Identification, Evaluation, and Management of Children With Autism Spectrum Disorder: American Academy of Pediatrics 2020 Clinical Guidelines

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The American Academy of Pediatrics recently published clinical guidelines for evaluation and management of children and adolescents with Autism Spectrum Disorder (ASD), nearly 12 years after the previous version. This article outlines salient features, highlights significant differences from the 2007 version, and discusses implications for Indian professionals dealing with affected families.

Keywords: *Dignostic tools, Investigations, Neuroimaging, Screening.*

The American Academy of Pediatrics (AAP) recently released clinical guidelines for the evaluation and management of children and adolescents with autism spectrum disorder (ASD) [1]. The previous 2007 guidelines covered both separately [2,3]. Many changes have occurred over the last 12 years: increasing prevalence; revised nomenclature and diagnostic criteria of Diagnostic and Statistical Manual of Mental Disorders, fifth edition (DSM-5) [4,5]; greater understanding of clinical profile [6], neurobiology and etiopathogenesis; advances in genetic testing [7]; evidence-based interventions; and a paradigm shift to family-centred therapy and holistic management throughout life. Understandably, there was a strong need for an update.

The increasing worldwide prevalence of ASD means primary care service providers (PCP) and pediatricians will encounter ASD routinely. Not only should we be competent enough to recognize, evaluate and establish diagnosis, we should be empowered to counsel, help families in decision making, and provide continual support. After outlining salient features of the 2020 guidelines and highlighting differences from the last one (***Tables*** **I** and **II**), implications for Indian professionals will be discussed.

Previously, increasing prevalence was attributed to growing awareness, improving surveillance and less misdiagnoses [2]. The present status (1in 59) of ASD in the US is also probably due to broadening of phenotype by DSM-5, universal surveillance and increased availability of services. Whether biological risk factors contribute to etiopathogenesis remains uncertain [1]. Earlier diagnosis is more common in higher socio-economic strata who have better access to services, while later identification is associated with milder manifestations. Clinical symptoms include core symptoms and co-existing conditions (medical, genetic, neuro-developmental, psychiatric and/or behavioral), the cumulative effect of which influence extent of social and functional impairment. The guidelines described these in-depth. They also emphasize the need for holistic evaluation and management to achieve best possible outcomes.

SCREENING AND DIAGNOSIS

The USA health system practices universal developmental surveillance with ASD-specific screening at 18 and 24/30 months. Earlier screening is indicated in high-risk individuals or when red flags for ASD are identified. Suspicion or parental concerns warrant in-depth evaluation. Establishment of diagnosis is primarily clinical, based on parental interview, personal observations and DSM-5 criteria. Though diagnostic tools are not mandatory, they help in extracting clinical information. Structured evaluation of behaviour, cognition, language, adaptive function, motor function, hearing, vision and sensory processing is recommended. Diagnoses established by the aforementioned comprehensive assessment in children under 30 months remain stable in ≥80% in adulthood.

Etiologic evaluation comprises of detailed history-taking and examination (anthropometry, dysmorphism, skin, neurologic and systemic). The indications for

Table I American Academy of Pediatrics Guidelines for Autism Spectrum Disorder (ASD)

Identification and evaluation; AAP, 2007 [2]	*Identification, evaluation and management; AAP, 2020* [1]
Clinical symptoms	
Cognitive impairment in 50%. Secondary ASD (10%) due to medical/genetic or environmental factors (more when severe delay and dysmorphism). Co-morbid conditions: seizures, gastrointestinal and sleep disorders, and challenging behaviors.	Cognitive impairment and minimally verbalin 30% each. Additional co-morbidities, other developmental/psychiatric (ADHD, motor coordination disorder, anxiety, mood disorders) and behavioral disorders (food refusal, pica, self-injury and aggression).
Screening and diagnosis	
Developmental surveillance existed, but few (8%) PCP practiced it. M-CHAT used.	Surveillance increased (75%). M-CHAT-R/FU used. Tools listed for younger ages.
Clinical diagnosis by DSM-4. Focus on category of severity, functional impairment & etiology (mainly by experts).	Clinical diagnosis by DSM5. ADI-R, ADO-S, CARS-2, SCQ and SRS may be used. Evaluation (*see text*) by PCP and experts.
Etiologic evaluation	
• High resolution karyotype; • DNA tests for FXS in all cases with GDD/ID; • *MECP2* analysis in Rettsdisorder.	• Discuss chromosomal microarray; • Discuss tests for Fragile X Syndrome; • Consider *MECP2* sequencing, if applicable • Consider WES and genetic referral.
Recurrence rate 2-8% in idiopathic ASD, higher/lower in secondary ASD.	Empirical, 4-14% if one previously affected child, 32-36% if ≥2 affected children.

Prepared from Hyman, et al. [1] and Johnson, et al. [2].ADHD Attention deficit hyperactivity disorder; ADI-R Autism diagnostic inventory-revised; ADOS-2 Autism diagnostic observation schedule, 2nd edition; CARS-2 Childhood autism rating scale, 2nd edition;, M-CHAT: Modified checklist for autism in toddlers, R/F Revised with follow-Up;MECP2: Methyl CpG-binding protein 2; SCQ: Social Communication Questionnaire; SRS: Social responsiveness scale; WES: Whole exome sequencing.

magnetic resonance imaging (MRI), electroencephalography (EEG) and metabolic testing remain individualized, with provision of more details. Genetic evaluation is recommended in all. The advantages of establishing genetic etiology include accuracy in counselling, possible specific therapy, avoiding unnecessary testing, and increased family acceptance.

Interventions

The goals remain minimizing core deficits, eliminating maladaptive behaviour, and maximizing functional independence. Intervention should be "individualized, developmentally appropriate and intensive" [1]. Periodic documentation of performance is required for monitoring response. The caveat that all interventions should be evidence-based has been added, with enumeration of characteristics of effective intervention.

Some sections *i.e.*, models of early intervention and education, psychopharmacology and complementary alternative therapy (CAM) are quite technical, since the basics were extensively explained in the previous guidelines. Hence, non-experts may not understand them unless they read the earlier version. Management of medical conditions, social skill instruction, speech and language therapy, motor therapy (including occupational therapy) and sensory therapies (the supportive evidence of which is still low) are given in greater detail.

Evaluation of maladaptive behavior and psychiatric conditions are separate and described with respect to the atypical development of ASD. The psychopharmacology section details principles of prescription and lists medications according to behavior-symptom cluster. The emerging role of psycho-pharmacogenetic testing is mentioned. According to the new guidelines, if a family opts for CAM, safety and effectiveness requires monitoring.

Working with Families

The USA'Medical home' model for primary care aims at "accessible, continuous, comprehensive, family centred, coordinated, compassionate, and culturally sensitive health care for all children and youth, including those with special needs" [8]. Though recommended for ASD since 2007, the process was not well-defined. The latest guidelines aim at better PCP and caregiver partnership, revolving around shared decision-making. Resources have been developed for pediatricians to enable them to deal with emerging issues, counsel effectively, provide parents with information and direct them towards advocacy and support groups. It is envisioned that this will result in easier handling of challenges, smoother transitions during adolescence (higher education/vocation, sexuality) and

Table II American Academy of Pediatrics Guidelines for Autism Spectrum Disorder (ASD)

Management; AAP, 2007 [3]	*Identification, evaluation and management; AAP, 2020* [1]
Interventions	
Principles, components and curricula used in early intervention are well explained.	This continues without alluding to the basics and hence may appear more technical.
Educational models have been named but not explained. Differences between programs by age (younger *vs* older) given.	More details are presented. Emphasis is on classroom models, less restrictive settings and development of social skills.
Speech and language approaches named but not explained. Brief mention of occupational and sensory therapy.	Details of language behavior and techniques included. Stress given to developing skills for conversing. Motor component new.
Management of concurrent medical conditions like seizures, gastrointestinal symptoms and sleep problems detailed	Management of feeding disorders, obesity, pica, dental health, wandering and motor disorders have been added
Challenging behaviors were included as a sub-group of medical problems and also in the section of psycho-pharmacology.	Behavioral and psychiatric disorders well described.Screening for behavioral and emotional problems (including depression > 12 y) advised.
Clinical approach to psycho-pharmacology explained step-wise.	Focus is on principles of prescription and drugs listed by behavior-symptom cluster.
CAM categorized as biological and non-biological groups	CAM grouped as natural products, mind and body practices, and others
Working with families	
PCP responsibilities include provision of longitudinal support to families, handling crises, providing emotional support and referring them for counselling, medical and/or mental health services if required.	Approaches have been devised at various levels for capacity building of PCP and promotion of professional-family partnership to provide patient and family centered care as well as promoting research.
Research and service needs	
Not included in the previous guidelines	Seven broad research areas identified

Prepared from Hyman, et at. [1] and Myeos, etal. [3]. CAM: Complementary alternative medicine; PCP: primarycare service provider.

adulthood (employment readiness, medical care, legal guardianship and living arrangements), and better understanding of ASD related rights and laws.

Research and Service Needs

Key areas identified to direct focus of funding include, basic and translational science (genetics, epigenetics, neurobiology, psychopharmacology), clinical trials for focussed interventions, epidemiological surveillance and implementation research for health care services.

Implications for the Indian Setting

These guidelines have brought our existing lacunae to the forefront. Few pediatricians routinely practice developmental surveillance. Though DSM-5 and indigenous Indian tools are used for diagnosis, and intervention centres have been established all over the country, there is wide variability in skills and availability of multi-disciplinary professionals dealing with ASD, inconsistency in practice protocols, and minimal quality checking. National Trust workshops are infrequent and primarily related to disability certification. Consensus statements and clinical practice guidelines framed by expert bodies [9,10] sensitize professionals, but do not focus on capacity-building.

Given these challenges, the provision of easily accessible, family centred, individualized and intensive, multi-disciplinary intervention according to these recommendations (but tailored to Indian settings) to all affected families is still a distant goal. The need of the hour is planning and implementing evidence-based concrete strategies that will enable professionals dealing with ASD to provide global standards of care to these children and their families.

Quality improvement, collaboration and integration is required among the health, education, social welfare and public health systems to provide evidence-based, universal care to children/adolescents and families affected by ASD. The 2020 guidelines outline strategies for capacity building of PCP to support this vulnerable population from suspicion of ASD, through diagnosis and service provision, to adulthood.

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