Outline

- My perspective and research
- Research ethics involving siblings
- Risk of therapeutic misconception
- Presuming competence and hearing autistic voices
- Future horizons and hope
My Perspective and Research

• Trained in genetics and epidemiology
• Funding from NHGRI
• Parent of two boys, one with autism
• Ethical issues in genetic research on complex traits, including autism
• Ethical issues in autism risk communication
• Ethical issues in whole genome and exome sequencing (research and clinical care)
Research ethics involving siblings of children with autism
Research on Children in Autism

• Studies are often family based
• Recruit affected children and unaffected (and undiagnosed) children
• Range of expectations/involvement
Research in Children

Federal requirements (OHRP):

– Research in children “can only be justified if the level of risk entailed in the research is very low, or if there is the potential for direct benefit to the child by participating in the research project”

– Research that does not fit these criteria may be allowed “if the research is likely to yield generalizable knowledge about the subject’s disorder or condition and the risk of research represents no more than a minor increase over minimal risk” for that group of patients
Benefits to Siblings

• Unaffected siblings do not receive direct benefit from research themselves, although their siblings and families may receive such benefits.

• Indirect benefits: access to early screening for ASD-related phenotypes, positive feelings/altruism (in the future).

• Possibility for unexpected findings that must be returned.
Risks to Siblings

- Minimal risk: giving a blood sample, parental questionnaires
- Psychological Risks
  - Interview study of adult siblings of children with Ataxia Telangiectasia who were enrolled in linkage studies when they were children (Fanos 1999)
  - Fear of blood draw, apprehension about carrier results, confusion around possibility of having A-T
  - Visit described as “a highly charged event” in their lives
- Other risks?
  - DNA sequence and phenotypic data shared in a database like dbGAP? behavioral assessment disclosure?
Risks to Autonomy

• Parental-child disagreement about participation:
  – Survey of 117 parent-child pairs where the child was enrolled in clinical research or receiving clinical care for asthma or cancer
  – 90.5% of pediatric patients believed they should be involved in making research enrollment decisions
  – 61.5% of adult parents believed that the children should be involved
  – Implications for healthy children enrolled in research
Newer Autism Sibling Research

• Pubmed search on “autism” and “siblings”
  – Endophenotypes
    • 3 articles on non-autism phenotypes or endophenotypes (complex immune dysfunction, dysfunctions in the prefrontal cortex, emotional problems)
  – Genotype-Driven Research
    • Phenotypic features of siblings of individuals with a CNV at chromosome 16p11.2, described as “apparently healthy” to “meeting criteria for ASD
What can we do?

• NOT a call to stop or limit research on siblings. There are good scientific reasons for these kinds of studies.
• Need to weigh risks and benefits specifically for unaffected or undiagnosed siblings
• Need to tailor consent forms and consider reconsent as the research objectives change to specifically address the unique risks to “healthy siblings,” opportunity for discussion
• Require reconsent of healthy siblings at age 18, especially for genetic studies
  – In a recent study, 46% of participants would want to be recontacted as adults for continued research use of samples collected when they were children (Goldenberg 2009)
• Consider potential burdens of future genotype-driven recontact
  – (McGuire & McGuire 2009)
Risk of Therapeutic Misconception
What is therapeutic misconception?

- A fundamental confusion among researchers and/or research participants between the goals of research, or generalizable knowledge, and the goals of clinical care, or improving the health of an individual person
What is therapeutic misconception?

- Needs to be framed in the current context of diagnosis and treatment of autism
  - Academic medical centers and hospitals
  - Researchers are also clinicians and wear both “hats”
  - Backdrop of a medical system that provides little for treatment
  - Desperate parents looking for guidance they can’t get from their pediatrician or through school and government services
  - Challenge in even getting access to services like ABA
Autism Research at Academic Medical Centers

- Parents are often invited to participate in research studies
- Involvement may be peripheral to their care
- Studies may provide services/evaluations for free, or on a faster timeline (e.g., diagnostic reports, genetic testing, access to therapies, provision of free behavioral interventions)
- Some families seek out studies for this reason
- A good thing: respect, reciprocity, access
Autism Research at Academic Medical Centers

• A bad thing?
• Families may misunderstand the procedures, risks and benefits of research
• Families may believe research will provide a direct benefit to their child
• This affects perception of risks and benefits
  – Study of parents whose children were enrolled in a randomized placebo controlled trial of risperidone. 25% were unaware that the treatment was randomized and not personalized. (Vitiello 2005)
Autism at Academic Medical Centers

- We need empirical research about:
  - whether parents of children with autism experience therapeutic misconception
  - what aspects of research and what kinds of studies
  - what to do about it

- Possible approaches:
  - Use of a qualitative interview guide focusing on understanding of the purpose of research in juxtaposition with motivation for participating (Kim 2009)
  - Focus on recruitment strategies and target populations
  - Specific discussion in informed consent
  - Discussions with family about the delineation of roles (clinical vs research) for the providers
Other Emerging Ethical Challenges

• Diagnosis:
  – Variability, how it is delivered, how families process over time, parental depression
• Access to diagnosis and care/treatment/intervention
• New DSM criteria and shifting diagnostic boundaries
• Emerging role of genetics, especially use of arrays and exome sequencing, in diagnosis and screening
• Risk communication around genetics and environment
• Respecting autonomy of children and adults with autism
“I didn’t want to think of Benji in terms of syndromes and categories and labels and diagnoses, in terms of his performance on tests and evaluations, in terms of his differences from a perceived norm or imagined ideal. One of the questions my experience with Benji raised for me is: how do you value your child in a culture whose benchmarks for achievement and whose standards for evaluating and assessing kids are so out of line with your own values and who your child is.”

-Priscilla Gillman
Communication, hope and respect

“There is nothing less romantic, literary, or lyrical than the language of pathology, diagnosis, symptom checklists. As I read through these checklists over and over again I was struck by the harshness, the crudeness of the terminology.”

-Priscilla Gillman

How do we offer hope and build resilience?
Questions and solutions

Questions
• Why did this happen?
• Is it my fault?
• If it’s not my fault, who or what is to blame?
• Can it be cured? Should it be cured?
• What will his/her life be like, as a child? As an adult?
• What should I do?
• How do I support myself and my family as we navigate this journey?

Solutions
• Lots of debates about causation, not a lot of answers
• Debates hinge on fear, anxiety, blame, trajectory, hopes and fears
• Lack of prognostic information, positive examples
• Lack of clear guidance about what to do
Autism Voices
Autistic Advocacy and Social Media

Question for autistic community members on executive functioning: Did you frequently forget critical school supplies, etc.? How did your parent support you (or not)? What are your recommended support strategies for parents of "forgetful" kids? –SR
A study published last week found that the brains of autistic children show abnormalities that are likely to have arisen before birth, which is consistent with a large body of previous evidence. Yet most media coverage focuses on vaccines, which do not cause autism and are given after birth. How can we help people separate real risks from false rumors?

Over the last few years, we've seen an explosion of scientific publications mentioning autism in 1943, over half have come since 2008.
PRESUME COMPETENCE BELIEVE IN YOURSELF
TREAT YOURSELF LIKE YOU ARE YOUR OWN BEST FRIEND
BREATHE DO YOUR THING
HUG THOSE YOU LOVE...EVERY SINGLE DAY
CHALLENGE THE PARADIGM TAKE A MILLION PICTURES
PLAY TRUST YOUR GUT
BELIEVE IN RANDOM ACTS OF SILLINESS KEEP ALL RECORDS AND REPORTS
BE INFORMED THINK IN PARALLEL
APPRECIATE THE BEAUTY AND STRENGTH IN OUR DIVERSITY TURN IT UP TO 11
QUESTION EVERYTHING RESEARCH
LOVE DEEPLY
OWN YOUR DIFFABILITY BUST A MOVE
KNOW THAT TYPICAL IS OVERRATED LEAN INTO IT
EMBRACE THE TEACHABLE MOMENTS LAUGH LAUGH LAUGH LAUGH LAUGH
CALL ON A FRIEND SLEEP WHEN YOU CAN
WE’RE MORE ALIKE THAN WE ARE DIFFERENT ALL OF A KIND

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WHAT DOESN'T KILL YOU MAKES YOU CRANKY STRONGER
STRONGER PISSED OFF STRONGER
GRUMPY STRONGER
(IT MAY TAKE A WHILE, BUT YOU'LL GET THERE!)

Seattle Children's®
HOSPITAL • RESEARCH • FOUNDATION
UW Medicine
SCHOOL OF MEDICINE
Autistic Self-Advocacy
“Since Owen turned three, the daunting, never-enough demands of autism have remained inelastic, bottomless. Not knowing what really works, or helps, makes identifying the inessentials all but impossible. You try everything. And we have: from changing his diet to gluten-free to auditory processing, where he spends hours doing high-speed computer tests while different noises ring in his ears. Lots of families run themselves into bankruptcy. Though divorce rates are no higher than the norm, families tend to either break apart of pull more tightly together. But every family knows the crush of constant pressure.”

-Ron Suskind, Life, Animated
Parenting and Advocacy

http://bcove.me/723bw3ay
“Presume Competence” – What Does That Mean Exactly?

Posted on March 7, 2013 by arianezurcher | 46 Comments

When my daughter was first diagnosed at the age of two and a half, presuming competence was not a concept I was ever told about or had heard of. And even if someone had suggested we do so, I’m not sure I would have fully understood what that meant exactly. So what does “presume competence” really mean? And how and why should we carry out a presumption of competence?

In an interview, Douglas Biklen explained: “Assume that a child has intellectual ability, provide opportunities to be exposed to learning, assume the child wants to learn and assert him or herself in the world.”

A key component to presuming competence is to become aware of the prejudice that currently exists regarding autism and how these ingrained beliefs harm not just our children, but ALL Autistic people. Like any prejudice, based on layers and layers of misinformation, misperceptions, and misunderstandings, we must be willing to acknowledge our own “beliefs” before we can begin to deconstruct them.
“No horizon is so far that you cannot get above it or beyond it.”

-Beryl Markham
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