

MEDICAL MEMO

Kevin Leehey, M.D. Child, Adolescent, and Adult Psychiatry; Board Certified

Autism, Asperger's, ASD, NLD, PDD - Part One

This broad and complex topic will be addressed in two issues. This Medical Memo, Part One, will cover history, diagnosis, causes and key features of these conditions.

Autism was first described in 1943 by Leo Kanner MD, a psychiatrist at Johns Hopkins in Baltimore. A year later, Austrian pediatrician Hans Asperger MD, unaware of Kanner's work, described essentially the same condition but with a higher percentage of the patients having more developed verbal and cognitive skills. Autism first appeared on its own in our official Diagnostic Statistical Manual DSM-III (3rd edition) in 1980 and was expanded upon in 1987 by adding Pervasive Developmental Disorder Not Otherwise Specified (PDD-NOS) as a diagnosis in DSM-III-R (Revised). In 1990 the Individuals with Disabilities Education Act (IDEA) federal law for the first time made Autism an official special education label in US public schools. Also in 1990 the Americans with Disabilities Act (ADA) facilitated the education of youth with Autism, intellectual challenges (known as MR = mental retardation), and other severe disabilities outside of institutions and into public schools and made them "count". Asperger's Syndrome (AS, now called Asperger's Disorder) did not appear officially as a diagnosis in DSM until DSM-IV in 1994. The newest version of DSM-IV-TR (4th edition Text Revision) uses Pervasive Developmental Disorders as the umbrella category with Autism, Asperger's, PDD NOS and 2 other even more severe and rare conditions Rett's Disorder and Childhood Disintegrative Disorder included as PDD subtypes. Although the term Autistic Spectrum Disorder (ASD) is now often used to include Autism, Asperger's and PDD NOS it is not an officially recognized DSM diagnosis. The term "High Functioning Autism" and describing Asperger's as "Mild Autism" are also not official diagnoses and are essentially equivalent. Although they do often accurately imply a better

prognosis than classic Autism "high functioning" and "mild" understate the severity of all PDD's. I'll use the term PDD rather than ASD to include the whole group because PDD is in DSM and ASD is not although both terms mean pretty much the same thing.

In 2007 the prevalence of Autistic Spectrum Disorder (ASD) in the US was estimated at 6.6 per 1000; which is 1 in every 150 eight year olds. This is 10 times the rate of Autism fifty years ago - a huge increase which has led to much conjecture as to why - toxins in the environment? vaccinations (especially MMR containing thimerosal with mercury)? other heavy metal exposure? dietary factors? yeast? nutrition deficits? new genetic factors? However, it is instructive to note that 50 years ago only Autism counted and now the whole spectrum is being counted in ASD. Additionally, both doctors and the public have learned much better how to recognize these conditions, especially since the addition of Asperger's to the DSM in 1994. Many cases of ASD that were missed before the late 1990's are no longer missed. Whether this explains all or just some of this increased prevalence is unsettled.

So what does cause Autism and its cousins? Some parents of youth with ASD suspect vaccinations were the cause or a contributing cause while many others suspect inheritance. The timing coincidence of a vaccination and the onset or awareness of ASD symptoms reasonably causes one to wonder but does not prove causation. The actual cause(s) of many PDD cases are not yet understood. We do know there is a complex interplay of genetic predisposition and environmental factors or triggers. The #1 cause by far is "idiopathic", which is Latin for "unknown". Thus 75 to 90% of the time there is no specific discernible cause. Of the 10 to 25% we can find a cause for the most common are Fragile X, Down's, Tuberculous Sclerosis, Fetal Alcohol, Angelman's, CHARGE,

Smith-Lemli-Opitz, Cornelia de Lange, or other identifiable or unknown genetic syndromes. All the PDDs (except Rett's) are far more common in males than females. Even cases of PDD without an identifiable cause seem to run in families - if one child has PDD the risk of another sibling having PDD is 5-10%. A few families run a much higher frequency though other cases show no increased family risk.

On a microscopic and biochemical level the brain development pathology (abnormalities) include abnormal numbers, functioning, and connections between various brain cell types in specific areas of the brain including the cerebellum, forebrain limbic system, temporal lobe, band of Broca, inferior olive, and brainstem beginning (at least) as far back as the first month in the womb. PDDs are currently believed to result from interacting multifactorial genetic factors (such as coding errors that lead to faulty protein and enzyme production that impair brain development) that may be influenced by environmental factors (eg, toxins, drugs and alcohol, certain medicines or hormones, maternal illness, infections like Rubella, injury/trauma, in utero stroke, etc) primarily during pregnancy but occasionally around the time of birth (eg, newborn encephalitis) and perhaps in the first 2 to 3 years of life (such as the so far not scientifically validated theories re nutrition or mercury/thimerosal in vaccinations, etc). To be safe, the FDA has acted and states on their website, "Thimerosal has been removed from or reduced to trace amounts in all vaccines routinely recommended for children 6 years of age and younger, with the exception of inactivated influenza vaccine. A preservative-free version of the inactivated influenza vaccine

Diagnostic Criteria for Autism

- A.** A total of six (or more) items from (1), (2), and (3), with at least two from (1), and one each from (2) and (3).
- (1) qualitative impairment in social interaction, as manifested by at least two of the following:
 - (a) marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
 - (b) failure to develop peer relationships appropriate to developmental level
 - (c) a lack of spontaneous seeking to share enjoyment, interests, or achievements of other people (e.g., by a lack of showing, bringing, or pointing out objects of interest)
 - (d) lack of social or emotional reciprocity
 - (2) qualitative impairments in communication as manifested by at least one of the following:
 - (a) delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime)
 - (b) in individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others
 - (c) stereotyped and repetitive use of language or idiosyncratic language
 - (d) lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level
 - (3) restricted repetitive and stereotyped patterns of behavior, interests, and activities, as manifested by at least one of the following:
 - (a) encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus
 - (b) apparently inflexible adherence to specific, nonfunctional routines or rituals
 - (c) stereotyped and repetitive motor mannerisms (e.g., hand or finger flapping or twisting, or complex whole-body movements)
 - (d) persistent preoccupation with parts of objects
- B.** Delays or abnormal or abnormal functioning in at least one of the following areas, with onset prior to age 3 years: (1) social interaction, (2) language as used in social communication, or (3) symbolic or imaginative play.

(contains trace amounts of thimerosal) is available in limited supply at this time for use in infants, children and pregnant women. Some vaccines such as Td, which is indicated for older children (= 7 years of age) and adults, are also now available in formulations that are free of thimerosal or contain only trace amounts. Vaccines with trace amounts of

thimerosal contain 1 microgram or less of mercury per dose.”

Short of extremes like wanton child neglect, abuse, or substance abuse while pregnant PDD's are not caused by faulty parenting. We can miss chances to help or improve the prognosis of a child with PDD but “bad” parenting does not cause Autism, Asperger's or the other PDDs.

Making the diagnosis of Autism, Asperger's or other PDD's is much more than simply counting the number of criteria your child or patient or student meets. As you review the DSM-IV-TR criteria for Autism included in this Medical Memo please note in section “B” onset must be before turning 3 years old and (as you can read in the DSM-IV-TR Autism narrative pgs 70-75) is often before turning 2. Please note in the all important section “A” that both (1) and (2) begin with “qualitative impairments in ...”. Many errors occur in viewing these items as quantitative, not qualitative, impairments. The core disorder or deficit in PDDs is not simply lack of (quantity) or even merely abnormal social skills but it is a certain type (or quality) of atypical, odd, idiosyncratic relatedness or worded differently, a particular kind (or quality) of non relatedness. It's a type of relationship that words, the criteria, don't really capture - it's a feeling you get in the exchange or rather absence of exchange of give and take interaction that is best understood via being around many Autistic Spectrum youth for long periods. It may be less obvious in adults. It is not, for instance, a huge interest in trains or dinosaurs, but an exclusive focus on a part like twirling the trains wheels or memorizing New York city's entire train schedule when you live in Tucson and also don't know and can't express your own feelings. This is why only well trained experienced clinicians should be making this diagnosis which has likely lifelong consequences. Personally, I believe requiring that at least 3 of the 4 criteria in each of A (1), (2), and (3) should be met to increase accuracy. Notice that based on these criteria the only difference between Autism and Asperger's is that in Asperger's the entire A (2) section re language is removed completely. The other 2 sections are identical. Nonetheless, language oddities are common in AS. PDD NOS does not even have criteria ! This is overly simplistic and leads to error. Another error source is taking too lightly the A (3) criteria for

restricted and stereotyped patterns. These abnormal movements, behaviors, and interests must be severe, not merely present to some degree, to be Autism or Asperger's. Otherwise, perhaps PDD NOS is a better diagnosis. Also notice that none of the commonly cited associated findings which occur more often in the PDDs than in the general population are part of the diagnostic criteria - findings like MR (mental retardation), seizures, clumsiness/cooridination problems, sensory integration problems, gastrointestinal problems, etc. These are referred to as “non-specific” because they occur in other conditions too and do not specify or point to one and only one diagnosis. Fever is also non specific in that it can occur with various types of infections (bacterial, viral, fungal, parasitic), cancer, auto immune disorders, and over heating, etc. Fever is a common sign of illness; but not a specific illness.

Classically (as in the old days, ie more than 10 years ago) roughly $\frac{3}{4}$ of youth and adults with Autism score below the 69 threshold for mental retardation in IQ testing and often have severely delayed, limited, and qualitatively disordered language while 25 % have seizures by the time they are adults. Although one can debate the validity of IQ tests for PDDs most persons with Autism will score below 90 (90 to 109 is the “normal” range) with higher nonverbal than verbal scores while most with Asperger's will score above 100 and do better on the verbal section. Many with Asperger's will have superficially “normal” language skills but often have notable tone, rhythm, verbosity, pedantic, and surprisingly concrete thinking despite good intellect (lack of ability to communicate and think well abstractly) problems. The severe impairment of classic Autism is often obvious quickly while the also severe life disruption of Asperger's becomes more apparent as developmental problems transitioning to middle school, high school, college, jobs, roommates, romance, etc caused by being so atypical and out of step in relationships with odd habits (stereotypies), atypical behaviors (eg, ignore hygiene, fashion, or basic manners and courtesy) and excessive but limited interests (eg, obsessive compulsive ritualistic patterns and interests) mushroom over the years and in crises. Motivation, pace, time, money concepts and abilities may be so out of step as to result in disability. Occasionally, the fortunate combination of high intelligence and exclusive

focus in a limited and even esoteric area results in socially rewarded “genius” in art/music, math, rocket science, astronomy, research, computer science, etc. Dustin Hoffman’s character in The Rain Man is classic Autism, not Asperger’s. His gifted extraordinary concrete “savant” skills do occur but rarely. Please read DSM-IV-TR pages 80-84 for more helpful examples and details about Asperger’s Disorder.

Nonverbal Learning Disorder (NLD or NVLD) was originally an educational label that does not appear in DSM-IV-TR and is thus not an official medical psychiatric diagnosis. It is very similar to Asperger’s and is an Autistic Spectrum Disorder (PDD) but is described from an educational, special ed, school psychologist perspective. Imagine we are in a courtyard looking at the same statue. You see it from the front, I see it from the side; someone else may view it from behind. We all see the same

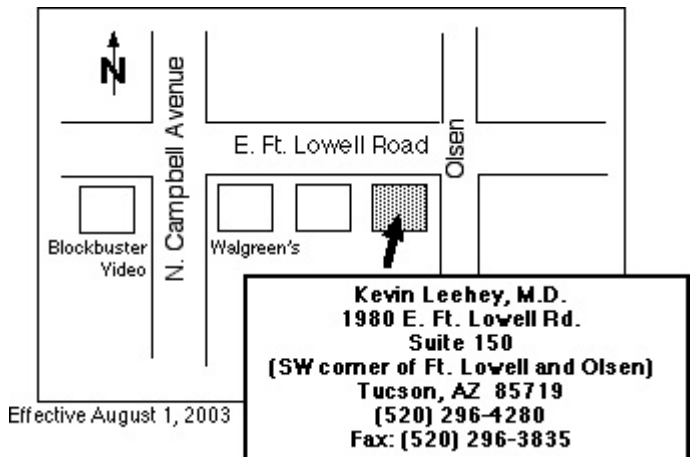
statue but describe it differently based on our own perspective; our own point of view.

Autism, Asperger’s, PDD, NLD, and ASD are all similar, are at least phenotypically related (that means their superficial appearance is similar although their underlying causes and inner workings or pathophysiology may differ a little or a lot). As our knowledge grows these terms may change thereby lessening the current confusing and overlapping gray areas. For now, although imprecise and even misleading, the terms matter greatly because of school special education eligibility, qualifying for services from agencies like Arizona’s Department of Developmental Disability (DDD), for insurance coverage for needed treatment and support services, and to help researchers get a more homogenous sample group for needed research to enlighten us all.

The second issue, Part Two, will cover treatment.

Kevin Leehey, M.D.
Child, Adolescent and Adult Psychiatry

Find this issue of Medical Memo, past issues, and other helpful information at Dr. Leehey’s web site:
www.leeheymd.com



This newsletter is for information only and does not substitute for talking with your psychiatrist, medical doctor, and/or therapist.