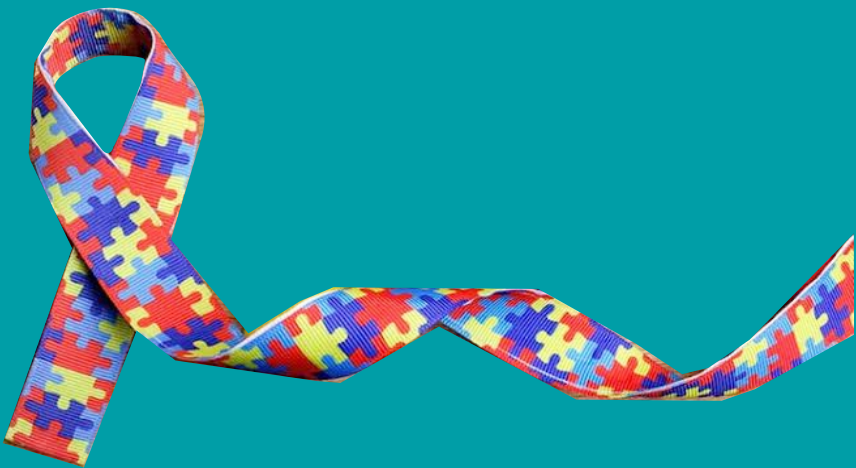




# Guidelines Autism Spectrum Disorder Screening, Diagnosis and Management



Developmental-Behavioral Pediatrics Group  
Pakistan Pediatric Association (DBP-PPA)

## Objective

The aim of these guidelines is to provide evidence based recommendations for screening, diagnosis and management of children with Autism Spectrum Disorder. This will help doctors & allied health professionals for early detection and timely referral leading to early intervention for improved prognosis.

## Target Users

These guidelines are intended to guide doctors, allied health professionals and even parents involved in the care of children with ASD.

## Introduction

Autism Spectrum Disorder (ASD) is a neurodevelopmental disorder with persistent deficits in reciprocal social interaction and communication across multiple contexts with restricted, repetitive patterns of behavior, interest or activities accompanied by sensory issues.<sup>1</sup> The symptoms begin in early developmental years and limit or impair daily functioning. In South Asia, the reported prevalence of ASD ranges from 0.09% in India to 1.07% in Sri Lanka, however no local epidemiological study was found on ASD prevalence in Pakistan.<sup>2</sup> In the Department of Developmental Behavioral Pediatrics, University of Child Health Sciences, The Children's Hospital, Lahore, 1 in 29 children were diagnosed with ASD, in the year 2022. In USA Center for Disease Control and Prevention (CDC), Autism and Developmental Disabilities Monitoring (ADDM) network found that an estimated 1 in 36 children were identified with ASD in 2020.<sup>3</sup>

Clinicians now see an increasing number of children with speech delays and social communication issues who need further evaluation. Early referral and diagnosis for prompt interventions changes the way in which the brain develops. In view of the above, there is a dire need to develop Guidelines for ASD diagnosis and management for a wide range of healthcare professionals in Pakistan to optimize the care of children with ASD to reach their maximum potential.

## Risk Factors

The clear etiology of ASD is not defined. The available evidence suggests that many factors put children at greater risk for developing ASD such as:

- Increased parental age (Maternal age >40 years, Paternal age > 50 years)<sup>4,5</sup>
- Prematurity (< 33 weeks of gestation)<sup>6</sup>
- Neonatal encephalopathy<sup>6</sup>
- Family history of developmental behavioral disorders like ASD, Social Communication Disorder (SCD) and Attention Deficit Hyperactivity Disorder (ADHD) &/or other mental illnesses such as depression, schizophrenia, anxiety<sup>7</sup>

## Identification of Red Flags

Early warning signs of ASD may begin to emerge in infancy as early as 6 months of age requiring early intervention services though the diagnosis can be made later. These red flags<sup>8</sup> can alert healthcare providers about possible risk of ASD in children.

- Avoids or does not maintain eye contact at 2 months of age
- Does not respond to name by 9 months of age
- Does not display facial expressions like happy, surprised and sad by 9 months of age
- Does not play simple interactive games like peek-a-boo by 12-18 months of age
- Uses none or few gestures by 12 months of age (for example, does not wave bye-bye)
- Does not share interests with others by 15 months of age (for example, shows you anything that they like)
- Does not point to show you something interesting by 18 months of age
- Does not notice other children and join them in play by 36 months of age
- Does not pretend like a teacher or any other character/role play during play by 48 months of age

## Identification, Assessment and Diagnosis

### Screening for ASD

Screening is important for timely detection and intervention of behavioral problems in children. All children need to be screened for developmental delays using indigenous screening tools (ShaMaq).<sup>9</sup> ASD screening should be done between 16 to 30 months of age by (M-CHAT R/F)<sup>TM, 10</sup>

### At Primary and Secondary Health Care Level

Healthcare Professionals should be aware of the red flags of ASD. Once they identify any child suspected or at risk with ASD after screening they should be referred to a Developmental Behavioral Pediatrician for early diagnosis and intervention.

### At Tertiary Care Level

A comprehensive evaluation by a multidisciplinary team led by Developmental-Behavioral Pediatrician is necessary for final diagnosis. Assessments include history, physical and developmental examination.

### History

When parents with a child present with concern of either speech delay or behavior issues. Ask the following

Does your child

- Respond to his/her name
- Give you eye contact
- Speak age appropriately
- Remain in his/her own world
- Prefer to play alone
- Play skillfully with toys
- Lead you by hand to get a desired object
- Has difficulty following a simple command

- Point to an interesting object
- Show unusual attachment with innate objects  
e.g. flashlights, key chains etc
- Chew food properly
- Has difficulty in having a haircut or nail cutting
- Covers ears in response to a loud noise

Also ask for history of:

- Associated behavioral problems (temper tantrum, aggression, hyperactivity)
- Associated sleep disturbances, gastrointestinal symptoms (diarrhea, vomiting), seizures, hyperactivity, obesity, anxiety, depression
- Birth and developmental history
- Three-generation family history with pedigree, history of neurodevelopmental disorders or any mental illness
- Screen media usage including age of starting screen media use along with daily time spent on screen by the child

## Criteria

ASD Criteria DSM V TR<sup>1</sup>, to be kept in mind while doing a detailed evaluation of symptoms

- A. Persistent deficits in social communication and interaction across multiple contexts
- Deficits in social emotional reciprocity (failure of normal back and forth conversation, reduced sharing of interests and emotions, poor response to name)
  - Deficits in nonverbal communicative behaviors (poor eye contact, deficits in understanding and use of gestures)
  - Deficits in developing, maintaining and understanding relationships (difficulty in making friends, lack of interest in peers, prefer to play alone)

- B. Restricted, repetitive patterns of behaviors, interests and activities (at least 2 of following)
- Stereotypes (hand flapping, lining up of objects, echolalia)
  - Fixated interests (strong attachment with a toy/object)
  - Insistence on sameness or adherence to routine
  - Hyper or hyposensitivity to sound, smell, taste, vision or touch
- C. Symptoms must be present in the early developmental period
- D. Symptoms cause clinically significant impairment in social, occupational and other areas of functioning
- E. The symptoms are not better explained by global developmental delay or intellectual disability

## Examination

General physical examination includes

- Anthropometry (height, weight, head circumference)
- Dysmorphic features
- Neurocutaneous stigmata

## Observation

Clinical observation is a key element in diagnosis of ASD. A child with ASD should be observed for

- Poor eye contact
- Poor response to name
- Poor pointing and imitation skills
- Play with toys in an unusual way (e.g. playing with car wheels)
- Aloofness
- Gibberish language
- Stereotypes (e.g. hand flapping, body rocking)
- Sensory issues like mouthing, toe walking, etc

## Developmental and Systemic Examination

Informal developmental assessment, detailed neurological examination including soft neurological signs along with other systemic examination needs to be done.

### Assessments

Multidisciplinary team including clinical psychologist, speech & language pathologist, occupational therapist, special needs educationist led by Developmental-Behavioral Pediatrician who have sufficient expertise and training to establish the diagnosis of ASD using formal assessment tools do the following developmental/psychological/speech/occupational assessments:

- Portage Early Education Program (PEEP)
- Griffiths Mental Developmental Scale II (GMDS II)
- Schedule of Growing Skills II (SGS II)
- Intelligence Quotient: Slosson Intelligence Test or Wechsler Intelligence Scale for Children (WISC)
- Childhood Adaptive Behavior Scale (CABS)
- Vineland Adaptive Behavior Scale (VABS)
- Diagnostic evaluation tool for ASD: Childhood Autism Rating Scale II (CARS II) or Autism Diagnostic Observation Schedule-2 (ADOS-2)
- Occupational/Sensory therapy evaluation
- Speech and language assessment including Picture Exchange Communication System (PECS)
- Behavior Rating Scale (BRS)
- Functional Behavior Analysis (FBA)
- Hearing Assessment

Following investigations can be done if indicated

- Complete blood count (CBC) for anemia
- MRI/CT Brain if any of the following is suspected
  - Abnormal neurologic examination (cranial nerve abnormalities, spasticity, ataxia)

- Unexplained microcephaly
- Neurodevelopmental regression
- EEG is indicated if there is a clinical suspicion of seizures

## Diseases Associated with ASD

Following are some of the conditions associated with ASD:

**Neurofibromatosis Type 1 (NF1):** NF1 is an autosomal dominant single-gene disorder characterized by cafe au lait spots, neurofibromas (skin nodules), skin freckling in axilla and groin etc. NF 1 has association with developmental disorders like ASD and learning disorders (LD).

**Tuberous Sclerosis (TS):** Tuberous sclerosis complex (TSC) is a genetic disorder that is characterized by hamartomas, skin lesions (ash leaf spots, shagreen patch), epileptic seizures and developmental delay/behavioral problems (ASD, ADHD).

**Rett Syndrome:** Rett syndrome is a genetic neurological and developmental disorder characterized by microcephaly, seizures and hand wringing movements.

**Others:** Fragile X syndrome, Angelman syndrome, Down syndrome etc. may have ASD features.

## Differential Diagnosis

**Social Communication Disorder (SCD):** Persistent difficulties in the social use of verbal and nonverbal communication resulting in functional impairment in effective communication, social interaction and academic/occupational performance.<sup>1</sup>

**ADHD:** It is a neurodevelopmental disorder characterized by persistent patterns of inattention and/or hyperactivity-impulsivity that interfere with functioning. Symptoms of inattention or hyperactivity-impulsivity must be present in two or more settings prior to 12 years of age. Intelligence of these children is mostly not affected.<sup>1</sup>

**Hearing Impairment:** These children have normal social interaction and nonverbal communication but lack in verbal



communication. They may or may not have accompanying behavior problems.

**Global Developmental Delay/Intellectual Disability:** Global developmental delay is reserved for children under the age of 5 years when a child fails to meet the expected developmental milestones in two or more developmental domains.

Intellectual disability is a disorder that includes both intellectual and adaptive functioning deficits in conceptual, social and practical domains. This term is used for children more than 5 years of age.<sup>1</sup>

**Developmental Language Disorder (DLD):** Persistent difficulties in the acquisition and use of language across modalities (i.e. spoken, written, sign language, or others) due to deficit in comprehension or production of language in early developmental period resulting in functional limitation.<sup>1</sup>

**Sensory Processing Disorder:** It is a disorder that can affect the way the brain processes sensory information. Children may be extra sensitive to or not react to sensory input (light, sound, touch, smell, taste) depending upon their sensory impairment.

**Screen Media Overuse:** Children may present with ASD like symptoms but show marked improvement after discontinuation of screen media and starting social interaction along with referral for early intervention.

## Management

Children with ASD require individualized behavioral and developmental interventions along with medical support as needed, by a multidisciplinary team lead by a Developmental-Behavioral Pediatrician to maximize their full potential. All children with ASD should be offered an early intervention program (EIP).

The multidisciplinary team consists of

- Developmental-behavioral pediatrician
- Clinical psychologist
- Speech and language pathologist

- Occupational therapist/ Sensory therapist
- Special needs educationist
- Developmental therapist
- Child care worker
- Others

## **Behavioral Interventions**

Interventions should be evidence based, structured, and appropriate to the developmental and functional level of the child with ASD. Interventions should be hospital/center based & community based having a home plan for parents. The main evidence based interventions used for children with ASD include Applied Behavior Analysis (ABA), Picture Exchange Communication System (PECS), Treatment and Education of Autistic and Related Communication Handicapped Children (TEACCH).

Eclectic approach is evidence based therapeutic modality in managing children with ASD. It involves a combination of therapies such as early readiness skills, ABA, PECS & Speech therapy, Occupational/Sensory therapy, Social Skill training and counseling. Individualized education program (IEP) is made for each child to increase socialization and reciprocal communication, language, play, academic and daily living skills and to reduce inappropriate behavior. Parental training for home based plan is equally important for continuation of therapies and generalization in the environment.<sup>11</sup> Children with ASD should receive 1:1 intervention for 25 to 30 hours in a week with parents as partners.<sup>12</sup>

## **Medical Management of Target Symptoms**

Developmental-Behavioral Pediatrician offers pharmacotherapy to children with ASD targeting specific symptoms.

**Table 1:** Pharmacological management of target symptoms<sup>13</sup>

Target Symptoms	Medication Class	Drugs with Evidence of Efficacy in ASD	Pre-Requisites	Side Effects
Hyperactivity/ Impulsivity	Psychostimulant	Methylphenidate	BP ECG	Decreased appetite, difficulty with sleep initiation, stomach aches, headaches
	Selective nor epinephrine reuptake inhibitors	Atomoxetine	BP ECG	Fatigue, nausea
Irritability/ Aggression/ Tantrums/Self-injury	Atypical antipsychotics	Risperidone	BP ECG	Drowsiness, weight gain, tardive dyskinesia, increased blood sugar, high triglycerides, nausea
	Selective Serotonin reuptake inhibitor	Fluoxetine		Nervousness, anxiety, dry mouth, heartburn, difficulty falling asleep, diarrhea
	Anticonvulsants	Valproic acid		Dry mouth, swollen gums, weight gain, headache, liver toxicity, bone marrow suppression
Repetitive Behavior/ Behavior rigidity	Atypical antipsychotics	Risperidone		
	Anticonvulsants	Valproic acid		
Anxiety		Fluoxetine		
Sleep dysfunction	Endogenous Chrono biotic hormone with hypnotic properties	Melatonin		Headache, dizziness, nausea

## Guidelines for Screen Time in Children

Children who are exposed to excessive screen time at a younger age may develop symptoms mimicking ASD. Therefore, children younger than two years should have no screen time. Recommended guidelines by American Academy of Pediatrics Should be followed for screen time in children.<sup>14</sup>

- For children under 18 months: Avoid screen based media except video chatting
- For children 18 months to 24 months: Parents may be encouraged to choose high quality programs and watch with their children. Letting children use media by themselves should be avoided
- For children 2 to 5 years: Limit screen time to one hour per day of high-quality programs
- For children 6 years and up: Establish consistent limits on the time spent using media and the types of media

## Quality Time for Children

Parents may be encouraged to give quality time to their children of all ages. During this time, parents need to interact and give undivided attention to their children while playing or reading books etc. This not only improves the parent child relationship but also the social, emotional and behavioral development of the child.

## Myths and Facts

**Vaccination:** A systematic review of 10 studies found that there is no association between MMR immunization and ASD.<sup>15</sup>

**Diet, Complementary and Alternative Therapies (CAM):** Gluten-free, casein-free diet,<sup>16</sup> chelating agents,<sup>17</sup> vitamin B6 and Magnesium,<sup>18</sup> vitamin B12<sup>19</sup> have no proven evidence in management of ASD.

**Virtual ASD:** Virtual ASD is not a recognized medical term. Excessive screen media exposure can cause ASD like symptoms in younger children, which is better categorized as Screen Media Overuse and not Virtual ASD.

## Implementing the Guidelines

Policy makers play an important role in the implementation of services for children with ASD. Therefore, implementation of these guidelines is encouraged and fully supported by Pakistan Pediatric Association (PPA) and policy makers. Empowering legislation exists in all four provinces that make it mandatory for the government to facilitate Persons with Disabilities.

## Legislation

The Government of Sindh has passed the Empowerment of Persons with Disabilities Act in 2018, which recognized and listed different neurodevelopmental disorders including ASD, ADHD, ID and Specific Learning Disorders in a separate Schedule for the first time in Pakistan's history.

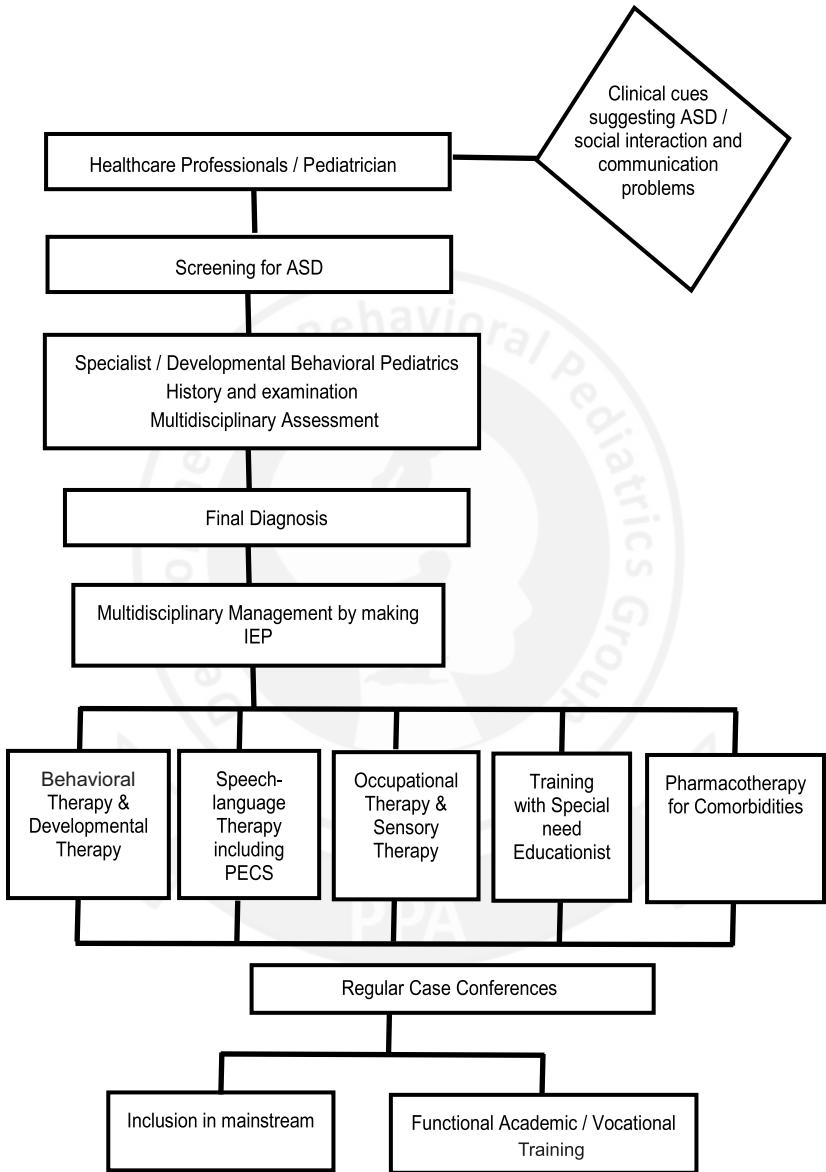
The ICT Rights of Persons with Disability Act 2020 does not define ASD and related conditions separately, but includes cover for transgender persons with disabilities.

The Punjab Government passed the Empowerment of Persons with Disabilities Act in 2022 similar to the Sindh Act, with a separate Schedule listing neurodevelopmental disorders and conditions.

KPK and Balochistan have not revised their respective Acts pertaining to Disability. However, existing Acts provide cover to all disabilities, including mental disabilities.

Therefore the Guidelines would pave the way for a greater understanding of all neurodevelopmental disorders and set benchmarks for detection, diagnosis and intervention of Autism Spectrum Disorder.

# Road Map for Health Professionals



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