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The SVIP Study: An Interdisciplinary Approach to Autism Spectrum Disorder
Owen Adams

My time in the autism research department at the University of Washington Center on Human Development & Disability (CHDD) increased my interest in both psychology and neuroscience. The research division I was involved with, the Simons VIP Study, was researching the connection between mutations in the 16p gene locus and autism spectrum disorder (ASD). The study took in families from around the country with 16p mutations who also had children with suspected ASD symptoms. During the family’s time at the CHDD, each family member participated in several psychological tests to detect symptoms of autism, such as the ADOS and ADI. Additionally, neurological and genetic testing was performed to search for any similarities in inheritance patterns. The study involved collaboration between many different research professionals, which was as important for the validity of the study as the well being of the families being researched. The advice given to the families at the end of each week-long research session was informed by multiple schools of thought, which is important for a disorder as complex as ASD. The SVIP study showed me that interdisciplinary collaboration with a personal focus is essential for research and treatment of autism spectrum disorder.

Much of my work involved data entry for psychological and neurological tests. Through this process, I learned about the details of neurological assessments for diagnosing developmental disabilities. The tests looked at physiological factors, like macules and sacral dimples, but also developmental indications of neurological deficits, like abnormal eye movement and speech. To interpret the neurologist’s notes about these physical factors, I often had to look up the meanings of complicated phenotypes, finding their relationships with different types of developmental pathology in the process. There was also an assessment of previous diagnoses, as ASD is commonly comorbid with attentional disorders and anxiety. Medication history is among the most important variables to assess because of the varied effects that ASD medication can have when combined with other drugs.

I also assisted with several sessions of electroencephalography and magnetic resonance imaging. The EEG studies were examining abnormalities in μ waves for children with autism and other developmental disabilities. The μ wave is active during movement for both typically developing and ASD-affected children. μ rhythm suppression is involved in analysis of other people’s movement, but has shown less suppression during observation of motor activity in children with ASD. The director of the SVIP study, Raphael Bernier, is mainly involved with research involving developmental changes in μ rhythm suppression, which is thought to be involved in the mirror neuron system. He was involved with a recent study that showed μ rhythm suppression increases with age with little difference between ASD-affected children and typically developing individuals. μ rhythm differences are important to consider in children with ASD, but may be more subtle than suppression of activity since selectivity is also important to consider. Also, μ rhythms reflect activity in a large swath of motor cortex including primary motor areas, premotor centers and parietal cortex [1]. Thus, it
may be more beneficial to combine EEG data with fMRI scans and genetic testing to assess symptom-based deficits in brain functioning.

The MRI scans looked for prior brain damage that could act as a confounding variable in the diagnosis of autism spectrum disorder. The MRI technicians mentioned that the MRI was also capable of DTI and fMRI scanning, which they used for other studies. In the SVIP study, though, MRIs were mainly used as a general physical assessment since the autism spectrum is not associated with any particular area of abnormal brain development. ASD affects distributed and variable areas of cortex, including social areas like the medial prefrontal cortex and STS as well as self-awareness regions like the temporal parietal junction and the precuneus. This cortical distribution of symptoms also causes difficulty in comprehensive treatments for ASD, meaning that most medications address singular symptoms like irritability and attentional difficulties [2]. ASD is a genetically linked developmental disorder, making genetic tests a more reliable assessment than neurological scans in tracking the pattern of inheritance.

Toward this end, we were asked by the director of the SVIP study to research the neurological phenotypes of different deletions along the 16p gene locus, particularly those that pertained to autism spectrum disorder. He asked the interns to conduct this research because he was trying to publish a study on the relationship between EEG data and the genetics of autism before a different research lab could put forth a similar study. I found six 16p gene deletions that had some correlation to autism spectrum disorder. Some were related to channels for neurotransmitters like GABA and glutamate; these seemed to have more relation to synaptic development. Other mutations caused genetic syndromes like tuberous sclerosis, which have shown some of the highest genotypic and phenotypic correlations with autism spectrum disorder.

Approximately 20% of parents have been to a genetic professional regarding their children’s ASD diagnosis and the recurrence risk for future children, making studies like SVIP important for disseminating information. [3] Genetic causes have been identified in only 10% of ASD cases, but tiered evaluation may increase identification rates to about 40%. Unfortunately, access to genetic counseling resources is stratified based on income, leading to widespread parental misperceptions about ASD etiology. Parent’s improperly informed ideas about the genetics of ASD may negatively impact their probability of having future children and lead to inaccurate blame on influences like vaccinations [4]. Without private genetic counseling, families participate in the SVIP study for resources such as these as well as financial compensation and pragmatic advice. At the end of each week of participation, the psychologists and neurologists met with the study director to discuss symptom severity for each family member. Beyond the purposes of the study, they assessed the likelihood of other psychological disorders for all family members involved in the study. They then met with the parents for an extended period of time to go over possible medication approaches and skills training resources. As a study with many longitudinal components, many of the families have developed lasting relationships with the researchers. Thus, the SVIP team acts as a sort of advocate for parents and children affected by ASD.

Inspired by the efficacy of the SVIP study, I decided to write my thesis on therapeutic and pharmaceutical treatments for autism spectrum disorders. I addressed the limitations of several anti-psychotic medications such as aripiprazole and
risperidone, two effective treatments for irritability with side effects like drowsiness and nausea and long-term detriments such as tardive dyskinesia. I also looked at the future of hormonal treatments like oxytocin therapy, which has shown immediate social benefits with unclear longitudinal effects. FDA-approved medications such as risperidone seem to be more effective with simultaneous parent training programs, meaning that medication should be coordinated with other facets of treatment. Furthermore, I found that genetic counseling is extremely important since early intervention greatly reduces the cost of future treatment and the severity of future symptoms. For these intervention elements to be combined effectively, researchers need to communicate extensively among themselves. Study leaders also need to reach out to community leaders for political support, introducing sociological and economic factors. This kind of interdisciplinary collaboration needs to be a part of neuroscience research, particularly when it involves a disorder as complicated as ASD. Studies such as SVIP provide hope for future assessment and treatment of complex developmental disorders by fostering dialogue among researchers, parents and community leaders.

References

