

Case Study

## Diagnosis and Differential Diagnosis of Autism Spectrum Disorder: A Case Study

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### ABSTRACT

Autism spectrum disorders (ASD) are a group of developmental disorders that can cause significant social, communication and behavioural challenges. ASD are spectrum disorders and they affect each person in different ways, and can range from very mild to severe. People with ASDs share some similar symptoms, such as problems with social interaction but, there are differences in when the symptoms start, how severe they are, and the exact nature of the symptoms. Differential diagnosis is the process of distinguishing disorders from other similar disorders; the diagnosis is often established by ruling out other disorders. Some ASD symptoms can overlap with symptoms of other mental health disorders, such as schizophrenia or attention deficit hyperactivity disorder (ADHD). This study focuses on differentially diagnosing Autism spectrum disorder from other similar disorders and other mental health disorders by analyzing one case in detail.

**Keywords:** *Autism Spectrum Disorder, Differential Diagnosis, Attention Deficit Hyperactivity Disorder*

The preferred current term for describing children and youth with autism-related disorders is autism spectrum disorders (ASD). This term is used to refer to the broad range of subtypes and levels of severity that fall on the spectrum of autism and pervasive developmental disorders. These are a group of related brain-based disorders that affect a child's behavior, social and communication skills. The term "spectrum" refers to the wide range of symptoms, strengths and levels of impairment that people with ASD can have. The ASDs include three of five disorders known as Pervasive Developmental Disorders (PDDs). These disorders include autistic disorder, Asperger syndrome, and PDD-not otherwise specified (PDD-NOS) as per earlier version of DSM. There are other forms of autism and pervasive developmental disorders on the spectrum, but the aforementioned are the primary subgroups of autism.

Emerging in childhood, it affects about 1 or 2 people in every 1000; and is 4 times more common in boys than girls (Gillberg, 1990). It is estimated by the Center for Disease Control that Autism Spectrum Disorder will impact boys at a higher rate than girls on an average of 4 to 5 times. Autism Spectrum Disorder occurs in all racial, ethnic, and socioeconomic groups.

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## **Diagnosis and Differential Diagnosis of Autism Spectrum Disorder: A Case Study**

In the last two decades the number of diagnoses of ASD has been increased drastically throughout the world. Autism spectrum disorders affect an estimated 4 to 6 out of every 1,000 children. The reported number of children with ASD has increased since the early 1990s. The reason for the increase is unclear. It may be because of many factors, including an increased awareness of what ASDs are, more screening tools and services, and/or changes in how ASD has been defined and diagnosed. In the past, only children with the most severe autism were diagnosed (the tip of the iceberg). Now children with milder symptoms are being identified and referred to intervention and educational programs.

Because most children with ASD will usually master the early motor skills such as sitting, crawling, and walking on time, delays in social and communication skills may not be as distinctively obvious to parents. Many parents of children with ASD can think of specific examples that suggest something was different, but nothing can be said so specifically that would be indicating a serious problem. Autism spectrum disorders are lifelong conditions with no known cure. However, children with ASD can progress developmentally and learn new skills. Some children may improve so much within short span of time that they no longer meet the criteria for ASD, although milder symptoms may often persist. They have a wide range of abilities, ranging from near- or above-average intellectual and communication abilities to severe mental retardation and an absence of spoken language (Myles & Simpson, 2003)

The signs and symptoms of ASD may include behaviors like;

- Repeating certain behaviors or have unusual behaviors
- Have overly focused interests, such as with moving objects or parts of objects
- Have a lasting, intense interest in certain topics, such as numbers, details, or facts
- Be upset by a slight change in a routine or being placed in a new or overstimulating setting
- Make little or inconsistent eye contact
- Tend to look and listen less to people in their environment
- Rarely seek to share their enjoyment of objects or activities by pointing or showing things to others.
- Respond unusually when others show anger, distress, or affection
- Fall or be show to respond to their name or other verbal attempts to gain their attention
- Have difficulties with the back and forth of conversations
- Use words that seem odd, out of place, or have a special meaning known only to those familiar with that person's way of communicating
- Have facial expressions, movements, and gestures that do not match what they are saying
- Have an unusual tone of voice that may sound sing-song or flat and robot-like
- Have trouble understanding another person's point of view, leaving
- him or her unable to predict or understand other people's actions
- People with ASD may have other difficulties, such as sensory sensitivity, sleep problems, digestion problems, and irritability.

### **DSM V and Autism Spectrum Disorder:**

The diagnosis of Autistic Disorder in DSM-V has 2 key criteria:

- Impairments in social communication and social interaction
- A restricted, repetitive range of interests, behaviors, and activities

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The DSM-5 describes three “severity levels” to be associated with a diagnosis of Autism Spectrum Disorder. The levels are described as:

- Level 3 “Requiring very substantial support”
- Level 2 “Requiring substantial support”
- Level 1 “Requiring support”

Individuals who have marked deficits in social communication, but whose symptoms do not otherwise meet criteria for autism spectrum disorder, should be evaluated for social (pragmatic) communication disorder as per DSM V.

### **Causes of Autism Spectrum Disorder:**

No one knows exactly what causes ASD. Scientists know from twin and other family studies that genetics play a major role but many chromosomal and gene abnormalities have been identified, none of these are present in all children with ASD. The changes that siblings might also have some form of ASD are 10 times higher than in the general population. Environmental factors may also play a secondary role, but this has not yet been proven.

There are no specific lab tests for ASD, so pediatricians must rely on information from parents and through observation. The condition is complex and symptoms are different for each child. When ASD is suspected as a cause of language and social delay, a full evaluation will be made. This will be done by doctor or psychologist who has expertise in the diagnosis of ASD or, preferably, by a team of specialists that may include developmental pediatricians, child neurologists, child psychiatrists, psychologists, speech and language pathologists, occupational or physical therapists, educators and social workers.

Children with ASD are affected by many factors that will shape their future. The sooner ASD is identified, the sooner appropriate intervention programs can begin, and the better the outcomes will be. However, the outcome will be limited depending on their intelligence, the severity of autistic symptoms, and associated medical problems such as seizures.

### **Assessment of Autism spectrum Disorders:**

- 1) Diagnosing an autism spectrum disorder is not a brief process. There is no single medical test that can diagnose it definitively; instead, in order to accurately pinpoint child’s problem, multiple evaluation and tests are necessary by multiple specialists. The team of specialists includes child psychologist, child psychiatrist, speech pathologist, developmental pediatricians, pediatric neurologist, audiologist, physical therapist and special education teacher.
- 2) Parent interview: In the first phase of the diagnosis evaluation, parent will give background information about child’s medical, developmental, and behavioral history.
- 3) Medical examination: the medical evaluation includes a general physical, a neurological exam, lab tests, and genetic testing. Child will undergo this full screening to determine the cause of his or her developmental problems and to identify any coexisting conditions.
- 4) Hearing test: Since hearing problem can result in social and language delays, they need to be excluded before an autism spectrum disorder can be diagnosed. Child will undergo a formal audiological assessment where he or she is tested for any hearing impairments, as well as any other hearing issues or sound sensitivities that sometimes co-occur with autism.

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- 5) Observation: Developmental specialists will observe child in a variety of settings to look for unusual behavior associated with the autism spectrum disorders. They may watch child playing or interacting with other people.
- 6) Lead screening: because lead poisoning can cause autistic like symptoms, the National Centre for Environmental Health recommends that all children with developmental delays be screened for lead poisoning.
- 7) Speech and Language evaluation: a speech pathologist will evaluate child's speech and communication abilities for signs of autism, as well as looking for any indicators of specific language impairments or disorders.
- 8) Cognitive testing: child may be giving a standardized intelligence test or an informal cognitive assessment.
- 9) Adaptive functioning assessment: Child may be evaluated for their ability to function, problem-solve, and adapt in real life situations. This may include testing social, nonverbal, and verbal skills, as well as the ability to perform daily tasks such as dressing and feeding him or herself.
- 10) Sensory motor evaluation: Since sensory integration dysfunction often co-occurs with autism, and can even be confused with it, a physical therapist or occupational therapist may assess child's fine motor, gross motor, and sensory processing skills.

### *Differential Diagnosis*

Differential diagnosis is the process of distinguishing i.e. disorder from other similar disorders. Because there are currently no medical tests to detect autism, the diagnosis is often established by ruling out other disorders.

### *DSM IV TR Category: PDDs*

<b>Autistic Disorder</b>	Severely disordered verbal <i>and</i> non-verbal language; unusual behaviors.
<b>Asperger's Syndrome</b>	Relatively good verbal language, with "milder" non-verbal language problems; restricted range of interests and relatedness.
<b>PDD-NOS</b>	Non-verbal language problems not meeting strict criteria for other PDD disorders.
<b>Rett's Disorder</b>	Rare neurodegenerative disorder of girls.
<b>Childhood Disintegrative Disorder</b>	Psychiatrists assume it as neurodegenerative disorders.

### *DSM (Diagnostic and Statistical Manual of Mental Disorders) V Category: ASD*

<b>Autism Spectrum Disorder</b>	Persistent deficits in social communication and social interaction across multiple contexts along with Restricted, repetitive patterns of behavior, interests, or activities occurring in early developmental period causing clinically significant impairment in social, occupational, or other important areas of current functioning.
<b>Rett's Disorder</b>	Rare neurodegenerative disorder of girls.

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### *ICD (International Classification of Diseases) 10 Category: PDDs*

<b>Childhood Autism</b>	Severely disordered verbal <i>and</i> non-verbal language; unusual behaviors.
<b>Atypical Autism</b>	Arises most often in profoundly retarded individuals and with severe specific developmental disorder of receptive language.
<b>Rett's Syndrome</b>	Rare neurodegenerative disorder of girls.
<b>Other Childhood Disintegrative Disorder</b>	Psychiatrists assume it as neurodegenerative disorders.
<b>Overactive disorder associated with mental retardation and stereotyped movements</b>	
<b>Asperger's Syndrome</b>	Relatively good verbal language, with "milder" non-verbal language problems; restricted range of interests and relatedness.
<b>PDD, unspecified</b>	Non-verbal language problems not meeting strict criteria for other PDD disorders.
<b>Other Pervasive Developmental disorders</b>	

### *Differential diagnosis of autism and other disorders*

#### **Autism and Mental Retardation**

<b>Factors</b>	<b>MR (Mental Retardation)</b>	<b>Autism</b>
<b>History</b>	Developmental delays in areas other than language are often noted	Usually, they show language deviancy.
<b>Communication</b>	Even though retarded children may not understand, he can gesture or mimic something and he appears to want to communicate.	Usually doesn't want to communicate with others in their environment.
<b>Inter-personal relations</b>	Overall, not grossly impaired. Social smile present and maintains eye contact.	Most of these behaviors are absent.
<b>Motor development</b>	Show some of the same unusual motor movements seen in autistic children.	Unusual motor movements like stereotyped movements of rocking, flapping his hand etc., tantrums and responsiveness to pain or self-stimulation.

#### *Autism and Attention Deficit Hyperactive Disorder*

<b>ADHD</b>	<b>Autism</b>
Onset in childhood before 7 years of age	Onset in infancy or early development but always before 3 yrs. of age
Behaviorally seem to be overactive.	Not overactive
Have difficulty monitoring attention	Have deficits in shifting attention
Prevalence- 2-3 % in children 3-5 % in school aged	Prevalence- 2-5 in 10,000 children.

***Differential Diagnosis of Autism and Schizophrenia***

<b>Autism</b>	<b>Schizophrenia</b>
Has failure or delay in the development.	Has loss of reality after development is better established.
Have deficiency in fantasy	Retreats from reality into fantasy
Symptoms like poor eye movements etc. are present	Symptoms like hallucinations and delusions frequently occur.
Ratio- 3:1	Ratio- 1:1
Show relatively steady course	Marked by remissions and relapses
MR is more common	Not common
Have high frequency of evidence of cerebral dysfunction and occurrence of epilepsy	Not of high frequency

Because OCD (Obsessive-compulsive disorder) also involves unusually repetitive behaviour it is important to highlight some key differences between OCD and people on the autism spectrum:

- Social development is not necessarily atypical in childhood in people with OCD
- Repetitive behaviours result in anxiety in people with OCD, so the absence of an anxiety response precludes OCD (but the presence of anxiety does not necessarily mean that someone must have OCD and not autism).

It is important to highlight the key difference between people with autism and those with personality disorders (and so help avoid misdiagnosis). Personality disorders do not typically involve the ‘obsessive’ narrow interests or resistance to change. In addition, although empathy deficits are present in both autism and psychopathy (or antisocial personality disorder), in people with autism it is the cognitive component of empathy that is impaired (‘theory of mind’ or recognising what others may be thinking or feeling) while affective empathy (having an appropriate emotional reaction to/caring about other’s feelings) may be intact. In contrast, whereas in psychopathy the cognitive component of empathy is intact (enabling them to deceive and manipulate others) affective empathy is impaired (they do not care about others’ suffering, for example). Autism can coexist with other conditions involving ‘rigid’ behaviour and cognition such as eating disorders or gender identity disorder, and a dual diagnosis might be appropriate if the difficulties related to autism predate the second diagnosis. Emotional difficulties such as social anxiety disorder or depression are also common in people with autism and are usually seen as secondary to the autism. This is because autism often develops first and can cause social difficulties including social isolation, which can then give rise to anxiety and depression. Individuals can also be diagnosed in childhood as having a language disorder, only later in life receiving a diagnosis of autism. (Bishop et al., 2008)

**REVIEW OF LITERATURE**

Huerta M., Lord C (2012) had done an investigation on the identification and evaluation of autism spectrum disorders, developmental factors in diagnostic process, latest research on diagnostic tools and their recommended use and best practices for clinical work.

Wong, V., Yu, Y., Keyes, M.L., McGrew, J.H (2017) had examined the experiences of parents receiving an autism spectrum disorder (ASD) diagnosis for their child. Mixed methods were used to give a detailed account of the sequences of events, parent’s experiences and activities associated with the ASD diagnosis. Parents waited nearly two and a half years

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and results showed that parents with lower general and autism-specific social support, poor physical health functioning and children with more severe communication problems reported longer wait times. Surprisingly, parents reported more positive than negative experiences from receiving the diagnosis. From this study we can say that differential diagnosis at early stage will make a huge difference in the child.

Todorov, S., & Arnaoudova, M. (2012) investigated on the differential diagnosis of Autistic disorder (AD) and Asperger's syndrome (AS) as it is quite difficult since most of the symptoms are clinically undistinguished. The crucial distinctive points between AD and AS appear to be the level of cognitive function and the time of onset. In autism onset is before 3 years and in AS it is mostly diagnosed between age four and eleven without significant delay in language or cognitive development whereas, in AD there will be cognitive impairment. In relevance to the need for our study, the author also quoted that correct diagnosis is of great importance for the choice of adequate therapeutic approach.

Matson L.J (2005) had done a study on the current status of differential diagnosis for children with autism spectrum disorders. Early intervention for autism spectrum disorder (ASD) has proven to be a successful strategy for remediating many difficulties experienced by ASD children. As a result, accurate diagnoses of children with this range of disorders are more critical.

Duijkers, J.C.L.M., Vissers, C.T.W.M., Verbeek, W., Arntz, A., Egger, J.I.M (2014) had conducted a study on the average intelligent patients with ASD and personality disorders who are expected to show different problems in social cognition. The results indicated that the ASD patients estimated themselves as more impaired on the ability to read emotions, but better on intrapersonal functioning than the PD (Personality Disorder) patients. Both patient groups showed more social cognitive impairment as compared to age and sex matched non-patient data. Measuring social cognition had contributed to a better understanding on the differentiation of ASD and PD.

### **METHODOLOGY**

#### ***Statement of the problem***

The problem of the study is to find out the needs and ways of differential diagnosis of autism spectrum disorders in terms of DSM IV-TR, DSM V and ICD 10.

#### ***Objectives of the study***

The main objective of the study is to elaborate on the need, importance and ways of differential diagnosis of autism spectrum disorders within other similar disorders and with other disorders. A case study is also done to reflect of the need for a multiple approach in assessment and the importance of multiple specialists in order to make a much relatable provisional diagnosis.

#### ***The present study***

The present study is aimed at differentially diagnosing the autism spectrum disorders within similar disorders and other disorders. By the use of a case study done in detail, the diagnostic process is further explained and differential diagnosis is also made. This study highlights on the various classifications of the Autism Spectrum disorders, differential diagnosis of ASD with other disorders, and the need for the multiple specialists and multiple tests in making an appropriate diagnosis.

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### *Need for the study*

A major public health problem in the care of children with autism spectrum disorder and other pervasive developmental disorders is the inconsistent diagnosis. The present study is done to emphasize on the need for differential diagnosis of the Autism Spectrum Disorders as it has always been confusing. Though DSM 5 and ICD 10 have different umbrella terms for the autistic disorders, the disorders included inside the spectrum are mostly overlapping. So the need for differential diagnosis is higher. It is also very much important to eliminate misdiagnosis of ASD with any other similar disorders. Delayed diagnosis of ASD is a serious problem, because early initiation of treatment increases the likelihood of a favorable outcome. Early detection leads to early intervention, and for youth with ASD, participation in specialized intervention programs can optimize long-term outcomes (Volkmar et al., 2005). Some young children with ASD who receive early intervention have shown more significant improvements in cognitive, social, and language functioning than older children who undergo the same interventions (Rogers, 1998). The case study is done mostly to highlight on the various areas that need to be assessed and analyzed, before a provisional diagnosis is made. The analysis was done with the help of multiple specialists and all their data's are combined, analyzed and based on the overall output the provisional diagnosis is made.

### **CASE STUDY**

*Case Name: XYZ*

*Age/sex: 12 years / male*

#### **Brief Case History:**

- complaint of language problems like, stereotypic expression, excessive echolalia
- Complaint of behavior problems like, sudden burst of laugh, stereotypic movements
- Prenatal & perinatal history reveals no abnormality.

#### **Medical History:**

- At the age of 8 years testing was done in Centre for DNA Finger Printing and Diagnostics and no chromosomal abnormality was observed in karyotypic 46xy
- No metabolic disorder has been noted at the age of 8 yrs.
- Fragile-x syndrome is also not present as per the reports reveals.
- At the age of 7 years the Electro Encephalogram (EEG) reveals potentially epileptic form of activity. After 7 years it was frequently noted. And the occurrence is for every one year. Lastly it occurred in January.
- The child is under medication for epilepsy.

#### **Cognitive Testing:**

- As per the IQ testing done at 6 years in NIMH, the reports reveal mental age of 5 years 7 months and Borderline Developmental Delay with Autistic Features with an IQ of 74.
- Again, the IQ testing was done 2 years later and the reports reveal IQ of 58.
- As per the testing done 2 years later in NIMH, the reports reveals a mental age of 6 years 2 months and the IQ is 51
- The scales that have been used to administer these tests are Developmental Screening Test (DST), Vineland Social Maturity Scale (VSMS), Binet-Kamat Test of Intelligence and Childhood Autism Rating Scale (CARS)

#### **Pre-requisites evaluation:**

- Biological
  - OPM examination: Normal both structurally and functionally.



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- Sensory status: Tactile, Hearing, Kinesthetic and Visual are good
- Vegetative skills such as chewing, blowing, biting, sucking, swallowing are present.
- Cognitive pre-requisites:
  - Means end relation: present, Use of object: present and Object permanence: present
- Social pre-requisites:
  - Language exposed at home and by the child is Telugu
  - Language stimulation given to the child is adequate
  - Parent child interaction is good
- Pre-linguistic skills:
  - Attention span, eye contact and smiling are fair.
- Imitation skills:
  - Gross body and speech is good.
  - Play Behavior is fair.

### ***Language Assessment:***

- Semantic Intentions, semantic relations and communicative functions are present.
- In supra segmental aspects stress is inappropriate.
- Concepts like money, time, shape, color and preposition are present in both comprehension and expression. Spaces are inconsistent in expression.
- Vocabulary: Nominal, action words, pronouns, noun modifiers and verb modifiers are present. Morphology and tense markers are poor. Phonological awareness is fair. Pragmatics skills like turn taking, topic maintenance and topic imitation are fair.

### ***Speech Assessment:***

- Articulation: At phonological level – uses all consonants and vowels. Phonemes are used consistently.
- Intelligibility: Rated to be 1 – everyone can understand the speech.
- Voice: pitch is appropriate; Intensity is normal; Quality and Breath control is normal.
- Fluency of the child is fluent.
- CARS rating shows a score of 36 (moderate autism)
- Attention level is at Level -3 (2 – 3 years)

**Provisional Diagnosis:** Delayed speech and language developmental delay with moderate autism

**Associated problem:** Mental Retardation

### ***Recommendations***

- psychological evaluation at regular intervals based on the condition of the child.
- Use of the differential diagnosis approach in diagnosing the autism spectrum disorders in regular clinical and hospital setup.
- Speech and language and psychological therapy.
- Use of multiple specialists and multiple tests in order to form a proper and criteria related provisional diagnosis.

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- Follow-up in regular intervals.

### RESULTS AND DISCUSSION

Overall, the study highlights on the need for differential diagnosis of Autism Spectrum Disorders in order to avoid misdiagnosis. This is done by using DSM IV-TR, DSM V and ICD 10 criteria. The case study helps us to have a clear understanding on the areas that need to be assessed and the need for multiple specialists in order to frame an appropriate provisional diagnosis. The case study shows that the tested child has moderate autism with delay in development of speech and language. Since, the level of IQ is fluctuating and not above average, the associative problem is said to be mental retardation. The more the specialists come together, the more the accuracy rate of the diagnosis will be. The importance and ways of differentiating Autism Spectrum Disorders with other similar disorders is also discussed in detail. The differential diagnosis of the ASD in an early age will be beneficial in framing a much suitable intervention strategy for the child. Though there are many similar disorders inside the broad term of ASD with similar symptoms, the need for proper diagnosis is also high.

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### ***Conflict of Interest***

The author(s) declared no conflict of interest.

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