What causes autism?

The etiology of autism has been an open question since the identification of the syndrome over 70 years ago. Leo Kanner (1943) wrote: “We must, then, assume that these children have come into the world with innate inability to form the usual, biologically provided affective contact with people, just as other children come into the world with innate physical or intellectual handicaps.” (p. 250) Beyond, that, he offered no speculation as to the cause of the syndrome.

In the succeeding 70 years, a variety of hypotheses have been put forward and the short answer to the question in the title is: “We still do not know.” But not for lack of trying . . . two recent papers, one negative and one positive, each add a piece to our knowledge about the etiology of autism.

Negative findings are valuable; it is useful to know what does not cause ASD, particularly when clarification of the hypothesized cause has important public health implications. Jain and colleagues (2015) seek to add one more nail in the coffin of the MMR vaccine-autism hypothesis, an idea that has been buried and re-buried over the past 15 years, yet like the un-dead keeps creeping back to terrorize parents and to sustain paranoia and conspiracy theory.

The study examined privately insured children with older siblings; they chose this subgroup because of previous findings that children with an older sibling with ASD are less likely to have received the MMR vaccine. The authors conclude: “In this large sample . . . receipt of the MMR vaccine was not associated with increased risk of ASD, regardless of whether older siblings had ASD. These findings indicate no harmful association between MMR vaccine receipt and ASD even among children already at higher risk for ASD.” (p. 1539)

The Jain et al. investigation can be added to the substantial body of research exploring a possible relationship between autism and vaccines. Every study with adequate methodology has yielded the same result; autism cannot be attributed to a reaction to MMR vaccine.

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A more promising line of research into the etiology of autism has been in the field of genetics. It is clear that there is a genetic component to the etiology of ASD but considerable effort devoted to genetics research has yielded limited success. At this point, a genetic cause for the syndrome can be identified in only about one-fourth of individuals with ASD (Huguet et al., 2013) and most researchers have recognized for some time that a single genetic (or other) cause for ASD will never be discovered (Happe et al., 2006).

A possible synthesis arising from the environment-versus-genetics debate is the hypothesis that ASD is a result of genetic vulnerability to differences in the intra-uterine environment. In a paper exploring one aspect of this hypothesis, Xiang et al. (2015) examined the effects of gestational diabetes mellitus (GDM). These researchers were particularly interested in gestational age at GDM diagnosis to determine whether GDM at any point during pregnancy increased the risk for ASD in the child. They found that the risk for ASD for children who were exposed to GDM which was diagnosed at 26 weeks or earlier was increased by about 40% (a Hazard Ratio of 1.42) compared with unexposed children. If GDM was diagnosed after 26 weeks, there was no increased risk. They concluded that “children in the group with GDM diagnosed at 26 weeks or earlier may have been exposed to untreated hyperglycemia during early critical brain developmental windows, which led to ASD risk after birth.” (p. 1432)

Even if these findings are replicated, they do not tell us how it is that hyperglycemia might lead to differences in brain development that lead to autism, nor for which individuals is that pathway most likely. But the Xiang et al. study offers an example of the kind of research that might prompt new and creative thinking, the kind of thinking that is essential if autism is ever to be better understood.

References


