Health Policy, Management & Evaluation
UNIVERSITY OF TORONTO

Identifying and communicating meaningful genetic results in ASD diagnosis & screening

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Overview

- Genetic research results in ASD
  - Placing genetic information in context
  - Considering the nature of the information
  - Obligations to provide updated information?

  - Genetics in clinical diagnosis
  - Population screening – a role for genetics?

Complex research context …

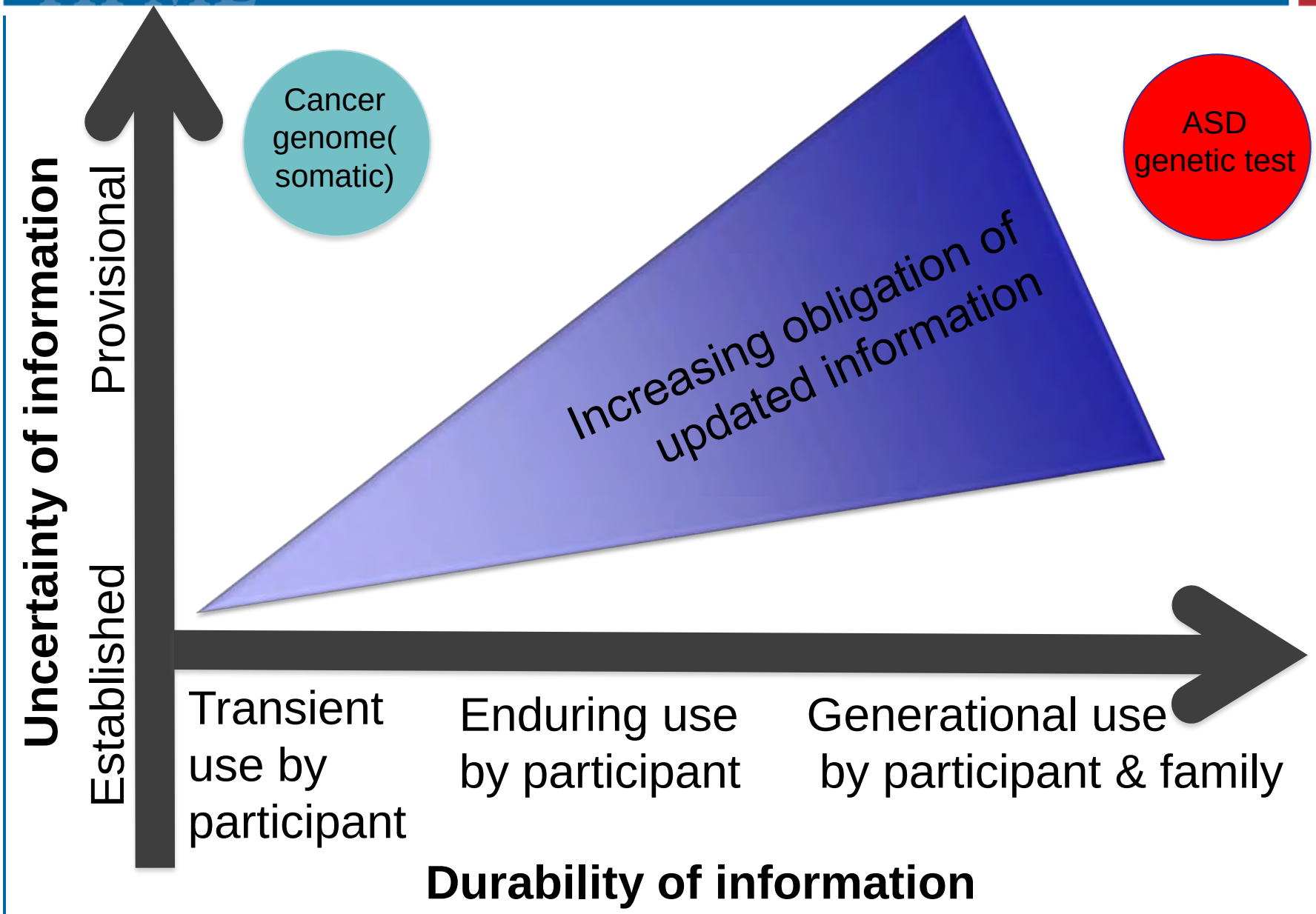
- Families with ASD diagnosis need care/info
  - Uncertainty of ASD
- Research can be a resource
  - Access to specialists
  - Access to diagnostic assessments
  - Access to information

ASD genetic results: meaning

- … are a relatively small part of overall needs
- Meaningful information would be valued
  - Instrumental value (extrinsic):
    - Reproductive risk
    - Personalized treatment
  - Non-instrumental value (intrinsic):
    - Understanding ‘why?’
    - Seeking legitimacy – a ‘real’ disorder

ASD genetic results: reporting

- Researchers' judgments to report …
  - Informed by science
  - Informed by values
  - Informed by interests
  - Informed by disciplinary norms/ epistemological assumptions
  - Informed by ontological assumptions


Uncertainty of information
Provisional
Established
Cancer genome(somatic)
ASD genetic test
Increasing obligation of updated information
Transient use by participant
Enduring use by participant
Generational use by participant & family
Durability of information

In sum …

- Genetic research serves many needs
  - For information
  - For care

  → Genetic information is a part, and not the whole
- Genetic information in ASD
  - Is highly provisional
  - Is highly durable

  → Obligation of information update

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ARTICLE

Consensus Statement: Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies

David T. Miller,[1,*] Margaret P. Adam,[2,3] Swaroop Aradhya,[4] Leslie G. Biesecker,[5] Arthur R. Brothman,[6] Nigel P. Carter,[7] Deanna M. Church,[8] John A. Crolla,[9] Evan E. Eichler,[10] Charles J. Epstein,[11] W. Andrew Faucett,[2] Lars Feuk,[12] Jan M. Friedman,[13] Ada Hamosh,[14] Laird Jackson,[15] Erin B. Kaminsky,[2] Klaas Kok,[16] Ian D. Krantz,[17] Robert M. Kuhn,[18] Charles Lee,[19] James M. Ostell,[8] Carla Rosenberg,[20] Stephen W. Scherer,[21] Nancy B. Spinner,[17] Dimitri J. Stavropoulos,[22] James H. Tepperberg,[23] Erik C. Thorland,[24] Joris R. Vermeesch,[25] Darrel J. Waggoner,[26] Michael S. Watson,[27] Christa Lese Martin,[2] and David H. Ledbetter[2,*]

The American Journal of Human Genetics *86*, 749–764, May 14, 2010

Genetics in clinical diagnosis

- As in research context
  - To explain causation in idiopathic cases

  →Durable information
- As in research context
  - Complex professional judgments

  →Provisional information

→ Obligation of updated information

Population screening – genetics?

- No current role for genetic testing
- But, likely to be complex addition
  - May increase diagnosis/ overdiagnosis challenge
- CF NBS instructive
  - CFTR vs. other biomarkers in pre-symptomatic diagnosis
  - The problem of “borderline” babies

Draft for Consultation

NHS

Screening for Autism Spectrum Disorders in Children below the age of 5 years

A draft report
for the UK National Screening Committee

Dr Martin Allaby
Dr Mohit Sharma

4 July 2011

- “Screening differs from routine clinical care because the process is initiated by the state or professionals, not by patients or parents. … In the context of screening, it is not appropriate for professionals or the state to initiate contact with the public unless there is very strong evidence that available treatments are effective.”

ASD and ELSI

- Avoid unnecessary exceptionalism
  - There are differences but also similarities
  - Evidence standards for common, not ultra-rare, disease
- Research on genetic tests in ASD
  - Comparing receipt to non-receipt of genetic info (interpretation and use in context)
  - Updated information: when required; how provided; how paid for

Many thanks

Parents and researchers who have provided information and insight

Robin Hayeems, PhD, HPME
Colleagues in the AGP
Genome Canada

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