2009 IACC Scientific Workshop:

Updating the Strategic Plan for Autism Spectrum Disorder (ASD) Research

September 30 - October 1, 2009

Bethesda, Maryland

Panel 2: How can I understand what is happening?

- Clinical Experts
  - *Pauline Filipek, Sarah Spence*
- Research Experts
  - *David Amaral, Emanuel DiCicco-Bloom*
- Family/Personal Experts
  - *Ashura Buckley, Denise Resnick*
- Coordinated by IACC Liaisons
  - *Alison Singer, Edwin Trevathan (Cathy Rice)*

Research Opportunities in the 2009 IACC Strategic Plan

- Multi-disciplinary, longitudinal, bio-behavioral studies of children, youths, and adults beginning during infancy that characterize neuro-developmental and medical developmental trajectories across the multiple axes of ASD phenotype and identify ASD risk factors, subgroups, co-occurring symptoms, and potential biological targets for intervention. Such studies could include:
  - *High-risk siblings of children, youths, and adults with ASD, children without a family history of ASD, and typically developing children.*
  - *Multi-disciplinary assessments of brain imaging, metabolic and immune markers, microbiomics, electrophysiology, and behavior.*

Panel 2: Research Opportunities (Continued)

- Research on females with ASD to better characterize clinical, biological and protective features.
- Human and animal studies that examine immune, infectious and environmental factors in the occurrence of ASD.
- An international public-private collaboration to expand current postmortem brain and other tissue resources (*e.g.,* skin fibroblasts) to increase the acquisition, quality, type and availability of biomaterials relevant to the pathology of ASD.
- Research on the unique strengths and abilities of people with ASD.

Panel 2: Noted Research Gaps in the 2009 IACC Strategic Plan

- Studies focused on underlying biology of co-occurring *conditions* (*e.g.,* seizures, sleep disorders, familial autoimmune disorders)
- Studies focused on underlying biology of co-occurring *syndromes* (*e.g.,* Rett, Fragile X, TSC) looking at similar or divergent pathways
- Clarification that the recommended study of females also includes studies focused on sex differences – encompassing clinical and basic research to explain the 4:1 ratio

Panel 2: Noted Research Gaps in the 2009 IACC Strategic Plan (Continued)

- Expansion of subgroup study to highlight
  - *Nonverbal (or minimally verbal) autism*
  - *Impaired sensory-motor function*
  - *Racial minorities*
  - *Regressive autism*
- Collection of a variety of biomaterials to study the "biological signatures" in patients with ASD
- Studies focused on Mitochondrial markers and metabolism using biologic samples

Panel 2: New Opportunities for Advancing Research and Knowledge about ASD

- Collection and *in vitro* study using new technology to induce pleuripotential stem cells (IPSC) for neuronal differentiation from skin fibroblasts
- Studies that associate specific genotypes with functional or structural phenotypes
- Expansion of the biobanking initiative to include other biomaterials (not just brains) in large studies with well-characterized individuals with ASD.
  - *This may be most efficiently done in association with ongoing, large scale studies (e.g. High Risk Baby Sibs, National Children's Study, CADDRE, ATN)*

Panel 2: Suggestions for Prioritizing Short- and Long-Term Research Objectives

- Skin fibroblasts for creation of IPSC and neuronal cell lines
- Biology underlying Non Verbal autism
- Studies that associate specific genotypes with functional or structural phenotypes
- Research on underlying biology of co-occurring syndromes (Rett, Fragile X, TSC)
- Research on co-occurring conditions, particularly familial autoimmune disorders (*e.g.,* diabetes mellitus, celiac, rheumatoid arthritis), seizures, sleep disorders

Panel 2: Suggestions for Prioritizing Short- and Long-Term Research Objectives

- Expand subgroup studies to highlight sensory-motor dysfunction, racial minorities, regression
- Studies focused on mitochondrial and metabolic markers
- Collect a variety of biomaterials to study “biological signatures” in patients with ASD
- Studies on females should include clinical and basic research that defines sex differences that contribute to the 4:1 ratio

Panel 2: Summary & Discussion Questions

- Panel was concerned that some research opportunities already included in the plan had received little or no funding (brain and other tissue biobanks, studies of females)
- Studies should build the evidence base that informs treatment, services and supports, i.e., be translational
- Need to improve efforts to effectively communicate current biology/risk factor research efforts to the autism community