



# Identifying and communicating meaningful genetic results in ASD diagnosis & screening

Fiona A. Miller, PhD

Associate Professor, HPME

Joint Centre for Bioethics

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[www.hpme.utoronto.ca](http://www.hpme.utoronto.ca)

# 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 ...

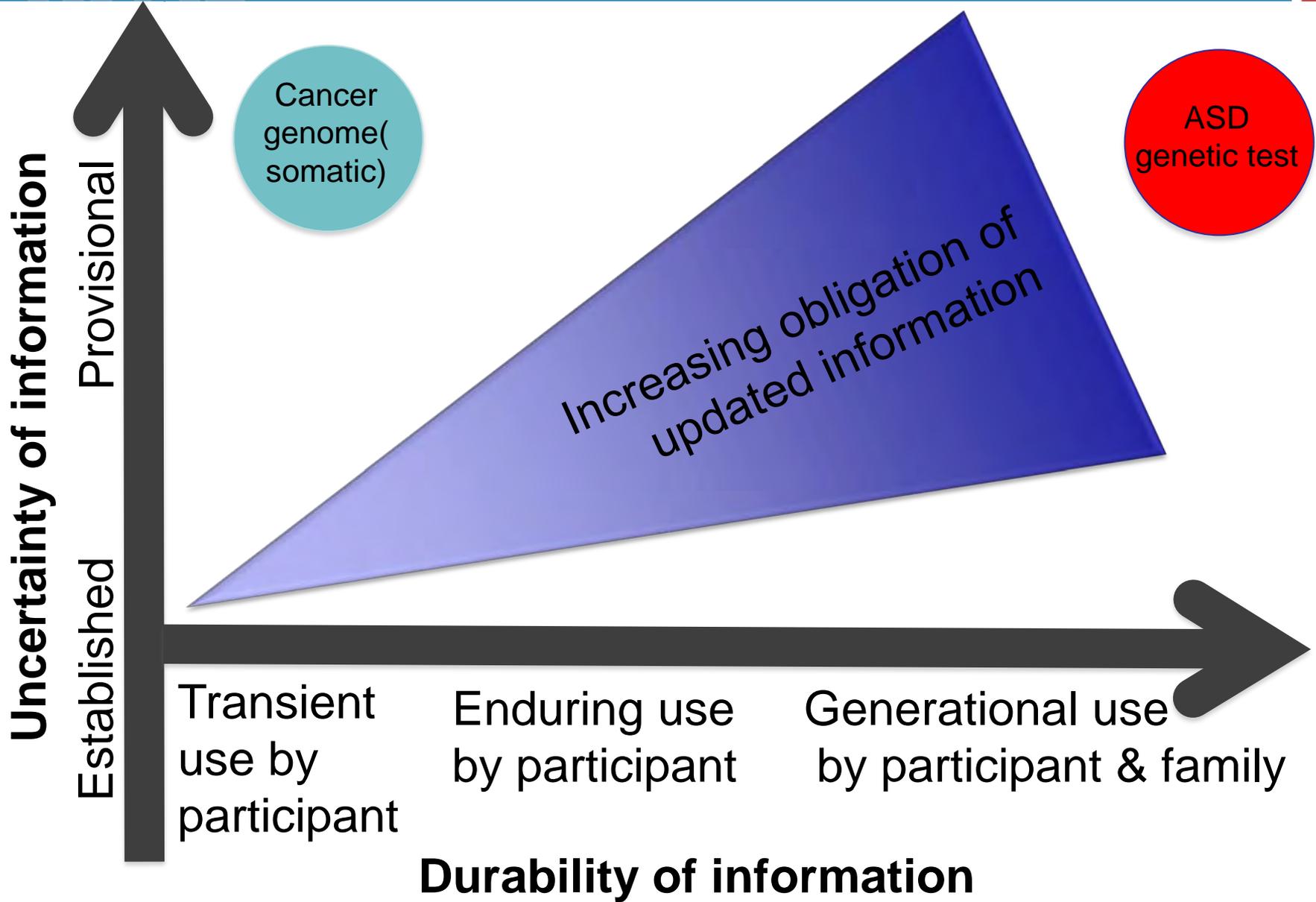
- Families with ASD diagnosis need care/info
  - Uncertainty of ASD
- Research can be a resource
  - Access to specialists
  - Access to diagnostic assessments
  - Access to information

# 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



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

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

# 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

- “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

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Genome Canada

HPME

