Practical Guide: Autism and Other Pervasive Developmental Disorders

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Foreword

Presently the most common and somewhat controversial mental health/behavioral conditions in children as well as others are autism and related disorders. Many television talk-shows deal with these conditions frequently, interviewing a number of "experts" on one side or the other of the many autism controversies, and many people think they "have all the answers," both to causation and prevention, as well as treatment. Widely publicized lawsuits against immunization companies have apparently spawned more lawsuits, and thereby more "experts" because of the demand for more plaintiff's expert witnesses. This present volume is designed to provide well-documented information for people who want to know the "real" truth, and I have carefully documented my references at the end of the book. I've also carefully assessed many of the issues personally, and hope this information will be helpful to those who read it.

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Chapter 1: General Information

Pervasive developmental disorders are characterized by severe and pervasive impairment in several areas of development, including reciprocal social interaction skills, communication skills, and/or the presence of stereotyped behavior, interests, and activities (DSM-IV-TR, American Psychiatric Association, 2000). The qualitative impairments that mark these conditions are distinctly different from what is expected by the person's developmental level or mental age. The DM-ID (Fletcher, Loschen, Stavrakaki, & First, 2007) points out the rapid evolving research in this field and suggests that many people are using a new label for this group of conditions, Autism Spectrum Disorders.

The list of conditions that are included in this group, according to DSM-IV-TR, include autistic disorder, Rett disorder, childhood disintegrative disorder, Asperger disorder, and pervasive developmental disorder not otherwise specified. They also note that this group of symptoms are sometimes observed with a wide group of other general medical conditions, such as chromosomal abnormalities, congenital infections, and structural abnormalities of the central nervous system.

According to DSM-IV-TR, *autistic disorder* is diagnosed when a total of six or more items from 1), 2), and 3) are present, with at least two from 1) and one each from 2) and 3):

1. qualitative impairment in social interaction as shown by at least two of the following:
    - 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
    - failure to develop peer relationships appropriate to developmental level
    - a lack of spontaneous seeking to share enjoyment, interests, or achievements with others (such as lack of showing, bringing, or pointing to objects of interest)
    - lack of social or emotional reciprocity

2. qualitative impairments in communication as manifested by at least one of the following:
    - 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
    - in people with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others
    - stereotyped and repetitive use of language or idiosyncratic language
    - 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 of the following:
    - intense, encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus
    - apparently inflexible adherence to specific, nonfunctional routines or rituals
    - stereotyped and repetitive motor mannerisms such as hand or finger flapping or twisting, or complex whole-body movements
    - persistent preoccupation with parts of objects
4. Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years: social interaction, language as used in social communication, or symbolic or imaginative play. (Also, the disturbance isn't better accounted for by Rett disorder or childhood disintegrative disorder.)

Diagnostic criteria for *Rett disorder* include the fact that the person has apparently normal prenatal and perinatal development, apparently normal psychomotor development through the first five months after birth, and a normal head circumference at birth. After the period of normal development, the following symptoms begin to develop:

- <u>deceleration</u> of head growth between ages 5 and 48 months

- loss of previously acquired purposeful hand skills between ages 5 and 30 months, with later development of stereotyped hand movements such as hand-wringing or hand-washing
- loss of social engagement early in the course, although often social interaction develops later
- appearance of poorly coordinated gait or trunk movements
- severely impaired expressive and receptive language development with severe psychomotor retardation

Rett disorder is usually associated with severe or profound intellectual disability, and seizure disorder may occur. This condition has only been reported only in females, and seems to be far less common than autistic disorder. (It also obviously appears to have marked neurologic problems.)

The essential feature of *childhood disintegrative disorder* is a marked regression in multiple areas of functioning following a period of at least two years of apparently normal developments. After the first two years of life but before age ten, the child has a significant loss of previously acquired skills in at lest two of the following areas: expressive or receptive language, social skills or adaptive behavior, bowel or bladder control, play, or motor skills. Usually acquired skills are lost in almost all areas. This condition is usually associated with severe intellectual disability. Its prevalence is unknown, but it seems to be very rare. Early studies suggested an equal sex ratio, but more recent data suggest that the condition is more common among males.

Essential features of *Asperger disorder* are severe and sustained impairment in social interaction and the development of restricted, repetitive patterns of behavior, interests, and activities. The disturbance must cause significant impairment in social, occupational, or other important areas of functioning. There are no significant delays or deviance in language acquisition, but more subtle aspects of social communication, such as give-and-take in conversation may be affected. The impairment in reciprocal social interaction is gross and sustained, and there may be marked impairment in the use of non-verbal behaviors, such as eye-to-eye gaze. People with Asperger disorder

do not have significant delays in cognitive development or in age-appropriate self-helps skills, and adaptive behavior other than social interaction. Intellectual disability is not usually observed. Symptoms of over activity and inattention occur frequently, and may be misdiagnosed as attention-deficit/hyperactivity disorder prior to the diagnosis of Asperger disorder. No definitive data for prevalence of Asperger disorder are available, but there appears to be an increased frequency when other family members have the condition.

The diagnosis of *pervasive developmental disorder not otherwise specified* is made when the individual has significant problems with some of the symptoms, but doesn't have enough symptoms to meet criteria for the other conditions in the autism spectrum.

The actual incidence of conditions on the autism spectrum is unknown. When the condition was first described in 1943, autism was thought to occur in fewer than three in 10,000 live births in the United States. Current estimates range from as few as 30 cases per 10,000 (about one in 300) to as many as 63 per 10,000. Obviously even the lower estimate shows that autism is not rare—it is more common than cancer, diabetes, spina bifida, and Down syndrome in children. U.S. Department of Education figures show a national increase of 544 percent in diagnoses of autism over a period of nine academic years, ending in 2001, with fantastic increases in some states—diagnoses in Alabama increased 1000 percent, Arkansas 2000 percent, and Kentucky, 2000 percent (Grinker, 2007). Obviously these increases in diagnoses do not mean the same thing as an increase in prevalence of the conditions. When services for these problems become readily available, more diagnoses are made in more people with problems. Also, many years ago people with higher intellectual functioning were not diagnosed, and many lower functioning people were just kept at home or placed in out-of-home residential care. Also, even in public institutions, the rules apparently stated that only people with intellectual disability were admitted, so low-functioning people with autism were listed as "mental retardation with autistic tendencies" (personal communication, Arkansas Children's Colony, 1965).

Chapter 2: Early History

While today autism is a very widely used term, what is understood by autism has changed a great deal since first being introduced in scientific dialogue nearly 100 years ago (Kumbler, Haack, & Herpertz, 2008). Autism is one example of the influence of the psychoanalytic school of Sigmund Freud on scientific psychiatry at the beginning of the 20th century. A bit later a Swiss psychiatrist, Eugen Bleuler, used the term "autism" to refer to a classical symptom of schizophrenia, but he wasn't very specific in his definition, so the term became used for other symptoms also. While today the term autism is exclusively used in psychiatry for autism spectrum disorders, it is used a bit more widely in everyday communication.

Apparently people with symptoms consistent with autistic disorder were present in nineteenth-century London, England, based on case histories from the notes of Dr. W. H. Kickinson at Great Ormond Street Hospital for Children—three cases meeting the DSM-IV criteria are described in detail (Waltz & Shattock, 2004). In 1920, Lightner Witmer, a pioneer in experimental psychology from Philadelphia, published the first detailed case report of a "psychotic" child, who always wanted to be left alone, but had good rote memory and excellent visual and spatial skills. He was diagnosed with schizophrenia.

By providing the first real description of infantile autism in 1943, Leo Kanner (1894-1981) substantially influenced the field of child psychiatry. His scientific career began at Berlin's Charite University Hospital, and he emigrated to the United States in 1924. He subsequently headed the first child psychiatry division within a pediatric hospital, at Johns Hopkins University. In 1935 he wrote the first book in English of child psychiatry, and championed a number of liberal causes, and emphasized an humanitarian approach to dealing with autistic children (Sanua, 1990). Dr. Kanner up-dated the life histories of the 11 children he first described in 1943 in 1954, and emphasized the importance for continued follow-up in these persons.

In 1944 a pediatrician from Vienna, Hans Asperger, described for the first time a number of boys with so-called "autistic psychopathy" who had higher intellectual capabilities than people with autistic disorder (Hippler & Klicpera, 2005). In retrospect,

many think that Michelangelo met the criteria for Asperger disorder or high-functioning autism, in view of his single-minded work routine, unusual lifestyle, limited interests, poor social and communication skills, and issues of life control (Arshad & Fitzgerald, 2004).

History of claims for causation of autism and of claims for various "very successful" treatments will be discussed in later chapters.

Chapter 3: What Causes Autism?

At the end of his second article, mentioned in the last chapter, Leo Kanner ventured a guess as to what caused autism—he assumed that these children came into the world with an innate inability to form affective contact with people, just as other children came into the world with innate physical or intellectual handicaps. He also noted that parents of autistic children had similar personality traits: "cold, bookish, formal, introverted, disdainful of frivolity, humorless, detached, and highly rational and objective. He felt that there was little hope for a cure (Offit, 2008).

Not everyone was as pessimistic as Kanner. The first to offer a cure for autism was Bruno Bettelheim, a Viennese-born psychoanalyst, who believed he had found the problem—bad mothers, whom he called "refrigerator mothers." If children were to recover, they needed to be taken from their homes and "thawed." He received a grant from the Ford Foundation and founded the Orthogenic school in the Chicago area. He published a book in 1967 in which he claimed to have successfully treated forty autistic children, all with dramatic results. University medical school pediatrics training programs at the time taught that this was the cause of autism (personal communication, University of Texas, 1959). A later, closer, look at Bettelheim's school showed his claims of success were fraudulent, as well as causing mothers to feel guilty and ashamed (Olfit, 2008).

In 1998 a gastroenterologist named Andrew Wakefield held a press conference in order to tell the world he had found the cause of autism. He told the group that he and his team at Royal Free Hospital (felt to be an excellent medical school) had inserted fiber-optic colonoscopies into eight autistic children and found lots of abnormalities in their intestines, which he felt to be due inflammation caused by the MMR (measles/mumps/rubella) vaccine. He said he had been approached in 1995 by very well educated, articulate parents who told him stories of their children's deterioration into autism. They said their children had developed normally for the first 15-18 months of life, when they received the MMR vaccine. After a variable period of time the children regressed, losing speech, language, social skills, and imaginative play. Wakefield said that he thought when the vaccine was injected, the vaccine virus

traveled to the intestine and caused infection and inflammation. Harmful proteins, now able to pass through a damaged intestine, entered the bloodstream and eventually the brain, causing autism. He also said that his findings would be published in the *Lancet*, an important British medical journal.

Simon Murch was the second author on the *Lancet* paper and a quite well-respected pediatric gastroenterologist. He was excited about the finding of intestinal problems in children with autism, but he knew that Wakefield hadn't proven his claim against MMR—he had only raised the possibility. He said that the link was unproven and measles is a killing infection, and he feared that this announcement might precipitate a scare and immunization rates would go down. This would mean that measles would return and children would die.

The day after Wakefield's press conference the British media exploded, with many frightening headlines. In response, thousands of British parents refused the vaccine for their children. During the next few years, journalists wrote more than 1,500 articles about Andrew Wakefield, the MMR vaccine, and autism. Even more British parents stopped vaccinating their children. In the months after Wakefield's' warning the proportion of children receiving MMR vaccine dropped from almost 90 percent to 50% in some areas of London. Small outbreaks of measles first appeared in upper-middle-class elementary schools in London, then other outbreaks followed, first in London, then in Scotland and Ireland. By 2002, hundreds of children in Ireland had fallen ill with the disease. One small Dublin hospital admitted 100 children with pneumonia and brain swelling caused by measles, three of whom died. Despite all this, a number of scientific "experts" supported Wakefield's findings.

At a U.S. congressional Committee on Governmental Reform meeting in 2000 in the Rayburn House Office building in Washington, a debate was set up for parents, Dr. Wakefield, and others to discuss the autism/vaccine question. After the parents, Dr. Wakefield, and his supporters spoke, Dr. Brent Taylor, a professor of community medicine and child health at the same university where Dr. Wakefield worked, spoke, and specifically stated that the belief that MMR is the cause of autism is a false hope. He showed his data, which showed that the autism rates in the United Kingdom had

clearly been rising before MMR was first introduced in 1988, and that children who had received the vaccine weren't at greater risk. Other experts challenged what he said, and basically said he was lying, which he wasn't, and which his data showed he wasn't. Further supportive discussions by Dr. Taylor were presented, including data from the World Health Organization. Continued press reports, largely supporting Dr. Wakefield, caused more parents world-wide to refuse to give MMR to their children, and even actors and sports celebrities were quoted as supporting Dr. Wakefield's findings.

Then, in 2004, Brian Deer, an investigative journalist in London found that Andrew Wakefield wasn't exactly what he appeared to be—and his science, his theories, and his career all came crashing down. He had stated that his investigations were approved by the Ethical Practices Committee, but they hadn't been. Very complex procedures had been performed on the children under general anesthesia, including spinal taps, fiber-optic scoping of the intestines, biopsies, and large quantities of blood drawn. Several children had trouble with the anesthesia, and one five-year old was in critical condition because his colon had been perforated. While he had reported on one fiscal supporter of his project, he neglected to mention that his largest supporter was a personal-injury lawyer. At least five of the eight children in the study were clients of the lawyer, and obviously the children's parents had a financial interest in finding a link between MMR and autism. Other lawsuits were also involved. (The lawyer involved was Richard Barr, who had considerable experience suing pharmaceutical companies.) Later editors of the *Lancet* apologized on BBC for not checking the written material as thoroughly as they should. The Royal Free Hospital team wrote a retraction in the *Lancet* stating that the previously published paper really showed no causal link between MMR vaccine and autism, as the data were insufficient. The investigative reporter later found that an investigative commission in London had given money to the lawyer Richard Barr--$20 million to his law firm—and had given $10 million to doctors and scientists, to be recommended by the law firm. Andrew Wakefield got $800,000 to support his research, which was far

more than he had admitted receiving earlier. He also had conflicts of interest in other ways.

After no real proof was found that MMR vaccine caused autism, the issue of mercury preservative in vaccines (thimerosal) became the new "cause," and many people thought that might be what was causing the condition, since most vaccines were being marketed in multi-dose vials where some sort of preservative seemed to be necessary. While no one had any proof that mercury substances caused autism, great interest developed. The mercury was removed in 1999, and in 2000 two mothers with children with autism, one of whom was a nurse, looked at their children's earlier immunization records and saw that their earlier vaccines had contained mercury. They published a paper on this in *Medical Hypotheses*, which has a circulation of about 200. Someone in the press saw it and began to write about the issue, and again, the flood-gates opened. Some researchers suggested that chelation therapy—giving a drug to get the mercury out of the body's system—might be helpful. (Some very bizarre theories were tested on children). Again, a number of well-known people, not researchers, including Robert F. Kennedy, Jr., and John Kerry came out supporting the idea that vaccines caused autism.

Finally, in 2004, after reviewing more than 200 epidemiological and biological studies of the relationship between thimerosal and autism, a committee of the Institute of Medicine made a statement to the press and the public: "The committee concludes that the body of epidemiological evidence favors rejection of a causal relationship between thimerosal-containing vaccines and autism. The committee further finds that potential biological mechanisms for vaccine-induced autism that have been generated to date are theoretical only." They recommended that autism research money should be spent on more fruitful leads. Many more articles were published in well-known scientific journals after this. Many parents and others were displeased about this research, and actually made physical threats against some of the authors (Offit, 2008).

In 1986, after a series of lawsuits that threatened to end vaccine manufacture for American children, Congress passed the National Childhood Vaccine Injury Act,

which set up the Vaccine Injury Compensation Program, where parents thought their children had been harmed by vaccines could sue the federal government for compensation, making their argument in front of federally appointed judges. As a consequence, the number of lawsuits against vaccine makers went down rapidly. In 2007 the judge at this court said that the first case was one of nearly five thousand children to have filed claimed under the Vaccine Act, and these had been grouped together in a joint proceeding known as the Omnibus Autism Proceeding. Interestingly, after lengthy discussions from various well-educated people who spoke in the child's behalf, and stated that of course her problems were due to the vaccines, one of the government defense team witnesses showed some early videotapes of the girl, and pointed out the features she presented that made her diagnosis of autism quite obvious—then showed that the videos were made significantly earlier than the child had received any immunizations whatsoever. In February of 2009 this court ruled against the petitioners (Stewart, 2009). Also, autism is present in cultures all over the world (Grinker, 2007) and in many of these areas children may not have the opportunity to even receive immunizations.

The American Academy of Pediatrics has developed a new series of audio interviews which offer sound advice for parents and others who have concerns about vaccines: www.cispimmunize.org/fam/soundadvice.html.

Also, autism is an extremely heritable disorder. The risk of an individual developing autism if he/she has an affected sibling is 22 times as great as in the general population (Glatt, Faraone, & Tsuang, 2007). When researchers first described the genetic basis of autism, they hoped that one gene was responsible, but, unfortunately, the genetic basis of autism is far more complex. Also, though all autism is genetic, not all autism is inherited—sometimes spontaneous changes occur in certain genes (Offit, 2008). (As autism genes have become better defined, people have become more and more excited about the possibility for specific treatments, but this appears to still be quite a long time in the future.)

While the discussion in this chapter had dealt mainly with causes of autistic disorder, as noted in Chapter 1, a number of medical conditions can give rise to the

cluster of symptoms in autism spectrum disorders, such as chromosomal abnormalities, congenital infections, and structural abnormalities of the central nervous system. As an example, when mothers have German measles (rubella) during pregnancy and their babies have congenital rubella syndrome, many of these children have symptoms that place them in the autism disorder spectrum.

Chapter 4: Treatment of Autism Spectrum Disorders

Although some parents and others have been skeptical of the scientists and public health officials who failed to find that vaccines caused autism, they haven't been anywhere near as skeptical of the vast array of autism therapies, all of which are claimed to work and all of which are based on theories that are ill-founded, poorly conceived, contradictory, or disproved (Offit, 2008):

- One individual claimed that vaccines caused an immune response against the sheath that lines nerve cells, and that drugs like steroids, which treat inflammation, could treat autism. (Autistic children don't have inflammation of their nerve sheaths.)
- Another person claimed that cod liver oil, which contains vitamin A, extended the field of vision of autistic children. (Autistic children don't have a restriction of their visual fields.)
- One man claimed that spinal fluid was getting trapped in the brains of autistic children and causing increased pressure. He said he could relieve the pressure by manipulating the head. (Autistic children don't have increased pressure on their brains.)
- Two individuals claimed that autistic children had more mercury in their bodies than nonautistic children and getting rid of it by chelation therapy could help. (Autistic children don't have excess mercury in their bodies; cells damaged by heavy metals aren't healed by chelation.)
- An osteopathic physician prescribed a chelation medication which is rubbed on the skin and has never been shown to cause the excretion of heavy metals and also injected children with filtered urine.
- One treatment which is very pervasive is diets free of casein (dairy products) and gluten (wheat, barley, and rye). Studies on these treatments have not shown efficacy, but they are still being used. (Studies have shown that autistic children deprived of calcium and vitamin D in dairy products have developed osteoporosis.)

- Injection of secretin, an intestinal hormone, was supposed to cause dramatic results. (Children injected with it intravenously were judged by their parents to have improved, but so were children who had been injected with salt water.)

All of these and many other treatments are diverse, expensive, and unproven, and some are even dangerous.

Only one medication is presently approved by the Food and Drug Administration for use in the treatment of children with autism and irritability, risperidone. Several other drugs are presently in testing stages for use for irritability and/or aggression, but nothing is being tested for the basic symptoms of autism (Stigler et al., 2009).

Very early diagnosis of autism is extremely important, since communication and behavioral interventions are probably by far the best types of treatment programs for children with this condition (Jellink, Patel, & Froehle, 2002). McConachie and group (2005) evaluated a training course for parents, designed to help them understand autism spectrum disorders and to facilitate social communication with their young child. Parents received either immediate intervention or delayed access to the course. Fifty-one children aged 24 to 48 months were involved. Outcome was measured seven months after beginning of the study in parents' use of facilitative strategies, stress adaptation to the child, children's vocabulary size, behavior problems, and social skills. The training course was well received by parents and had a measurable effect on both parents' and children's communication skills. A number of clinical studies are presently underway assessing the types of intervention that seem most effective, and research in this area will be on-going (Vismara, Columbi, & Rogers, 2009; Aldred & Green, 2009).

Altemeier and Altemeier (2009) remind us that although autism spectrum disorders are 90% genetic, they respond dramatically to intensive early training, probably at least partially because many of the genetic variations associated with ASDs involve activity-dependent regulation in the brain, or synapse (connection) development that depends on after-birth learning and experience, and, also, brain plasticity (the ability to change structurally) declines after six years of age.

Chapter 5: Current Research

Present research programs on the genetics of autism spectrum disorders and very early-life training programs have been mentioned earlier. Dr. R. R. Grinker, a cultural anthropologist mentioned earlier, recently received a grant from the National Alliance for Autism Research to conduct the first-ever epidemiological study of autism in Korea (Offit, 2008).

Researchers are looking for 1,200 women who have a child with autism and who are pregnant with another child, as part of a large study of the disorder, primarily funded by the National Institutes of Health, along with money from Autism Speaks, an advocacy group. After birth they will monitor the babies until they are three years old (Dooren, 2009).

(Books that have been written by family members and persons with autism themselves are listed as a supplement to the list of cited references.)

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Books of Interest

Barnbaum, D. R. (2008). *The ethics of autism: Among them, but not of them.* Bloomington, IN: Indiana University Press. (contains a number of personal reports and pictures done by individuals with autism)

Gardner, N. (2008). *A friend like Henry: The remarkable true story of an autistic boy and dog that unlocked his world.* Naperville, IL: Sourcebooks, Inc. (contains a mother's story of the problems getting services for her son, and a later report written by the son)

Grinker, R. R. (2007). *Unstrange minds: Remapping the world of autism.* Cambridge, MA: Basic Books. (a father, a daughter, and a search for new answers)