News, literature, and events in the ethical, social, and legal implications of psychiatric, neurologic, and behavioral genetics.

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Braingenethics Update

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In the Literature

Genetic Testing Preferences in Families Containing Multiple Individuals with Epilepsy

By Janice O. Okeke et al.

This study aims to examine genetic testing preferences in families containing multiple individuals with epilepsy. One hundred forty-three individuals with epilepsy and 165 biologic relatives without epilepsy from families containing multiple affected individuals were surveyed using a self-administered questionnaire. Interest in epilepsy genetic testing may be high in affected and unaffected individuals in families containing multiple individuals with epilepsy, especially when testing

Commentaries

The Height of Folly: Are the Causes of Stature Numerous but at Least 'Finite'?

By Ken Weiss

This blog critique questions the conclusions of a study recently published in Nature Genetics that claims that the number of causes of stature variation is finite. Weiss argues that there is no known upper limit to genome size (as some huge-genomed species show), and hence no limit to the number of insertions that may occur sometime or somewhere.

has implications for improving clinical care.

The High Heritability of Educational Achievement Reflects Many Genetically Influenced Traits, Not Just Intelligence
By Eva Krapohl et al.
Krapohl et al. identify the general ingredients of educational achievement using a multivariate design that goes beyond intelligence to consider a wide range of predictors, such as self-efficacy, personality, and behavior problems, to assess their independent and joint contributions to educational achievement. Authors use a genetically sensitive design to address the question of why educational achievement is so highly heritable. This study concludes that the high heritability of educational achievement reflects many genetically influenced traits, not just intelligence.

Eating in the Absence of Hunger but Not Loss of Control Behaviors Are Associated with 16p11.2 Deletions
By Richard Gill, Qixuan Chen, Debra D'Angelo, & Wendy Chung
The ~600-kb BP4-BP5 16p11.2 deletion has been consistently associated with obesity. Authors studied two heritable disinhibited eating behaviors, eating in the absence of hunger (EAH) and loss of control (LOC), to better characterize the relationship between the deletion and obesity. They conclude that 16p11.2 deletion may influence specific obesity-associated disinhibited eating behaviors: EAH due to external trigger and EAH due to boredom.

He concludes that it is misleading and incorrect to say that the number of causes, even just the genetic causes, of stature variation is truly finite.

Serving Epigenetics Before Its Time
By Eric T. Juengst, Jennifer R. Fishman, Michelle L. McGowan, & Richard A. Settersten Jr.
Society prizes the rapid translation of basic biological science into ways to prevent human illness. The authors of this paper argue that a premature rush to take murine epigenetic findings in these directions makes impossible demands on prospective parents and triggers serious social and ethical questions.

The Benefits of "Binocularity"
By Erik Parens
Just as we need two eyes that integrate slightly different information about one scene to achieve visual depth perception, we need to view ourselves through two lenses to gain a greater depth of understanding of ourselves.

In the News

Epigenetics Paper Raises Questions
By Kate Yandell
GENETICS published a commentary by Gregory Francis criticizing a *Nature Neuroscience* paper claiming that mice can inherit smell sensitivities that their parents acquired during life. Francis

Neuropeptide Precursor VGF is Genetically Associated with Social Anhedonia and Underrepresented in the Brain of Major Mental Illness: Its Downregulation by DISC1

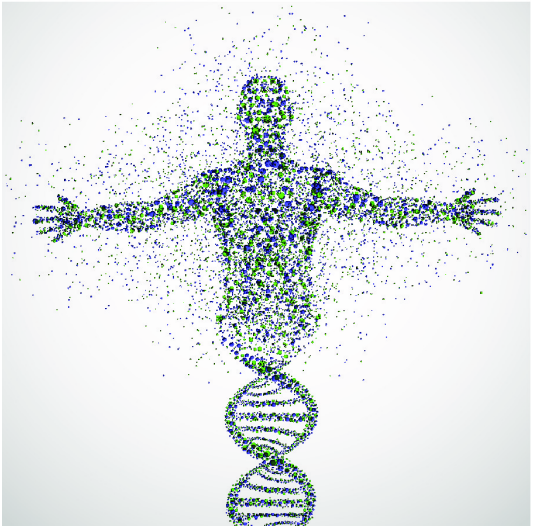
By Adriana Ramos et al.

In a large Scottish pedigree, disruption of the gene coding for DISC1 clearly segregates with major depression, schizophrenia and related mental conditions. Thus, study of DISC1 may provide a clue to understand the biology of major mental illness. A neuropeptide precursor VGF has potent antidepressant effects and has been reportedly associated with bipolar disorder. Ramos et al. show that DISC1 knockdown leads to a reduction of VGF, in neurons, and they propose that VGF participates in a common pathophysiology of major mental disease.

Refining Analyses of Copy Number Variation Identifies Specific Genes Associated with Developmental Delay

By Bradley P. Coe et al.

Coe et al. created an expanded copy number variant (CNV) morbidity map from 29,085 children with developmental delay in comparison to 19,584 healthy controls, identifying 70 significant CNVs. 26 candidate genes were resequenced in 4,716 additional cases with developmental delay or autism and 2,193 controls. An integrated analysis of CNV and single-nucleotide variant (SNV) data pinpointed 10 genes enriched for putative loss of function. This combined CNV and SNV approach facilitates called into question the statistical power of the study, but others are not convinced that his criticism invalidates the study.



Center Seminars:

"Ethical Issues with Alzheimer's Disease Research Involving Human Subjects"

Dena Davis, JD, PhD, Lehigh University

November 12, 2014, 4:00 – 5:00 pm

Alzheimer's disease research with human subjects poses distinct ethical challenges. These challenges arise from the societal fear of the disease, the predictive qualities of Alzheimer's biomarkers, and the fluid cognitive status of research subjects. This presentation lays out four of these challenges and suggests some possible responses.

Room 405A-B, Educational

discovery of new syndromes and genes involved in neuropsychiatric disease despite extensive genetic heterogeneity.

Effect of Enhanced Information, Values Clarification, and Removal of Financial Barriers on Use of Prenatal Genetic Testing A Randomized Clinical Trial

By Miriam Kuppermann et al.

Prenatal genetic testing guidelines recommend providing patients with detailed information to allow informed, preference-based screening and diagnostic testing decisions. This study examines the effect of a decision-support guide and elimination of financial barriers to testing on use of prenatal genetic testing and decision making among pregnant women of varying literacy and numeracy levels. Full implementation of prenatal testing guidelines using a computerized, interactive decision-support guide in the absence of financial barriers to testing resulted in less test use and more informed choices.

Newborn Screening: Complexities in Universal Genetic Testing

By Nancy S. Green, Siobhan M. Dolan, and Thomas H. Murray

Newborn screening (NBS)—in which each newborn infant is screened for up to 50 specific metabolic disorders for early detection and intervention—is the first program of population-wide genetic testing. New technologies and new economic and social forces pose significant ethical and clinical challenges to NBS. Two primary

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"Epigenetics and the Maternal Body: Ethical and Social Dimensions"

Sarah Richardson, PhD, Harvard University
November 19, 2014, 4:00-5:00 pm

Epigenetics research situates the maternal body as a central site of epigenetic programming and transmission and as a significant locus of medical and public health intervention. Is there a potential for this research to heighten public health surveillance of and restrictions on pregnant women and mothers? How might this new research participate in the often-troubled history of notions of the supreme role of the mother in normal and pathological development?

Rm 405A and B, Educational Center, Irving Institute for Clinical and Translational Research, 10th Floor, Presbyterian Hospital (PH) Building, 622 W. 168th Street

In the Literature

Genetic Testing of Children for Diseases That Have Onset in Adulthood: The Limits of Family

challenges concern (1) accommodating clinical and ethical standards to rapid technological developments in NBS and (2) preparing public health systems to respond to the medical advances and social forces driving expansion of NBS programs. The authors describe and analyze these challenges through consideration of 3 disorders: phenylketonuria, medium-chain acyl-CoA dehydrogenase deficiency, and cystic fibrosis.

Toward a Jurisprudence of Psychiatric Evidence: Examining the Challenges of Reasoning from Group Data in Psychiatry to Individual Decisions in the Law

By Carl E. Fisher, David L. Faigman, & Paul S. Appelbaum

Psychiatry presents the challenge inherent in all scientific evidence of reasoning from group data to an individual case, which is termed the "G2i problem." But psychiatry, unlike many scientific fields that come to court, also confronts the G2i problem in its daily practice, since mental health professionals routinely diagnose and treat individuals based on aggregate data. Authors employ the G2i lens to examine the admissibility of psychiatric expert testimony, both as regards general research findings – or "framework evidence" – and the application of those general findings to specific cases – or "diagnostic evidence." G2i inference provides a useful lens by which the interactions of psychiatry and law can be better understood and managed.

Book Review

Interests

By George H. Hardart & Wendy K. Chung

Two recent policy statements, one from the American Academy of Pediatrics and one from the American College of Medical Genetics, reach very different conclusions about the question of whether children should be tested for adult-onset genetic conditions. This article analyzes these 2 different positions and suggests ways that the seeming conflicts between them might be reconciled.

Regulatory changes raise troubling questions for genomic testing

By Barbara J. Evans, Michael O. Dorschner, Wylie Burke, & Gail P. Jarvik

By Oct. 6, 2014, many laboratories in the US must begin honoring new individual data access rights created by recent changes to federal privacy and laboratory regulations. These access rights are more expansive than has been widely understood and pose complex challenges for genomic testing laboratories. This article analyzes regulatory texts and guidances to explore which laboratories are affected.

Gatekeepers or Intermediaries? The Role of Clinicians in Commercial Genomic Testing

By Michelle L. McGowan et al.

Several commercial laboratories offering genomic risk profiling have shifted to more traditional "direct-to-provider" (DTP) marketing strategies, repositioning clinicians as the intended recipients of advertising of laboratory services and as gatekeepers to personal genomic information. This

Principles of Psychiatric Genetics

Edited by John I. Nurnberger Jr. & Wade H. Berrettini Reviewed by Ming T. Tsuang & Joyce van de Leemput

Although the release of the first draft of the human genome sequence came in 2001 (bringing subsequent genome-wide association studies and a boost for genetics and genomics studies in identifying genetic causalities), the actual foundations for the field of psychiatric genetics were laid in the late 19th century. Despite the depth and duration of this field's history, identification of genetic components of psychiatric disorders has been lagging.

study assesses how and why early clinical users of genomic risk assessment incorporate these tools in their clinical practices and how they interpret genomic information for their patients. Authors concluded that the DTP service delivery model cannot guarantee that providers will have adequate expertise or sound clinical judgment.

To contribute a news item, an academic article, or an event on the ethical, legal, and social implications of psychiatric, neurological, and behavioral genetics research please email us**.**

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