

## MEETING REPORT

# Exploring the ELSI Universe: critical issues in the evolution of human genomic research

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### Abstract

A report on the National Human Genome Research Institute's Ethical, Legal, and Social Implications Research Program 2011 Congress, 'Exploring the ELSI Universe', Chapel Hill, North Carolina, USA, 12-14 April 2011.

**Keywords:** Empirical research, ethics, genomic research

The 2011 Ethical, Legal, and Social Implications (ELSI) Congress, 'Exploring the ELSI Universe', hosted by the Center for Genomics and Society in Chapel Hill, kicked off with an inspiring introduction by Eric Green (Director, National Human Genome Research Institute (NHGRI), National Institutes of Health, USA) who outlined the place of ELSI research in the NHGRI's new strategic plan. The 3-day interdisciplinary meeting, which was attended by hundreds from many different countries, and fields as varied as genomics, bioethics, the social sciences, philosophy and law, offered an opportunity to reflect on the myriad of ethical, legal and social issues in genomics and to plan for the future of genomic medicine. Green challenged attendees to contemporize ELSI research and stressed the importance for ELSI researchers to balance a foundational knowledge base with knowledge gained from acting as 'research SWAT teams' collecting empirical evidence to inform best practices. The conference presented the novel ways ELSI research is helping to guide the field of genomics in plotting the course from 'base pairs to bedside'. Here we cover some of the conference highlights, including forward-looking proposals to repurpose research governance and oversight, significant efforts in empirical research and creative methodological approaches.

### ELSI in the 21st century

The opening plenary session of day 2 provided accounts of the ongoing role of ELSI researchers in the development

and implementation of research governance and oversight. Jane Kaye (University of Oxford, UK) presented a pioneering vision of a future in which research governance and oversight should be aligned with how research is now being conducted in global networks (no longer one research project, by one researcher, in one jurisdiction). As part of this change, she advocated a move towards coordinated, global, non-duplicative efforts to advance ELSI research using the same approach and scale as the Human Genome Project (see <http://www.publichealth.ox.ac.uk/helex/> for more information on the Global ELSI Initiative). Employing information technology (IT) innovations was also advocated by Malia Fullerton (University of Washington, USA) who explained how these strategies can provide mechanisms to engage participants while promoting transparency of governance and oversight.

Research governance and oversight structures, even if enhanced through novel IT solutions, must be supported by evidence-based policies. Mark Rothstein (University of Louisville, USA) pointed out how difficult it is to influence policy and noted that ELSI has been most effective in shaping legislation the closer it stays to its central mission of genomics. Concrete ways in which ELSI research can contribute to policy development were described by Pearl O'Rourke (Partners HealthCare Systems, Inc., USA). In order to produce research data to inform policies and educate and engage various stakeholders, ELSI researchers must continually monitor and assess emerging issues in a broad research portfolio. Although the overarching theme of this meeting was to conceive of ways to contemporize and propel the field forward, Kathy Hudson (National Institutes of Health, USA) reviewed previous research ethics atrocities that now serve as reminders of the tremendous improvements achieved in research ethics and regulations. Hudson underscored the need for new, empirically informed policies to revolutionize the implementation of existing protections and regulations. Through innovative research, ELSI researchers will lead policy makers in the creation of modern governance structures engineered to meet the ethical challenges of genomic medicine.

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### **Empirical data abounds at ELSI**

With empirical evidence needed to develop well-informed policies, conference panels and themed sessions were accordingly dominated by talks presenting the rich and compelling data being generated. Major ethical issues under study in the field included topics such as health equality, behavioral genetics, human microbiome research and biobanking, to name a few.

In looking at health equality research, David Williams (Harvard University, USA) presented data illustrating health inequalities faced by racial minority groups in the USA; these disparities persisted after controlling for confounding factors. Wylie Burke (University of Washington, USA) spoke of the complexity of genomic research, especially with regard to the interaction of genetics and the environment. Burke and Williams urged scientists to forgo thinking of genes as the cause of disparities, yet to use genomic tools as another way to understand the biological effects of our social experience.

Two other pressing areas drawing much attention were data sharing and the return of genetic research results. To elucidate the intricacies of these complex issues, ELSI researchers reported on their explorations of the distinct voices of various stakeholders (for example, genomic researchers, institutional review board (IRB) members, research participants and the general public).

With regard to data sharing, findings from several studies indicated that participants want to be active participants with some control over the use of their genetic data. Susan Trinidad (University of Washington, USA), for example, reported data from focus groups where the majority of participants appreciated being re-contacted for permission prior to the sharing of their genetic and health information with other researchers, as this was viewed as a sign of respect.

In an effort to treat participants as partners in research, there is growing consensus that at least some individual genetic research results (IRRs) and incidental findings (IFs) ought to be shared with participants. An overview by Susan Wolf (University of Minnesota, USA) discussed how IRRs and IFs arise in all phases of genomic research, and highlighted ethical challenges to be addressed, such as establishing criteria that make these results returnable. A survey of genome-wide association study (GWAS) investigators presented by Rachel Ramoni (Harvard Medical School, USA) found that while most GWASs do not allow for the return of IRRs, nearly all GWAS investigators expressed that there are cases when results should be returned. Janet Williams (The University of Iowa, USA) described IRB chairs' struggles in making determinations about returning genomic IFs, and noted their concerns with the interpretability of IFs, the potential for risk and harm, and a lack of existing guidelines. Initial results from a survey of IRB chairs by Laura Beskow

(Duke University, USA) also stressed the need to establish guidelines, particularly in emerging areas such as genotype-driven recruitment where eligible participants are identified based on genetic information learned about them in a previous study.

### **New ways to explore the ELSI universe**

A high degree of innovation will be required of ELSI researchers to engender trust, advance the field, and help facilitate ethical genomic research. Benefits of the interdisciplinary nature of the ELSI research program are apparent in the range of thought and methodological approaches currently used and proposed for future studies. For example, Rachel Haase (University of North Carolina at Chapel Hill, USA) presented fresh approaches to qualitative methods using data analysis software to assess word frequency and interpret word usage. Inventive approaches to community engagement presented by Wendy Marsden (University of Edinburgh, UK) also described advancements in technological methods that link participants in patient networks to genetic research.

One session featured a play entitled 'It's Not That Simple! Genomic Research and the Consent Process,' which was cleverly scripted by Karen Rothenberg (University of Maryland School of Law, USA) and Lynn Bush (Columbia University, USA). Lines were read by attendees; notably, the Oscar-worthy performance of Jim Evans (University of North Carolina at Chapel Hill, USA) did not disappoint! The writing team's ability to transform language from genuine genetic research informed consent documents into a flawless portrayal of a family's struggle as they face enrollment into a genetic research study showcased the talents present. The conference's final sessions also reflected the imaginative minds at play, as three open-forum workshops on the future of the 'E,' the 'L' and the 'S' in ELSI research provided a unique platform for attendees to express their final thoughts and reiterate proposed directions for the field.

### **The future of ELSI**

The closing talk by Eric Juengst (University of North Carolina at Chapel Hill, USA) imparted the future of the 'T' in ELSI research. Juengst described the historical and ongoing changes in the ELSI research portfolio and stressed that the goal of realizing genomic medicine, which was emphasized in Green's introductory remarks, might itself pose some ELSI challenges that we should remain alert to. He also proposed international collaboration to avoid unintended social consequences of genomic medicine in other parts of the world. Juengst encouraged ELSI researchers to engage the more challenging and controversial issues in genomics, rather than simply trying to 'smooth the path' for genomic medicine.

This tension between facilitating ethical genomic research while providing more critical assessment of the advances being made was a major theme of the ELSI Congress and will be an important challenge facing ELSI researchers. A recent spate of requests for applications from the NHGRI, which seeks novel integrative approaches to incorporate ELSI research into genomic research to capture real-time assessments of genetic information being delivered directly to participants and applied to clinical care, affords ELSI researchers an opportunity to practice balancing these roles of facilitators and critical assessors. ELSI researchers must be ready to evaluate and anticipate ethical issues that may arise in this new era of genomic research to ensure a successful path forward; many at the ELSI Congress seemed prepared to take on that challenge.

#### Abbreviations

ELSI, ethical, legal, and social implications; GWAS, genome-wide association study; IF, incidental finding; IRB, institutional review board; IRR, individual research result; IT, information technology; NIH, National Institutes of Health; NHGRI, National Human Genome Research Institute.

#### Competing interests

The authors declare that they have no competing interests.

#### Acknowledgements

We thank Laura Achenbaum and Stacey Pereira, and ELSI Congress presenters mentioned in the manuscript, for their thoughtful editing and input.

Published: 21 June 2011

doi:10.1186/gm254

**Cite this article as:** Oliver JM, McGuire AL: Exploring the ELSI Universe: critical issues in the evolution of human genomic research. *Genome Medicine* 2011, **3**:38.