

Early Diagnosis for Autism in Infants

Rhymes Walton

Human and Organizational Development, Cognitive Studies
Vanderbilt University

1. Introduction

In America today, roughly 147,867 people are diagnosed with Autism. Since one out of every 2000 people is diagnosed with some degree of Autism Spectrum Disorder (ASD), it is an important disorder to research and understand in order to identify autism at an earlier age and to possibly create necessary environmental changes to counter the onset of developmental delay. In this paper, the biological and environmental aspects of ASD will be discussed in relation to the potential for early diagnosis and intervention of autism in infants. Previous research on family and sibling studies will be used to examine the biological basis for ASD while Applied Behavioral Analysis will be discussed to examine how treatment and changing the at-risk infant's environment can improve his or her development.

2. Genetics

The link between genetics and diagnosis of Autism has been clearly established through family and sibling studies that analyze heredity in diagnosis of ASD. Bernalova and Buxbaum (2003) attribute the predisposition to have autism to be at least 90% genetic as seen through twin, family, and sibling studies. Since Autism is so highly genetic, parents who have one autistic child are five percent more likely to have another autistic child, and much more likely to have a second autistic child if the older sibling is a girl. This is because the male to female ratio of being diagnosed with ASD is four to one (Bernalova & Buxbaum, 2003).

As Rutter (2000) discusses, these genetic findings are important for a variety of reasons. First of all, they focus the root of autism on neurodevelopment rather than attributing Autism to poor parenting as previously assumed. Also, further genetic research could result in finding protective factors for females to see why males are more inclined to be autistic. Additionally, the genetic component of Autism is important because researchers and doctors can focus on identifying a phenotype to help intervene when children show at-risk behavior.

Since the younger siblings of autistic children are at higher risk for developing autism, sibling studies have been conducted to try and isolate characteristics that could lead to early diagnosis of autism. Furthermore, since overt symptoms of Autism do not fully manifest until the child is two or three years old, an infant diagnosis could result in earlier treatment the potential to counter the at-risk child's developmental delay. Two recent studies that focus on younger siblings of autistic children who are at risk for developing autism (Sib-ASD) and younger siblings of typically developing children (Sib-TD) will be examined.

3. Sibling Studies

The first study, conducted by Yirmiya, Gamliel, Pilowsky, Feldman, Baron-Cohen, and Sigman in 2006, compares Sib-ASD and Sib-TD at 4 and 14 months of age on a variety of social and language tasks. At four months, twenty-one infants (8 girls, 13 boys) were observed and coded for mother-infant interaction during free play, infant attention and reaction to the “still face” paradigm, and the infant’s reaction to the mother calling his or her name. The Sib-ASD infants were matched with paired controls that accounted for gender, age, birth order, sex of older sibling, and mental and motor scores. Later, at fourteen months, thirty babies (11 girls, 19 boys) were evaluated for verbal and nonverbal communication and cognition. These babies were also matched with paired controls from the Sib-TD group.

Although most of the results were the same for both groups, Yirmiya et al. (2006) identified discrepancies in early social engagement that might have contributed to differences in communicative and cognitive skills at 14 months. The Sib-ASD group was more neutral to the still face and was much less upset than the Sib-TD group (Yirmiya et al., 2006). This could imply that the infants at risk for autism had already developed an aversion to faces and social interaction so they were less likely to be disturbed by the experimenter’s still face. Later, at fourteen months, the Sib-ASD group had significantly fewer nonverbal gestures and had a lower language score on the Bayley scale (Yirmiya et al., 2006). While this is an important finding that could indicate potential language developmental difficulties, this group mean was primarily lowered by six Sib-ASD infants who showed a five month language delay. A surprise result was that more Sib-ASD infants responded to the mothers calling their names than the typically developing babies (Yirmiya et al., 2006). Although one would expect the Sib-TD group to orient to their names more, these children may have been more engaged in the task or play at hand when their names were called while the Sib-ASD were not fully engaged and easily distracted.

The results from the study conducted by Yirmiya et al. (2006) showed attention and reaction differences in the Sib-ASD infants and the Sib-TD infants as young as 4 months of age that were heightened by 14 months of age, as seen through Bayley’s scale. This research is important, as it has the potential to help predict the onset of Autism by observing the infant’s inherited genes and allowing for possible environmental changes to compensate not only for having an autistic sibling and the parenting struggles involved with raising an autistic child, but also the higher genetic likelihood of the sibling becoming autistic.

The second sibling study to look at infants who were high risk for developing autism was conducted by Noland, Stone, Walden, Sheridan, and Reznick in 2006. Since children with autism have lower scores for working memory and social awareness, Noland et al. (2006) tested younger siblings of autistic children to try and discover early behavioral risk markers for autism. Infant siblings of autistic children and siblings of typically developing children were analyzed for their looking response for a social (person) and non social event (toy) with a modified peek-a-boo game that tests working memory.

First, fifteen pairs of Sib-ASD and Sib-TD between the ages of six and nine months were matched for gender and age. A modified peek-a-boo game was then conducted and coded for the infant’s “target looks” back to where the stimulus (toy or face) had last appeared (Noland et al., 2006). To have correct “target looks,” the infant would have to be engaged enough in the game to remember the last emergent point after having a light and a bell as a brief distraction.

After the data were analyzed, significant differences in the Sib-ASD and Sib-TD groups were found. Sib-ASD infants had more correct target looks during the non social trial with the toy than the Sib-TD infants (Noland et al., 2006). In the social trial, however, Sib-TD infants had more correct looks than Sib-ASD (Noland et al., 2006). Therefore, the Sib-ASD group is not only looking at the social stimuli less than the Sib-TD, but is also much more in-tune to tracking and predicting the location for the non-social object than the Sib-TD infants. These results can support the case that infants at risk for autism have tendencies to avoid social stimuli and prefer non-social interaction at as young as six and nine months. Biological theories apply to the study of autism because, even in infancy, there are risks for autism that are apparent long before diagnosis for Autistic Spectrum Disorder can occur, but identifying these early signs in at risk infants can lead to early intervention.

Environment

Although the onset of Autism is greatly determined by genetics and biology, changing the environment in which the at-risk infant lives can potentially reduce the severity of the child's ASD. In a study by Lauritsen, Pedersen and Mortensen (2005), the genetic background of autism was analyzed in light of the children's social/familial environments. 943,664 children were followed from age ten to seventeen to study risk factors of the 818 children who went on to develop autism (Lauritsen et al., 2005). Risk factors of importance included the location the child and his or her parents were born, the parental age and ethnicity, and the family's history of psychiatric disorders.

While the highest risks were associated with genetics either by having an autistic sibling or with having a mother who was diagnosed with a psychiatric disorder, environmental risks were notable as well. Lauritsen et al. (2005) found a significant link between urbanization and older paternal age with autism as well as having a risk of 1.4 if the mother was born in another country. All of these risks could contribute to a less engaging home environment for the child.

The parental stresses associated with having an autistic child could lead them to react more distantly with a less interactive relationship with the sibling. This could compound the genetic influences on siblings of autistic children to almost encourage autistic tendencies of withdrawal and lack of communication. Similarly, if a mother has a disorder such as depression, then rather than encourage pro-social behavior she might encourage withdrawal in this case as well. The next risk, urbanization, could result in parents working during the day and might also provide fewer safe opportunities for the child to explore his or her environment. Additionally a sense of community and cohesiveness is generally attributed to a small town while independence and self-sufficiency attributed to largely urban areas. The final two risks identified by Lauritsen, Pedersen and Mortensen (2006) of older parental age and having a mother who was born outside the country could contribute to lesser communication between the parent-child dyad. An older father might not be as involved in childrearing and a mother from a foreign country might have language barriers to overcome which could be a disadvantage to the child's language development. Although the greatest risk for autism is genetic, the theory of environmental influences greatly applies since the environment can significantly impact the child and his or her diagnosis of Autism Spectrum Disorder.

4. Applied Behavioral Analysis

Applied Behavioral Analysis is relevant to infant diagnosis of autism because the purpose of identifying autism in infancy is to try and adapt the environment to help improve the child's development. In a study by Smith, Eikeseth, Klevstrand and Lovaas (1997), Applied Behavioral Analysis resulted in increased IQ score and increased language development of autistic children. Smith et al. (1997) compared eleven children diagnosed with either autism or mental retardation to a comparison group of ten children. All of the children were younger than 46 months old with had an initial IQ under 35 and there were no significant differences between the groups. The experimental group received thirty hours of one-on-one treatment over the course of the next two years while the comparison group had minimal treatment. At the conclusion of the study, the experimental group yielded an increase in IQ with a standard deviation of 13.14. The comparison group, on the other hand had an overall decrease in IQ (Smith et al., 1997). Additionally, the experimental group saw significant strides in increased language and communication after extensive treatment (Smith et al., 1997). The importance of this study is that it shows the effect that extensive treatment can have on helping control autistic characteristics and how deliberately changing a child's environment can lead to positive developmental outcomes.

5. Application and Conclusion

Genetic theories and twin studies shed light to help determine causes of autism while environmental theories and Applied Behavioral Analysis are useful to be proactive in intervening to help improve social and language development. Diagnosis of autism and environmental and behavioral intervention in infants is important and applicable in our society. In the American public education system children are attending public school as young as three years of age. It is important for these schools to have significant knowledge about genetic and environmental influences on Autistic Spectrum Disorder, especially in "identifying patterns of strengths and weaknesses that can be used in tailoring behavioral goals and designing developmentally appropriate interventions for very young children" (Stone, 1999, p.188). With continued research infants at risk for developing autism could potentially undergo extensive treatment and be spared the developmental delays associated with Autistic Spectrum Disorder.

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