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## Commentary

The Autism Speaks symbol is a puzzle piece. *Treating the Whole Person with Autism: The Proceedings of the Autism Speaks National Autism Conference* puts several of the more important pieces of the puzzle in place. It is my task as a developmental and behavioral pediatrician with a long-standing interest in autism spectrum disorders to frame what I hope will be a practical perspective on the picture that is emerging.

### ND Disorder in the DSM 5

There may be no single piece of the puzzle more important than the very definition of autism as recently revised by the new DSM 5 in May of this year.

Though many of us have been anxious about the new criteria, Susan Swedo, MD, and Audrey Thurm, PhD, do a good job of calming us down and convincing us (with their nifty color-coded chart) that the children with ASD identified in the past will still meet criteria with the exception of a few cases of PDD NOS. The new criteria are not only inclusive but also practical. With parents, I much prefer using the term “autism spectrum disorder” than the intimidating and misleading “pervasive developmental disorders.”

I do have one reservation—call it a pet peeve. The *delay in language* criteria in the DSM IV is now relegated to a mere “specifier” in the DSM 5. I have always taught pediatric residents to use any *delay in language milestones* as a quick screen for ASD: “If the child doesn't have one word phrases by one year or two word phrases by two years, think autism.” Maybe the new AAP emphasis on screening for ASD at the 18- and 24-month well-child visits will make my clinical pearl moot.

There were three aspects related to the new DSM 5 that were not, in my opinion, emphasized enough in the proceedings. The first is the importance of the *severity criteria*, which were clearly mentioned but were not listed in the appendices. I strongly suggest reading these. Parents always ask me how severe their child's autism is. These severity criteria, based on the amount of support the child requires, help answer that critical question. The second aspect needing emphasis is the completely new inclusion of *sensory reactivity* as part of restrictive and repetitive patterns of behavior. This piece of the picture has been missing for far too long. Sensory–motor issues profoundly affect the lives of both the child and family. Moreover, professionals and parents can do so much to address these sensory/regulatory issues.<sup>1</sup> And the third aspect unaddressed is the lack of mention of standardized tests [e.g., the Autism Diagnostic Observation Schedule (ADOS)] that have become a staple of the diagnostic process in most clinical settings. Without confirmatory testing, it is rare any more that children meeting the DSM criteria on a purely clinical basis would get insurance coverage to pay for intervention services especially under the Autism Speaks laws that have been passed in over 30 states.

### Autism Genetics for the Real World: What Science Tells Us and What it Means for the Child With Autism

The genetics of autism is puzzling for many, but David Miller, MD, PhD, nicely frames the picture with a basic introduction and definition of terms. He has some trouble putting the whole picture together because as it turns out—with over 400 different genes implicated—the genetics of autism is a bewildering puzzle indeed.

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Traits of autism (e.g., analytic, obsessive, and detail-oriented progenitors) can often be found in the proband's genogram.<sup>2,3</sup> If one child in the family has ASD, there is between a 5% and 20% recurrence risk that a sibling will have it. If one identical twin has ASD, there is a 70% (the DSM 5 mentions up to a 90% chance) chance the other will have it. These risks must always be discussed with the family.

Though loss or gain of a gene copy has been found among individuals with autism "...none accounts for more than 1% of all cases of autism." In other words, the large majority of children with ASD will have no identifiable genetic abnormalities. Nonetheless, when chromosomal microarray (CMA) analysis has been performed on children with ASDs, approximately 20% will be positive.

So, Dr. Miller recommends *routine* testing with a study for Fragile X, and CMA. I am convinced, though reluctantly, because now I see single gene mutations that have rarely been identified before. You cannot know for sure if it explains the child's autism, so you have to test the parents. I just had a case of a completely normal parent having the microdeletion. It turns out, this time, it was the wrong piece to the puzzle. Caveat emptor and make sure the parents are aware of the perplexing ramifications. Finally, Dr. Miller does not mention that CMA (at a cost of over \$2000) is *not* at this time routinely covered by insurance companies and so one must seek prior authorization. My final piece of advice is to always consider referring a child with ASD to a geneticist if the child has a positive family history, three or more minor dysmorphic findings or one major dysmorphic finding, hypotonia, and/or a severe dose of autism.

## Advances in Medicines

Which medication we should use for children with ASD is often a puzzlement, as Jeremy Veenstra-VanderWeele, MD, notes early in his report that there is "no solid evidence that any medication benefits *core* symptoms in ASD." His review of brain-based research was excellent. Clinical medication trials on Fragile X, Tuberous Sclerosis, and other rare syndromes, if successful, may put a few small but (at least for these particular children) important pieces into the puzzle. The "secretin" fad of the 1990s was a cautionary tale about magical, reductionistic thinking, and I would be wary about the same kind of thinking when it comes to oxytocin, touted as the medical treatment for the social deficits of autism.<sup>4</sup> Dr. Veenstra-VanderWeele wisely acknowledges that such medications might be used in the future as adjunctive to behavioral and developmental interventions.

On the other hand, the so-called "pseudo-specific" medications—SSRIs, Risperdal, and stimulants—can, in my experience, be very beneficial for children with ASD when used sparingly for the right indications.

Though the literature on SSRIs for ASD is discouraging,<sup>5</sup> I am convinced from years of experience that low-dose *SSRI treatment* (2–5 mg q day of fluoxetine, for example) can be very helpful for children with ASD (6 years and older) with *severe* anxiety that impairs daily functioning. Good research on this aspect of care is yet to be done.

*Atypical antipsychotic* prescribing for children with ASD in the U.S. has burgeoned<sup>6</sup> because, in our immediate-gratification-society, it is much easier to give a drug than take the time to puzzle things out. I use the atypicals only for children who are unresponsive to good behavioral, developmental, and/or psychological strategies.

Finally, as discussed in this section, medications for ADHD are not nearly as effective for children with ASD.<sup>7</sup> This is not particularly perplexing. Children with autism tend to be non-compliant with normative expectations; they have their own interests (regarding which they can focus on just fine, thank you); and their distractibility tends to be related to a core autism feature, namely, self-absorption. So I am cautious in prescribing ADHD medications. When I use stimulants, I often do a two-week trial with teachers "blind" (I have them fill out short Connor's forms) to the child's medication regimen, which varies from no medications, to low dose, to higher doses, according to a calendar I give to the parents.

## Addressing Medical Issues From Childhood to Young Adulthood

Dr. Paul Carbone creates the picture of a child with ASD in an ideal medical home setting where the child receives comprehensive, compassionate, and culturally sensitive medical care, with coordination between multiple professionals; with insurance coverage for speech and language, occupational therapy, and intensive behavioral

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