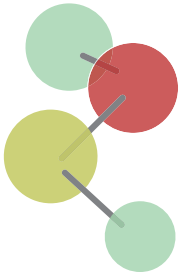

Autism Science Foundation

Profound Autism Research

Strategic Plan

searching. solving. sharing.

GOAL 1:

UNCOVER BIOLOGICAL MARKERS TO UNDERSTAND THE CAUSES OF PROFOUND AUTISM

- **Studies to discover the causes of profound autism,** including both genetic factors and a wide spectrum of environmental influences which may support resilience or confer susceptibility to symptoms of profound autism. These insights will help identify new pharmacological and medical interventions to ameliorate symptoms of profound autism as well as prevent profound autism and improve quality of life.

- **Research to uncover the neurobiological etiologies of catatonia, aggression and self-injury.** Aggression, catatonia and self-injury are highly prevalent in this population and have a major impact on the quality of life for the individual and their caregivers. These comorbidities can unfortunately be fatal, and our lack of knowledge about their etiology has limited our ability to prevent them. New interventions will enable more individuals to live in the community and participate in community activities.

GOAL 2:

DEVELOP PERSONALIZED TREATMENTS AND INTERVENTIONS FOR PEOPLE WITH PROFOUND AUTISM

- **Research to develop personalized treatments for profound autism using organoid and stem cell models.** Organoids and assembloids (aka "brain in a dish") have just started being used to identify targets for intervention. Because they are developed from a person's unique DNA, these high-throughput models can recapitulate the neurobiological features of profound autism at an individual level and provide insight into specific drug targets which can ameliorate symptoms in that individual.

- **Research to develop small molecule and genetic therapies for profound autism starting with genetically defined/monogenic autisms.** Those with rare genetic variants associated with autism have a higher rate of profound autism than those without an identifiable genetic marker. There have been great advances in targeting and manipulating expression of autism genes through gene therapies. These therapies have the potential to be life-changing, but they are still in very early stages of discovery and are still being developed on a variant-by-variant basis. In addition to better techniques, researchers need to identify critical windows of treatment, discover better methods of administration and improve access to a broader portion of the community.

- Clinical and basic science research to understand the gender disparity in autism diagnosis. **There is a 4:1 ratio of males to females in non-profound autism but no sex difference in profound autism diagnoses.** Females may be protected against some forms of autism but more vulnerable to others. A closer look at factors related to the female sex (genetics, hormonal differences) will help identify targets for intervention as well as new ways to support females with a profound autism diagnosis, as their needs will be different than those of males.

GOAL 2: CONTINUED ...

DEVELOP PERSONALIZED TREATMENTS AND INTERVENTIONS FOR PEOPLE WITH PROFOUND AUTISM

- **Studies to support or test new technologies (wearable devices, passive data capturing, apps, virtual reality, tele-assessments) to better understand the biological markers of profound autism and also to be used as innovative supports.** The use of wearable devices for medical purposes has exploded in the past decade, providing caregivers, educators and physicians access to information collected from a variety of settings (home, school) over longer periods of time than previously possible. This information can be used not only to improve our understanding of precursors of negative behaviors, but to improve the timing and effectiveness of interventions.

GOAL 3:

DETERMINE WHETHER EXISTING ASD INTERVENTIONS ARE APPROPRIATE FOR THOSE WITH PROFOUND AUTISM; DEVELOP AND TEST NEW TREATMENTS

- **Randomized controlled trials of communication interventions for individuals who are minimally or nonverbal.** Communication is the key domain influencing quality of life and long-term outcomes, but there is a paucity of high-quality research on communication interventions for those who are minimally verbal. Interventions which promote language communication will help individuals lead more fulfilling lives. In addition, we need innovative approaches to creating alternative/augmentative communication systems that are adapted to the individual needs of people with profound autism.

- **Randomized controlled trials of treatments that include only those with profound autism,** specifically focused around improving adaptive functioning and language, and reducing self-injurious and aggressive behaviors, including pharmaceutical studies and ECT studies. Also, **randomized controlled trials that include those with profound and non-profound autism to measure if individuals with profound autism respond differently.** In addition, we need longitudinal studies to identify early markers of profound autism and predictors of developmental trajectories.

- **Comparative effectiveness research on real-world treatment packages,** such as residential treatment, or combined medication and behavioral treatments, especially for those with intense behaviors, such as physical aggression and self-injury. Thousands of individuals are in residential placements, and millions receive combinations of medication and behavioral treatment. A better understanding of how different interventions and supports work together is essential to delivering treatments that are best suited to a specific person's needs.

GOAL 4:

DEVELOP AND IMPROVE TOOLS TO DIAGNOSE PROFOUND AUTISM AND MEASURE EFFECTIVENESS OF INTERVENTIONS

- **Develop assessment tools** specifically for profound autism. A research consensus definition should be published shortly, and this new assessment should test for the items in the new definition. A new assessment tool will provide clarity and consistency in applying the criteria to diagnosis and research studies.
- **Develop a tool** to collect information on cognitive function in the population of people with profound autism. Current tools cannot accurately assess IQ in this population. This is critical to evaluating an individual's abilities and disabilities so that the correct supports and interventions can be provided.
- **Develop rigorous assessment tools** for co-occurring psychiatric and medical disorders in people with profound autism. Co-occurring psychiatric conditions, such as anxiety, catatonia, depression and psychosis, and medical problems like GI dysfunction and epilepsy, are highly prevalent and create substantial morbidity and mortality. There are no validated tools for assessing these conditions in people with autism who are minimally verbal or have intellectual disability. A large percentage of individuals with profound autism also have severe and debilitating comorbid conditions that make symptoms worse and which are masked by cognitive and language disabilities. Better understanding of the risk factors for comorbid psychiatric conditions can drastically improve the quality of life for families.