Two Major Concerns in Autism
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Editor's Note:

A March 2012 report from the Centers for Disease Control and Prevention (CDC) showing that the prevalence of autism spectrum disorders (ASDs) in the United States has risen by 78% since 2002, along with a controversial Diagnostic and Statistical Manual of Mental Disorders, fifth edition, (DSM-5) proposal to revise diagnostic criteria, have drawn recent attention to ASDs. Medscape spoke with Fred Volkmar, MD, Chair of the Child Study Center at the Yale University School of Medicine and Chief of Child Psychiatry at Yale-New Haven Children's Hospital, about the implications of these important issues.

Autism Up 78%: Reality or Increased Diagnosis?

Medscape: Hello Dr. Volkmar. A new CDC report[1] shows a 78% increase in ASD prevalence since 2002. How do you explain these findings? Do the data represent an actual increase, or rather increased diagnosis?

Dr. Volkmar: A long story made short: There are a number of possible factors that could account for an apparent increase that might or might not be real. Of course, one possibility is that to some extent there might be a real increase. However, while that can't be ruled out, it is probably a less likely possibility.

There is more public awareness of autism. There's more attention. There's often a pressure on the part of parents for an autism spectrum label. Often there's a slippery slope equating autism with autism spectrum and there's often a push for getting the label for school services -- sometimes quite appropriately and sometimes less so. This is a problem that sometimes has been referred to as diagnostic substitution in the literature: having multiple possible labels and picking the one that will get you the most services. All of those things factor into it. And there is some reason to think that there could, at least in theory, be some potential for the actual increase based on genetic studies. Usually in terms of seeing such a fast increase, you would think this is going to be more about method than reality. But again, it's hard to know.

In terms of the CDC paper, we would have a better handle on all this had they broken it down by diagnostic categories. The great beauty of the report is that it involved multiple sites around the country. The downside is there's not quite enough diagnostic detail -- how many of the diagnoses were spectrum and how many classical autism? I think also there's a general problem in epidemiologic studies; that is, not seeing the child and looking at records vs seeing the child. All of these issues must be factored into the findings. Eric Fombonne has discussed these issues in some detail.[2]

Medscape: Could the CDC study have overestimated prevalence owing to study design?

Dr. Volkmar: Yes, it's possible. To get the best data, children would have actually been seen and evaluated recently. Or at least a subsample of the children would've been seen to compare results of direct assessment with record review.

Medscape: You mentioned that there are genetic data supporting an actual increase in autism prevalence. Could you elaborate?

Dr. Volkmar: Data suggest that there's a potential for subtle changes in sperm as men get older. To make a long story very short, when women are born they have their full comple ment of eggs. They're not going to develop any more during their life. Men, on the other hand, are constantly regenerating sperm once spermatogenesis begins. And as men get older, there's an increase in the rate of small mutations. There's some suggestion from studies both here in the United States at the Simmons Foundation and also in Iceland and other locations that older fathers might account for some degree of a true increase in rates of autism.[3-6]
Environmental Factors

Medscape: Where do the data stand on potential environmental influences in autism and ASDs?

Dr. Volkmar: The interesting thing about environmental studies is that they've been much less productive than we expected. However, there are a number of interesting studies here. For example, there were a group of children who grew up in the horribly depriving orphanages in Romania, when Romania was a communist country. These were just dreadful institutions and the children looked very autistic. The interesting thing is they were followed over time as researchers thought, "Oh, we might have an environmental model of autism related to severe deprivation." As those children were placed in more supportive family environments, they got better. I'm not saying they became perfectly normal, but it was a very different kind of course.

There was an eminent child psychiatrist named Stella Chess who wrote about children with congenital rubella, a congenital infection that often leaves babies blind and deaf. She wrote about them having what looks like possible autism. The interesting thing again with those children is that over time they looked less so, and of course children with very severe problems like blindness and deafness associated with congenital rubella are difficult to assess. And so we've learned to be a little careful about this because it's a slippery slope. Sadly, it's a little like the joke about asking Mrs. Lincoln how the play was. Because there's so much else going on, this is just not adding very much. And the trouble is that the issues of diagnoses are actually, especially in classical autism, most complicated at the bottom and top of the IQ range. The middle range is less complicated.

In the very impaired child with mental retardation, what's now called intellectual deficiency, there are many autistic-like behaviors, such as motor mannerisms and unusual movements. Often those children don't have social difficulties that are more profound than their intellectual disability. That's one of the great complexities with dealing with all this.

And, of course, there was a lot of interest in the last few years about that work suggesting that perhaps the measles-mumps-rubella vaccination had some role in autism. That paper has been withdrawn, as I understand it, from The Lancet. It is surprising that more hasn't panned out about the environmental causes.

One area that has some potential relevance is whether very premature infants are more likely to have autism or ASDs. There is a small body of work on this but it is growing. In some ways it is interesting in that it may parallel some of the older work on animal models of autism using lesion studies. That is, there may be some potential for linking damage in some areas of the brain to behaviors and developmental features similar to those seen in autism. On balance, however, while there has been some research on environmental factors in autism, there is much less evidence here as compared with genetics.

Medscape: Do most experts believe there probably is, at least in some cases, an environmental component to autism?

Dr. Volkmar: Well, there's certainly some potential here. Even if you look among identical twins, the concordance for autism is not perfect, although it is very high. But again even here, keep in mind that identical twins in utero may be exposed to different things. So there is certainly a potential role for environmental factors, but the data for now are lacking.

And Finally, The DSM-5 Controversy

Medscape: Let's touch on the topic that seems to be on everyone's minds these days: the DSM. Can you summarize what's being proposed in the way of autism and ASDs for inclusion in DSM-5 vs the current DSM-IV criteria?

Dr. Volkmar: Well, first of all, there's no question that periodically things need to be redone, and that's just the nature of business. DSM-IV has been around since 1994. It has been remarkably productive, and research has advanced. On the other hand, there are some things that clearly do need to be thought through and changed. One might argue at the very high level that perhaps the way to do this is not to have to change the entire book every time you redo it but to redo sections on a periodic basis. Updates could be published in loose-leaf hardcopies and online.

As far as autism goes, there are some aspects of what's being proposed that are very interesting and innovative, particularly those looking at dimensions of function/dysfunction. The question of moving from this funny term 'pervasive developmental
disorder" -- which was coined back in 1980 -- to "autism spectrum disorder" is being discussed, which is in some ways in keeping with how people are using the term. So there are aspects to the proposal that are not bad. And also we have a number of very good diagnostic instruments now that have been in the past key to the categorical DSM and International Classification of Diseases (ICD) criteria. And I would point out that one of the other great advantages of the DSM-IV system is that DSM-IV and its international counterpart, ICD-10, essentially are effectively the same. So that means whether you're doing work in Switzerland, Swaziland, or New Haven, you're pretty much working with the same system.

DSM-IV retained what had historically been described since about 1978 -- that is, the grouping of autism features into 3 areas: social disability, communication disability, and unusual behaviors. The proposals in the DSM-5 are to combine social and communication into one criterion set and have another set that has to do with sensitivity to the environment. One of the potential disadvantages of this is that at the end of the day this really needs to be a research-based question and proposal -- it's not a question of opinion, it should be a question of what the data showed.

They're proposing moving to a classification called "autism spectrum disorder," which on the face of it implies a broader view. And that, in fact, was my first worry upon hearing about this -- that it would broaden criteria and potentially dilute services. Paradoxically, my worry is now the opposite. There are multiple studies that have looked or are looking into this at the moment,[9-11] but in reviewing the data we collected as part of DSM-IV,[12,13] we are worried that the more cognitively able -- people with IQs of 70 and above -- would actually be somewhat more likely to lose a diagnosis of ASD. This could greatly complicate their lives in terms of getting services and also complicate things for those of us who do research, in terms of following children over 10, or 15, or 20 years who suddenly are no longer supposedly on the spectrum. In going back to matching the new proposed criteria with those we used in DSM-IV, we discovered that even the more highly able people with classical autism lost that label and may be at risk for losing services. Many of the children who previously would have been said to have what are called atypical pervasive developmental disorder and pervasive developmental disorder not otherwise specified also lost their label and thus likely their eligibility for services, as did a majority of the children with Asperger syndrome.

Medscape: Some American Psychiatric Association representatives have reported that the new criteria will not dramatically alter diagnosis rates. Can you comment on this?

Dr. Volkmar: Yes, I've heard people argue this several ways. One group of people says, "Look, it's not going to change anything," which would be interesting. That would be fine except if it's not going to change anything, why mess with it?

Other people say we'll lose people who have been cured. Now the idea of being "cured" is a tricky business -- my worry is that because we're doing a better job of identifying children early and intervening earlier, children are as a group doing better. In April we had a daylong conference for high school students, their parents, and teachers about sending children with ASDs to college. Now that's not a problem I would have had to cope with very much 20 years ago. That's a new phenomenon and it's because they're doing better. Now, are those kids "cured?"

I did see some unpublished data from someone who was basically claiming this. Those data had to do with kids who are in college with good IQ scores. But the part of the data that she didn't make such a point of addressing -- but which I asked about -- was their real-world skills, which turned out to be an order of magnitude down from where their IQ is. And yes, these kids are very bright, but they may not be able to cope with the world. They may not be able to be out there and be independent, to drive, to balance a check book. I have one young man who does math equations in his head, who's got an IQ of about 140, who literally can't go into McDonalds and get a cheeseburger. It's just too much for him. You could argue he's cured. But I would argue that, well, that's ridiculous. If you mean by cured he's got a high IQ, then yes, but the problem is he's quite disabled.

And so I worry a little bit that when we talk about this "cured" business, we're minimizing the service needs of our more able children. They're doing better and better, which is fantastic, but the problem is how can we best support them? And many of them get depressed and anxious as they get older because they're out on their own and their resources no longer exist in the usual ways. I worry a little bit that we're going to abandon them. And I worry that school districts could potentially institute occasional evaluations using the new criteria and basically say, "This kid no longer qualifies; no reason to give service."

Medscape: By how much could the prevalence of autism decrease according to your data?
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Dr. Volkmar: Among the more cognitively able children (those with IQs above 70) our data suggest it could be as high as 40%; this would be less of an issue in individuals with lower intellectual levels.

Medscape: It's almost paradoxical because usually psychiatrists come under fire for overdiagnosing.

Dr. Volkmar: Yes. And that's certainly the issue for some of the other proposals like the new substance abuse criteria, which could greatly increase rates. But again, if you're somebody who's an epidemiologist, or even somebody like me, who works in a very specific area, you have to step back and say to yourself, "does it make sense that something's going to change radically in either direction overnight?" It makes sense to make changes as change is needed, but the idea of turning the whole book upside down at this point seems to me a little nutty.

Medscape: How would you approach updating the DSM ASD criteria?

Dr. Volkmar: I think at this point there are going to be conflicting data. And I think before we have a radical change, which is going to complicate not only the lives of researchers but especially the lives of parents and children, we should really take a step back and say, "okay, let's really think through what is needed here." And I worry a little bit that the way the process has gone, we have lost of sight of the patients and clinicians who are trying to use this in community and practice settings. I hope I'm wrong about this but fear I'm not.

Medscape: In terms of potentially cutting people out of access to care, what specific treatments are you most concerned about?

Dr. Volkmar: There are good data supporting a number of therapies. And this goes back to a report from the National Research Council back in 2001 called Educating Children with Autism. There are very good data that say that structured and behavioral interventions do work for many children — though not necessarily for everybody — and that the earlier we intervene, the better. We understand more and more about how treatment is working. We understand more and more about how having a social disability like autism can result in an idiosyncratic way of learning about the world. I'm not saying right or wrong, just idiosyncratic and not shared with the majority of people. You don't share with others what is and isn't relevant to learn about, so by the time you get to first grade you're in deep trouble. And now we're getting to these children earlier and earlier with a number of evidence-based treatments that really help these kids. It's not like there's necessarily a single right and wrong one, but there are many good, evidence-based treatments that really help with development and normative learning. There's behavioral therapy, special education, communication therapy, technology-based treatments — there are a ton of options, which is great. So as I said, the problem is that as kids are getting better, I worry that we're going to essentially disqualify some of the kids who most need services and who've done the best and should continue working to do even better. That seems extremely unfortunate.

References


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