Leading Change in Disability Rights, Ethics and Genetics:

Generating Conversation for Policy and Practice

Tuesday, November 19, 2019

8:30 - 10:15 a.m.

Panelists

Kara Ayers, Ph.D., OH-Cincinnati UCEDD

Sunday Francis, Ph.D., MN LEND

Leila Jamal, ScM, PhD, CGC, National Institutes of Health

Stephanie Meredith, KY UCEDD

Goals of Presentation

- Background
- Intersection of topic within AUCD network
- Future directions for AUCD network
- Centers' role in future activities

Genetics and the Disability Community: A complex history and a hopeful future

Kara Ayers, PhD
University of Cincinnati UCEDD and LEND
Center for Dignity in Healthcare for People with Disabilities

Lived Experience with Genetic Disabilities

"It is not enough that people with disabilities have a place at the tables where genetic technologies and policies that govern them are being discussed. Those with lived experience must also have a role in decision-making and leadership in these spaces."

History of Genetic Science and the Disability Community

OCTOBER 29, 1926.

HEREDITY IS BIG PROBLEM

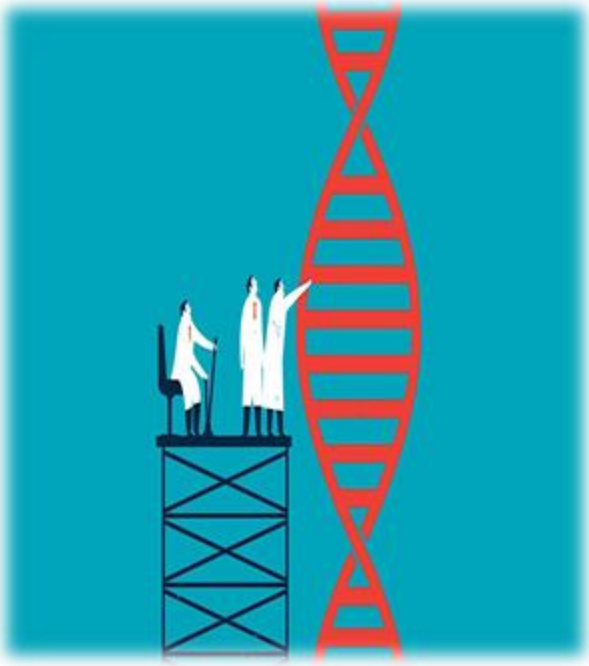
Home for Feeble-Minded Is Filled With Those Whose Parents Were Not as Carefully Selected as Dairymen Breed Cattle

- Eugenics was (is?) a social movement that argues the human race should be improved if "undesirable" traits were not passed on to future generations.
  - Sterilization is one result of the eugenics movement.

Genetics and Parenting with a Disability

- Parents and prospective parents with genetic disabilities have an increasing number of choices related to family-planning.

Is provider neutrality possible?



- Providers, like genetic counselors, aim to provide non-directive (or neutral) information.
- Medical education expects neutrality but is it realistic with the reality of ableism?

What's often left out of these discussions?

- The role of disability identity in people's decision-making about prenatal testing
  - Those who endorsed a social model of disability were more likely to pursue testing.
- The role of implicit bias among providers in recommending genetic technology

Considering Long-term and Short-term Consequences of Genetic Technologies

- Individual considerations and the need to include a wide spectrum of options
- Speed of advancement
- Actual access and availability of these technologies to most or all
- Unintended or unexpected consequences

Kara Ayers, PhD

Assistant Professor

Associate Director, UCCEDD

Cincinnati Children's Hospital Medical Center

University of Cincinnati University Center for Excellence in Developmental Disabilities (UC UCEDD)

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Genetic Research

(in Autism Spectrum Disorder)

Sunday M. Francis, PhD

University of Minnesota

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ASD Research: From Micro to Macrosystems

- Genetics
- Gut microbiome

- Functional coupling of brain and behavior (e.g., eye-tracking, imaging)
- Development of screening and diagnostic tools
- Biomarkers

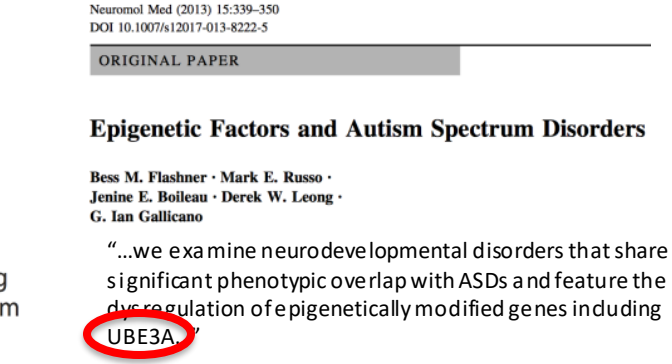
- Prevalence
- Differences across cultures & populations
- Research Registries (i.e., FIND Network, Simons Clinical Network)

- ASD research happens at the very small scale to the very large scale

ASD Genetics

Examples from the Literature

- Some examples of genes found to have connections to ASD



“Social” Neuropeptides

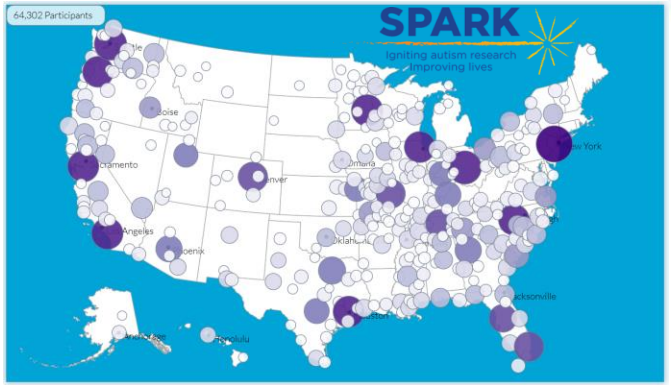
ASD Genetics + Biomarkers

- Research has connected these genes with diagnosis and other characteristics of ASD

Francis et al, 2016

Taking the Micro Macro

- Studying the genetics in large groups can be helpful
- Large genetic studies are underway
  - SPARK - a nationwide genetics study to collect samples from 50,000 families
  - Larger studies - increase statistical power and can address heterogeneity issues



From SPARK Website July 2017
Dark Purple 4,700-10,000

NEUROVIEW | VOLUME 97, ISSUE 3, P488-493, FEBRUARY 07, 2018

SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research

The SPARK Consortium

npj | Genomic Medicine

Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes

Pamela Feliciano,[✉#1] Xueya Zhou,[#2] Irina Astrovskaya,[#1] Tychele N. Turner,[#3] Tianyun Wang,[3] Leo Brueggeman,[4] Rebecca Barnard,[5] Alexander Hsieh,[2] LeeAnne Green Snyder,[1] Donna M. Muzny,[6] Aniko Sabo,[6] The SPARK Consortium, Richard A. Gibbs,[6] Evan E. Eichler,[3,7] Brian J. O'Roak,[5] Jacob J. Michaelson,[4] Natalia Volfovsky,[1] Yufeng Shen,[2] and Wendy K. Chung[✉1,8]

The Scientist

One Researcher's Journey

- Graduate Research
  - Neuroplasticity in the motor cortex
- Postdoctoral Research
  - Investigating ASD and other NDDs from genetics to big data modeling
  - Role of neuromodulation in the understanding of perinatal stroke
  - MN LEND fellowship

The Scientist

As Communicator & Advocate

- Begin early within training to connect beyond other researchers, ask socially-minded questions, and increase awareness
- Connecting with communities through informal and formal programs
- Understanding the language and images preferred by communities
  - 2018 MN State Fair Study
- Listening to communities

The Team

CAN | Lab

Converging Approaches to Neurodevelopment

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canlab.umn.edu

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Bernadette Gillick, PhD, MSPT, PT

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The Funding

NIH National Institutes of Health

Turning Discovery Into Health

SFARI SIMONS FOUNDATION AUTISM RESEARCH INITIATIVE

UNIVERSITY OF MINNESOTA

Driven to Discover℠

LEND | Leadership Education in Neurodevelopmental Disabilities

DEPARTMENT OF DEFENSE

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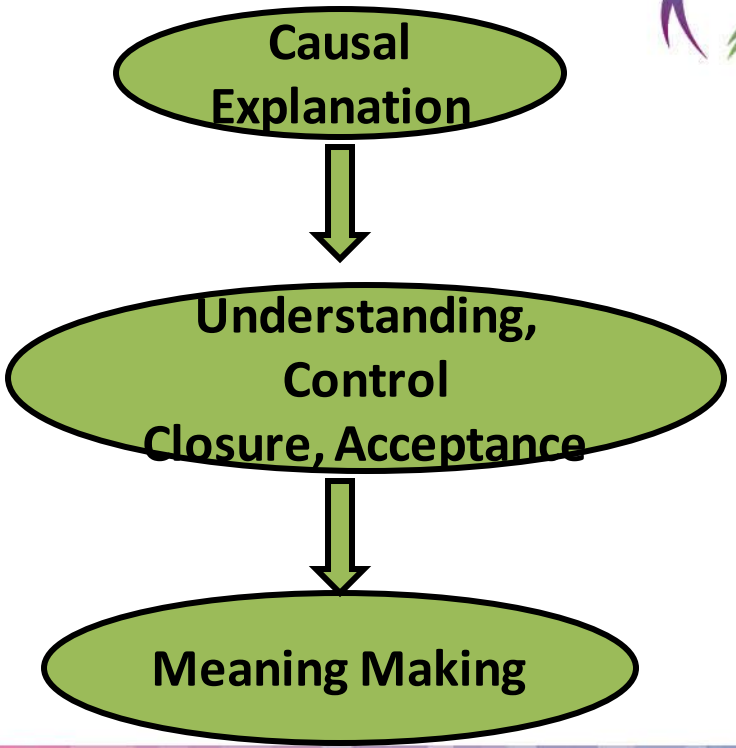
Clinical Practice

Leila Jamal, ScM, PhD

About Me

- Research genetic counselor at NIH
- Affiliated Researcher, NIH Department of Bioethics
- Previously worked in pediatric neurology and adult oncology

Two Ways to Use Genetic Information



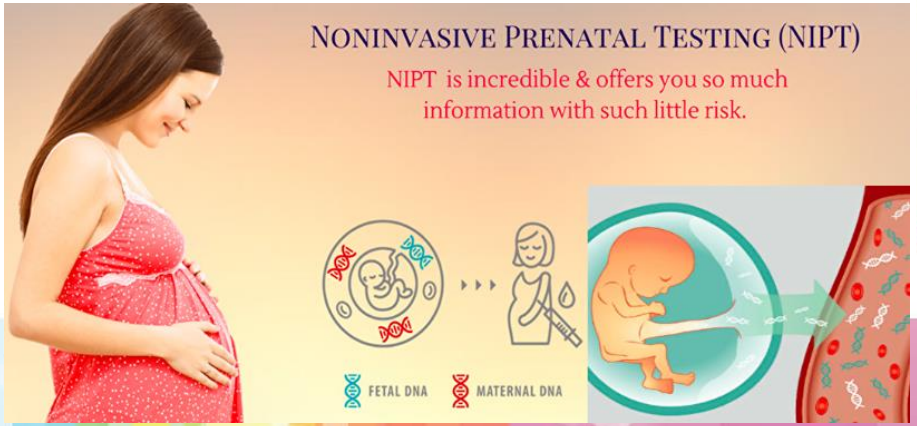

MAY 27, 2013
The IRS Mess / Syria's YouTube War / The End of Alimony
TIME
THE
ANGELINA
EFFECT
Angelina Jolie's double mastectomy puts genetic testing in the spotlight. What her choice reveals about calculating risk, cost and peace of mind
BY JEFFREY KLUGER & ALICE PARK
time.com

A Changing World

The New York Times

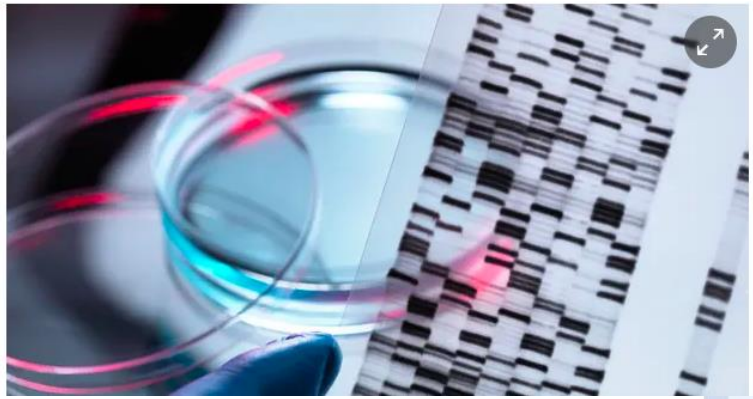
The Online Gene Test Finds a Dangerous Mutation. It May Well Be Wrong.

Third-party analysis of raw DNA is not as rigorous as that done in a certified laboratory. But many consumers don't understand that their results are not conclusive.



New gene editing tool could fix most harmful DNA mutations

'Prime editing' more precise than Crispr-Cas9, but still needs time before use on humans



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Genetics: *Original Research*

Noninvasive Prenatal Whole Genome Sequencing

Pregnant Women's Views and Preferences

Haley K. Sullivan, BS, Michelle Bayefsky, BA, Paul G. Wakim, PhD, Kathi Huddleston, PhD, Barbara B. Biesecker, PhD, Sara Chandros Hull, PhD, and Benjamin E. Berkman, JD, MPH

More than half of respondents wanted (at minimum) clear recommendations from clinicians when deciding which prenatal whole genome sequencing results to receive.

The most frequently cited reason for wanting medical prenatal whole genome sequencing results was "to prepare financially, medically, or psychologically for a child with special needs."

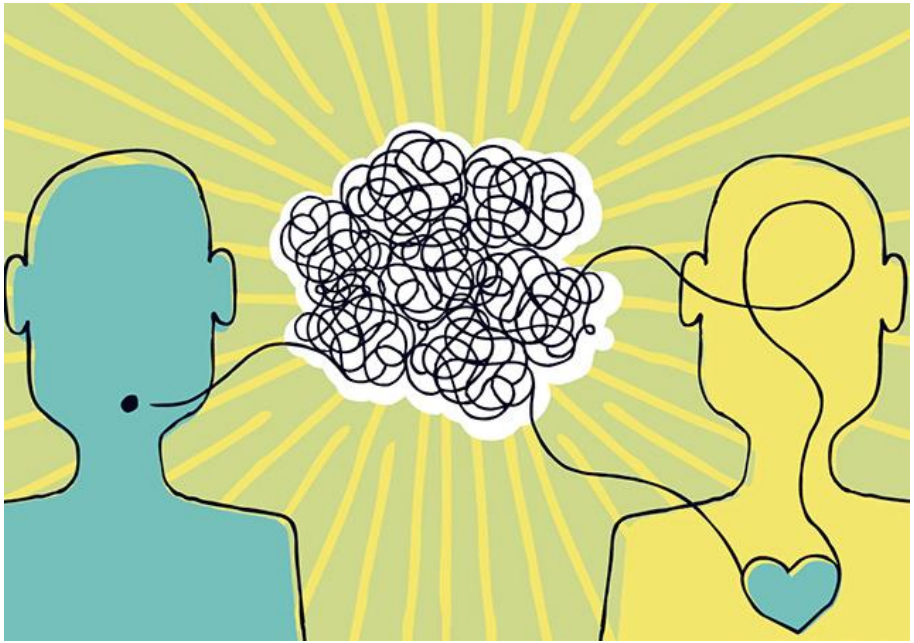
Questions

- How can we provide scientific expertise without reinforcing ableism?
- How can we simplify and explain test options without undermining patient autonomy?
- How can we engage relevant communities in our work, early and often?

Key Considerations

- What is the topic of genetic counseling?
- How complex is the genetic information?
- How potentially impactful is the genetic information?
- What is the nature of the relationship between counselor +patient?
- What professional role is the counselor in?
- What are the interests + contributions of family members in the counseling process?

Above all else, listen…



Family Policy and Practice

Stephanie Meredith,

Director, The Lettercase National Center for Prenatal and Postnatal Resources, University of Kentucky's Human Development Institute

The Diagnosis Experience

First Point on the Life Course

Research by Skotko, Berrier & Levis

Disparity Between Genetic Technology & Educational Infrastructure

Prenatal Screening Technology

- Industry and federal funding (FASTER study)
- Rapid adoption (cell-free DNA), profit driven
- Marketing mechanisms
- Limited regulation

Educational Infrastructure

- No funding for Prenatally & Postnatally Diagnosed Awareness Act
- Developing solutions is slow & deliberate w/consensus
- Dissemination challenge
- Limited access to genetic counseling

Outcomes for Families

- Nelson-Goff Study (2013) 35% Negative and 11% Positive

"I don't ever remember her saying 'sorry' at any point. She gave us the facts and resources … The genetic counselor at the MFM group was also very kind … She gave us some of the common health concerns, resources and hope (ie. talked about how things have improved so much for individuals with Ds since even 10 years ago and encouraged early interventions). Overall our experience was very positive."

Outcomes for Families

"It was 8 days before we had the appointment...an excruciating 8 days where I barely slept, cried every time I was alone, and did countless (unhelpful) google searches to get any information I could, both medical and related to living with a child with DS. I wish there would have been more info available to me and my husband, because a lot of what is available on Google is scary and not necessarily accurate."

The Challenges

- Making sure patients accurately understand the technology and have given informed consent
- Moving conversations about ethics from theory to practice. *Ethics and best practice should drive the engine instead of potential profit or excitement about technology*
- Embedding the disability rights perspective into medical and genetics culture through resources, training, culture, and policy
- Cost-benefit justifications

What is at stake is the future fabric of humanity.

AUCD Network Can Be the Power for Change

- Provide forums for interdisciplinary conversations about genetics, bioethics & disability issues

AUCD Network Can Be the Power for Change

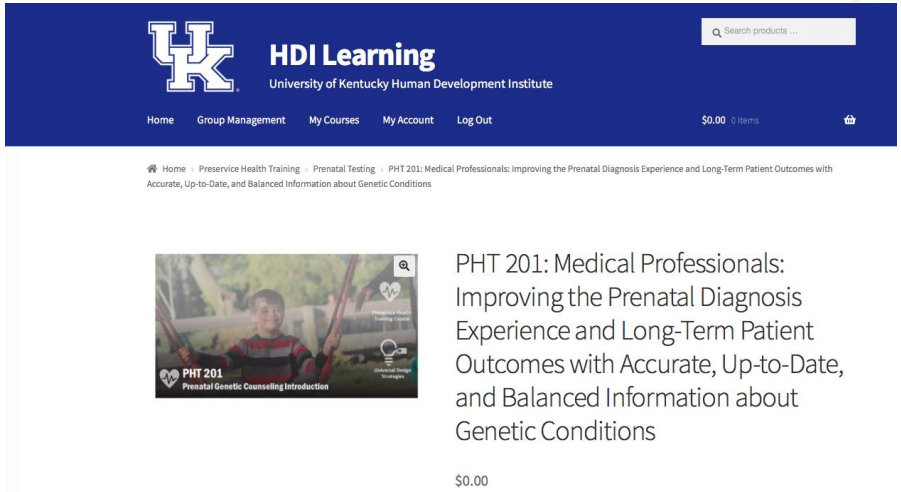
- Stay current & educate young professionals about the intersection of genetics & disability identity in the news & history

AUCD Network Can Be the Power for Change

- Develop educational resources and partner with advocacy & medical organizations for dissemination

AUCD Network Can Be the Power for Change

- Develop & disseminate training for genetics and medical professionals that is embedded with the disability rights perspective



AUCD Network Can Be the Power for Change

- Be a policy leader where genetic and disability issues intersect

DISCUSSION

A Discussion of Ideas

How do we intersect our personal perspective in our work?

What are the emerging technologies and concerns?

What does authentic engagement of people with disabilities and families mean for this field?

How do we grown from history and become more neutral about disability?

A Discussion of Ideas: AUCD

Are there future opportunities for our Centers?

- Research
- Training
- Policy

How can we (AUCD, Centers) engage the disability perspective?

Identify next steps for your Center, AUCD network.

Questions