

Etiology of Autism Spectrum Disorder (ASD):  
Familial Perceptions versus Empirically Validated Data

Jacqueline Lubin, M.Ed., Ed. D. Candidate

May 2015

The etiology of autism spectrum disorder (ASD) has been a widely debated issue for several decades. However, the exact cause of autism is still unknown (Russell, Kelly & Golding, 2009). Research has suggested that ASD may be caused by genetic and/or environmental factors (Glasson et al. 2004; Newschaffer et al., 2007). Others have proposed that it may be a combination of three factors-genetic factors, environmental factors and neurological development (King, 2015). This wide umbrella of factors has caused uncertainty among parents and family members and so have led some to deduce their own etiology. Some parents believe that ASD is caused by a combination of biological and environmental factors (Dardennes et al., 2011; Goin-Kochel & Myers, 2005; Mercer, Creighton, Holden & Lewis, 2006), while many others hold the opinion that there is a significant connection between vaccines and autism spectrum disorder (Bazzano, Zeldin, Schuster, Barrett & Lehrer, 2012; Gerber & Offit, 2009; Mercer et al., 2006; Russell et al., 2009). Although many studies around the world have shown that there is no relationship between vaccines and ASD, many parents adamantly hold the view to the contrary (Bazzano et al., 2012; Gerber & Offit, 2009).

Presently, there is desynchronization between what parents and family member perceive as causal factors and what research has proven to be possible indicators. This is of great concern because parents choose interventions based on what they perceive to be the cause of ASD (Dardennes et al., 2011). The implication of this is that parents may unintentionally opt for interventions which may be unsuitable for their child. This research paper will seek to highlight and discuss the factors that parents, family and professionals in the field perceive as causes of ASD and the factors that research have identified as triggers (or not) to ASD.

### **Familial Perception on the Causes of ASD**

Parents and family members have identified many factors as the causes of autism. Many of these factors are based on parental instinct and not empirical data. As parents and family members are the principal intervention agents for children with ASD, their opinion must be heard when identifying etiology. Dardennes et al. (2011) explained that “exploring parental causal beliefs may be an important step in care formulation” (p. 1144).

Dardennes et al. (2011) studied the relationship between parents’ perception of the causes of autism spectrum disorder and the treatment that was selected. They surveyed 89 parents of children with ASD. The main etiologies of ASD highlighted by parents were genetic factors, brain abnormalities and environmental factors such as prenatal problems and food allergies. Only 7% of parents cited vaccines as a cause of ASD. The researchers concluded that parents chose treatment based on the cause they perceived to have led to the diagnosis of ASD. Parents who believed that ASD was caused by mother’s sickness during pregnancy were more likely to use medication. Parents who believed it was caused by food allergy were more likely to use special diets and detoxification. If parents perceived the etiology to be a result of a traumatic event, the probability of the Picture Exchange Communication System (PECS) and behavior therapy being used decreased. Parents also associated a negative relationship with vitamins and brain abnormalities (Dardennes et al., 2011).

Russell et al. (2009) sought to investigate the common person’s view about the causes of ASD. One hundred and five people from the UK, North America and other industrialized nations took part in the study. Participants were self-selected after they responded to a UK epidemiological team’s press release about available funding to study the environmental causes of ASD. These included people with ASD, family members of people with ASD and

professionals in the field of ASD. The results revealed that 96% of participants highlighted environmental factors as the cause of ASD. The range of environmental factors highlighted by participants included: technology used in the medical field (e.g., ultrasounds, baby being induced, contraceptive pill, high level of mercury due to mothers' dental fillings, vaccines); changes in way of life (e.g., working mothers, increase use of television, food preservatives, gluten in diet); everyday exposure to technology (e.g., water and air pollution, indoor molds, radioactivity, carbon monoxide); and medical problems (previous miscarriage, dry birth, child born after twins). Approximately 66% believed that vaccinations was a major cause of autism. This study highlighted that the average person believes that ASD is caused by man-made factors that are closely related to improvement in technology (Russell et al., 2009).

Mercer et al., (2006) further explored parents' perception about the causes of ASD. Participants included 41 parents of children with ASD from USA and Canada. More than 90% of parents believed that their child's ASD diagnosis could be attributed to genetics. Forty-three percent stated that it was prenatal factors (e.g., advanced age of mother, smoking, medications, sickness during pregnancy) while 68% highlighted that it was perinatal factors (trauma at birth, induced labor, premature birth, early labor). Forty percent of parents ascribed vaccines as a major contributor to ASD. Fifty-one percent believed that diets, such as food containing gluten and casein, were causes. Mercer et al. (2006) concluded that parents may seek their own etiologies as a coping strategy.

Goin-Kochel and Myers (2005) conducted a study to examine etiologies that parents assign to congenital and regressive ASD. Congenital ASD refers to a child displaying characteristics of ASD from birth or immediately after whereas regressive ASD refers to a child displaying initial signs during later developmental years. Three hundred and twenty-seven

parents of children with ASD from New Zealand, United States, Canada, England, Ireland and Australia participated in the study. Parents of children with children with congenital ASD noted biological factors as the main causes while parents of children with regressive ASD stated environmental factors as the principal cause. The results showed that one quarter of the parents believed the etiology to be biological, such as genetics, and family history. Other etiological factors highlighted by parents included vaccines, medications, environmental toxins, maternal illness, medical condition, brain development, newborn illness, and prenatal difficulties. Goin-Kochel and Myers (2005) elaborated that 40% of parents stated strictly environmental or strictly genetic causes for ASD. Some parents believe it was a combination of factors, noting specifically vaccines in combination with other external factors and biology. However, a very small percentage (3.7%) noted other environmental toxins apart from vaccines. It is not surprising that parents of children with congenital ASD attributed the diagnosis to genetics while those with children with regressive ASD accredited it to environmental factors (Goin-Kochel & Myers, 2005). This may be because parents perceive that congenital ASD happened at conception while regressive ASD was as a result of external factors (as the child was believed to be “normal” before regressing).

The literature revealed that parents and family members perceive a vast array of etiological factors for ASD. Many believe that ASD is caused by both environmental and biological triggers. However, many still hold the view that vaccines are major contributors to ASD. In many instances, their perception determines the intervention chosen and utilized.

### **Empirically Validated Investigations into the Causes of ASD**

To date, no study has conclusively proven the cause of ASD. Many have attempted to prove and disprove perceptions and hypotheses, but none have determined the exact etiological

factors. Research shows that genetic factors play an important role (Grafodatskaya, Chung, Szatmari, Weksberg, 2010; Lo-Castro, Benvenuto, Galasso, Porfirio & Curatolo, 2010; Newschaffer et al., 2007), but the extent to which it does is unknown. Contrary to the perception of parents and family members, there has been no scientific proof that vaccines are linked to autism spectrum disorder (Uchiyama, Kurosawa & Inaba, 2007). Gerber and Offit (2009) reported on several studies conducted in various parts of the world, which have shown no correlation between the vaccine and autism. A recent study has confirmed that there is no relationship between vaccines and ASD, even among children who are genetically predisposed (Jain et al., 2015).

Jain et al. (2015) sought to find out whether there was an association between measles-mumps-rubella (MMR) vaccine and ASD by investigating the pattern of MMR immunization and ASD diagnosis among younger siblings of children with and without ASD. The study included 95,727 children with older siblings: 994 participants had been diagnosed with ASD and 1929 had an older sibling with ASD. One main criteria for inclusion in the study was that both children with ASD and older siblings had to be enrolled in a health plan for at least five years. The evaluation revealed that the receipt of MMR vaccination was not linked to ASD outcome. In fact, there was no significant relationship between the receipt of MMR vaccines and increased risk of ASD in both sets of children (older siblings with and without ASD). Jain et al. (2015) concluded that “there was no harmful association between MMR vaccine receipt and ASD even among children already at higher risk for ASD” (p. 1539).

Bazzano et al. (2012) examined the extent to which parents continued their child’s vaccine schedule after the diagnosis of ASD. The researchers also sought to find out if perception about the cause of ASD impacted decisions. The study included 198 parents of

children with ASD from Los Angeles County. The data revealed that 49% of parents believed that vaccines were the cause of the ASD diagnosis in their children. More than half changed the vaccine schedules of their children after diagnosis. The results indicate that scientific data have not convinced parents that vaccines are not linked to ASD. Therefore, there is a gap between parents' perceptions of etiological factors of ASD and empirical data.

Uchiyama et al. (2007) investigated whether the measles, mumps and rubella vaccine (MMR) was the cause of regressive ASD. This study was conducted in Japan where the MMR vaccine was used solely between 1989 and 1993. Children who had been diagnosed with ASD were placed in three groups- Pre-MMR Generation (100), MMR Generation (54 received the MMR vaccination, 132 did not receive the MMR vaccination) and Post-MMR Generation (n= 483). The researchers surveyed parents to find out children's medical history and present status as well as examine whether children with ASD had regressed developmentally and behaviorally. The data revealed that "within the MMR era, the rate of regression among children who received the MMR vaccination was not higher than those who did not" (Uchiyama et al., 2007, p. 214). The results further revealed that the rate of regression among children in the Pre-MMR Generation and Post-MMR Generation was not greater than those in the MMR Generation. Hence, Uchiyama et al. (2007) confirmed previous findings that there was no link between the MMR vaccines and regressive ASD.

Glasson et al. (2004) explored the relationship of obstetric factors to ASD. They compared 2259 cases of children with and without ASD in Western Australia. Participants were divided into three groups: individuals with ASD (465); siblings without ASD (481); and control group (1313). The researchers compared obstetric factors such as parental demographic, pregnancy data, labor and delivery information, and infant characteristics (birth order, weight,

gestational age). All data were obtained from Maternal and Health Research Database, which is a governmental data collection agency. The analysis of data revealed that “individuals diagnosed within the autism spectrum are more likely to have experienced obstetric difficulties during pregnancy, labor delivery, and neonatal period compared with people without an autism diagnosis” (Glasson et al., 2004, p. 624). The findings also indicated that increased paternal and maternal age, cesarean sections, epidural caudal anesthesia, fetal distress, no labor, poor birth condition, and threatened abortions were more prevalent in cases where individuals had ASD as opposed to the control group. Individuals with ASD were most likely to be first born. It must be noted that siblings had less obstetrics complications than individuals with ASD, but more than the control group. Glasson et al. (2004) concluded that environmental factors may be a major determinant of ASD as siblings experience similar obstetric conditions as individuals with ASD. This suggests that individuals with ASD react differently to external factors and may have a lower tolerance for prenatal and postnatal experience.

The literature revealed that empirical data show that an eclectic group of factors may be the cause of autism spectrum disorder. Research has shown that vaccines may not be the cause but many parents do not buy-in to that explanation. However, scientific data have revealed that prenatal, perinatal and postnatal factors are major contributors to ASD (Glasson et al., 2004).

### **Discussion**

The debate over the exact cause of autism spectrum disorder continues. The data revealed a large difference between what parents/family believe to be the cause and what research has proven to be (or not be) the cause. The wide debate for parents seems to be with regard to vaccines. Despite several studies proving that vaccines are not linked to ASD, parents are convinced that there is a relationship. Researchers will have to seek to provide parents with more



concrete data on how vaccines are not triggers to ASD. This gap in beliefs and scientific data need to be lessened if appropriate interventions are to be chosen for children with ASD. More sensitization and education will have to be done about vaccines as parents may inadvertently choose inappropriate interventions based on beliefs. Educators will have to be knowledgeable about the data on vaccines so as to educate parents (especially at IEP meetings). This could be one way empirical data can reach the parents and be discussed in a forum of professionals.

Although genetic factors seem to be a major cause of ASD, empirical research has not been able to conclusively identify specific genes that cause of ASD (Lo-Castro et al., 2010). Therefore, more research is needed to investigate genetic abnormalities. As technology improves, more biological testing may be done to pinpoint the genetic causes.

Additionally, more research is needed on the environmental factors that have been seen as risk factors. The prenatal, perinatal and postnatal factors need to be investigated more thoroughly so as to identify which ones are closely related to ASD. The new belief is that three factors strike at once to cause autism- genes, environmental factors and a period of time in the child's brain development (King, 2015). This is an interesting finding as several studies have revealed that the etiology of ASD is a combination of factors. Further investigations are needed to find out which environmental factors, and at what point in brain development do a child acquire ASD.

Research alone will not change parents' perception and therefore, extensive sensitization will need to be done to convince parents and family members of the results. In conclusion, although the exact etiological factors for ASD remain elusive, the quest for answers need to continue to improve the quality of life for children with ASD. The answer lies with extensive research and open minds.

### References

- Bazzano, A., Zeldin, A., Schuster, E., Barrett, C., & Lehrer, D. (2012). Vaccine-related beliefs and practices of parents of children with autism spectrum disorders. *American Journal on Intellectual and Developmental Disabilities, 117* (3), 233-242.
- Dardennes, R. M., Al Anbar, N. N., Prado-Netto, A., Kaye, K., Contejean, Y., & Al Anbar, N.N. (2011). Treating the cause of illness rather than the symptoms: parental causal beliefs and treatment choices of autism spectrum disorder. *Research in Developmental Disabilities, 32* (3), 1137-1146. doi: 10.1016/j.ridd.2011.01.010
- Gerber, J. S., & Offit, P. A. (2009). Vaccines and autism: A tale of shifting hypotheses. *Vaccines, 48*, 456-460.
- Glasson, E.J., Bower, C., Petterson, B., de Klerk, N., Chaney, G., & Hallmayer, J.F. (2004). Perinatal factors and the development of autism. *Archives of General Psychiatry, 61*, 618-627.
- Goin-Kochel, R. P., & Myers, B. J. (2005). Congenital versus regressive onset of autism spectrum disorders: Parents beliefs about causes. *Focus on Autism and Other Developmental Disabilities, 20* (3), 169-179.
- Grafodatskaya, D., Chung, B., Szatmari, P., & Weksberg, R. (2010). Autism spectrum disorders and epigenetics. *Journal of the Academy of Child & Adolescent Psychiatry, 49* (8), 794-809.
- Jain, A., Marshall, J., Buikema, A, Bancroft, T., Kelly, J., & Newschaffer, C. J. (2015). Autism occurrence by MMR vaccine status among us children with older siblings with and

- without autism. *Journal of the American Medical Association*, 313(15), 1534-1540. doi: 10.1001/jama.2015.3077
- King, B.H. (2015). Promising forecast for autism spectrum disorders. *Journal of the American Medical Association*, 313(15), 1518-1519. doi:10.1001/jama.2015.2628
- Lo-Castro, A., Benvenuto, A., Galasso, C., Porfiro, C., & Curatolo, P. (2010). Autism spectrum disorders associated with chromosomal abnormalities. *Research in Autism Spectrum Disorders*, 4, 319-327. doi: 10.1016/j.rasd.2009.10.006
- Mercer, L., Creighton, S., Holden, J.J.A., & Lewis, M. E. S. (2006). Parental perspectives on the causes of an autism spectrum disorder in their children. *Journal of Genetic Counseling*, 15 (1), 41- 50. doi:10.1007/s10897-005-9002-7
- Newschaffer, C.J., Croen, L.A., Daniels, J., Giarelli, E., Grether, J.K., Levy, S.E. ... Windham, G. C. (2007). The epidemiology of autism spectrum disorders. *Annual reviews of Public Health*, 28, 235-258.
- Russell, G., Kelly, S., & Golding, J. (2009). A qualitative analysis of lay beliefs about aetiology and prevalence of autistic spectrum disorders. *Child: Care, Health and Development*, 36 (3), 431-436. doi: 10.1111/j.1365-2214.2009.00994.x
- Uchiyama, T., Kurosawa, M., & Inaba, Y. (2007). MMR-Vaccine and regression in autism spectrum disorders: Negative results presented from Japan. *Journal of Autism Developmental Disorder*, 37, 210-217. doi: 10.1007/s10803-006-0157-3