Autism - Open Access
ISSN: 2165-7890

Berman, Autism Open Access 2015, 6:1
DOI: 10.4172/2165-7890.1000158

Perspective Article | Open Access Journal

# Autism Spectrum Disorder-A Paediatric Dentist's Perspective

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**Rec date:** September 02, 2015; **Acc date:** December 14, 2015; **Pub date:** December 21, 2015



## Introduction

Most recent reports published by the Autism and Developmental Disabilities Monitoring network under the auspices of the Centres for Disease Control and Prevention predict that one in every 150 children will suffer from some kind of autistic spectrum disorder (ASD), with a 4-to-one prevalence of boys over girls [1]. The etiology of ASD is currently unknown, and there is no known cure [2]. In a 2009 study involving phone conversations with 78,000 parents of US children who were identified by a health professional as having an ASD, the prevalence of ASD amongst 3 to 17 years old was estimated to be as high as 1:100 [3].This article discusses the diagnosis, etiology, and clinical aspects of ASD that are pertinent to dental professionals who treat these patients.

## Statistics

The alarming rise in the statistics of ASD and the multifaceted efforts of parent-led advocacy groups has put the ASD issue front and centre for scientific researchers, healthcare providers, educators, and the broad expanse of society [4]. Bombarded with news reports concerning a host of social and health issues, the sheer mass of global statistics becomes an impersonal and overwhelming blur. In the case of autism, the statistics have taken on real meaning with a palpable relevance and challenge for the dental profession.

Whether the statistics represent an increased incidence of autism or whether society has simply become more aware of the condition (or both) is a matter of conjecture. There is also the question of whether the parameters for labelling a child autistic have become so broad that the alarming statistics are distorted [1,5,6]. However, there is no question that those who actively work with children are encountering patients with increasing frequency that have been diagnosed with autistic disorders. Dentists treat very young patients in an up-close, personal atmosphere where communication is absolutely essential. Often, dentists may be the first healthcare providers to recognize that a one or 2-year-old child has some type of extraordinary pervasive behavioral disorder that a parent, fearing the worst, may have suspected instinctively and emotionally but never faced objectively. In many instances the idiosyncratic behavior is mistakenly attributed to immaturity or the "terrible 2s."

## Diagnosis

ASD is a "catch-all" term that refers to a highly variable range of neurodevelopmental disorders observed in young children [7]. Currently, there are no empirical biological tests (e.g., blood tests or brain scans) for ASD that are reliable. Parents, caregivers, teachers, or healthcare workers may notice some aberrant behaviors, language delays, odd speech patterns, or mood swings that indicate a need for a consultation with various specialists. By the time a child has reached 24 months of age, 80% of parents of children with an ASD are noticing the lack of babbling or meaningful gesturing, repetitive hand flapping, failure to respond to their name, excessive single word usage without spontaneous phrases, an aversion to hugging or touching, and difficulty in making eye contact. All of these symptoms and others may be mild, or severe enough for parents to seek professional advice, hoping for a remedy [8,9].

The American Academy of Pediatrics recommends a well-child screening for ASD at 18 to 24 months of age using formal autism screening tests [10]. Its most recent policy statement suggests that "early identification of developmental disorders is critical to the well-being of children and their families. Further, it is recommended that developmental surveillance be incorporated into every well child visit and any concerns be addressed promptly. The sooner the disorder is identified, the soon-er that interventions can be instituted" [11].

The definitive diagnosis of ASD is usually made by pediatricians, psychologists, or psychiatrists who institute a process of analysis which involves a developmental and clinical history, tests for cognitive function, and assessment of receptive and expressive language skills. Evaluation of each of these components requires a great deal of time and patience, especially if the child is very young or is reluctant to cooperate. Pervasive developmental disorders, Asperger's syndrome, Rett syndrome, and childhood degenerative disorders are all considered a part of the ASD group, but the distinction between the various entities is not always clear.

### Pervasive developmental disorder-Not otherwise specified

The diagnosis of pervasive developmental disorder is made when some autistic symptoms are evident but the entire set of criteria is not present. Generally these children are higher functioning, have fewer communication problems, and often respond very positively to interventions.

### Asperger's syndrome

This condition (named for Hans Asperger) is one of the higher functioning forms of ASD and is distinctive because these children generally speak with clarity and have the ability to have normal conversations [12]. Some authorities prefer to consider Asperger's syndrome separate and apart from autism. A definitive diagnosis or differentiation may be nearly impossible to establish. Persons with Asperger's think very literally without subtlety or nuance…concrete rather than abstract. They may speak obsessively about a particular subject of special interest to them and be reluctant to move on to another topic. Their ability to socially interact is noticeably impaired, therefore establishing or maintaining friendships with their peers is negatively impacted [13-15].

### Rett syndrome

Rett syndrome is a degenerative disorder, which has its onset in infancy, and all of the autistic symptoms are more severe in nature

both physically and mentally. The etiology is thought to be a genetic mutation, predominately affecting girls. The self-infliction of pain, nonverbal temper tantrums, lack of speech, and lack of interest in their surroundings are often accompanied by severe physical limitations as well. The condition is rare but devastating [16].

## History and Nature of Autism

Autism, as an entity, was first described by Hans Asperger in 1938 [17]. Leo Kanner of Johns Hopkins first used the term "autism" in the modern sense in the 1940s, referring to it as "early infantile autism" [18].

Based on the Greek word "autos," meaning "self," autism was extrapolated to mean "alone, preoccupied with self, a withdrawal into private fantasy," the kind of self-isolation where any stimulation from the outside world may be an intrusion [19].

The diagnosis of ASD is based on both testing and behavioral observation of an extensive list of characteristics such as impairment of social interaction and communication, restricted interests, and focus and repetitive behavior. These characteristics become more evident when in the course of treating these children clinicians observe the lack of eye contact, echolalia (repetition of the last word you say), self-infliction of pain, addiction to a specific routine and sometimes expert recall of complicated minutiae out of context with the moment. About 30% to 50% of these children do not develop natural speech patterns [20,21].

In some instances children affected by ASD will possess an amazing aptitude for music or art beyond the average, but at the same time they may be incapable of managing the more mundane tasks of everyday life, such as dressing themselves, brushing their teeth, etc. [22,23].

The social deficits are what may be most notable in individuals with ASD. While they may desire the company of others, they frequently lack the intuition of a "give and take" relationship with another person, making it difficult for them to interact or sustain long-term friendships. It's not that they want to be alone they simply cannot process the more subtle emotions or recognize the emotional cues that we take for granted. Hurting another person's feelings is something they may not grasp [24,25]. Sometimes the brain of an autistic child is described as a control centre that is overloaded with too many simultaneous messages coupled with the inability to receive long distance calls. They may not do well with the "what ifs" in everyday life. They need to know "what is!" They won't appreciate or understand sarcasm or irony. They stand apart from people with typical behaviors, as if looking into a strange world. Dr. Temple Grandin [26]. A noted autistic author remarks, "Her inability to understand the social communications between normal people left her feeling like an anthropologist on Mars."

One of the pervasive symptoms associated with ASD individuals is their compulsive repetitive behaviour [21,22] Repetitive behavior is generally measured on a repetitive behavior scale which evaluates purposeless movement (such as hand flapping or body rocking), compulsive behavior (arranging objects in a certain pattern), ech-olalia (repetitive speech), and incessant humming. Ritualism is characteristic: the compulsion with always does things in the same order, the same chair with the same person, one certain toy, one particular television program, etc. [27]. Self-abuse, ranging from biting their hands or hitting themselves in the head as if they were oblivious to pain, is an oft encountered symptom. Parents will say that their child has no definitive awareness of where his body is in space. In the more severe situations, various chew toys or gloves are employed in an attempt to mitigate the physical damage [27,28]. Dentists should not confuse the self-infliction of pain as a sign that they can treat these patients without local anaesthetic. These patients do feel pain, but their interpretation of that external pain is unclear. Some autistic authors have related that the self-infliction of pain gives them a "sense of reality."

It should be noted that the variations in symptoms and behavior make accurate diagnosis of the various ASD entities very difficult. One child may be nonverbal, using gestures or screams, whereas another child may talk incessantly and compulsively about a favorite subject. This kind of behavior should not be mistaken for a lack of intelligence. Many of these children have surprising powers of observation and hidden talents despite their seeming inability to relate socially to the everyday world. "Autistic savants" exhibit these characteristics to the extreme [22].

## Etiology

The etiology of ASD is an enigma. Highly regarded researchers are of the opinion that there is probably more than one cause since the disorder can have such disparate manifestations. Genetics, environmental poisons, neurologic psychopathy, dietary deficiencies, and allergies have all been implicated [5]. At one time the villains were thought to be "refrigerator moms," referring to mothers who were not able or willing to bond with their newborn babies [29]. The theory was that a mother's coldness toward her infant could cause that child to retreat from the world.

Hypotheses revolving around allergies, food poisoning, vitamin deficiencies, heavy metal poisoning, pesticides, and dietary factors have launched the marketing of a plethora of faux solutions such as fat-free diets, fat-laden diets, low-sugar diets, gluten-free regimens, blood transfusions, hyperbaric treatments, chelation, and antioxidant supplements. Web sites and other sources too numerous to mention are replete with so-called miracle treatments and often unfounded interventions. In the absence of evidenced-based solutions there seems to be an abundance of "experts," professional and nonprofessional, willing to experiment on these children with the approval of well-intentioned parents.

There are those, with no evidence whatsoever and referencing pseudoscience, who still preach loudly against vaccinations and mercury exposure as being the culprits. Significantly, in February 2010, The British journal Lancet issued a formal retraction of a flawed study published in 1998 linking autism to the measles, mumps, and rubella (MMR) vaccines [30]. Specifically, the editors stated, "We fully retract this paper from the published record. It should never have been published." In addition, the co-authors of the study have been discredited and in some cases their licenses to practice have been revoked [30]. No reliable health agency or evidenced-based scientific study has associated vaccines with ASD [31,32]. Moreover, Thimerosal, an ingredient of the vaccines, was preemptively re moved from most vaccines in 2001, and yet the incidence of ASD continues to accelerate [33] (Note: There is also no evidenced-based information that would implicate dental amalgam in the etiology of ASD, and therefore there is no justification for the removal of existing dental amalgam restorations based on health implications [34,35].

The most current scientific evidence indicates a strong genetic predisposition to ASD, even to the extent that if one child is affected

the tendency will be for the siblings to have the disorder as well [36,37]. In the case of identical twins, there is a 90% chance that both children will be affected (8% in the case of fraternal twins) [36,38]. Genetic testing is now recommended for parents who have an autistic child in order to determine the likelihood that subsequent children will also have the disorder.

Additional perspectives frequently add to the complexity of the causation of ASD. The results of a study involving 1,237 Swedish children suggest that children of parents who were hospitalized for a mental disorder were 70% more likely to be diagnosed with ASD [39]. The preponderance of reliable scientific reasoning is that once predisposed, certain environmental factors may trigger the onset of symptoms [40]. Regressive Autism Complicating the etiology puzzle is the observation that the majority of children with ASD exhibit early onset of the symptoms, whereas about 30% of the children seem to be normal when evaluating all the development markers (talking, walking, gesturing, smiling, eye contact, socialization, etc.) and then suddenly at 2 to 3 years of age a regression takes place [41]. Initially, it may seem like a hearing loss, but then there is a noticeable decline in word skills and social play. The children begin to lose their acquired language and social awareness and motor functions as well.

They may no longer want to be hugged or held. Temper tantrums, aggressiveness, and extended crying spells become the routine [2,42]. This period of time ironically coincides with the age that toddlers are receiving their appropriate vaccinations (DPT, MMR, etc.), and therein lies the continuous speculation about a cause-effect relationship between vaccines and autism. Again, despite the controversy, no link has been found to vaccines [5,43].

It should be further noted that numerous studies are underway to investigate the possibility that regressive autism has an autoimmune basis [41]. There have even been studies which indicate a significant association between advancing paternal age and ASD. Offspring of men aged 40 years or older were 5.75 times more likely to have ASD [44,45].

### Treatment for autistic spectrum disorder

Given the fact that the etiology and the increased incidence of the various ASD disorders are scientifically puzzling, treatment modalities tend to be wide ranging and very much trial and error, especially since there is no cure [5,8,11]. The nature of autism renders family members and their supporters vulnerable to highly dubious etiological theories and interventions, many of which can be characterized, at best, as pseudoscientific. But parents searching for answers will grasp for straws with the hope of finding the miracle cure [46].

Once the prescribed (previously mentioned) screening tests are completed, recommendations are made regarding appropriate interventions. The main goals of the interventions are to lessen the child's behavior deficits and family distress and to increase quality of life and functional independence.

Nondrug Interventions Under the care and supervision of pediatricians, neurologists, and psychologists, nondrug treatment may include applied behavior analysis (ABA), speech therapy, occupational therapy, and structured teaching.

ABA, initially introduced by Lovaas, is a frequently employed and extensively studied approach to treating children with autism [47-49]. It is considered by many researchers and clinicians to be the most effective nondrug therapeutic approach [50,51]. ABA incorporates several techniques designed to improve a child's social, motor, and verbal behaviors. An example of an ABA technique would be to say to a child, "Please pick up the paper." If the child complies, you say, "Good job, here's a sticker!" If the child doesn't respond, you place his hand on the paper and try again. ABA has its strong advocates among parents and therapists who in uncontrolled studies report "noticeable progress." However, there are also highly regarded authorities who disparage this treatment because the children tend to respond robotically without developing any sense of integration. Is the perceived behavior modification too literal and concrete? Does this methodology prepare a child for real life situations? Be that as it may, ABA is the standard of nondrug intervention.

Further complicating the ABA option is the cost of implementing the program that involves teachers, therapists, and most of all, parents. To be effective, the therapy requires a time commitment of 25 to 30 hours a week and a financial obligation of thousands of dollars per year [52,53].

Controversies with the ABA formula also arise when more aversive techniques are utilized, i.e., yelling or restraining. But common sense would indicate that sometimes it is necessary to get a child's attention in a more "in-your-face" manner.

One of the dilemmas that parents often face is the inability to determine from moment to moment and day to day whether a child's behavior is "autistic." Is the child simply acting out or misbehaving? Does that child merely need discipline like any other child?

### Drug interventions

Evidenced-based, controlled studies are the state of the art when researching and implementing treatment for any disease entity. Asking a specific question and looking for that specific answer would be ideal; however, ASD is not a single disease, therefore controls, double blind, and duplication, all of which are part of good research, are not a reality at this point in time. So much of the information being circulated on the Internet and through other media is anecdotal, at best. This state of affairs is especially applicable to drug interventions for children with ASD.

It often becomes necessary to introduce drug regimens to help mitigate negative behavior or improve focus sufficiently to allow a child to function at home or in school [54]. More than half of the children in the United States diagnosed with ASD are being treated with one or more mood-altering drugs. Tranquilizers, antidepressants, anticonvulsants, stimulants, and antipsychotics are prescribed alternately or simultaneously with the intent to modulate the child's mood swings, help the child focus, and enhance social interaction [55].

In 2006, the FDA approved Risperdal or Risperidone (an antipsychotic drug used primarily as a treatment for schizophrenia and for bipolar disorder) for the treatment of irritability in children and adolescents with ASD. It has become an especially popular medication when prescribed for children whose behavioral demeanour includes violent meltdowns and extreme aggression [56].

The clinical results are mixed because of the unpredictable interaction of the various drugs, and titrating the optimum dosages can be problematic. In many cases not only are the drugs ineffective but the listed side effects are often profound and can produce the very anxieties, temper explosions, and social dysfunction that the medication was intended to mitigate [57]. It should be noted that the validity of many treatment interventions is not supported by scientific studies. Prescription of these drugs is often driven by commercial marketing or parent request [46,57].

Even if certain ineffective therapies may be harmless, they waste the child's valuable therapy time, deplete the parents' financial resources, and perhaps most importantly, they delay the implementation of more effective alternatives. Moreover, the initiation of more bizarre interventions without regard for the far-reaching side effects does not seem justified. Prime examples would be chelation to purge heavy metals or hormonal therapy to suppress testosterone. But, from an overwhelmed parent's perspective, the perceived benefit of any particular therapy regimen for a child with an ASD apparently outweighs the lack of evidence-based science. Something is better than nothing. Or is it?

Parents of children with "special needs" deserve special understanding. Whether contending with cancer, juvenile diabetes, congenital heart disease, cerebral palsy, cleft palate, deafness, blindness, brain tumors, Down syndrome, or birth defects, these parents travel a tough road physically and emotionally. In the case of autism, the stress and strain are monumental. They are usually faced with a bewildering diagnosis, hit-and-miss treatment options, overwhelming financial obligations, lack of personal and psychological support, and challenging educational and child care options [58,59].

The financial burdens are sometimes insurmountable, involving the cost of medical care, day care, extra therapies, education, and special classes. Family income is often seriously diminished because the responsibilities at home limit working hours and prevent the pursuit of employment opportunities and advancement on the job [60,61]. Frequently, dentists meet parents who simply "hang on" to a particular job because of the need to maintain the insurance benefits, as limited as they may be.

Mothers are more affected than fathers in regard to work issues because, in general, mothers usually assume the major day-to-day childcare, doctor appointments, school, therapy sessions, etc. When money gets tight, families go further into debt, magnifying the financial burden [52,60].

### Formal education issues

Another complication encountered by ASD children, their parents, and caregivers is the difficulty with structuring the child's formal and informal education. The ideal situation would be for the children to attend public schools where state and federal taxes cover the major expenses. In the case of special needs children, the cost of providing the special teachers (5:1 student-teacher ratio), assistants, and facilities can be out of control for the public school system and an impossible burden for the individual family [62]. The Individuals with Disabilities Education Act is a federal mandate on all states to provide an Individualized Educational Program for every student diagnosed with a disability with the added proviso that this education be provided in the least restrictive environment (LRE). The LRE is defined as mainstreaming with peers wherever possible. Added to the mix is the Free Appropriate Public Education Act [62,63]. This maze of well-intentioned legislation by its very nature (least restrictive, individualized, free, and appropriate) opens up so many issues which are subject to a wide range of interpretation. Frequently, complications arise from disagreements in diagnosis, failure by the system to provide what is promised, and overly optimistic expectations by parents, resulting in acrimonious debate and sometimes costly lawsuits.

Most devastating of all to parents who spend every waking moment searching for answers and trying to love and care for a child is the reality that often their son or daughter may not reciprocate the same affection or share their emotions. They live every day with the hope of the unsolicited hug or kiss and instead they suffer the unpredictable explosion of disruptive behavior that the parent is powerless to control. They have to be content with the few sporadic moments of joy when the child seems to miraculously do something really positive.

What must it be like to wake up every morning thinking that your child is progressing, only to find that yesterday's experiences are lost in a void and you're starting from scratch again? This phenomenon is very important for dentists to remember when initiating dental treatment. To paraphrase the Beatles song, a "slow down, you're going too fast" approach is needed.

Imagine trying to watch television when each and every day the image is blurred and the channels you receive come and go without the benefit of a remote control. The unpredictability of the behavior saps the energies, patience, and strength of even the most dedicated individuals and loving families. Not surprisingly, clinical depression can be a common phenomenon amongst parents of autistic children [64-66].

Families often speak about "never being able to take a break" or "no rest for the wicked." Remember that the autistic child 2 or 3 years of age may grow into a 6-foot tall, 200-pound 15year-old. One of the most heart wrenching experiences for these parents is the realization that they can no longer physically care for who once was their "little baby" and they now have to "institutionalize" him. Their emotions vacillate from guilt to relief and back again…the guilt of "surrendering" and the relief of now being able to go out to a movie or for dinner, or to go on vacation...the opportunity to feel some normalcy in their lives.

Younger siblings of ASD children seem to mimic the slower cognitive development of the older sibling during their preschool years and then they seem to close the gap when compared with families with "normal" children. There is a tendency for parents to neglect the "normal" sibling because of the intensive attention a child with ASD requires. Often, the siblings of special needs children grow up to be the most empathetic, self-effacing adults, and in other instances they harbor a deep resentment because of the inadvertent parental neglect. Parents struggle to find "alone time" with their normal child whenever possible [67-69].

Interestingly, fathers and mothers often react differently to the challenge of coping with a special child. Fathers can sometimes feel helpless and guilty, and in their frustration at not being able to make their child better or fix the problem they will choose to leave. Mothers, although feeling equally guilty and frustrated, tend to shoulder the day-to-day responsibility, resolving to care for the child no matter what and make the best of it. Fathers tend to look for the miracle cure somewhere. Regulatory problems are associated with maternal stress, whereas externalizing behaviors are associated with paternal stress [59,60,70].

It is also important to keep in mind that because of the wide range of behavior patterns in the world of ASD, it is often difficult for parents to set parameters of discipline. They're never sure if the child is acting out like any other child would in a given situation or whether the reaction is a manifestation of the autism.

The tension among mother and father, the autistic child, and the normal siblings creates an unsustainable situation. The relationships between husbands and wives become so strained that divorce sometimes is the only viable option. The divorce rate for first marriages in the United States is estimated to be 50%, whereas the divorce rate

for families with autistic children is said to approach upwards of 80% (wide range of unverifiable statistics).

## A Dental Clinician's Perspective

After working for many years with special needs children and their parents, this author says without equivocation that the central issue for almost all parents is for their children to be "normal." It means a lot to them if a child is mainstreamed in certain classes in school, can play with schoolmates, participate in sports, sing a song and yes, even sit in the dental chair and be treated like a normal child. It should be in this context that dental professionals undertake the dental treatment of these special children. It is not just about the teeth, it is about the children, the parents, and the dental professional…separately and together.

## Summary

Those who actively work with children are, with increasing frequency, encountering patients who have been diagnosed with autistic disorders. Often, dentists may be the first healthcare providers to recognize that a 1or 2-year-old child has some type of extraordinary pervasive behavioral disorder that a parent, fearing the worst, may have suspected instinctively and emotionally but never faced objectively.

Currently, there are no empirical biological tests (e.g., blood tests or brain scans) for ASD that are reliable. The definitive diagnosis of ASD is usually made by pediatricians, psychologists, or psychiatrists who institute a process of analysis which involves a developmental and clinical history, tests for cognitive function, and assessment of receptive and expressive language skills.

The etiology of ASD is an enigma. Highly regarded researchers are of the opinion that there is probably more than one cause since the disorder can have such disparate manifestations. Genetics, environmental poisons, neurologic psychopathy, dietary deficiencies, and allergies have all been implicated. Pervasive developmental disorders, Asperger's syndrome, Rett syndrome, and childhood degenerative disorders are all considered a part of the ASD group, but the distinction between the various entities is not always clear.

Given the fact that the etiology and the increased incidence of the various ASDs are scientifically puzzling, treatment modalities tend to be wide ranging and very much trial and error, especially since there is no cure. Dental professionals who treat patients with ASDs should be knowledgeable about the special needs of not only these patients, but also of their parents.

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