

Autism Spectrum Disorder: A Case Study

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ABSTRACT

Autism spectrum disorders (ASD) comprise a group of neurodevelopmental abnormalities that begin in early childhood and are characterized by impairment of social communication and behavioral problems including restricted interests and repetitive behaviors. Several genes have been implicated in the pathogenesis of ASD, most of them are involved in neuronal synaptogenesis. Autism is a very rare diagnosis, hence an underdiagnosed condition in clinical practice. It is a condition in which patients, severe speech and language impairment. These patients, on clinical testing, are found to have normal IQ. Unlike space occupying lesions, radiologic evaluation, hearing evaluation reveals normal results. Patients with autism require a different management approach to their speech-language and cognition problems. The management approach included the following; speech-therapy, occupational therapy & psychological counselling etc.

Keywords: Intelligent quotient (IQ), speech-therapy, pathogenesis, radiological, occupational therapy

INTRODUCTION

The term “autism” was first used by a Swiss psychiatrist Eugen Bleuler around 1911 .It is a developmental disorder of variable severity that is characterised by difficulty in social interaction and communication. It is also characterised by restricted or repetitive patterns of thought and behaviour. Autism spectrum disorders (ASD) comprise a group of neurodevelopmental

abnormalities that begin in early childhood although the first diagnosis may sometimes occur later in life and are characterized by problems in communication and social behavior. According to the 5th edition of the diagnostic and statistical manual of mental disorders (DSM-5) diagnostic criteria that has recently been released, major ASD manifestations include impairment in social communication and behavioral problems such as fixated (restricted) interests and repetitive behaviors; delay in language and age of onset are not emphasized in DSM-5 diagnostic criteria. According to DSM(V), Autism Spectrum Disorder is defined as Persistent deficits in social communication and social interaction across multiple contexts, as manifested by the following:

- Deficits in social-emotional reciprocity, ranging, for example, from abnormal social approach and failure of normal back-and-forth conversation; to reduced sharing of interests, emotions or affect; to failure to initiate or respond to social interactions.
- Deficits in nonverbal communicative behaviors used for social interaction, ranging, for example, from poorly integrated verbal and nonverbal communication; to abnormalities in eye contact and body language or deficits in understanding and use of gestures; to a total lack of facial expressions and nonverbal communication.
- Deficits in developing, maintaining, and understand relationships, ranging, for example, from difficulties adjusting behavior to suit various social contexts; to difficulties in sharing imaginative play or in making friends; to absence of interest in peers. Autism spectrum disorder (ASD) is a developmental disorder that affects communication and behavior. Although autism can be diagnosed at any age, it is described as a “developmental disorder” because symptoms generally appear in the first two years of life.

PATHOPHYSIOLOGY OF AUTISM

The exact cause of autism is not clear. It could stem from problems in parts of the brain that interpret sensory input and process language. Autism runs in families, so certain combinations of genes may increase risk of child. A child with an older parent has a higher risk of autism. Other risk factors include drugs or chemicals, anti-seizure medications, maternal metabolic conditions such as diabetes and obesity, genetic causes. The exact pathomechanism of ASD is not known so far, while several factors have been implicated in its pathogenesis of autistic disorders. Among them, the genetic cause has long been implicated to be a strong evidence based on etiology. In cases of some co-occurring or associated conditions with ASD such as tuberous sclerosis, fragile X syndrome, Rett syndrome and some other. Siblings of autistic offspring have a higher incidence of autism than general population and twin studies have also indicated strong role for inheritance. There is wide range of phenotype but more genetically homogeneous. ASD patients present with less phenotypic heterogeneity. Nevertheless, with the discovery of several genes as well as interactions of multiple genes in one individual, epigenetic factors, and effects of environmental modifiers on these genes in ASD, genetic causes including the diagnosable medical conditions, single-gene defect, and cytogenetic problems comprise 25% of the ASD patients so far. Therefore, a number of clinical phenotypes and associated co-morbidities have become the characteristic features of ASD. Although some studies indicated a role for mitochondrial DNA mutation in ASD that may possibly lead to impairment of mitochondrial energy metabolism, more research is needed for definitive answers. Mitochondrial dysfunction has been implicated in several neurological disorders and it may have a role in ASD. Mitochondria has antibacterial immunity and would be important in case of infections especially that of the GI tract in ASD children. Male to female ratio in ASD: The reason for 4/1 male to female ratio in ASD is not very well understood but it is very important. Recent studies implicate some epigenetic phenomena such as sex-specific effects of Y-linked genes,

balanced, as well as skewed X-inactivation, escaping X-inactivation, and parent-of-origin allelic gene among others in the etiology of ASD and heterogeneity in gene regulation at allelic level as well as total gene expression. One of the neuropathological findings in ASD includes the presence of focal cortical dysplasias due to possibly the heterochronic division of germinal cells leading to abnormal migration of daughter cells to their target regions. Abnormal neuronal migration leads to circumscribed foci of thin cortical areas in ASD human brain especially in frontal lobe containing smaller pyramidal neurons and interneurons. These pathological findings have been attributed to the sensory and motor deficits as well as the epileptic seizures seen in ASD. Autism-epilepsy phenotype has recently been shown to be associated with macrocephaly, a pathologic condition due to accelerating brain growth in early development leading to ASD. There are evidences that the overall size of the brain is increased in some cases of ASD.

SIGNS AND SYMPTOMS OF AUTISM

Little or no response to name call, very few or no words, babbling or social skills, persistent preference for solitude, delayed speech and language development, unusual and intense reactions to sounds, lights, tastes, textures or colors, repetitive behaviours like flapping, rocking, spinning, etc., resistance to minor changes in routine, avoidance of eye contact, lack of eye contact, narrow range of interest or intense interest in certain topics. Repeating words or phrases, rocking back and forth, high sensitivity to sounds, touches, that seem ordinary to other people, problems undertaking or using speech, gestures, facial expressions, or tone of voice, trouble adapting to changes in routine, not looking or listening to other people, lack of attention and concentration.

CAUSES OF AUTISM

The exact causes of ASD is unknown, but studies suggest that genes can act together with influences from the environment to

affect development in ways that lead to ASD. Although scientists are still trying to understand why some people develop ASD and others don't, some factors that increase the risk of developing ASD include: Having a sibling with ASD, older parents having certain genetic conditions (For example, people with conditions such as Down syndrome, fragile X syndrome, and Rett syndrome are more likely than others to have ASD). Being born with a very low birth weight, not all child who have these risk factors develops ASD.

DIAGNOSIS OF AUTISM

The clinicians diagnose ASD by looking at a person's behavior and development. ASD can usually be reliably diagnosed by the age of two. It is important for those with concerns to seek out an assessment as soon as possible so that a diagnosis can be made, and treatment can begin. Diagnosis in young children is often a two-stage process.

- **Stage 1:** Every child should be checked by a pediatrician or a clinical psychologist. Additional screenings might be needed if a child is at high risk for ASD or developmental problems. Children at high risk include those who have a family member with ASD, have some ASD behaviors, have older parents, have certain genetic conditions, or who were born at a very low birth weight. Parent's experiences and concerns are very important in the screening process for young children. Sometimes the clinician will ask questions to parents about their child's behaviors and combine those answers with information from ASD screening tools and with his or her observations of the child. Children who show developmental differences during this screening process will be referred for a second stage of evaluation.
- **Stage 2:** This second evaluation is with a team of doctors and other health care professionals who are experienced in diagnosing ASD. This team may include: Paediatrician,

Clinical psychologist, Child psychiatrist, & Speech-Language Pathologist.

CASE REPORT

A case, aged 6 years boy came to the department of speech language pathology (SGT Hospital, Gurugram, Haryana) with the complaint of inability to speak and communicate with the peers and family. He was born full term following a normal pregnancy and delivery. His newborn screen and neonatal hearing test were normal. His mother also had concerns that he was not speaking any words, and his doctor said, "Let's wait and see." Earlier investigation/ treatment on 03-04-2019, detailed speech and language evaluation was done and found to be delayed speech and language and poor eye contact. Speech development: delay first word: achieved at age of 2.5 years, phrases and first sentence: not achieved yet. Social and behavioural history: recognises parents: yes, socialises with peers/elders: no, Irritable: no, distractive: yes, aggressive: yes, restless: yes, refuses to go to strangers: no, makes eye contact: yes(very limited), indulges in parallel play: yes, exhibits temper tantrum: yes, span of attention and concentration: poor, hyperactive: yes.

Comprehension

The child comprehended single one step commands on repetition, comprehends family members, lexical items (plate, glass, fan) apple, categorical perception is not achieved. The child does not have comprehension of adjectives, prepositions.

Expression

The child has expressive vocabulary of 5-10 words. Predominantly uses non-verbal mode to communicate (pointing, gestures). The stimulibility in the child is fair.

Educational history

Age of admission: 2 years, still in paly group. Performance in school: Very poor.

He had been asked to leave his previous schools because of behavioral problems. His mother reported that his current preschool teacher recommended that you should consult with the pediatrician and got his opinion about his behavior. Since he began preschool, his teachers reported that he is extremely hyperactive, does not follow instructions, and largely ignores the children in the class. She reported that he had good health recently and his speech milestones were delayed. She denied any history of developmental regression and added that he had made some nice progress since he began receiving speech therapy from SGT Hospital, Gurugram, Haryana. He will now use single words like “papa” and “mama” to make requests. He was usually calm down when his father played games on the mobile. He described him as a sweet and loving boy, but he also shared that he will have prolonged tantrums when denied what he wanted something and that tantrums will often occur when they were attempting to leave their house. During these tantrums, he will frequently cry and bite his hand, which frightens him. For some time, father concerned that he did not like to play with his similar-aged cousins during family get-togethers, but he thought that this was because he was an only child and didn’t like to share. He had been eagerly awaiting his first day of preschool so that he could spend more time around other children. He mentioned that it had been hard for the family, and he found that his preschool report was to be embarrassing and discouraging. Parents were frustrated and wanted the best for him.

He will verbally request (one word) at least ten desired items or activities at home each day. He will request desired items (one word) along with exhibiting interfering behavior (crying, screaming, dropping to floor) and will demonstrate no more than three occurrences of interfering behavior. Through discussion and observation, it was determined that he would benefit the most from speech therapy to request the following: his parents said that they would like to increase the number of speech therapy session and type of interactions they have with him. The following goals were developed by the speech pathologist.

Long term goal

To make the child comprehend and express at some extent so that he can lead his normal life through vocational training and other mode of dependency.

Short term goal

- To work on speech and language throughout the session by labeling objects and actions at least five times each session for the duration of two months.
- Read bed time stories to him daily for the duration of two months.
- Play concept development games during daily activity time three times per day for the duration of two months.
- Give opportunity to him to request a desired item verbally for a minimum of five times a day for two months.
- To work on attention and concentration during the therapy session.

Progress report

- His attention span has been increased upto 7 seconds during conversation more than that during play activity during therapy session.
- Vocabulary increased from 2 words to 10 words.
- He follows the commands after 3 consecutive request.

MEDICAL HISTORY

Audiological findings: BERA test was done on 10.04.2019. Report reveals that:

The child is having bilateral hearing sensitivity within normal limits.

Radiological findings: C.T. Scan-Head: Normal study.

Pathological findings: Haematology Report;

<u>Name of test</u>	<u>Results</u>	<u>Units</u>
Haemoglobin	10.2	gm/dl
TLC/DLC	13000	/cu.mm.
Polymorphs	75	%
Lymphocytes	15	%
Monocytes	05	%
Eosinophils	04	%
Basophils	01	%
PCV	35.1	%
MCV	73.6	Fl
MCH	21.4	Pg
MCHC	29.1	gm%
Total RBC Count	4.77	millions/cu/mm
Platelet Count	2.72	lakh/cu.mm

Blood Group: A” Positive

Psychological findings: IQ assessment has been done on 08.04.2019

DST score is 37, that is, DQ of 37, Moderate intellectual disability

VSMS score is 52, that is SQ of 52, shows mild impairment

CARS - 2, CARS score is 41 shows severe symptoms of autism.

TREATMENT

A doctor may use medication to treat some symptoms that are commonly associated with ASD. The symptoms are the following: Irritability, aggression repetitive behaviour, hyperactivity, attention problems anxiety and depression. Speech therapy, occupational therapy, special educator, and clinical psychologist also play important role during intervention process.

DISCUSSION

People with ASD may be referred to clinicians who specialize in providing behavioral, psychological, educational, or skill-building interventions. These programs are typically highly structured and intensive and may involve parents, siblings, and other family members. These programs may help people with ASD: Learn life skills necessary to live independently, reduce challenging behaviours, increase or build upon strengths, learn social, communication, and language skills. There are many NGOs related social services programs and other resources that can help people with ASD. Here are some tips for finding these additional services: autism support group will be helpful for them. Sharing information and experiences can help individuals with ASD and their caregivers learn about treatment options and ASD-related programs.

CONCLUSION

Conversations and meetings with health care and other related professionals. This information helps when it's time to make decisions about which programs might best meet an individual's needs. Keep copies of doctors' reports and evaluations. This information may help an individual qualify for special programs like speech therapy, special education, psychological counselling, occupational therapy along with medical intervention by paediatrician or psychiatrist.

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