

Ethical Considerations for Responsible Child-Specific Neurotechnologies

Paul S. Appelbaum, MD

Dollard Professor of Psychiatry, Medicine & Law

Columbia University

Background

- I will draw on our study of reactions of parents of autistic offspring to genetic test results for autism
- Why did we do this study?
 - Probands and families were thought to be susceptible to:
 - Genetic essentialism (i.e., belief that people's essential characteristics are embedded in their genes), and
 - Genetic determinism (i.e., belief that genes alone determine human outcomes)
 - Hence, concern that positive genetic results would lead people to see their children as “defective” and their conditions as hopeless



- SPARK "Simons Foundation Powering Autism Research" aims to understand the genetic causes of autism
 - To date, includes over 100,000 people with autism and 175,000 family members
 - Over 45,000 probands and their parents have had exome/genome sequencing
 - Extensive clinical phenotyping through parental interview and chart review
 - Formal developmental assessments
 - Return of definitive diagnostic results to families with genetic counseling
 - PI: Wendy Chung, MD, PhD

Methods

- Survey sample of parents (also adult and adolescent probands) before and 1 month and 1 year after return of genetic results, either diagnostic or non-diagnostic
- Compare change in scores on measures between groups that did and did not have causal findings
- Qualitative interviews used for in-depth exploration of responses by families that received diagnostic genetic results

Sample

- 847 parents (37% response rate)
 - 148 received diagnostic results; 97% *de novo* variants
 - 699 told no current diagnostic findings
- 44.1 yrs. (mean)
- 83.6% White
- 65% college-educated
- 48.4% annual income >\$100,000

Lesson 1: Test Results Can Make Parents More Tolerant But Also Less Hopeful

- Many parents described now having greater patience with their children, because the genetic results somehow meant that their child's difficulties were "real" and "not their fault"

"It definitely solidified granting her a little more grace. So, when she drops her milk four times, she really doesn't mean to. She just has horrible fine and gross motor skills."
[Mother 12]

On the Other Hand...

- Such lowered expectations of the child's future self could also reduce hope and generate a sense of sadness.

“I always felt as though she had the potential to be some kind of ‘normal’ as they say. And the test results just wiped out that little inkling and hope that I had always held on to...And that's sad.”

[Mother 13]

- Thus, variation identified via laboratory testing concretizes a child's limitations for parents, puncturing what are often states of denial—with mixed impact

Lesson 2: Test Results Can Reduce Parental Self-blame

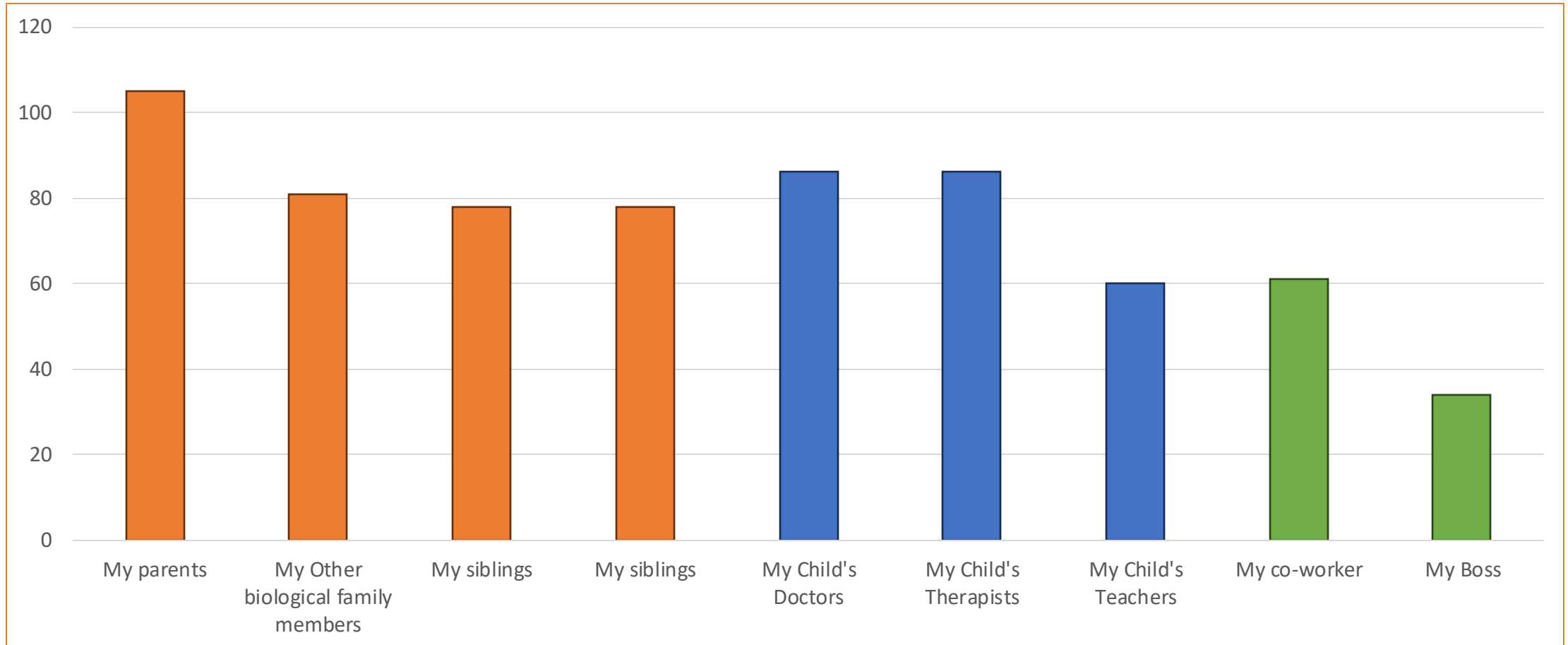
- Parents with diagnostic genetic results perceived more of a role of chance in their child's autism and less of a role for causes that might implicate their own actions, including exposures during pregnancy
- As one father said:

“It was a relief, actually, because neither my wife nor I carry the mutation, so it kind of took a little pressure off the thinking that we have contributed to his autism. Because in the general population, the occurrence of autism is higher in the scientist and medical population.” [Father 4]

But...Never Underestimate the Potential for Parents to Blame Themselves

- Yet ambiguities about what caused the de novo pathogenic variants also left room for continuing self-blame:
“I took an antibiotic early in my pregnancy before I knew I was pregnant. Just those niggling fears: ‘Did that cause it?’ I might have had a glass of wine before I knew I was pregnant...Could that have caused it?” [Mother 1]

Nonetheless, Results Were Shared Surprisingly Widely



Why Share? Test Results May Affect Others' Views of Parental Responsibility

“My parents are a lot older, so still kind [of] have the mentality of, ‘It’s because you spoil him...’ But I can now say, ‘Hey, it’s not just because of my mothering.’ So, the genetic test has helped with that...[My parents’ views are] still hard to deal with sometimes, but at least they have a better understanding.” [Mother 1]

Lesson 3: Many of the Impacts of Data From New Technologies May Derive From Misunderstandings of Their Implications

- Behavioral and psychomotor difficulties experienced by children in this study were no less “real” and no more “their fault” before genetic testing than after
- Children’s likely achievements in education and relationships were no different before the testing than after
- Parents were no more to “blame” for their child’s condition before the testing than after
- Yet, we found an impact on all of these views, despite universal post-test genetic counseling—probably reflecting essentialist/deterministic views of genetics

Potential Implications of These Findings for Child-Specific Neurotechnologies

- Concrete results from neuromonitoring, neuroimaging and other technologies could affect parents' perceptions of the "reality" of their child's difficulties, leading to increased tolerance but also decreased hope
- Such testing might also impact parental views of their own "responsibility" for their child's condition—a reason why parents may very much want to know what the test results show
- We cannot assume that parents share our scientific understanding of the significance and future implications of test results

And hence...

- Education of parents (and older children) about the nature of the findings and especially the causal inferences (if any) that can be drawn from them and the prognostic implications that might be deduced would appear to be an important part of any research effort in this field
- Since conclusions drawn by parents may be idiosyncratic, their understanding should be assessed and misinterpretations addressed
- But they will very much want to know the answer to the question: “What did the test show?”