Recurrence Risk for Autism Spectrum Disorders: A Baby Siblings Research Consortium Study

Valeria Li Tang¹*
¹Santa Cruz Cooperative School, Santa Cruz de la Sierra, Bolivia

Abstract. Autism spectrum disorder (ASD) is defined based on the core symptoms of typical autism. It is considered to be a congenital mental disorder and has nothing to do with acquired family education. Children with this disease have difficulties in social interaction, communication, and repetitive behaviours [3]. Since it is a severe disease that hinder the development of children, the risk factors of this disease are worth being investigated. One known risk factor is the gender. However, other risks are not defined yet such as the recurrence risk in siblings. Therefore, the author tried to figure out this risk. This paper reviewed a study done in 2011 about the recurrence risk for autism spectrum disorders in siblings. A longitudinal study was used, meaning that the same test subjects were examined over a period of time. The autism diagnostic observation schedule (ADOS), the standard diagnostic test for ASD was used in this study to determine whether a child is diagnosed with ASD. The results were estimated through parameters of a model numerically by an iterative fitting process. It was found through this research that there was indeed a correlation between children who had siblings with ASD and having ASD themselves.

1 Introduction

Autism spectrum disorders have long existed as one of the most prominent neurodevelopmental disorders to manifest in children. Around 1 in every 160 children are reported to have some sort of autism spectrum disorder, with speculation that numbers are actually much higher due to the limitations in diagnoses between low-income countries [1]. The main causes of autism spectrum disorders are not totally clear, as a combination of environmental and genetic factors can contribute towards the manifestation of ASDs in children [2]. One of the speculated causes is reported to be sibling relationships, in which there is a higher risk of recurrence for ASD between siblings. This study was conducted as a means of proving a genetic factor to ASD and how that correlates with its subsequent psychopathology.

The study [5] utilized a prospective longitudinal study of infants at risk for ASD, in which the participants were infants with older siblings with ASD. This also means that the same subjects were studied over the course of a period of time instead of being studied once. The recruitment process consisted of various methods, including clinics and agencies as well as word of mouth and fliers posted around the community. Online resources such as mail and media were also used, ensuring greater diversity and less chance of a biased result. The study specified in recruiting children younger than 18 months, as the longitudinal study would require the children to be younger in order to study them through time. The researchers utilized the autism diagnostic observation schedule (ADOS) (used at 36 months) and the Mullen Scales of Early Learning (used at 68 months) to measure symptoms of ASD in the children. Some variables in this study include the race and ethnicity of the children, the education of the parents, and the birth order of the child. However, it should be noted that the researchers state that the mix of race, ethnicity, and socioeconomic status were to maximize heterogeneity and did not change the potential outcome of the study. Relative risk was measured using SAS Proc Genmod, a method to generalize linear models.

2 Analysis

2.1 Review of the research

As shown in Figure 1, human brains are composed of many synapses, axons, dendrites and cell membranes. The ASD is considered to be a disorder of the information processing of the brain. Of the 132 infants in the study who met the criteria for ASD right away, 54 received a clinical diagnosis of autistic disorder and 78 received a clinical diagnosis of a non-specified pervasive developmental disorder [5]. Race and ethnicity were revealed to have no effect on the risk a child had. Gender, however, did predict the outcome, where 26.2% of male infants received an ASD diagnosis versus the mere 9.1% of female infants [4]. The relative risk for gender was almost three times as higher for male infants as it was for female infants. Children with affected siblings also reported experiencing a risk twice as large than children
with only 1 affected sibling, with 32.2% of children with older affected siblings experiencing ASD to the 13.5% of children. In the end, it was reported that boys who had older siblings with ASD experienced an increased risk of experiencing ASD symptoms. This conclusion in turn supports that genetics are indeed a factor that affect ASD.

2.2 Analysis on the research results

A few things should be noted with this study. First, it is the current largest study of ASD sibling recurrence to date. This study reported much higher numbers of recurrence of risk than previous ones had, in that previous studies reported recurrence to be around 3-14% when the results of this study reported that at 18.7% of children with at least 1 sibling with ASD [6]. However, it should be noted that this study was published in 2011, which can suggest that some of the information reported needs to be updated. The largest factors seem to be gender and number of siblings. The researchers also state that overreporting is also a threat to the estimation of recurrence risk, as time and resources posed as limitations for the researchers. Since the study was capped off at 68 months, the age where Asperger's disorder and other forms of mild ASD are diagnosed is promptly also capped off, meaning that the true recurrence risk might be higher.

2.3 Enlightenment of the research

It is also important to note that gender as a factor should really be taken with a grain of salt. As new research continues, it has been revealed that ASD has a long history of gender inequality in that ASD is very commonly misdiagnosed in girls as being something else. Symptoms of ASD vary between males and females, and for a while it was even thought that only boys could get ASD. As such, seeing that this study was conducted in 2011, gender differences regarding ASD should be duly noted.
3 Discussion

3.1 Genetic component

It is not a surprise that ASD has some sorts of a genetic component. Most developmental disorders have been reported that there are genetic factors involved, though an even bigger cause of these disorders comes from other factors such as parental health and behaviours, birth complications, and even instances during infancy where the child was exposed to dangerous/damaging things. However, to deny the existence of genetics completely would be doing a disservice to a lot of families [7].

ASD in general has shown to have a strong genetic base; twin studies have shown that the heritability reaches upwards of 90%. However, there is evidence to suggest that the genetic factor comes from not heritability but from mutations; for example, this is why a family with generations of non-ASD members can eventually spawn a child with ASD [7]. And given the rates at which ASD are diagnosed between children of different racial backgrounds, sexes, and cultures, it is unclear whether or not it is more heritable than mutation-based.

3.2 Limitations

The studies mentioned in this paper are mainly from the early 2010s, meaning that research in ASD has increased dramatically since then. It is important to factor in the time period, as researchers and mental health experts are still normalizing things like different testing for boys than for girls as ASD has been shown to manifest differently between boys and girls.

4 Conclusion

In the end, it was found that children with older siblings who have ASD are more likely to have ASD themselves than children who do not. The elevated risk is important for a few reasons. Firstly, this study demonstrates that parents with children who have ASD should be aware of the elevated risk of future children they decide to conceive. Second, this serves as additional information that can be utilized when studying the epidemiology of ASD, as the results from this study suggest a strong genetic factor at play. This study also plays into creating more awareness about ASD in general. People cannot care about what they do not know about, and so more studies involving ASD are incredibly important to make sure that awareness is raised so children with ASD can receive the support they need.

The limitations of this paper is that this paper reviews not much research and the main research this paper covers is made in 2011. In recent years, this maybe much more researches in this field and there might be more evidence of this topic. Therefore, in the future study, the author needs to review more current literature to further verify this result and make new findings.

Acknowledgment

First and foremost, I would like to show my deepest gratitude to Professor Chiang-shan Li, who has provided me with valuable guidance in every stage of the writing of this thesis. Further, I would like to thank Ms. Yvonne for her review and suggestions to my paper. At last, I would like to say thank you to my friends and families for their encouragement and support. Without all their
enlightening instruction and impressive kindness, I could not have completed my thesis.

References

1. Autism spectrum disorders. (n.d.). Retrieved May 27, 2020, from https://www.who.int/news-room/factsheets/detail/autism-spectrum-disorders

2. S. Hansen, D. Schendel, R. Francis, G. Windham, M. Bresnahan, S. Levine, E. Parner, E. (2019, March 06). Recurrence Risk of Autism in Siblings and Cousins: A Multinational, Population-Based Study. Retrieved July 12, 2020, from https://www.sciencedirect.com/science/article/abs/pii/S0890856719301728

3. Recurrence Risk for Autism Spectrum Disorders: A Baby Siblings Research Consortium Study. (2011). Pediatrics. doi:10.1542/peds.2010-2825d

4. T.K. Grønborg, MSc. Autism Spectrum Disorders Recurrence in Siblings. 1 Oct. 2013, jamanetwork.com/journals/jamapediatrics/article-abstract/1728998.

5. S. Ozonoff, et al. “Recurrence Risk for Autism Spectrum Disorders: A Baby Siblings Research Consortium Study.” American Academy of Pediatrics. American Academy of Pediatrics, 1 Sept. 2011, pediatrics.aappublications.org/content/128/3/e488.short.

6. "Sibling Recurrence Rate of Autism Spectrum Disorders Is 18.7%." Evidence-Based Mental Health, Royal College of Psychiatrists, 1 May 2012, ebmh.bmj.com/content/15/2/31.

7. C.M. Freitag. The genetics of autistic disorders and its clinical relevance: a review of the literature. Mol Psychiatry. 2007