access reagent, instrumentation and information platforms, in partnership with academic and industrial groups. He co-founded the IMAGE and SYSTEMOSCOPE International Consortia to also tackle related ethical, legal, and social issues with emphasis on interdisciplinarity and public education. Dr. Charles Auffray is the co-author of 237 original publications, including 175 in peer-reviewed scientific journals and 10 books, which have received over 12,000 citations. He is a member of several international societies, review committees, and editorial boards, and the co-organiser of a series of international conferences on Functional Genomics and Systems Biology.



## **Linda AVEY**

**Founder of Alzheimer's Research Foundation  
Co-Founder (Former), 23andMe**

Linda has over 20 years of sales and business development experience in the biopharmaceutical industry in San Francisco, Boston, San Diego, and Washington, D.C. Prior to starting 23andMe, she developed translational research collaborations with academic and pharmaceutical partners for Affymetrix and Perlegen Sciences. Linda also spent time at Spotfire helping scientists understand the power of data visualization and at Applied Biosystems during the early days of the human genome project. The advent of high density genome-wide scanning technologies brought huge potential for significant discoveries. However, the lack of sufficient funding to enable adequate studies prompted Linda to think of a new research model. These ideas led to the formation of 23andMe. Her primary interest is the acceleration of personalised medicine, using genetic profiles to target the right drug to the right person at the correct dose. Linda graduated from Augustana College with a B.A. in biology.



## **Timothy CAULFIELD**

**Canada Research Chair in Health Law and Policy  
Senior Health Scholar, Alberta Heritage Foundation for Medical Research  
Professor, Faculty of Law and School of Public Health  
Research Director, Health Law Institute**

Timothy Caulfield has been Research Director of the Health Law Institute at the University of Alberta, since 1993. In 2001 he received a Canada Research Chair in Health Law and Policy. He is also a Professor in the Faculty of Law and the School of Public Health. Over the past several years, he has been involved in a variety of interdisciplinary research endeavours that have allowed him to publish over one hundred and fifty articles and book chapters. He is a Senior Health Scholar with the Alberta Heritage Foundation for Medical Research, the Principal Investigator for Genome Canada project on the regulation of genomic technologies, the theme leader in the Stem Cell Network and the Advanced Foods and Materials Network (National Centres of Excellence) and has several projects funded by the Canadian Institutes of Health Research.

Professor Caulfield is and has been involved with a number of national policy and research ethics committees, including Canadian Biotechnology Advisory Committee, Genome Canada's Science Advisory Committee, the Federal Panel on Research Ethics and the Royal Society of Canada's Expert Panel on the Future of Food Biotechnology (2001). He is a member of the Royal Society of Canada and the Canadian Academy of Health Sciences. He teaches biotechnology in the Faculty of Law and is the editor for the Health Law Journal and Health Law Review.



## **David COX**

Senior Vice President and CSO  
Biotherapeutics and Bioinnovation Center  
Pfizer Inc.

David serves as Chief Scientific Officer for the Target Generation Unit of Pfizer's Biotherapeutics and Bioinnovation Center. This new unit brings together human genetics, systems biology, and cell biology, combining internal capabilities with outside collaborations, to focus on increasing preclinical target validation. David is a co-founder of Perlegen, and was most recently Chief Scientific Officer of the company since its formation in 2000. David was Professor of Genetics and Pediatrics at the Stanford University School of Medicine as well as the co-director of the Stanford Genome Center. He obtained his A.B. and M.S. degrees from Brown University in Rhode Island and his M.D. and Ph.D. degrees from the University of Washington, Seattle. He completed a 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. David is certified by the American Board of Pediatrics and the American Board of Medical Genetics. He was an active participant in the large scale mapping and sequencing efforts of the Human Genome Project while carrying out research involving the molecular basis of human genetic disease. David has been a member of several commissions and boards, including the National Bioethics Advisory Commission (NBAC) and the Health Sciences Policy Board of the Institute of Medicine. He is presently a council member of the international Human Genome Organisation (HUGO). He has authored over 100 peer-reviewed scientific publications and has served on numerous editorial boards. Dr. Cox's honors include election to the Institute of Medicine of the National Academy of Sciences.



## **Philippe DUCOR**

**Associate Professor in the Department of Commercial Law  
Director of the Interdisciplinary Master in Life Sciences Law  
Geneva University  
Partner in Geneva business law firm**

Philippe Ducor is a professor at the University of Geneva Law School and the director of the Master in life sciences law program, where he teaches technology transfer and biotechnology law. He also practices law as a partner with a Geneva business law firm.

Philippe is fully trained in medicine and law (both at PhD level, including professional medical and legal certifications) and obtained degrees from the Universities of Geneva and Stanford. His areas of expertise cover intellectual property as applied to technology, esp. patent law, licensing, IP strategy and policy, unfair competition, pharma/biotech law, health law and medical malpractice law. His work has been published in legal and scientific journals, such as Nature and Science.

Among other associative activities, Philippe is a member of the Scientific Advisory Board of the Brocher Foundation in Geneva.



## **Gerardo JIMENEZ-SANCHEZ**

Director General

National Institute of Genomic Medicine, Mexico

Instituto Nacional de Medicina Genómica

Dr. Gerardo Jimenez-Sanchez was born in Mexico City. He obtained his Medical Doctor degree from the National Autonomous University of Mexico (UNAM). He did his residency in Pediatrics at the National Institute of Pediatrics and earned his Ph.D. Degree in Human Genetics and Molecular Biology from the Johns Hopkins University in Baltimore, MD, USA.

Dr. Jimenez-Sanchez is Professor of Genomic at the UNAM and Resident Investigator of the Mexican Health Foundation (FUNSALUD). He is also Director General of the National Institute of Genomic Medicine and affiliate member of the Institute of Genetic Medicine at Johns Hopkins University.

His actual work focuses on the development of a national platform in genomic medicine for Mexico. This includes the conduction of major scientific research projects, including the production of the first haplotype map for the Mexican Mestizo population. Dr. Jimenez-Sanchez is a certified pediatrician and a member of the Mexican Academy of Pediatrics, the National Academy of Medicine, the Mexican Society of Biochemistry and the American Societies of Human Genetics, Inborn Errors of Metabolism and Gene Therapy, the European Society of Inborn Errors of Metabolism and the Human Genome Organisation (HUGO).

Dr. Jimenez-Sanchez work has resulted in the publication of articles and chapters in specialised journals and books. He received the Research in pediatrics Award of the Society for Pediatric Research in 1999. Along with his colleagues David Valle and Barton Childs, he did the first medical analysis of the human genome, published with the first draft of the human genome in 2001. He received the National Award in Clinical Investigation "Dr. Miguel Otero" from the Government of Mexico. In April of 2003, he was appointed Silanes Professor in Genomic Medicine. He is the regional editor for Latin America and the Caribbean of the Journal Genomic Medicine and charter member of the international Public Population Project in Genomics (P3G Consortium).



## **Klaus LINDPAINTNER**

Vice President and Director, Roche Genetics  
F. Hoffman-La Roche Ltd.

Klaus Lindpaintner graduated from Innsbruck University Medical School with a degree in Medicine and from Harvard University with a degree in Public Health. He pursued postgraduate training and specialisation in Internal Medicine, Cardiology, and Genetics in the United States and Germany and holds board certifications in these specialties. He practiced interventional cardiology and pursued academic research in the area of cardiovascular disease, molecular genetics, and genetic epidemiology, most recently as an Associate Professor of Medicine at Harvard Medical School. He joined Hoffman-La Roche in 1997 and serves currently as Director of the “Roche Molecular Medicine Laboratories” in Basel, and Global Head, Molecular Medicines Policy and External Affairs, coordinating the company’s efforts and activities in implementing biomarker research based on genetics, genomics, proteomics, and associated disciplines across the value chain from early discovery to late stage clinical trials. He has co-authored more than 200 scientific papers, and currently holds honorary and adjunct professorships at the University of London Guy’s and St. Thomas, Jiao Tong University in Shanghai, Stanford University, and the University of Vienna. He has served on numerous working groups and advisory panels for trade organisations, regulatory authorities, and non-governmental organisations on issues related to ethical and societal impact of novel technologies in biomedicine.



## **Jeantine LUNSHOF**

Assistant Professor  
European Centre for Public Health Genomics  
Maastricht University and  
Department of Molecular Cell Physiology  
VU University Amsterdam

Jeantine Lunshof is a philosopher and bioethicist. She is an Assistant Professor at the European Centre for Public Health Genomics, Maastricht University, The Netherlands. Further affiliations are with the Department of Molecular Cell Physiology, VU University, Amsterdam, The Netherlands Institute for Systems Biology (NISB), and with CESAGen at Cardiff University, UK. Since 1992, Jeantine has been involved in international research collaborations on ethical and public-policy issues in human genetics. Her current work focuses on ethical questions related to pharmacogenomics, genome-wide association studies and personalised medicine, and on the philosophy of systems biology, in particular its conceptual and normative issues. Jeantine has been an ethics consultant to the Personal Genome Project at Harvard Medical School, Genetics Department, since early 2006. Recent publications address the challenges that advances in the genomics sciences and technologies pose to traditional biomedical ethics. She is an affiliate member of the NIH Pharmacogenetics Research Network (PGRN), and a Member of the Public Population Project in Genomics (P3G) Consortium. She serves on a number of Editorial Boards.



## **Partha MAJUMDAR**

**Professor and Head, Human Genetics Unit, Indian Statistical Institute  
Chief Programme Officer, National Institute of Biomedical Genomics  
Director, TCG-ISI Centre for Population Genomics, Kolkata**

Partha Majumdar is currently Professor and Head for the Human Genetics Unit, Indian statistical Institute, Kolkata, and Director of the TCG-ISI Centre for Population Genomics, Kolkata. His major scientific interests and contributions have been in the field of human population genetics and genetics of complex human disorders. He has over 200 scientific publications to his credit. He is an elected Fellow of all the three science academies of India. He has served on the Board of Directors of the International Genetic Epidemiology Society (IGES), and was the founding Chair of the Ethical, Legal and Social Issues (ELSI) Committee of IGES. He is a recipient of many awards and medals, including the G.D. Birla Award for Scientific Research – 2002, Shri Om Prakash Bhasin Award in Biotechnology – 2001, Ranbaxy Research Award in Applied Medical Sciences – 2000, and the New Millennium Science Medal, ISCA and CSIR, Government of India, 2000.



## **Mark McCARTHY**

Robert Turner Professor of Diabetes  
Oxford Centre for Diabetes, Endocrinology and Metabolism

Mark McCarthy is Robert Turner Professor of Diabetes at Oxford University, based at the Oxford Centre for Diabetes, Endocrinology and Metabolism (OCDEM) and at the Wellcome Trust Centre for Human Genetics. His work on the genetics and genomic basis of type 2 diabetes seeks to define the relationship between genetic variation, environment, intermediate phenotypes, and clinical disease, in the belief that such information will translate into a significant impact on clinical care for people with diabetes, through more rational and effective deployment of available preventative and therapeutic modalities.

He obtained his medical degree at the University of Cambridge and postgraduate clinical training in general medicine, diabetes and endocrinology at the London Hospital. Following research training with – amongst others – Newton Morton and Eric Lander, he took up an appointment as Senior Lecturer (subsequently Professor in Genomic Medicine) at Imperial College in 1995, moving to Oxford as the Robert Turner Professor of Diabetes in 2002. In the past decade, his group has become established as one of the leading international teams working on the genetics of T2D, with expertise extending from physiological characterisation through to high-throughput genomic analysis and related issues in computational biology. Prof McCarthy currently leads the T2D component of the Wellcome Trust Case Control Consortium, and has been intimately involved in many of the recent discoveries generated by the application of genome wide association approaches to diabetes, obesity and related traits. His group is increasingly involved in efforts to understand how such information can be translated into advances in clinical management, and in extending these studies beyond the European context. He has a strong commitment to the wider research community, having served as a member and chair of the Diabetes UK Research Committee, and on Wellcome and MRC panels. He serves on the Editorial Board of Diabetes, Diabetologia, PLoS Medicine, Human Molecular Genetics and several other journals.



## **Helen NISSENBAUM**

Professor  
Media, Culture and Communication  
New York University

Helen Nissenbaum is Professor of Media, Culture and Communication, and Computer Science, at New York University, where she is also Senior Faculty Fellow of the Information Law Institute. Her areas of expertise span social, ethical, and political implications of information technology and digital media. Nissenbaum's research publications have appeared in journals of philosophy, politics, law, media studies, information studies, and computer science. She has written and edited three books and a fourth, *Privacy in Context: Technology, Policy, and the Integrity of Social Life*, is due out in 2009, with Stanford University Press. The National Science Foundation, Air Force Office of Scientific Research, Ford Foundation, and U.S. Department of Homeland Security have supported her work on privacy, trust online, and security, as well as several studies of values embodied in computer system design, including search engines, digital games, and facial recognition technology. Nissenbaum holds a Ph.D. in philosophy from Stanford University and a B.A. (Hons) from the University of the Witwatersrand. Before joining the faculty at NYU, she served as Associate Director of the Center for Human Values at Princeton University.



## **Olufunmilayo OLOPADE**

**MBBS, FACP**

**Walter L. Palmer Distinguished Service Professor**

**Director, Center for Clinical Cancer Genetics and Global Health**

**Associate Dean for Global Health**

**The University of Chicago**

Dr. Olufunmilayo Olopade is a highly skilled hematology oncologist with proven expertise in cancer risk assessment. She conducts comprehensive evaluations of family history and other risk factors for patients in the Cancer Risk Clinic. Once she identifies a person at high risk, Dr. Olopade designs an individualised prevention plan that may include periodic screening, preventative drugs or other options for risk reduction. She is an expert on individualized treatment for breast cancer. Dr. Olopade takes an interdisciplinary approach to breast cancer care with treatment plans tailored to each person's risk factors. She also focuses on quality of life concerns for young breast cancer patients--addressing topics related to pregnancy, fertility and employment.

As an international leader in breast cancer research, Dr. Olopade continues to help scientists gain a greater understanding of the disease. Her current research interests include identifying the source of ER-negative breast cancer--an aggressive form of the disease, which is resistant to hormone therapy. Dr. Olopade aims to improve screening standards and early detection for moderate- and high-risk populations. She has a special interest in women of African descent, who are at higher risk for the more aggressive breast cancer and more likely to be diagnosed at a younger age. Dr. Olopade has lectured on topics such as breast cancer and cancer genetics at several national and international conferences.



## **Raj S. RAMESAR**

Professor and Head  
Division of Human Genetics  
MRC Human Genetics Research Unit  
Institute for Infectious Diseases and Molecular Medicine  
Faculty of Health Sciences  
University of Cape Town and Allied Hospitals

Raj Ramesar is Professor and Head of the Division of Human Genetics at the University of Cape Town and its Allied Hospitals in South Africa. This facility has wide-ranging clinical responsibilities from the quaternary and tertiary care levels, to extensive rural outreach programmes, in addition to diagnostic and research capabilities.

His interest is in using the exciting developments in the field of genomic sciences to investigate human biodiversity. Africa offers the opportunity to use population lineages in all of their richness towards identifying aspects of human biology that have to do with both health and disease. As the Director of the MRC Human Genetics Research Unit, the emphasis of his research has been on disease susceptibility in South African populations, progressing from the commonly recognised inherited diseases, to those that are more complex yet more common and relevant to a large burden of disease. In this regard, his most recent research enterprise is embodied in a large scale project entitled: 'Human Diversity and Health'.

As Director of the national Colorectal Cancer Research Consortium his focus has been on the genetics of familial colorectal cancers, and the most effective translation of laboratory findings to the field for optimum benefit of patients and their kin. In this regard Raj recently received the (Vice Chancellor's) Alan Pifer Award for 'outstanding research in cancer genetics which shows relevance to the advancement of South Africa's disadvantaged populations'. Apart from being on the editorial board of several international journals, Raj serves on the Executive of the African Society for Human Genetics, and is its Liaison Officer to the International Federation of Human Genetics Societies.



## **James TOUPIN**

### **General Counsel**

### **United States Patent and Trademark Office (USPTO)**

James A. Toupin became the General Counsel of the United States Patent and Trademark Office (USPTO) in January 2001. He has executive responsibility for the Office of the Solicitor, the Office of General Law, the Office of Enrollment and Discipline, the Board of Patent Appeals and Interferences, and the Trademark Trial and Appeal Board. In this position, Mr. Toupin supervises the provision of legal advice and court representation on all intellectual property and administrative matters for the USPTO. He serves as a member of the USPTO Executive Board, Management Council, Executive Review Board, and Performance Review Board. Mr. Toupin also coordinates with the Department of Justice, Department of Commerce, and other agencies in developing the U.S. position on major intellectual property cases before the Supreme Court and Court of Appeals for the Federal Circuit. In 1987, he was appointed Assistant General Counsel for Litigation of the United States International Trade Commission (ITC), and subsequently to Deputy General Counsel. In those positions, he supervised defence of the Commission's actions in U.S. courts and international tribunals. The domestic court representation included defence of the Commission's determinations on patent and other intellectual property matters in the Federal Circuit and of the Commission's determinations in antidumping and countervailing duty investigations in the U.S. Court of International Trade and the Federal Circuit. While at the ITC, Mr. Toupin assisted in the negotiations leading to the adoption of unfair trade practice provisions in the U.S.-Canada Free Trade Agreement and the North American Free Trade Agreement (NAFTA) and the Agreement Establishing the World Trade Organization and the Dispute Settlement Understanding for the WTO Agreements. He subsequently represented the United States in proceedings under those Agreements. He has been a member of the Advisory Committee of the Court of International Trade, as well as of the judicial conference organising committees of the Court of Appeals for the Federal Circuit.

Mr. Toupin began his professional career at Covington and Burling in Washington, D.C., specialising in intellectual property and unfair competition law and administrative agency litigation. From 1985 to 1987, he worked for the Washington office of the California law firm of Memel, Jacobs, Pierno, Gersh and Ellsworth, where he concentrated on international trademark registration and licensing. A native of San Francisco, Mr. Toupin earned a bachelor's degree in with distinction from Stanford University, and a juris doctor from the Boalt Hall School of Law at the University of California at Berkeley, where he was an editor of the California Law Review . He is a member of the California and District of Columbia Bars.

## **ABSTRACTS – INTERNATIONAL SPEAKERS**

### ***The Medical Genome***

#### **Stylianos E. ANTONARAKIS**

For about 50 years, since the discovery of DNA, people were dreaming of the day that the complete diploid sequence of the genome of each individual would be possible. Well, this day is almost there! And the initial sequencing of a few individual genomes, confirmed the earlier prediction that there is a tremendous variability both in terms of SNPs and CNVs. The important question and challenge is now apparent: How could the knowledge of the genome sequence improve the health of individuals and the health of the population?

The therapeutic answer could be summarised as follows: the genome analysis will provide knowledge on disease mechanisms that in turn will result in intelligently targeted treatments. This is however a slow process that will take many decades per disorder.

The diagnostic answer could be naively summarised as: one can predict the development of a disorder by knowing the individual predispositions. To arrive at scientifically sound risk assessment may also require many decades of research, large numbers of individual genomes sequenced, and careful phenotypic evaluation and follow up. In addition, the expectations of risk prediction need to be realistic and not exaggerated. Genetic counseling becomes a moving target since new knowledge introduces additional dimensions to the overall risk.

The jump from a single variant information (the experience of which clinical genetics in the last 20 years), to the total genome variation (the new reality) is enormous. The clinical utility of the “Medical Genome” requires a knowledge-based process that undergoes rigorous validation similar to the medical standards for other diagnostic and predictive tests.

## ***Redefining intellectual property in the transition from genomics to systems medicine***

**Charles AUFFRAY**

High-throughput technologies for DNA sequencing and for analyses of transcriptomes, proteomes and metabolomes are advancing rapidly, providing the foundations for deciphering the structure, variation and function of the human genome of a large number of individuals, and relating them to their health and disease states. The wealth of data produced is being analysed and modelled using increasingly powerful computational and mathematical tools enabling the development of systems approaches for deciphering the functional and regulatory networks underlying the behaviour of complex biological systems in normal and perturbed conditions. Systems biology approaches are beginning to transform the way diagnostic tests and novel drugs that target multiple components of networks and pathways perturbed in diseases are developed. They will enable medicine to become predictive, personalised, preventive and participatory. The development of an international network of systems biology and medicine centres would facilitate the transition from genomics to systems medicine and help reduce the gap in healthcare between developed and developing countries. In order to overcome the bottlenecks that limit the development of diagnostic and therapeutic products, academic and industrial researchers, patient organisations and charities, and regulatory and funding institutions should redefine the basis for a more open and efficient sharing of the knowledge collected in large-scale clinical and experimental studies, which are often poorly designed and exploited. In the process, it will be essential that the basic elements of description of biological systems should remain available to all, and that the requirements for granting intellectual property rights rely on the clear demonstration of useful applications.

### **Reference Articles:**

Systems medicine: the future of medical genomics and healthcare  
Auffray, C., Chen, Z. and Hood, L. (2009) *Genome Medicine* 1:2.

Sharing knowledge: a new frontier for public-private partnerships in medicine  
Auffray, C. (2009) *Genome Medicine* 1:29.

## ***Personal Genetics - Lessons Learned and the Opportunities Ahead***

**Linda AVEY**

The personal genetics industry is less than two years into establishing its footing in this new--and many would argue, controversial--era of individual access to genetic information, based on consumer choice. Proponents of this nascent field herald the benefits of self-empowerment--that knowledge, even if it's incomplete, is better than ignorance. Skeptics, on the other hand, claim that it's too early in the evolution of our understanding of the role genetics plays in our health, that it isn't standardised<sup>1</sup>, and that the lay public is generally unable to comprehend this limitation, much less the statistical basis of genetic risk. There is also the concern that the medical establishment is unprepared to deal with the onslaught of patients armed with this rather complex and dense information. Both sides of the debate will be explored.

What tends to be ignored is the value of these personal genetics services as research platforms. The early mission of 23andMe was to create the ability to not only arm individuals with their genetic data through a dynamically updated online interface, but to empower them to be part of research studies of their own choosing. The model for this concept is being established<sup>2</sup> and will expand into disease-related phenotypes as cohorts are introduced. The value of combining disease groups into a centralised, standardised analysis platform is yet to be proven but the goal is to challenge existing, "siloeed" approaches to disease research. By enabling patient-centric, socially-networked communication, the goal is to bring research into a "2.0" construct.

### **Reference Articles:**

*Nature* 461, 724-726. An agenda for personalised medicine. Pauline C. Ng, Sarah S. Murray, Samuel Levy, J. Craig Venter, 7 October 2009.

ASHG'09 abstract. Web-based, participant-driven studies yield novel genetic associations for common traits. N. Eriksson, J. M. Macpherson, J. Tung, L. Hon, B. Naughton, S. Saxonov, L. Avey, A. Wojcicki, I. Pe'er, J. Mountain, 2009.

## ***What – if Anything – is Special about ‘Genetic Privacy’?***

**Alastair V. CAMPBELL, Jacqueline J L Chin**

A general problem of privacy arises when technology delivers impressive powers of data capture, storage, processing and retrieval. Ubiquitous online networking, high-speed extraction and analysis of personal information at ever lower cost for use by individuals, corporations and governments have now made disclosure of personal information essential to individual participation in everyday life. Likewise, the progress of genomics requires statistically significant and representative data on human populations. The benefits of creating genetic databanks for various uses, such as the medical, genealogical, forensic and recreational, include economic growth and the development of commercial interests, advancement of science, promotion of public health and public safety. But proliferation of public and private sector genetic databases and genomic research worldwide is also widely perceived as a threat to individual and group privacy interests.

In this paper, we shall attempt to examine how expanded uses of genetic information do raise issues of privacy in a range of contexts, but we also suggest that some putative ‘special’ privacy claims arising from the availability of genetic knowledge are false, and do not warrant special ethical attention. Rather, the privacy of personal information is a central ethical requirement, crossing the boundary between genetic and non-genetic information sources.

### **Reference Articles:**

Neil C. Manson (2006) What is genetic information, and why is it significant? A Contextual, Contrastive Approach. *Journal of Applied Philosophy*; 23 (1): 1-16

Anita L. Allen, Genetic privacy: emerging concepts and values, in Mark A. Rothstein (ed.) *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era*, New Haven and London, Yale University Press, 1997

## ***Are Gene Patents the Problem?: A Look at Emerging Evidence***

**Timothy CAULFIELD**

For decades there has been concern that gene patents are hurt the research environment - particularly upstream academic work. Indeed, patenting has been on of the most dominant areas of ethical and legal inquiry in the field of genetics. In general, the concern is that patents lead to a more secretive ethos and make research more expensive and inefficient. This concern, often called the "anti-commons" effect, has led to a great deal of policy debate and some concrete policy actions. But do gene patents really hurt the genetic research?

Emerging evidence from throughout the world indicates that this may not be the case and that our policy efforts have been focused on the wrong concern. In this presentation I will: 1) review available empirical data on the impact of gene patents, including relevant work by our team at the University of Alberta; 2) argue that the real issue, at least in the context of data withholding, is commercialisation pressure more broadly; and 3) suggest that existing policy efforts should be more evidence based and refocused.

## ***Redefining Privacy, Choice and the Internet***

**Ruth CHADWICK**

The concept and protection of privacy is coming under pressure in a number of different 'spheres'. The genomics sphere is one in which discussion of challenges to privacy has been considerable, but there are analogous discussions in other areas including those concerning biometrics and surveillance; and privacy on the internet. Following E.M.Forster's 'only connect' this presentation argues that important insights may be gained by bringing the spheres together.

## ***Genetic Determinism and Real Life: The Application and Misapplication of Human DNA Sequence Variation***

**David COX**

Broad public interest in the predictive power of genetic information, combined with access to large amounts of genetic information via the internet, has created a revolution in consumer genetics. However, consumer desire to use genetic information to improve the quality of life is often hampered by lack of sufficient scientific evidence relating genetic differences to clinical outcomes, as well as by a lack of specific options that can be utilised to avoid adverse outcomes. This talk will focus on the opportunities and challenges of using genetic information to improve life in a setting of strong preconceived opinions and limited knowledge.

## ***Genomics Initiatives in Developing Countries***

**Abdallah S. DAAR**

In 2002 the World Health Organization declared that all nations, including developing nations, need to harness the advantages of emerging genomic sciences and associated technologies to ensure global health equity. The McLaughlin-Rotman Centre for Global Health has a major interest in exploring how cutting edge science and technology goes from “the Lab to the Village” and our research over the past decade has largely focused on the various issues along this path. We have studied health biotechnology national innovation systems, followed by studies of individual companies, and then individual technologies in countries in several developing countries. Among the issues we have explored are the ethical, economic, environmental, legal and social challenges generated by life sciences research; the challenges in implementation and adoption of solutions; and the mechanisms of enhancing innovation in developing countries with the potential for improving local and global health and local economies.

This presentation will focus on our studies of genomics initiatives in India, Thailand, South Africa and Mexico. It will also highlight recent initiatives in other parts of the world, including other parts of sub-Saharan Africa and the Middle East.

### **Reference Articles:**

Béatrice Séguin, Billie-Jo Hardy, Peter A. Singer and Abdallah S. Daar (2008) Genomic medicine and developing countries: creating a room of their own. *Nature Reviews Genetics* Vol 9 June 2008 487-493

Billie-Jo Hardy, Béatrice Séguin, Federico Goodsaid, Gerardo Jimenez-Sanchez, Peter A.Singer, and Abdullar S. Daar (2008) The next steps for genomic medicine: challenges and opportunities for the developing world *Nature Reviews Genetics*, October 2008, S23-27

## **« Open access » aspects of DNA patenting**

### **Philippe DUCOR**

Patents are generally viewed as the opposite approach to open access. Indeed, because they provide exclusivity patents are valued in industries where developing a new product is costly and difficult, and copying it once developed is comparatively easy. The pharmaceutical/biotechnology industry is notorious for its appetite for all forms of exclusivity, including patents.

However, patents are not only about exclusivity. The underlying policy goal of the patent system is to strike a fine balance between (i) preserving the incentive of innovators (by providing exclusivity) and (ii) maximising the use of the relevant technology within society (notably through exclusivity limitations and restrictive patentability requirements). The “fine balance” is particularly difficult to find for DNA patents, due notably to the dual nature – structural and informational – of DNA and the finite number of naturally-occurring human genes.

As a result of the ongoing debate about DNA patenting and the European directive 98/44/EC, a number of European countries have implemented statutory provisions aiming to limit the exclusivity afforded by DNA patents. Although not a member of the EU (and not formally held by directive 98/44/EC), Switzerland has amended its patent law on several points relevant to DNA patents, such as: scope of DNA product patents (art. 8b and 8c Swiss Patent Statute =SPS), experimental use exemption (art. 9 al. 1 litt. b SPS), compulsory licensing for research tools (art. 40b SPS), compulsory licensing in case of anti-competitive behavior related to genetic diagnostics (art. 40c SPS).

In addition to exclusivity limitations, well understood patentability requirements may also play a role: not everything is patentable. In particular, realisations considered patentable a few years ago may be considered obvious and unpatentable today, notably in rapidly evolving fields such as genomics. In this respect, the recent reversal of US case law regarding obviousness of human DNA over the corresponding amino acid sequence is particularly relevant (In re Kubin, Fed. Cir. 2009).

#### **Reference Articles:**

Joseph Straus, An Updating Concerning the Protection of Biotechnological Inventions Including the Scope of Patents for Genes - An Academic Point of View OJ EPO Special Issue 2003, pp. 166-189 [http://archive.epo.org/epo/pubs/oj003/07\\_03/se2\\_07\\_03.pdf](http://archive.epo.org/epo/pubs/oj003/07_03/se2_07_03.pdf)

United States Court of Appeals for the Federal Circuit, In re Marek Z. Kubin And Raymond G. Goodwin, 2008-1184 (Serial No. 09/667,859), decided: April 3, 2009 <http://www.ca9.uscourts.gov/opinions/08-1184.pdf>

## **Genomic Medicine in Mexico: Progress and challenges related to equity and justice.**

### **Gerardo JIMENEZ-SANCHEZ**

Mexico faces important demographic and epidemiological transitions with significant implications to patterns of disease, disability, and death. On the one hand, there are problems of underdevelopment and, on the other, the emerging challenges of the chronic and degenerative diseases of the industrialised world. For these diseases, prevention becomes a key strategy for alleviating a major burden to the economy and health of the population. Genomic medicine has become a priority to the Mexican government as a means of finding new strategies to tackle common diseases (Jimenez-Sanchez G. *Science* 2003. 300(5617): 295). In 2000, strategic planning for genomic medicine began, from a feasibility study and a multi-institutional consortium effort, to the creation of a National Institute of Genomic Medicine (INMEGEN) by the Mexican Congress in 2004 (Séguin B, *Nat Rev Genet.* 2008 Oct;9 *Suppl* 1:S5).

INMEGEN is designed to develop world-class translational research focused on national health problems. Most Mexicans are Mestizos resulting from admixture of Amerindian, Spaniard and African populations. The admixture process has led to particular genomic ancestry structure. To optimise the use of human genome information to improve healthcare in Mexicans, we are systematically evaluating genomic variability of the Mexican population (Silva-Zolezzi I. *PNAS* 2009). We are including additional Mexican Amerindians and increasing SNP density to better understand the admixture process in Mexicans, and develop more suitable tools to analyse the genetic bases of complex diseases in this population (<http://diversity.inmegen.gob.mx>). In addition to the construction of a haplotype map of the Mexican population, INMEGEN is developing several genome-wide association studies for common diseases, such as diabetes, obesity, cardiovascular disease, and cancer, as well as other translational medicine projects that include biomarkers discovery for several kinds of cancer, pharmacogenomics, and nutrigenomics (Jimenez-Sanchez, G. *Genome Res.* 2008; 18 (8): 1191-1198). The design of INMEGEN includes an intellectual property unit and a business incubator to develop goods and services to improve healthcare for the Mexican population. These projects have included a strong component of community participation, resulting into local pride and support of the idea of individualised medicine ([www.inmegen.gob.mx](http://www.inmegen.gob.mx)).

Special attention has been devoted to ethical, legal and social implications (ELSI) related to developing genomic medicine in Mexico, including the development of an ELSI Research Center that has developed a comprehensive strategy for community engagement into research projects, including personal consent for sample collection. In addition, INMEGEN has a permanent interaction with the Mexican Congress to advise on emerging needs and opportunities for legislation on issues related to fair access and privacy, among many other issues. Soon INMEGEN will begin offering genomic services related to pharmacogenomics, for this a six-level rate strategy has been established based on individual financial capacity with a different degree of governmental subsidy to ensure equal access to genomic services.

This efforts have created knowledge and culture about genomic medicine in Mexico leading develop a more modern regulatory framework to ensure that genomic medicine successfully contributes to improve healthcare in the Mexican population (Hardy BJ, *Nat Rev Genet.* 2008 Oct;9 *Suppl* 1:S23).

**Reference Articles:**

Jimenez-Sanchez G, Silva-Zolezzi I, Hidalgo A, March S. Genomic medicine in Mexico: initial steps and the road ahead. *Genome Res.* 2008 Aug; 18(8):1191-8

Billie-Jo Hardy, Béatrice Séguin, Federico Goodsaid, Gerardo Jimenez-Sanchez, Peter A.Singer, and Abdullar S. Daar (2008) The next steps for genomic medicine: challenges and opportunities for the developing world *Nature Reviews Genetics*, October 2008, *Suppl* 1:S23-7

Béatrice Séguin, Billie-Jo Hardy, Peter A. Singer and Abdullar S. Daar Genomics, public health and developing countries: the case of the Mexican National Institute of Genomic Medicine (INMEGEN). *Nat Rev Genet.* 2008 Oct;9 *Suppl* 1:S5-9

Silva-Zolezzi I, Hidalgo-Miranda A, Estrada-Gil J, Fernandez-Lopez JC, Uribe-Figueroa L, Contreras A, Balam-Ortiz E, del Bosque-Plata L, Velazquez-Fernandez D, Lara C, Goya R, Hernandez-Lemus E, Davila C, Barrientos E, March S, Jimenez-Sanchez G. Analysis of genomic diversity in Mexican Mestizo populations to develop genomic medicine in Mexico. *Proc Natl Acad Sci U S A.* 2009 May 26;106(21):8611-6 Epub 2009 May11.

## ***Personal Genomics and Privacy***

### **Bartha-Maria KNOPPERS**

Will the era of open access databases and personal genomics via the internet spell the end of traditional concepts of privacy? Privacy in the context of health information is translated and protected under medical confidentiality if within the confines of a physician-patient or researcher-participant relationship. Both longitudinal databases and "virtual" personal genomics strain the confines of deontological and legal parameters of confidentiality. What are the convergent and divergent elements of privacy in these new contexts? And, what are the mechanisms to protect privacy, however defined?

## ***Future of Health Care Industry – Strategies for Personalised Medicine***

**Klaus LINDPAINTNER**

Throughout the course of its history, medical progress has always been driven by a more differentiated understanding of disease biology. Recent advances in science and technology have delivered to us new tools, allowing more fundamental and mechanistic insights into cell biology. This allows us now to also reach a more refined understanding of inter-individual differences in drug response on two levels. On one, such differences are independent of particular disease states, but are related to individual characteristics that affect the body's processing of xenobiotics. On the other, these differences are intimately related to disease biology, leading us to what amounts to a "rewriting of the textbook of medicine" based on molecular understanding. Both, in due course, may then allow a shift from today's largely empiric and palliative to a causally targeted and pharmacopoeia, where choice and regimen of administration of a medicine can be tailored in a much more differentiated fashion, providing more effective and safer treatment options. Ultimately, the associated expense to health care systems will need to be justified by demonstration of appropriate clinical utility, in the sense of incremental cost-efficacy. This will require a systematic, staged, and case-specifically adjusted approach for optimal balance of risk-benefit, and sustainable success of the concept.

## ***Genomic Sequencing: The Potential Impact of the Technologies on Science and Society***

**Edison T. LIU**

The success of the Human Genome Project, completed in 2003, was primarily due to the development of high throughput sequencing approaches and advanced computational capabilities. Earlier sequencing approaches relied on primer extension and fluorescent dye termination using DNA polymerase and specific nucleotides terminators (also called Sanger sequencing). The subsequent terminated fragments were then separated by capillary electrophoresis and the position of the specific nucleotide terminator deduced from the fragment sizes. The completed version of The Human Genome Project had less than 400 gaps covering 99% of the genome with an accuracy of more than 99.99% .

In the last few years, a dramatic change in sequencing technologies have allowed for improvements by five orders of magnitude in speed and reduction in cost. These "second generation" technologies have now superseded Sanger-based capillary electrophoresis sequencing and are the basis for the generation of data for genome-to-systems investigations. The fundamental shift that distinguishes this second generation sequencing is the reliance on reading the DNA code by assessing the incorporation of each individual complementary nucleotide – sequencing-by-"synthesis" (as compared with sequencing by fragment length). Alternatively, sequencing-by-hybridization is used whereupon precise sequences are deduced by specific hybridization of oligonucleotide probes. Secondly, this sequencing-by-synthesis is augmented in scale by arraying each sequencing reaction in a massively parallel fashion. What then that is limited is the length of sequencing. Until recently, the sequencing lengths have been limited from 25-250 base pairs. But here, the computational algorithms for sequence assembly allows for the "stitching" of these fragmented sequences into contiguous sequences – called "contigs".

The advance of these sequencing technologies has allowed the timely and cost-effective sequencing of whole genomes including human genomes. Several projects such as the 1000 Genomes Project seeks to assess the complete catalogue of genetic variation in humans through whole genome sequencing. The availability of genome sequences will change the face of biomedical research and of medicine. Individual genomes that do not change in life can be assessed at one time. Therefore one's complete genetic profile is accessible potentially as a clinical test. As with other technologies, genome sequencing of this magnitude raises clear legal and social challenges. In the same way that personal computers and the internet has changed our social activities and redefined privacy, the same will be true of personal genomic information.

### **Reference Articles:**

Turner DJ, Keane TM, Sudbery I, Adams DJ. Next-generation sequencing of vertebrate experimental organisms. *Mamm Genome*. 2009 Jun;20(6):327-38. Epub 2009 May 19. PubMed PMID: 19452216; PubMed Central PMCID: PMC2714443.

Ansorge WJ. Next-generation DNA sequencing techniques. *N Biotechnol*. 2009 Apr;25(4):195-203. Epub 2009 Feb 3. Review. PubMed PMID: 19429539.

Maclean D, Jones JD, Studholme DJ. Application of 'next-generation' sequencing technologies to microbial genetics. *Nat Rev Microbiol.* 2009 Apr;7(4):287-96. Review. PubMed PMID: 19287448.

Fullwood MJ, Wei ei, Liu ET, Ruan Y. Next-generation DNA sequencing of paired-end tags (PET) for transcriptome and genome analyses. *Genome Res.* 2009 Apr;19(4):521-32. Review. PubMed PMID: 19339662.

## ***Redefining privacy: public genomes and open consent***

**Jeantine LUNSHOF**

We are used to defining privacy in terms of something that is to be protected: privacy of individuals as the object of concern, in need of strategies to protect it from harm from outside. For some time, we have been holding the belief that 'genetic privacy' is an exceptional case deserving a particularly high degree of protection.

Recently, individual human genetics has been acquiring an extension in the form of personal genomes and in the Personal Genome Project (PGP) these were designed as public genomes. Public genomes imply a redefining of privacy: privacy as shaped by the individual's decision about information sharing and not by the well-intended concerns of others. In the PGP, open consent documents the decision to publicly share personal information with the awareness that anonymity does not exist. The accessibility and the sharing of personal genome and phenome data in the public playground of internet require a rethinking of normative concepts and long-held beliefs.

### **Reference Articles:**

Lunshof JE, Chadwick R, Vorhaus DB, Church GM. From genetic privacy to open consent. *Nature Rev Genet* 2008;9:406-411

Prainsack B, Reardon J, Hindmarsh R, Gottweis H, Naue U and Lunshof JE. Misdirected precaution. *Nature*, 2008;456:34-35

## ***Ethical Dilemmas in the Conduct of Genomic Research: When Technological Developments Encounter Economic and Educational Underdevelopment***

**Partha MAJUMDAR**

The phenomenal technological developments in the field of genomics, including whole-genome sequencing, has empowered researchers to undertake studies on humans that have important implications for health and disease. These technologies are being increasingly used in the developing world, where there is lack of commensurate economic and educational development. The data being generated by these technologies and the modalities of such data-generation through sample-sharing are enhancing the complexities of implementation of even the most basic ethical tenet – that of “informed consent” – in societies with low levels of literacy and with poor access to financial resources. I shall argue that under such circumstances, individual freedom (that is, freedom from intrusive interference by others, including the state) may be better guaranteed through societal and other governance arrangements in the conduct of genomic research. The safeguard of privacy and confidentiality, hallmarks of the quintessential ‘Western’ idea of justice and democracy, may have to be relaxed through public reasoning and engagement in order to better ensure individual freedom in the context of genomic research in a developing country.

## ***What will the new sequencing technologies deliver for science and society?***

**Mark McCARTHY**

The advent of genome wide association analysis has led to identification of many hundreds of loci influencing common diseases and traits, and many insights into disease pathogenesis. These advances have whetted the collective appetite for large-scale genetic discovery efforts, and seen the first attempts to deliver personalised genomic information. Second- and third-wave sequencing technologies promise much more of this, and it is no longer science fiction to imagine individual genome sequencing as a standard research and medical tool in the next decade or so. However, such fanciful visions have to be tempered against the current reality: as of 2009, whole genome sequence data remain expensive to obtain, lack sufficient accuracy for many purposes, and pose fearful analytical challenges. If we are to get from "here" to "there", a large number of obstacles (logistical, technical, analytical, financial, ethical, social) have to be addressed. My own research group is facing these challenges as we migrate from the relative security of genotypes to the brave new world of high-throughput sequence, and I will summarise some of our (and others') efforts in this respect.

## ***Genetic Exceptionalism: A Reassessment***

**Thomas H. MURRAY**

Genetic Exceptionalism expresses a conviction: that of all types of health related information about persons, genetic information requires special treatment and special protection. The alleged sensitivity of genetic information rests on a set of assumptions, among them:

- that genetic information is highly predictive for a person's life course (the "genetic diary" idea)
- that genetic information could be readily distinguished from other sorts of health related information both ethically and practically
- that other parties were likely to gain access to genetic information about a person that the individual himself or herself would not be able to obtain, and finally,
- that this information could and would be used adversely against the individual.

The assumptions made by genetic exception were first challenged in the 1990s. In particular, that analysis questioned the claims made about the predictive power of genetic information, and about the distinctiveness of genetic information among all types of health related information about persons. The recent rise of "recreational genomics" and the prospect of inexpensive mass sequencing in the near future may restore some balance in access to genetic information about individuals. The possibility that others may use genetic information against the interests of individuals has been mitigated by legislation such as GINA in the USA. But the fundamental claim made by genetic exceptionalism, that genetic information should be given special status and special protection, has not been successfully defended. We are better off if genetic information is treated as just another type of health related information about a person.

## ***Privacy, technology, policy, and the integrity of social life***

### **Helen NISSENBAUM**

My talk presents an overview of contextual integrity, an account of privacy that calls for the appropriate flow of personal information. This account contrasts with conceptions of privacy that present it as a right to shut off access to information (secrecy), or a right to control personal information. These conceptions, which frequently inform law, regulation, and technology design, are problematic because they constantly require privacy to be traded off against some other social good, such as security, efficiency, or free speech. But they also miss the complexity most people recognise and accept in the ways they share information and allow information to be shared with other individuals and institutions. Contextual integrity accommodates this complexity, modeling it with informational norms, which govern the flow of information. These norms vary as a function of the social context, the capacities in which senders and recipients act, the types of information in question, and the principles under which information flows from party to party. Personal information plays a crucial role in well-functioning societies, but its flow must be appropriately constrained not only for the benefit of individuals but for the integrity of social life itself.

#### **Reference URLs:**

<http://www.sup.org/book.cgi?id=8862>

<http://www.nyu.edu/projects/nissenbaum>

## ***Advances in breast cancer: pathways to personalised medicine.***

### **Olufunmilayo OLOPADE**

With an increasing global burden of breast cancer, cancer control efforts in the post-genome era should be focused at developing novel risk assessment and treatment strategies that will further reduce the morbidity and mortality associated with the disease. The discovery that mutations in the *BRCA1* and *BRCA2* genes increase the risk of breast and ovarian cancers has radically transformed our understanding of the genetic basis of breast cancer, leading to improved management of high-risk women. The immediate challenge is to learn how to use the molecular characteristics of an individual and their tumor to improve detection and treatment, and ultimately to prevent the development of breast cancer, not only in developed countries, but throughout the world.

#### **Reference Articles / URL:**

Breast cancer risk associated with BRCA 1 and BRCA2 in diverse populations. Nature Reviews Volume 7 December 2007

Population Differences in Breast Cancer: Survey in Indigenous African Women Reveals Over-Representation of Triple-Negative Breast Cancer. Olufunmilayo I. Olopade et al. *Journal of Clinical Oncology*, Vol 27, No 27 (September 20), 2009: pp. 4515-4521  
URL : <http://jco.ascopubs.org/cgi/doi/10.1200/JCO.2008.19.6873>

## ***Parent of Origin Effect: Will Africa be Relegated to the Role of a Neglected Parent Watching its Successful Children Bicker Over Their Inheritance***

**Raj S. RAMESAR**

Emerging countries are at very disparate stages of engaging with the products of the Human Genome Project. In the East, and in some parts of Latin America (notably Mexico and Brazil), there has been a ready engagement with genomics and other high-throughput technologies in an unprecedented manner. This is very likely the result of an active engagement of academic institutions with support from government. The involvement of government and academia is crucial in reaching threshold momentum for ready uptake in education and training, and for engaging with disciplines beyond just human genetics and health research, to e.g. food security and agriculture. I refer to this as a scaffolded approach, where a participatory or supportive government may further engage other facets of the state, e.g. Trade and Industry, Science and Technology, Agriculture and Health to engage with academia in long term strategic initiatives aimed at enhancing any state's development.

In Africa, a wide range of genetic studies have been undertaken (Sirugo et al., 2008), and several large-scale genome-based projects are currently underway, mostly marshaled from outside of the continent, and which, as part of their original design, set out to contribute significantly to training and capacity development. There is little evidence however, that the instruments of capacity development are sustainable beyond the actual focused interest of foreign researchers. This is because African governments, institutions and researchers have not adequately engaged with one another about the strategic importance of genomics, and have not capitalised on the foreign investment. More recently, with the establishment and growth of the African Society for Human Genetics, work is currently underway to facilitate intra- and inter-national cooperative projects which aim to harness the opportunities offered through the diverse indigenous African populations (Hardy et al., 2008).

With the above background, the debate about which African genomes will be sequenced soonest, and how to engage with whole genome data, at a society level, is largely academic. There has been great interest in studying the genomes of Africans, largely to understand human origins and diversity, not because this has been requested by Africans. There is little evidence to suggest that whole genome sequencing is going to be beneficial to individuals in the short term, particularly without adequate interpretive skills available. It is likely, that studies involving African subjects will be carried out, on and off the continent. However, and philosophically, very few Africans have ever been considered as more than research subjects, they seldom asked to be involved, and have not engaged in the net benefit that the rest of humanity and the respective researchers have derived, and will derive. The emergence of the 'whole genome sequence' era is a good time to reassess our reasoning to involve samplings of all identifiable groups of humanity in research, without a commitment to engage with relevant governmental and academic institutions, to bring to people, albeit via a very long and hard road, the benefits of the whole genome sequence, such as these might be.

### Reference Articles:

Hardy B-J, Séguin B, Ramesar R S, Singer P A, Daar A S. (2008). South Africa: from species cradle to genomic applications *Nature Reviews Genetics* Suppl 1:S19-23

Sirugo G, Hennig BJ, Adeyemo AA, Matimba A, Newport MJ, Ibrahim ME, Ryckman KK, Tacconelli A, Mariani-Costantini R, Novelli G, Soodyall H, Rotimi CN, Ramesar RS, Tishkoff SA, Williams SM. (2008). Genetic studies of African populations: an overview on disease susceptibility and response to vaccines and therapeutics. *Human Genetics*. 123(6):557-98

## **ABSTRACTS - POSTERS**

**Poster No : PP-01** (*Poster Presentation on 1 November 2009*)

### ***Genetic Counselors in the Era of Full Genomic Sequencing: What are their Legal Duties?***

**Ma'n Hilmi ABDUL-RAHMAN**

With the accessibility of rapid, inexpensive and complete genomic sequencing, individuals are expected to make lifestyle changes based on information about their genetic risk. Professional guidelines maintain that appropriate counseling needs to be put in place to help them. Primary care physicians are not always aware of the new developments in the field of genetics, and referrals to geneticists sometimes incur long delays. Hence, genetic counselors are increasingly expected to provide individuals and families with information on genetic risk in order to allow them to make informed decision-making.

In hospitals, genetic counselors usually operate within a multidisciplinary team and while the physicians and nurses working with them are fully recognised as professionals, genetic counselors in many countries have yet to be acknowledged by laws regulating professions. This legal uncertainty has enormous implications regarding the obligations of genetic counselors towards the individuals who seek their counsel and their families, especially on topics such as privacy. In the context of complete genomic sequencing, genetic counselors will face new challenges in several respects: the extent and the approaches taken for information-giving during consultations; the legal relationship between them and the individuals who may not yet be “patients”; and the protection of confidentiality.

Using Quebec’s civil law as a case-study, a comparative analysis of laws and regulations framing the physicians’ and the genetic counselors’ responsibilities will be undertaken. Although bearing in mind the exclusivity of medical practice, this analysis will set the context for a discussion of the issues shaping the genetic counselor’s duties towards individuals/patients in the context of complete genomic sequencing by providing innovative solutions emanating from the regulatory landscape governing physicians.

**Poster No : PP-02** (*Poster Presentation on 2 November 2009*)

## ***Bioethics law in revision: what challenges for genetic testing regulation?***

**Gabrielle BERTIER**, E Rial-Sebbag, S Julia, P Ducournau, A Cambon-Thomsen

Since 1994 in France, a bioethics law regulates genetic testing. It was revised in 2004 and a new revision is planned for 2010. Debates and consultations at various institutional, professional and social levels are underway.

In this paper we describe 1. the principles underpinning the law, the present legal regulation and organisation of genetic testing, 2. the points of debate, 3. factors that hamper efficient policy making in this domain.

The law restricts the production and use of genetic information to medical, scientific and judiciary domains. The profession of 'genetic counselor' was created in 2004. The main legal principles involved were separately identified in 2004. They include respect for human dignity, for privacy, for medical confidentiality, and right to access to one's origin. The balance between these fundamental principles is fiercely shaken by the developments of genetic technologies, and their prompt spreading throughout all social spheres.

Five reports from national public bodies<sup>2</sup>, and a general public consultation were used as sources for this analysis.

In the medical context, the debates focus primarily on modalities of disclosure of information in families, and on the conditions of validation of genetic tests, and the extension of their scope of use in procreation technologies and multifactorial diseases. Interestingly, the large scale production of genetic information was not a prominent feature in the general public debate.

In the scientific context, secondary uses of samples and data and the power of identification of genetic information deriving from them are the main topics discussed.

The use of genetic identification technologies by the judiciary system –family gathering in immigration policies, and postmortem genetic identification–, is also an issue in the discussion. Direct to consumer (DTC) genetic tests are discussed separately, as their regulation has a national and an international level. Indeed, their use both in and out of the medical issue raises many social and ethical questions. Besides, the ability of a national law to regulate such a global phenomenon is also identified as a main challenge. Even if opinions differ on the matter, all bodies insist on the importance to regulate the quality of the tests, and to provide validated information.

Proposals sometimes differ significantly among the bodies consulted. For instance, about family disclosure of genetic information, if the citizens consulted in the 'Etats Généraux' view medical confidentiality as imperative, both the 'Conseil d'Etat' and the National Biomedical Agency propose simplifications of procedures of disclosure in specific cases, and discuss the patient's responsibility.

This analysis showed that a number of questions remained mainly discussed in professional arenas, such as the scope of action of genetic counselors, whereas others, like that of the availability of genetic tests on the internet raised interest among all actors of the debate. However, empirical data are still lacking in this domain in France.

**Poster No : PP-03** (*Poster Presentation on 3 November 2009*)

### ***Launching of the Swiss National Task Force on Public Health Genomics***

**Murielle BOCHUD**, Deborah Bartholdi, Lazare Benaroyo, Thomas Binz, Angela Brand, Ivan Curjuric, Florence Fellmann, Martin Götz, Idris Guessous, Medea Imboden, Nicole Probst, Tobias Schulte in den Baumen, on behalf of the Swiss National Task Force on Public Health Genomics.

Rapid advances in genomics and molecular biology research have occurred during the last decades. Genomics is rapidly changing our understanding of health and disease, including complex diseases (e.g. diabetes, obesity, cancer, dyslipidemia) and the pathways involved. Healthcare systems and the society will need to adapt to these novel findings and to develop personalised healthcare. Public health genomics (PHG) aims to provide responsible and effective integration of genome-based knowledge and technologies into public policy and into health services for the benefit of population health (<http://www.phgen.nrw.de>). In 2005, the General Directorate for Health and Consumer Protection of the European Commission (DG SANCO) decided to fund the Public Health Genomics European Network (PHGEN). Members of this network advise the European Commission on health policies and on the integration of genome-based knowledge within the 'health in all policies' doctrine. As public health needs to act locally, PHGEN has established National Task Forces (NTF) on PGH in most European Commission Member States. The NTFs are in Europe the platform to establish national infrastructures and to guarantee sustainability. Comparative research between the different European countries can be done and best practices can be developed.

On August 28 2009, a panel of experts launched the Swiss NTF on Public Health Genomics at the University of Irchel in Zürich. There were 22 participants from various backgrounds and institutions: genetics, public health, epidemiology, ethics, law, public administration, GUMEK representatives, sociology and biostatistics. The event was supported by the Federal Office of Public Health, the Swiss School of Public Health Plus, the University Institutes of Social and Preventive Medicine in Lausanne and Zürich.

The points of view of geneticists, ethicists and public health specialists have been discussed and contrasted. Several issues have been raised during the meeting, among which the need to increase genomic literacy of lay people and health care professionals, the need to participate in the establishment of a general legal framework for genomic-based research, the need to encourage research on ethical and social issues in genetics, genomics and epidemiology. The precise structure of the Swiss NTF is to be determined in the coming months. Participants have been invited to submit a list of topics that they consider as relevant. The next meeting is planned in 2-3 months with the aim to identify priorities of the Swiss NTF and guide future actions of the NTF.

**Poster No : PP-04** (*Poster Presentation on 1 November 2009*)

***Securing Stable Participation in Biobanking by Safeguarding Subjects' Collective Altruism***

**Kuan-Hsun CHEN**

This paper aims to demonstrate the necessity of respect for subjects' collective altruistic expectations in the context of population-based biobanking and argues that scientists benefiting from subjects' donations should make a proportional effort to meet such altruistic expectations. It focuses on the case of Taiwan Biobank and analyses the present friendly public attitude toward the biobanking with current survey findings. It dissects the public's altruism and employs games to deduce how subjects will react if their collective expectations are not to be met. The analysis finds that it is not proper to treat or assume volunteers as pure altruists. They do anticipate rewards from their participation, but they anticipate rather the society than themselves to be the recipient of the rewards. If their collective altruistic expectations are not to be realised, they will participate no longer. The collective altruistic expectations can considerably influence the stability of subjects' participation. Therefore, a normative mechanism to safeguard subjects' constant altruism is necessary. Besides, without such a normative mechanism, the costs to recover the lost collective altruism will be shifted onto the later scientists, which is deeply unjust. It concludes that a legal framework to ensure scientists make proportional contribution to fulfill subjects' collective altruistic expectations is crucial for the biobank governance.

**Poster No : PP-05** (*Poster Presentation on 2 November 2009*)

***Key Metaphors of the Human Genome Project: Possibilities and Limitations***

**Myra CHENG**

This poster examines three critical metaphors emerging in genomic discourse over the course of the Human Genome Project (HGP). These metaphors include the human genome as significant text, computer software and the foundation of future scientific research in the life science. Since the commencement of the HGP, the human genome has been conceptualised as the 'Book of Life' and 'Book of Nature' (Nerlich and Hellsten, 2004). A variation of this textual metaphor is the reference to the genome as a computer program consisting of digitised biological information. Popular science writers have asserted that such ideas are not merely metaphors but rather the literal or 'plain truth' (Dawkins, 1986; Ridley, 1999). By contrast, other commentators have questioned the accuracy these claims (Marr, 2000; Venter, 2001). Notably, the informational metaphors were superseded by alternative constructions following the disappointing outcome of the HGP. For example, in their Nature article, Francis Collins and colleagues described the HGP as 'the firm bedrock foundation' upon which 'the true promise of genomic research for benefiting mankind can be realised' (Collins, 2003: 284). This poster explores the potential and shortcomings of contemporary scientific visions through an analysis of key genomic metaphors.

**Poster No : PP-06** (Poster Presentation on 3 November 2009)

**Acceptability of an Opt-Out Electronic Medical Record (EMR) DNA Research Resource**

**Ellen Wright CLAYTON**, Kyle Brothers, Dan Morrison, Jill Pulley, Dan Masys

BioVU combines de-identified clinical information from electronic medical records of patients at Vanderbilt University Medical Center (VUMC) with DNA extracted from left-over clinical blood samples (>63,000 samples to date, increasing by ~2,000 per month). Although not required by US law, information about BioVU is provided to patients at clinic check-in and in phlebotomy areas, and patients can choose not to have their residual blood included. We have solicited feedback regarding BioVU from thousands of people in middle Tennessee over 10 years and report here the results of three recent studies:

1) Interviews of 77 adult patients seeking care at Vanderbilt clinics in summer 2009

Slightly more than half had heard of BioVU, citing the opt out forms (n=5), brochures (n=5), and posters (n=6) in the clinic, and other sources of information (n=19). Although only ~1/3 knew that residual samples could be used for research, almost 90% supported the idea of BioVU, generally citing the importance of research. Reasons offered by those opposing BioVU included not knowing how their DNA would be used, worries about anonymity, and religious beliefs. Only one interviewee subsequently opted out.

2) Web-based survey of ~4000 faculty and staff at Vanderbilt University in December 2008

(response rate ~20%)

Statements regarding DNA databanks	% agreeing or strongly agreeing
"DNA databanks are fine as long as all identifying information is removed"	93%
"DNA databanks with all identifying information removed are fine as long as people can choose to opt of having their DNA included"	95%
"DNA databanks with all identifying information removed are fine as long as written permission from patients is required for their DNA to be included"	89%
"DNA databanks with all identifying information removed are fine as long as an ethics review panel approved research with DNA in the databank"	92%

Employees who prefer not to get health care at VUMC were more likely to criticise non-consent options.

Opinions about data sharing – “How would sending deidentified genetic information to national database affect your willingness to participate?”	% responses
More willing to donate	19%
Less willing to donate	12%
Would not affect willingness to donate	70%

3) Interviews of 77 English speaking parents of children seeking care at Monroe Carell, Jr. Children’s Hospital at Vanderbilt (MCJCHV).

These interviews were conducted in general pediatric and subspecialty clinics as part of assessing the acceptability of extending BioVU to children. 80% of respondents were women.

	% yes
Support medical research generally	99%
Support medical research in children	86%
Would permit own child to participate in minimal risk research	83%
Would permit own child’s information to be in BioVU	91%

In response to open-ended questions about possible benefits of BioVU, parents volunteered helping others (73%) and not throwing blood away (15%). The majority identified no concerns about BioVU, even when probed specifically (10% worried about privacy of genetic information, 21% denied this concern). Only 2 parents criticised opt-out process.

Conclusion. In three different populations, ≥ 90% respondents view an opt-out EMR DNA research resource to be acceptable, findings consistent with our previous studies.

**Poster No : PP-07** (*Poster Presentation on 1 November 2009*)

## ***Attitudes and Risk Perception of Pregnant Women Toward Prenatal Diagnosis in the Philippines***

**Eva Maria CUTIONGCO-DE LA PAZ**

Prenatal diagnosis has been a vital part of reproductive risk screening. The goals of prenatal diagnosis include providing a range of informed choices to couples at risk of having a child with an abnormality; providing reassurance and reducing anxiety among high-risk groups; allowing couples to begin a pregnancy with a knowledge that the presence or absence of the disorder in the fetus can be confirmed by testing; allowing couples the option of appropriate management for the birth of a child with a genetic disorder; and enabling prenatal treatment of an affected fetus. It is important to recognize that prenatal diagnosis remains controversial, particularly when the diagnosis leads to a decision to abort the pregnancy. The ethical dilemma is in trying to balance respect for autonomy of parents' reproductive decision-making with an appraisal of whether aborting the unborn child affected with a disability compatible with life, is either fair or beneficial to disabled persons. Currently, prenatal diagnosis is practiced in a very limited way in the Philippines. The most widely used prenatal diagnostic procedure is that of prenatal ultrasonography which uses two-dimensional ultrasound for congenital anomaly screening during the second trimester of pregnancy. Maternal serum screen as a non-invasive form of prenatal diagnosis has not been routinely offered in the country. Amniocentesis has been done in exceptional circumstances to provide anticipatory care and guidance to obstetricians and pediatricians in preparation for the birth of a child affected with a genetic condition. Chorionic villi sampling has also not been routinely offered in any institution. Although cost may be one of the major issues in performing these invasive procedures of prenatal diagnosis, the more prevailing concern is that termination of pregnancy is not allowed by law in the country ( Article II, Section 12 of the 1987 Constitution of the Philippines). Despite these limitations in the practice of prenatal diagnosis, health professionals as well as patients must continue to be attentive of advances in both genetic research to make the most educated, well informed and ethically well-defined decisions possible. A survey was conducted among pregnant women in the prenatal clinics at a large referral center in the Philippines. The objective was to determine the knowledge and attitudes of pregnant Filipino women toward prenatal diagnosis and their perception of their risk for the possibility of the birth of a fetus affected with a genetic condition and to determine whether prenatal diagnosis would be a form of testing these pregnant women would avail of. The results of the study will be presented.

**Poster No : PP-08** (*Poster Presentation on 2 November 2009*)

### ***How To Deal With Whole Human Genome Sequences?***

**Julie GAUTHIER**, Béatrice Godard, Pierre Drapeau and Guy A. Rouleau

Genomic factors play an important role in neurological and psychiatric disorders such as Autism Spectrum Disorders (ASD), Schizophrenia (SCZ) and Mental Retardation (MR). The recent development of next-generation sequencing technology offers the ability to sequence an individual's complete genome. As part of our ongoing S2D project which aimed at the identification of disease causative genes for ASD, SCZ and MR by re-sequencing synaptic genes, our goal is now to use whole exome or whole genome sequencing technologies to identify such genes. Because we plan to re-sequence entire genomes, we will identify a very large number of variants, most of which will not be immediately interpretable. Coping with such information and finding ways to present the relevant results to the patients is a very challenging issue. As part of our S2D project we studied several questions related to ethical duties of researchers and physicians as well as the rights of patients and relatives. In this regard, we are investigating the ethical, legal, and social implications of different issues such as: 1) A duty to communicate research results? 2) Expanding the researcher/physician's duty of care: a duty to recontact? 3) Disclosure of genetic information to family members: a duty to warn? Our preliminary data based on "Parents' attitudes toward the return of research results" question, indicates that parents (97%) would like to receive information, whether for general findings of the research project or positive individual finding or even negative individual finding . Related issues were also addressed, such as benefit and disadvantages of receiving information, Individuals charged with communicating findings, time, means and impact of communicating findings. Sequencing of individual genome in a clinical context, will not only allow early detection of specific diseases but will also provide information on the individual make-up for other health issue which may result in new therapies and hope for patients. However, considering the speed at which genetic technologies are integrated into clinical practice, the questions of whether researchers and physicians should inform their patients about new information that might be useful to them and whether physicians should inform family members of genetic information regarding the family are becoming increasingly important.

***Genetic discrimination: principles and policies***

**Alison HALL**, Caroline Wright, Stephen John and Alison Stewart

Over the last decade, our acquisition of genetic knowledge has gathered pace. The sequencing of whole human genomes, together with increasing accessibility of genomic information, has revolutionised the field. However, this knowledge has challenged existing conceptions of what the term 'genetic' means, particularly with respect to its ability to convey information that is more sensitive or predictive than other types of health-related data. These developments suggest that 'genetic' and 'genomic' factors are not fundamentally different from other types of biomarker and that the term 'genetic' is merely a convenient label to indicate a type of material (i.e.DNA) or category of information.

Within this context, it is all the more puzzling that 'genetic exceptionalism' – the belief that there is something special about genetic information – persists across many of the relevant professional disciplines as well as the general public. Moreover the principle of genetic exceptionalism seems to underpin emerging legislation aimed at preventing 'genetic discrimination'.

The aim of our project is to use a cluster of related research questions around a central question "Can the concept of 'genetic discrimination' do any useful work" to critically evaluate this area. We consider that a structured, comprehensive and analytic approach to this topic is needed in order to provide a rational basis for policy development .

The research questions are as follows:

1. *What do we mean by 'genetic' information/ material and why might it require 'exceptional' treatment?*  
This element of the project will examine the basis for genetic exceptionalism, particularly with regard to the predictive power of genetic information. It will include analysis of the use of genetic information in health and other contexts (e.g. forensics).
2. *What is the philosophical basis for the concept of 'genetic discrimination'?*  
We will examine the philosophical credibility of the concept of genetic discrimination , asking in what normative sense (epistemic or ethical, or both) and in what contexts it might be considered to be 'wrong'.
3. *What useful work do groups calling for 'genetic discrimination' legislation think that it would or could do?*  
This includes a critical analysis of the motivations for legislation, including arguments adduced, underpinning assumptions and relevant contextual issues such as social and political history and public opinion . Our focus will be primarily upon legislation which seeks to prevent genetic discrimination in the context of employment and insurance.

4. *What legislation currently exists specifically aimed at preventing 'genetic discrimination'?*

This element will explore existing legislation across different jurisdictions, its definitions, scope, remit and sanctions, citing relevant legal cases that have prompted such legislation.

5. *Could legislation prohibiting 'genetic discrimination' achieve desirable social and political outcomes?*

Other types of equality legislation – such as that prohibiting racial or disability discrimination – also suffer from definitional problems but are nevertheless thought to 'do useful work'. Through a comparative analysis we will ask whether legislation seeking to prevent 'genetic discrimination' is uniquely problematic or could similarly benefit society.

Finally, we will make a series of policy recommendations relating to 'genetic discrimination' legislation within the UK.

**Poster No : PP-10** (*Poster Presentation on 2 November 2009*)

***Large-Scale Genotyping Projects in the Developing World: Genomic Sovereignty***

**Billie-Jo HARDY, Béatrice Séguin, Peter A. Singer, Abdallah S. Daar**

In recent years, governments in Mexico, India, South Africa and Thailand have both funded or proposed to fund large-scale human genotyping initiatives. These countries provide compelling reasons for pursuing these studies: the potential to address local health needs and reduce health care costs; the opportunity to stimulate economic development through investments in genomic sciences, and the availability of unique population resources. In an effort to capture the value of these investments and promote an equal stake in international collaborations, some of these countries have developed guidelines and laws to protect local human genomic material as a sovereign resource. Critics suggest that this approach will impede international collaborations – reducing access to external funding which can be vital in these countries. We are conducting an in-depth study of the theme, genomic sovereignty, through an exploration of the empirical and theoretical understandings of the concept. We focus on how it may contribute to each country's aim of achieving health equity through investments in genomics, its relation to heritage, pride and patrimony, and its potential limitations. The debate is an important one as the knowledge generated from these large scale studies may need to be interpreted in larger international collaborative efforts before it can lead to health benefits. Qualitative case study methodology is being employed and the primary data source consists of interviews conducted with key informants. Ultimately we hope this research will result in a resource that can be built on by other groups studying how infrastructure and capacity are being established around genotyping initiatives in emerging and developing countries.

**Poster No : PP-11** (*Poster Presentation on 1 November 2009*)

***Managing the nebulous distinction between genetic research and clinical genetics: A triage tool to assist Research Ethics Boards (REBs)***

**Kathy HODGKINSON**, Daryl Pullman

This paper discusses the often nebulous distinction between genetic research and clinical genetics practice, and develops a triage tool to assist Research Ethics Boards (REBs) in determining how best to evaluate genetic research protocols. This tool is designed to assist REBs to assess when a genetic study should be treated much like a standard clinical trial in which research results should NOT be treated as clinically significant for individual patients, from studies in which a research result should be treated like a clinical test that MUST be shared with the patient (and at risk family members) in order to initiate appropriate clinical interventions. In the latter cases initial REB approval of a genetic research protocol may be contingent on the assurance of the availability of appropriate clinical follow-up.

It is standard practice in clinical trials research to keep research results distinct from clinical practice. This is because clinical trial information generally requires large samples and becomes relevant only when enough data has been accrued to attain statistical significance. Although large samples sizes are necessary in population based gene association studies, in many genetic studies results from a small sample—a single family, or even a single individual—can have immediate clinical implications. The distinction, therefore, between research results and clinical application, so important in the context of clinical trials, should not be used as the standard for genetic studies of serious diseases with high recurrence risks.

The key consideration here is that information obtained through genetic research often differs from results generated in the course of a standard clinical trial. The tool we have developed will assist REBs to triage genetic research protocols based on considerations of the severity of the disease, the recurrence risk to family members, the possibility of amelioration through treatment or screening, and the apparent gene penetrance. Using these criteria we have created a continuum that covers genetic research that cannot be separated from clinical care at the one extreme, and genetic research which should not be considered anything but research at the other. Various “grey area” types of projects are mapped between these two extremes.

In proposing this model we discuss the parallel issues of privacy and confidentiality when dealing with information that has relevance not only to the initial patient, but also their family members. Issues of duty to warn are also addressed. In so doing, we propose methods by which these issues should be handled by genetic researchers and clinicians, as well as research ethics boards.

**Poster No : PP-12** (*Poster Presentation on 2 November 2009*)

***The personal genomic and its cultural perspective in China***

**Ching-Li HU**, Chunmei Zhang

After the completion of the genomic sequencing and the HapMap project, the international HGP has entered a new period aiming at studying the genetic mechanisms of human diseases and other phenotypes. The new ethical issues personal genomic arise with the developing of sequencing technology which need to be discussed deeply.

In this paper ,we will address some ethical issues, such as how to deal with the huge biological samples with informed consent, how to interpret the relationship between scientific basic research and its commercial applying, how to think about genetic determinism, how to make ethical guidelines to prompt the new gene technology, etc. in a Chinese cultural perspective. All the ethical issues analysed will be based on the scientific works and ethical reflections of the Chinese National Human Genome at Shanghai. . It is emphasised that all the discussion on such ethical problems should be realised under the different cultural context, which will be benefit for the progress of personal genomic. On the one hand, our ethical analysis shall be followed with the international ethical principles; on the other hand, Chinese culture shall be absorbed so as to make our bioethical guidelines with Chinese characteristic features. The conclusion is that the purpose of cultural perspective on personal genomic is to make the harmonious relationship between technology and society, bioethics and new biology in China.

**Poster No : PP-13** (*Poster Presentation on 3 November 2009*)

## **Legal uncertainty in molecular genetic testing: more genes, more patents, more problems?**

**Isabelle HUYS**, Berthels, N., Matthijs, G., Van Overwalle, G.

The controversy on patenting and licensing of genes and diagnostic methods has lingered on over the last years. In an attempt to enrich the debate, we collected empirical data. We studied the patent landscape surrounding genetic diagnostic testing for 22 monogenic inherited disorders, most frequently tested in Europe, in order to assess (1) the nature, extent and scope of patents in the genetic diagnostic field in Europe and the US, (2) the patent ownership, and (3) the impact of the patents in view of the best practices in the field.

The results show significantly fewer claims on *genes per se* than initially suggested by others. Interestingly, most sequence claims are directed to cDNA. The few gene claims cause wide legal uncertainty as to their scope. Numerous *method* claims have been identified, tending to be most blocking. In both the genes and the method claims, the unclear character of claim language is most problematic.

We anticipate that the patent issue will become more dense and complicated when genetic testing will move from single-gene disorders to large scale genotyping for multifactorial diseases, and eventually to individual, whole-genome sequencing.

**Poster No : PP-14** (*Poster Presentation on 1 November 2009*)

***New Challenges for Ethics in the Era of Personal Genome Sequencing, Global Data Sharing and Web 2.0***

**Carol ISAACSON BARASH, Karin Gregory**

Direct to consumer genetic testing can be viewed as a strategic solution to consumer concerns about privacy violations and genetic discrimination risks. As such, the emergence of the \$1000 genome is not surprising and confirms a growing market. Personal genome sequencing is predicted to be both widely available in the imminent future, and highly desirable to individuals, newborn screening programs, possibly providers, and derivatively employers and payers. However, the confluence of large amounts of different types of individual data increasingly available in the growing numbers of bio-repositories, large scale international research collaborations, and web 2.0 formats, including but not limited to patient social networking sites, arguably raises unprecedented ethical, legal and policy challenges to consent, privacy, autonomy and the ability of principles of justice to serve as a basis for ensuring global benefit sharing.

While emerging molecular technologies would not exist were it not for the public's, knowingly and unknowingly, donated specimens, clinical benefits are likely to be differentially, and if not unfairly, distributed. Consent, privacy and autonomy face challenges given a host of possible scenarios. Findings subsequent to an original consent may be ones that an individual prefers to not know; yet they may be impacted nonetheless through scientific reporting. The right to not know on the part of future generations may be compromised by family member's decisions to know. A physician's possible duty to warn may be problematic in light of documented gaps in an ability to understand results and treat accordingly. How privacy laws will treat data shared on patient social networking sites or web mediated health care delivery remains uncertain. Cross cultural and ethical differences within large-scale databases compounds these complexities. Recent evidence that anonymized data can be relatively easily reidentified add to concerns about whether these fundamental principles can be appropriately, and proactively, protected. The public's low genetic literacy only exacerbates these concerns.

In a global context, ethically defensible strategies for balancing the competing interests of stakeholders, i.e. consumer's right to know, or not know, (and by implication family members and future generations), government's right to protect its citizens, and corporate interests, as well as responsibilities for assuring safe and effective products, pose significant policy considerations. It is important to distinguish scientific from ethical and legal issues so as to clearly frame the policy debate, particularly because policy decisions will have far reaching public health consequences, given the increasing global interconnectedness of research. This presentation provides an analysis of core ethics concepts and legal frameworks challenged by the confluence of these emerging science and technology advances and strategic policy suggestions for consideration by the global community.

**Poster No : PP-15** (*Poster Presentation on 2 November 2009*)

***Study of Ethical Aspects of Prenatal Testing for Neurodegenerative and Nervous-muscle Hereditary Diseases in Sakha (Yakutia).***

**Sardana KONONOVA**, Oksana Sidorova, Sardana Fedorova, Vera Izhevskaya, Elza Khusnutdinova

The use of prenatal diagnostics of late-onset autosomal dominant diseases is connected with problems of ethical character. In the report experience of prenatal diagnostics of two hereditary diseases most widespread in Yakutia: spinocerebellar ataxia I type (SCAI) and muscular dystrophy (MD) is discussed. Sakha (Yakutia) is one of Russia regions where SCAI and MD widely spread (frequency is 38,6: 100000 and 21,3:100000 accordingly). Patients: a total 38 pregnant women at risk for SCAI (n=26) and muscular dystrophy (n=12) who underwent genetic counseling. Data were collected from personal interview about sociodemographic and medical history, genetic knowledge, attitudes and motivations toward prenatal testing. All individuals were sakha nationality. Significantly more SCAI women had a higher university education than MD (76,9% and 33,3%,  $p < 0,05$ ). Also, pregnant women at risk for SCAI were more likely than individuals with MD to perceive that prenatal diagnostics would influence their life. Of SCAI group 14(53,8%) women requested prenatal testing then of MD group -10 (83,3%). More for SCAI risk pregnant women did not want prenatal testing than MD (12 (46,2%) and 2(16,2%),  $p < 0,05$ ). Results show a difference concerning pregnant women from families with SCAI and MD to prenatal testing and abortion with the revealed mutation fetus that demands different approaches to genetic counseling of pregnant women at these kinds of pathology. Series of reasons of low conversion of families with SCAI and MD are likely to be for carrying out the prenatal diagnostics. First, poorly-informed families about prenatal diagnostics, confirmed by questionnaire's data of the sakha rural population (52% don't know about existence of medical genetic service in Yakutia, 91% don't know about prenatal diagnostics as a method of genetic research). Second, difficulties of a money character, so as not every family has a possibility to get to Yakutsk for a medical-genetic counseling. Another possible reason is the ethnical features of sakha character: in interpretation of such a philosophical category as a destiny, that is patience in confronting of destiny's strokes as hereditary disease transmitting in the generation. The use of molecular genetic methods requires qualitatively new approaches in the practice of the medical- genetic service. We consider, that the complex preventive measures are necessary in Yakutia attracting psychologist, social workers, philosophers, lawyers.

**Poster No : PP-16** (*Poster Presentation on 3 November 2009*)

## ***Tracing the Use and Source of Racial Terminology in Representations of Genetic Research***

**Christen RACHUL**, Colin Ouellette, Darren Lau, Timothy Caulfield

The concept of race remains tremendously controversial. Recent population based genomic research has intensified the debate, as race (or proxies for the concept of race) have been used to describe populations under study and in the media report of research results. There is concern that the use of racial terminology in this context will have a number of adverse implications, including legitimising biological views of race, increasing racist attitudes, and giving rise to poor health outcomes. The use of race and related terms by genomics researchers introduces a number of methodological concerns and, as has been noted, are often used with less than ideal precision. Racial terminology in scientific literature lacks standardised definitions and often relies on cultural and political notions of race for stratifying research cohorts. Attempts at establishing precise terminology have resulted in slippage back to more generalised notions of race. Racial terminology can also hold different meanings dependent on geographic, social, historical and personal context. In addition, biomedical research often relies on participant self-identification of race, which may be a less than accurate indication of genetic similarity. Given the potential for adverse implications and methodological problems, the continued use of racial terminology in genomics research is questionable. In this study, we trace the use of racial terminology in peer-reviewed scientific articles, press releases of these articles and finally in the news articles prompted by the press releases. Both quantitative and qualitative data from textual analyses demonstrate the wide variation in the use of racial language by both biomedical researchers and the media. Results indicated that racial terms (eg. 'black', 'white', 'asian', 'ethnicity', 'ancestry') were used throughout the articles, regardless of the nature of the publication or author expertise. Based on the results of this study and previous research, it is becoming increasingly apparent that the use of racial categories in genetic research is fraught with problems, not the least of which is the ill-defined terminology and often unjustified methodologies that may have serious social and health-related implications. While the newspaper articles and press releases sometimes provide a limited report of research, the genetic research community is also responsible for accurately defining research terms and identifying research populations.

**Poster No : PP-17** (*Poster Presentation on 1 November 2009*)

## **Ethical aspects of Array Comparative Genomic Hybridisation (CGH) for mental retardation diagnosis and genetic counselling**

**Julia SOPHIE**, A Vigouroux, , E Rial-Sebbag, G Bourouillou, P Calvas. A. Cambon-Thomsen

The recent rapid emergence of new technologies in genetic medicine contributes to the diagnosis and the understanding of the molecular basis of numerous diseases. The high throughput and the sensitivity of these technologies will create an unprecedented volume of information for patients, counsellors, and health care providers, raising also unique ethical challenges.

Although the applications of many of these techniques are still limited to research, Array Comparative Genomic Hybridisation (CGH) has evolved to a standard application in clinical genetics, especially in individuals with syndromic or non-syndromic mental retardation. CGH allows a whole genome analysis at a resolution, 10-10 000 times higher than that of routine chromosome analysis by karyotyping. This method has revolutionised the present clinical practice in the detection and diagnosis of human chromosome abnormalities in mental retardation.

As any next generation sequencing method, CGH is likely to reveal complex and heterogeneous information with different level of prediction, making the test reporting complicated.

Since 2005 the Toulouse medical genetics hospital department has examined 176 cases of mental retardation using CGH. New questions appeared when considering results, their interpretation and their communication. They will be presented according to two main dimensions: uncertainty of results and incidental findings.

**Problem of uncertain results:** Micro array reveal far more novel variants of uncertain significance. Our current understanding of detectable variation is still at an early stage, and the knowledge base is rapidly changing. How to evaluate the clinical utility and patient benefit, how to give accurate counselling in term of reproductive recurrence risk for the parent and the family? Should we re contact the family when further knowledge is available?

**Incidental finding:** As any high resolution scan of the entire genome, CGH bears the risk of detecting unexpected data for example unintended predictive diagnosis of a cancer prone syndrome. The detection of adult onset condition in a child has clinical, legal and ethical implications. If the ethical dilemma raised by incidental finding have been discussed before, the "incidental" becomes "usual" representing new challenges for physicians and counsellors. How pre test counselling could anticipate such complex situations? How to respect the patient autonomy and the right to know or to not know? Should we exclude genomic loci involving adult onset conditions in paediatric sample?

CGH is currently becoming an indispensable tools in the diagnostic of idiopathic or syndromic mental retardation this technology challenges drastically our practice of genetic counselling. Ethical issues of genetic disease diagnostics and various health applications have already been addressed in numerous reports. Specific reference documents addressing the ethical issues of genetic testing in relation to CGH are still lacking.

## ***The search for a “reasonable” expectation of genomic privacy***

**Jennifer K. WAGNER**

It is commonly held in the United States that each individual has a right to health privacy generally. Mixing one part health privacy with one part genetic determinism, the American public has cooked up the notion of genetic/genomic privacy. HIPAA, GINA and other legislative postures encourage this notion. Yet geneticists recognise (and other scholars are beginning to recognise as well) that not all genetic or genomic information is of equal medical importance (regardless of the merits of phrases like “junk dna” or “dna deserts”) and that genetic and genomic information tells only part of the complicated story of how normal or disease traits and conditions come into being. Likewise, geneticists recognise that not all genetic or genomic information is of equal value when looking to DNA to identify a person individually or to affiliate a person with a group or population of interest. We must acknowledge that embracing a right to “genomic privacy” in a medical (research or clinical) context implicates expectations of privacy in other contexts, like searches and seizures by law enforcement.

The American public has been quick to embrace the personal genomics industry. It has been described by some as America’s “next obsession.” The American public is eager to find out their personal genomic information (which has been referred to with varying degrees of optimism as a “genetic diary,” “genetic blueprint,” or even “genetic horoscope”). Individuals reportedly tattoo their genomic information to their arm, hang their genomic data on the walls of their home and office as art, post their genomic sequence on blogs and social networking sites, and share their genomic information with friends, family and even strangers. Voluntary participants of genomics research have now seen (though they may not have recognised) the changing trend from a default standard that collected research data will be held confidential to the more realistic expectation that collected research data will be shared with a number of research collaborators, possibly worldwide.

GINA, the Genetic Information Nondiscrimination Act, was adopted to encourage participation in genomics research, encourage use of medical professionals to administer genetic/genomic tests (and thereby discourage direct-to-consumer tests), and minimise fears of genism (that is, genetic discrimination). While it may be too soon to tell if the public is buying GINA’s promises (one scholar describes the likely change as being merely a switch “from Fear Factor to Fantasy Island”), the expectation of genomic privacy is one that is constantly and certainly changing in perhaps unexpected ways.

This poster reviews, generally, the transient nature of the “right to privacy” since its 1890 inception in the United States and discusses the evolving expectation of privacy in genetic and genomic information. The discussion reveals how the treatment of privacy of genomic information in research and clinical settings necessarily changes how privacy of genomic information is treated in criminal law settings and briefly discusses practical issues in the application of genomic sequencing in the criminal justice system.

## ***A framework for regulating direct-to-consumer genetic tests***

**Caroline WRIGHT**, Alison Hall and Ron Zimmern

The Foundation for Genomics and Population Health (PHG Foundation) is an independent, non-profit, health policy research and development organisation based in Cambridge, and is the leading UK centre for public health genomics. Its overarching purpose is to foster and enable the application of biomedical science, particularly genome-based technologies, for the benefit of human health. Among its specific objectives is the promotion of a social and regulatory environment that is receptive to innovation, without imposing an undue or inequitable burden.

Following extensive work on the evaluation of genetic tests and complex biomarkers – building on the eponymous ACCE framework – we have recently turned our attention to direct-to-consumer (DTC) genetic testing services that purport to offer individual genetic risk assessments for numerous diseases. Such services commonly utilise genome-wide arrays to directly assess common DNA sequence variations. Energised by the sudden explosion in genomic risk profiling services available DTC via the internet, there has been a concomitant rise in calls for tighter regulation of this 'consumer genomics' movement. However, there is a lack of consensus as to the extent to which regulators should be involved, and the role of legislation versus self-governance or voluntary guidance within an appropriate regulatory framework.

In order to develop an overarching regulatory framework for DTC genetic tests, which is both practicable within a global market and robust enough to protect the consumer, we have undertaken a conceptual analysis of the key issues. We drew heavily upon the following distinctions: firstly, that between an assay and a test; secondly, that between a product and service; and thirdly, that between direct harm caused by the test itself and indirect harm caused by the result of the test.

Our analysis suggests that 'genetic' information varies enormously in terms of its sensitivity and predictive nature, such that it is neither sufficiently homogeneous nor different from other kinds of health-related information to warrant special or 'exceptional' protection. We also note that the principle of individual autonomy is being accorded greater significance in society than ever before, fostered by the rise in information technology, scientific knowledge and consumer healthcare. Combining these guiding principles and distinctions with the ACCE evaluation framework suggests that an appropriate regulatory response to burgeoning DTC genetic testing services would consist of just five key requirements, which we believe to be both feasible and sufficient:

- (1) a proportionate set of consent procedures such that citizens are unambiguously informed about the nature of the information they will receive as a result of testing and its possible implications;
- (2) formal laboratory accreditation, so that citizens can have confidence in the assay results (i.e. analytical validity) that are generated;
- (3) evidence that the claimed gene-disease association is valid (which is a necessary, though not sufficient, component of clinical validity);
- (4) appropriately qualified staff to interpret the test result and provide support to consumers in order to maximise personal utility; and
- (5) strengthening consumer protection legislation to prevent false or misleading claims, including unsubstantiated and overhyped assertions concerning clinical utility.

**Poster No : PP-20** (*Poster Presentation on 2 November 2009*)

## ***Estimating New Zealand Maori Genomic Ancestry Using a Minimal Set of Ancestry Informative Markers***

**David ECCLES HALL**, Rodney Lea and Geoffrey Chambers

We have utilised a bootstrap method to generate a set of ancestry informative markers for the investigation of Māori-European admixture within a subgroup of Māori from New Zealand.

Genome-wide Association Studies (GWAS) aim to determine the degree of association between single genetic markers and a heritable trait. Most often, these studies look for associations relating to susceptibility for particular diseases, but some have also looked at intermediate traits (endophenotypes) that are not directly associated with disease. Here we present a bootstrapping method that can be used to complement GWAS. The aim of this method is to produce a signature set of genetic markers that approximate the total genetic contribution to variation of a particular heritable trait.

This method has been tested with a trait that is 100% heritable - New Zealand Māori genomic ancestry. Genotype data from 30 Māori individuals and 90 European individuals were compared at 300k autosomal polymorphisms. After bootstrapping and evaluating the effectiveness of marker sets of different sizes, we composed a validated set of 10 genetic markers that estimates individual Māori ancestral fraction with high accuracy (median = 98%, IQR = 95-99). These markers have been used to determine the variation of ancestral fraction within a tribe of Māori from Hawkes Bay, New Zealand. This calculation of genetic variation is part of a broader health and ancestry study that is currently underway in this population. Using the set of 10 ancestry-informative markers, we estimate the proportion of European admixture within this Māori population to be  $28 \pm 3\%$ . Other survey questions and blood tests carried out as part of the health and ancestry study have enabled us to identify a number of clinically relevant traits that correlate well with the ancestry score within the Hawkes Bay Māori population.

Care must be taken when extrapolating beyond the populations from which GWAS discovery groups are derived, as overfitting and unexpected admixture can complicate the outcome of the genetic score. Although these issues need to be taken into account, we hope that clinicians will consider using a genetic score when predicting risk for heritable diseases. When genetic data are available, this score will be better than a qualitative ethnicity question, because the genetic basis of the test is closer to the genetic basis of the disease.

## **ABSTRACT INDEX**

<b>ANTONARAKIS, Stylianos E.</b>	.....	48
<b>AUFFRAY, Charles</b>	.....	49
<b>AVEY, Linda</b>	.....	50
<b>CAMPBELL, Alastair V.</b>	.....	51
<b>CAULFIELD, Timothy</b>	.....	52
<b>CHADWICK, Ruth</b>	.....	53
<b>COX, David</b>	.....	54
<b>DAAR, Abdallah S.</b>	.....	55
<b>DUCOR, Philippe</b>	.....	56
<b>JIMENEZ-SANCHEZ, Gerardo</b>	.....	57
<b>KNOPPERS, Bartha-Maria</b>	.....	59
<b>LINDPAINTNER, Klaus</b>	.....	60
<b>LIU, Edison T.</b>	.....	61
<b>LUNSHOF, Jeantine</b>	.....	62
<b>MAJUMDAR, Partha</b>	.....	63
<b>MCCARTHY, Mark</b>	.....	64
<b>MURRAY, Thomas H.</b>	.....	65
<b>NISSENBAUM, Helen</b>	.....	66
<b>OLOPADE, Olufunmilayo</b>	.....	67
<b>RAMESAR, Raj S.</b>	.....	68

PP-01	.....	70
PP-02	.....	71
PP-03	.....	72
PP-04	.....	73
PP-05	.....	74
PP-06	.....	75
PP-07	.....	77
PP-08	.....	78
PP-09	.....	79
PP-10	.....	81
PP-11	.....	82
PP-12	.....	83
PP-13	.....	84
PP-14	.....	85
PP-15	.....	86
PP-16	.....	87
PP-17	.....	88
PP-18	.....	89
PP-19	.....	90
PP-20	.....	91



