



IMFAR 2012 Toronto:
Biology-Based Intermediate Phenotypes in Autism:
Hope or Hype?

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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
- Pharmacologics- 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 life time