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CANADIAN INSTITUTES OF HEALTH RESEARCH

**Expert Review Team Report
for
Institute of Genetics**

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Chair, Expert Review Team
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Summary

Overall assessment

- Since CIHR's creation, a clear improvement is evident in Canadian health research performance, both from the perspective of knowledge generation and science management. Continued improvement has also been seen past the 2006 international review.
- The overall assessment of the Institute of Genetics (IG) for the 2006-2010 period is extremely positive. The Institute has developed a clear vision and a strong leadership (both past and present) and is regarded as a flagship institute for CIHR. The IG has been particularly successful in engaging its partners and stakeholders. The research community, stakeholders and partners all consider themselves well-represented by IG.
- IG has demonstrated that it is a cross-cutting institute that has penetrated multiple disciplines. IG is co-leading two of four CIHR initiatives that are moving forward to business cases. This attests to IG's ability to lead strategic directions of CIHR as a whole.
- In the last 10 years, IG has built up important research capacity, which has moved health research in the field of genetics in Canada into an internationally competitive position. Efforts should now be made to ensure sustainability of individual careers, as well as established multi-disciplinary research teams and facilities, thereby avoiding building capacity without utility. This observation may also have relevance to other CIHR institutes.
- IG has become an accepted interface between basic researchers and the clinical health research community. Care should be taken to ensure efficient governance procedures. One possible change could be the allocation of (limited) funds towards creating new strategic collaborations or new joint projects with other institutes or non-CIHR agencies that require quick and incisive action.
- IG has made great strides towards involving various stakeholders. However, better web-based communication and increased media exposure to the public at large are still needed to allow IG to realize its mandate fully.
- Healthcare will become increasingly genetically informed. CIHR has successfully provided scientific evidence to Health Canada to advise on the clinical efficacy of specific health care innovations. IG is therefore in a good position to actively support informed changes to health care policies related to genetics on the basis of scientific evidence. These efforts should be further supported by CIHR.
- Regulatory issues are increasingly seen to be an impediment to health care innovation. Patient organizations would like to see CIHR take an active role in

- The ERT strongly encourages both IG and CIHR to further and proactively engage the international scientific community and participate in international initiatives.
- Bioinformatics, Computational and Systems Biology have not been sufficiently integrated so far in the CHIR/IG strategic roadmap. We recommend IG and Genome Canada to take a lead and coordinate efforts to quickly put into place programs for advancing initiatives in personal genomes and bioinformatics.
- Both topics have great urgency and there should be a coordinated plan by the end of 2011. For personal genomes, this would include involvement with Health Canada, and for bioinformatics, the involvement of the Natural Sciences and Engineering Research Council of Canada (NSERC) and possibly other organizations to further develop and facilitate a national bioinformatics strategy.
- The analysis of gene-environment interactions bears great potential not only for public health, but also for environmental research. IG should consider including environmental aspects, i.e. gene-environment interactions, in its portfolio.

Section 1 – Institute mandate

The Institute of Genetics' (IG) mandate is to support research on the human genome and on all aspects of genetics related to human health and disease, including how information contained in the genome is modified or altered through interactions with physical and social environments. The interpretation of the mandate was later expanded to recognize the implicit inclusion of basic biochemistry, cell biology and research using model organisms and today, IG is generally viewed as the institute that advocates for fundamental research using animal and cell culture models aimed at gaining basic insights into biological processes of relevance to human health.

“...the present mission is to support research on the human and model genomes and on all aspects of genetics, basic biochemistry and cell biology related to health and disease, including the translation of knowledge into health policy and practice and the societal implications of genetic discoveries.”

CIHR Institute of Genetics – Internal Assessment for 2011 International Review, pg 1

Section 2 - Status of this area of research in Canada

Since CIHR's creation, improvement is evident across the board in Canadian health research science both from the perspective of knowledge generation and science management. Continued improvement has been seen past the 2006 international review, and with implementation of the previous ERT.

The number of publications has impressively increased and Canada is playing increasingly a competitive role in international health research. This is clearly evident from the work of IG-scientists. These have made many important discoveries and can be seen to be leaders in the field, in a number of topics such as stem cells, neurodegenerative diseases, autism, and many others ranging from basic biochemistry to applied human genetics.

In the last 10 years, CIHR has built up important research capacity, which was one of the driving forces to move health research in Canada into an internationally competitive position. The challenge however will be to maintain this position and capitalize on the achievements obtained so far.

Over the preceding period, a number of clinical and experimental genetics scientists have been able to establish their scientific careers through support of a career-development program launched by IG. This has proven highly effective and successful. Efforts should now be made to sustain individual careers, as well as established multidisciplinary research teams and facilities thereby avoiding building capacity without utility.

International joint programming and cooperation will be an increasing characteristic of competitive research. IG scientists already play an important role in several international initiatives, notably the Structural Genomics Initiative and the International Human

Epigenetics Consortium. CIHR should support a proactive role in the internationalization efforts of the IG and develop a national strategy to position Canada appropriately in the increasingly global research landscape.

Section 3 - Transformative Impacts of the Institute

The overall impression of IG is very positive. IG has a clear vision and strong leadership (both past and present) and is regarded as a flagship institute for CIHR. IG's focus on engaging partners and its readiness to engage stakeholders are further commended. It is noted that at the present time, the research community, stakeholders and partners all consider themselves well-represented by IG. Numerous initiatives with other CIHR institutes have been realized attesting to the broad and integrative vision of the Institute of Genetics, including its directors and its individual scientists.

IG has started out as the focal point for basic research within the CIHR, but has impressively extended into the more applied domains of genomic medicine and public health. IG has demonstrated that it is a cross-cutting institute that has successfully penetrated multiple disciplines. IG is co-leading two of four CIHR initiatives that are moving forward to business cases. This attests to IG's ability to lead the strategic directions of CIHR as a whole.

IG has made great strides towards involving various stakeholders. The Institute's Priority and Planning Committees (IPPCs) are seen as an effective tool to ensure that researchers influence and identify priorities to develop future initiatives. The process of building a scientific community has been continuously developed and seems to work very efficiently. However intra- and inter-organizational communication could still be improved. Particularly, better web-based communication and increased media exposure to the public at large are still needed to allow IG to realize its mandate fully.

IG has instigated a number of strategic initiatives. These are mostly well received by the community. IG scientists mention the Inventions and Technology Application - Tools Techniques and Devices for Research and Medicine Initiative is particularly notable for its return on investment.

Progress in genomics technologies has seen an exponential development. Automatisation and miniaturization will continue to drive the field, along with a strong convergence of biology and medicine with information and communication technology (ICT). High performance computing and large scale data storage are becoming an integral part of health research. There is some concern of the ERT that a coherent strategy and roadmap for the future of ICT and bioinformatics has not been implemented yet.

As a result of their complementary expertise IG and Genome Canada have great potential for joint cooperation in this area. We recommend IG and Genome Canada to coordinate their efforts and to put into place programs for funding initiatives in personal genomes and bioinformatics.

Both topics have great urgency and there should be a coordinated plan by the end of 2011. For personal genomes, this would include involvement with Health Canada, and for bioinformatics, the involvement of NSERC and possibly other organizations to further facilitate a national bioinformatics strategy.

Section 4 - Outcomes

Overall, IG has achieved numerous breakthroughs and publications. Output is very good with many IG scientists performing at the cutting-edge of science as is clearly evident from the list of top-50 IG researchers who all qualify as leaders in their fields.

Action on the recommendations from the 2006 review has led to fewer and more focused initiatives and this is appreciated by IG's scientists. IG has instigated a number of strategic initiatives. These are mostly well received by the community.

The rapid developments in genomic and medical technology bring along new business and economic opportunities. IG is encouraged to strengthen its efforts for an efficient knowledge transfer and for cooperation with industrial partners.

Section 5 - Achieving the Institute mandate

IG has made significant progress towards achieving its mandate. The Institute has performed particularly well in terms of building capacity and in fostering cross-discipline collaboration. The involvement of engineers is a notable element of the latter. The training program for physician scientists from the clinical genetics discipline is an example of the former.

CIHR and IG have successfully extended their activities towards the application domain of health services and population health. IG has become an accepted interface between basic researchers and the clinical health research community.

The current management structure with the scientific council setting priorities and determining the Institute's goals, and the IPPCs allowing individual scientists to participate through a bottom-up process seems to be effective. The IPPCs are seen as an effective tool by which researchers influence and identify priorities to develop future initiatives.

There exists however a risk of over-regulating the policy-setting process. Care should be taken to ensure that the Scientific Director can negotiate decisive actions nimbly, when this is required. One possible change could be the allocation of (limited) funds towards creating new strategic collaborations or new joint projects with other institutes or non-CIHR agencies that require quick and incisive action.

Section 6 - ERT Observations & Recommendations

In the last 10 years CIHR has built up important research capacity which has moved health research in Canada into an internationally competitive position. Efforts should now be made to ensure sustainability of these achievements. This applies particularly to individual careers, as well as established multi-disciplinary research teams and facilities, thereby avoiding building capacity without utility.

Action on the recommendations from the 2006 review has led to fewer and more focused initiatives and this is appreciated by IG's scientists. IG has made great strides towards involving various stakeholders. However, better web-based communication and increased media exposure to the public at large are still needed to allow IG to realize its mandate fully.

Healthcare will become increasingly genetically informed. CIHR has successfully provided scientific evidence to Health Canada to advise on the clinical efficacy of specific health care innovations. The acceptance within the scientific community puts IG in a good position to actively support informed changes to health care policies related to genetics on the basis of scientific evidence. These efforts should be further supported by CIHR.

Regulatory issues are increasingly seen to be an impediment to health care innovation. Patient organizations are already involved in discussing regulatory issues concerning innovation of disease management with Health Canada with the objective of removing barriers for innovation. They would like to see CIHR take an active role in these discussions enabling rapid implementation of innovative treatments, for instance for rare diseases. ERT strongly supports these efforts.

IG scientists play an important role in several international initiatives. CIHR seems to lack a clear international collaboration strategy. The ERT strongly encourages both IG and CIHR to proactively support a further engagement of Canada with the international scientific community.

Bioinformatics, computational and systems biology have not been sufficiently integrated so far in the CHIR/IG strategic roadmap. IG and Genome Canada have great potential for joint cooperation in this area given their complementary expertise. We recommend IG and Genome Canada coordinate efforts to quickly put into place programs for advancing initiatives in personal genomes and bioinformatics. Both topics have great urgency and there should be a coordinated plan by the end of 2011. For personal genomes, this would include involvement with Health Canada, and for bioinformatics, the involvement of NSERC and possibly other organizations to further develop and facilitate a national bioinformatics strategy.

The analysis of gene-environment interactions bears great potential not only for public health, but also for environmental research. IG should consider including environmental aspects, i.e. gene-environment interactions, in its portfolio.

Appendix 1 - Expert Review Team

Chair - Professor Han G. Brunner

Professor of Medical Genetics

Head - Department of Human Genetics, Radboud
University Nijmegen Medical Centre, The Netherlands

Expert Reviewer - Professor Jim R. Lupski

The Cullen Endowed Chair in Molecular Genetics

Professor, Department of Molecular and Human Genetics and
Department of Pediatrics

Baylor College of Medicine, Houston TX USA

International Review Panel – Professor Rudi Balling

Director - Luxembourg Centre for Systems Biomedicine

University of Luxembourg

Appendix 2 - Key Informants

Session 1 – Review of Institute

1. **Dr. Paul Lasko, IG Scientific Director**
2. **Dr. Michel Bouvier, Chair – Institute Advisory Board**
Professor of Biochemistry, Institute for Research in Immunology and Cancer
University of Montréal
3. **Dr. François Rousseau**
Professor, Department of Medical Biology
Université Laval

Session 2 – Consultation with researchers

1. **Dr. Howard Lipshitz**
Professor and Chair, Department of Molecular Genetics
University of Toronto
Senior Scientist, Program in Developmental & Stem Cell Biology
Hospital for Sick Children Research Institute, Toronto
2. **Dr. Christopher Yip**
Professor, Department of Biochemistry
Department of Chemical Engineering and Applied Chemistry
University of Toronto
3. **Dr. Kym Boycott**
Medical Geneticist, Regional Genetics Program, Investigator
Children's Hospital of Eastern Ontario

Session 3 – Roundtable with stakeholders

1. **Dr. Durhane Wong-Rieger**
President
Canadian Organization for Rare Disorders
2. **Dr. Cindy Bell**
Executive Vice-President, Corporate Development
Genome Canada