

Podcast: Autism and the Environment

Anne Johnson: [music] Welcome to Environmental Health Chat, a podcast about how the environment affects our health, from the National Institute of Environmental Health Sciences Division of Extramural Research and Training. I'm your host Anne Johnson.

Let's start with a number: 1 in 68. That, according to a recent CDC study, is the prevalence of autism among children in the United States today. Autism is being identified more and more, about double what it was a decade ago.

Autism is a spectrum disorder, meaning it includes a range of neurological and behavioral conditions. Some common characteristics include trouble with social interaction and communication or displaying repetitive behaviors or engaging in a restricted range of activities.

Better detection methods probably account for some of the increase in autism's prevalence simply because more people with autism are being diagnosed. But scientists believe there are other factors in play that are actually causing more people to become autistic.

One of the many scientists investigating autism's causes is Dr. Valerie Hu. She's a professor of biochemistry and molecular medicine at the George Washington University School of Medicine and Health Sciences.

She's also the mother of a son with autism and she says she is driven by a desire to help people like her son better handle the struggles of living with autism.

Hu: You know, I think all parents hope for a better future for their children. We don't really want to change who they are, but we want to make life a little bit easier. I think, personally, and I see that for my son, that social deficits are really very difficult, because so much depends on our ability to interact with others. What we're hoping for is to understand autism better so that we can help individuals by targeting therapies at whatever pathways might be dysfunctional to help, you know, make life better.

Johnson: So Valerie has set about finding those biological pathways that might be dysfunctional in people with autism. But the causes of autism appear to be extremely complex. Like I said before, it's not just one disease, it's a spectrum of characteristics that all have to do with how the brain is wired.

Hu: Overall, I think we know very little about the causes of autism. In fact, the more we study it the more contributors we find, both at the genetic level and now, I think, more and more, we're going to find environmental contributions.

Johnson: She thinks the key may be in how environmental factors interact with genes that affect brain development. There are hundreds of genes that have been associated with autism, but untangling how each one of them might contribute to the disorder has been a monumental task. Valerie and her colleagues have been looking at a gene called ROR-alpha, or RORA for short. RORA encodes a protein that appears to act as a master switch for lots of other genes.

Hu: We performed a study that asked “What are all the potential targets of RORA—in other words, what genes might be regulated by this one gene RORA?” And we found that there was a potential for it to regulate over 2,500 genes. And among the 2,500 other genes, over 400 genes were already considered autism candidate genes. So what that suggests is that RORA can act like a domino such that anything that might shake it a little bit or knock it over is going to have a huge impact on the genes that it regulates, including hundreds of autism candidate genes.

Johnson: Valerie also found that RORA seems to be less active in the brains of people with autism than in people without autism. It seems to be especially suppressed in people with severe language impairments, the largest and most severely affected subgroup of those on the autism spectrum. So what causes RORA to be activated or suppressed—what’s the disturbance that sets off that first shaky domino? Hormones including testosterone and estrogen appear to play a major role. Valerie says that clue may help connect autism to environmental factors.

Hu: The fact that the sex hormones can affect the level of expression or activity of this gene, RORA, suggests that endocrine-like substances called endocrine disruptors, which are really everywhere in our environment, might also affect the level of activity of this gene. And if this happens during critical windows of development, then it’s possible by deregulating the level of RORA, one might predispose the individual towards autism.

Johnson: With a new grant from NIEHS, Valerie is now looking for endocrine disrupting chemicals that might suppress RORA.

Hu: If we find that these chemicals, these environmental agents, can actually change this very important gene, you know that would be kind of a warning signal, like we need to be more vigilant about not letting these types of chemicals get into our environment.

Johnson: She says we’re a long way from knowing whether or not such chemicals actually lead to autism. But it’s a research avenue worth pursuing if it could one day help prevent autism in future generations. Valerie says learning more about RORA also might help scientists find ways to correct RORA deficiency and prevent some of the brain development problems associated with autism.

Thanks again to Dr. Valerie Hu for sharing these insights. We have links to more information on autism and endocrine disruptors at our website, niehs.nih.gov/podcasts.

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