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Biology-Based Intermediate Phenotypes in Autism: Hope or Hype?

Nancy Minshew, M.D.
Professor of Psychiatry & Neurology
University of Pittsburgh School of Medicine
minshewnj@upmc.edu
The Hope

- Individualized diagnosis & treatment:
  - highly accurate
  - comprehensive characterization
  - accounts for variability in expression
  - accurately predicts trajectory
  - accurately predicts response to treatments
  - defines risk before onset
  - identifies mechanisms that lead to improved outcomes
  - captures change over time
The Challenge

- Diagnostic precision: reliability in application of DSM criteria among clinicians is .3-.4
- Highly complex, highly variable, low prevalence disorder
- Response to treatments highly variable and unpredictable
- Behavioral treatments intensive, global and long
- Pharmacologic-adjunctive, substantial adverse reactions
- Environmental engineering grossly unappreciated
- Few community interventions have evidence base
- Large gaps in intervention
The Nominees

- The search for subtypes
- At first behavioral & uni-dimensional
- Endophenotypes- biologically based, single features
- Imaging: structural, functional, other derivatives that reflect network/systems function
- Genotypes, microRNAs & proteomics
- Progress highlights potential for future but also complexity; feasibility clear, time frame extended
The Hype

• Over interpretation of findings in scientific literature- the push to tell the great story which require leaps; under acknowledgement of complexity and future steps needed to achieve ultimate goal
• The Press- the 30-60 sec drama
• The Circulating Myths
• The often costly, new treatments that spring up and purport major progress without evidence base
• The explanation is simple. The brain is not simple and the “answer” will not be simple.
The Help

- Definition of impairments and abilities much improved
- DSM 5.0 will provide platform for centralizing existing advances documented with text, references, and video examples to improve identification and characterization
- Autism Speaks and SFARI websites have provided centralized knowledge and tools supported by extensive expertise
- Research has made extra-ordinary advances in understanding autism at all levels
- Translation to treatment is evolving
The Speed Bumps

- Still need a centralized and integrated repository of established advances that constitute standard of practice, what issues are under construction
- Relevant to age x ability subgroups
- Specialized for parents, teachers, pediatricians, neurologists, psychiatrists, speech pathologists, occupational and physical therapists, peers, neighbors, etc.
- A standardized method for storing key information across a lifetime