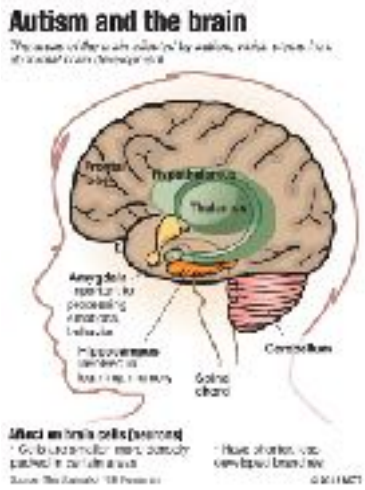
Dr Anjana Thadhani, Consultant Developmental Paediatrician,
Niramay Guidance Clinic (Chembur & Kharghar), KEM Hospital, B J Wadia Hospital

Aman, a 4 yr old male child usually spins around the furniture and difficult to control. When on bed he rocks himself to sleep. His interaction with his parents is less for his age. He is unable to follow simple commands at home and at play school. He just repeats words and uses jargon which his mother cannot understand. He occasionally gets aggressive with peers and screams without any clear reason. To an outsider, Aman looks like a typical overactive child full of energy.

Simran is 6 years old and is a very quiet child. Her mother says that she is a very shy child and still hides behind her mother if an outsider comes to her house. At school she follows instructions and is doing well. She does not participate in group activity at school and has no friends. During her visit to the doctor she had poor eye contact and demonstrated minimal interaction.



These children belong to the Autism Spectrum Disorder which is not so uncommon anymore. Most of these children have behaviour which is extreme and affects their learning and play. Doctors, school teachers and parents have become increasing aware of this condition in the last decade.

AUTISM SPECTRUM DISORDER is a neurodevelopmental condition which mainly affects areas of social interaction and communication. These children also may have some motor movements like hand flapping, rocking which are repeated over and over again. The manner in these children behave is quite different and represent a spectrum of symptoms from mild ones which are considered as shy to the real aggressive ones.

PATHOPHYSIOLOGY: Recent findings suggest the root of autism lies in an overabundance of brain connections, known as synapses. Normal functioning brains disconnect unnecessary synapses after infancy while autistic brains leave a large percent of synapses intact.

It has also been hypothesized that excessive protein synthesis is one core pathophysiological mechanism of intellectual disability and autism, caused by mutations in genes that regulate protein synthesis in neurons.

PREVALENCE : The Prevalence in US has been on rise with earlier statistics being 2-6 per 1000 population to the recent of 1 in 50 (NIMH). More boys than girls are consistently found to be affected with ASDs, with male-to-female ratios ranging from 2:1 to 6.5:1. It is difficult to estimate the numbers in our country as there is no mandatory reporting of Autism in India as for diseases like Malaria or Tuberculosis. However there is an alarming increase in the number of cases of autism in the past few years.

CLINICAL PRESENTATION: It is a Spectrum disorder with variable presentation. Age of presentation is before 36 months however most of the children present commonly between 1 ½ to 3 years. Most common initial presentation is usually with Speech & language delays with or without Behavior problems. Older children may have Academic underachievement with Unusual behaviours. Most of the parents report that these children were "Different" from birth. It is important to address the parental concerns.

Impairments

- **Language and communication** – impaired and deviant language and communication – verbal and non-verbal & **Social interaction** – impaired, deviant and extremely delayed social development
- **Restricted, repetitive, and stereotyped patterns of behavior, interests, and activities.**

These impairments would present as :

1. Difficulties in Language and speech

- Language skills are slow to develop or speech is delayed.
- Loss of previously acquired words or phrases, used to say few words or babble, but now doesn't.
- Echoes or repeats words and phrases without any meaning.
- Those with speech, fails to initiate or sustain conversation
- Pronoun reversal

2. Communication difficulties

- Is not interested in other children of own age group & Seems to prefer to play alone, solitary play.
- Has poor eye contact and seems to be in his/her "own world."
- Does not respond to his/her name and may seem to be deaf (despite normal hearing).
- Inappropriate laughter
- Inappropriate Non verbal communication, No Pointing rather takes mother's hand to indicate needs.
- Doesn't follow directions and usually cannot explain what he/she wants.
- Sudden withdrawn, self-abusive, or indifference to social overtures

3. Atypical Behavior

- Odd stereotypic & repetitive movement patterns, hand flapping, spinning, rocking.
- Casting, throwing, banging toys seen in older children

- Inappropriate play, lack of pretend play, lack of sharing, turn taking etc.
- Spends a lot of time stacking objects, lining things up, or putting things in a certain order.
- Shows unusual attachments to objects or schedules
- Hyperactivity, running around all the time.
- Severe Aggression, bitting, hitting & self mutilating behavior in extreme cases.

SCREENING & DIAGNOSIS: Children with autism are diagnosed based on standardized criteria, it is however a clinical diagnosis. The children have to meet these criterion, based on developmental and behavioral characteristics, for the diagnosis of ASD.

- M-CHAT- R can be used for screening purposes on children in the age group of 16 to 30 months.
- DSM – V (Diagnostic Statistical Manual) ASD –Diagnostic criteria
- CARS – (Childhood Autism Rating Scale) quantitative tool, gives the severity of autism, hence good for predicting prognosis.

INVESTIGATIONS

- **There are n**o specific Blood or Radiological investigations for autism. Investigations for associated medical disorder, are done based on clinical suspicion only.
- 5-10% cases of autism have an identifiable medical disorder. The remaining 90% are idiopathic.
- Inheritance is likely multifactorial. Current research evidence available gives families empirical recurrence risks only.
- There are no prenatal or carrier state diagnostic tests.

MANAGEMENT - The primary goals of treatment are to maximize the child's functional independence and improve the quality of life. These therapies aims at facilitating development and learning. It is important to promote speech and socialization and reducing maladaptive behaviors is dealt with first.

A: **MEDICAL MANAGEMENT** includes routine preventive care, including immunisations and treatment of acute illnesses. As ASD could be coexist with medical conditions like Epilepsy, Tuberous Sclerosis, Cerebral palsy, Fragile X, Down syndrome, Neurofibromatosis, the management essentially includes management of these conditions. Behavior problems in autism can be managed well with medications like Risperidone that help in managing the aggressive behaviours. Medications can be started early with gradual titration of dosage. Methylphenidate and Atomoxetine are commonly used for hyperactivity with significant improvement.

B: CONVENTIONAL THERAPIES include

- Adaptive Behavioral Therapy
- Occupational therapy mainly Sensory Integration
- Speech therapy
- Special education

Parent training in behavior management is essential and an important part of management. It is also imperative to sensitize teachers and and other care givers. Long term support to siblings and extended family with constant counseling helps to develop better coping skills for the entire family. Parents and home based program are the backbone of effective treatment plan for Autism. During the long intervention period the parents are involved in the therapy.

Academic support is needed as these children as they have problems with the receptive language and following instructions. Most show learning difficulties in languages or could be associated with ADHD and Learning Disability. Special educator helps these children learn new concepts.

Autism is a lifelong condition and hence parents tend to try newer modalities like stem cell therapy, Hyperbaric Oxygen etc for quick results or out of frustration in case of severe cases. These are experimental and their use is not supported by appropriate and convincing medical evidence or research and hence we as clinicians should discourage parents when consulted regarding the efficacy of such treatments.