Autistic spectrum disorders: Diagnostic and therapeutic challenges in Mexico

María Elena Márquez-Caraveo, Lilia Albores-Gallo

SUMMARY
Autistic Spectrum Disorders (ASD) are developmental disorders with impairments in three broad domains: social interaction, communication and stereotypic movements and repetitive behavior. Their symptoms are complex, bizarre and most of them persistent, causing maladaptive and poor psychosocial adjustment.

Early detection and diagnosis is a priority in ASD; parents are the first to notice early autism symptoms: 50% observe signs in the first 12 months of age. Despite initial observations of atypical development, there are significant delays in seeking proper medical attention and correct diagnosis; less than 38% of families receive their diagnosis through health services. Educational and health primary care providers need training in developmental milestones with focus on language and socio-communicative domains. Health policy planners should facilitate rationale referral when key symptoms such as language alterations are detected. The use of valid instruments and surveillance approaches versus awareness through red flag symptoms is discussed.

Psychosocial interventions are the most important treatment, with ABA and TEACHH techniques recommended; pharmacological treatment (atypical antipsychotics, antidepressants, drugs for hyperactivity, sleep problems and anxiety) must be directed to treat comorbid conditions and combined with behavioral interventions.

Key words: Autistic disorders, treatment, psychosocial interventions, pharmacological therapy, Mexico.

INTRODUCTION
Autism is classified in the Pervasive Developmental Disorders category according to ICD-10 and DSM-IV. The term ASD (autism spectrum disorders) was proposed by Lorna Wing to cover a spectrum ranging from severe autism with profound mental retardation to high functioning autism. Therefore, ASD definition in this article will include: autism (infantile and atypical), Asperger’s syndrome/disorder and pervasive developmental disorders not otherwise specified (PDD-NOS).

These complex developmental disorders are defined by a triad of impairments in reciprocal social interaction, communication and the presence of repetitive or overly restricted behaviors.

ASD have a high level of comorbidity with intellectual and learning disabilities. The underlying deficit of ASD has been related to social cognition processes such as joint attention. This term defines the individual’s early ability to coordinate attention between a social partner and an object or event.

This impairment has been assumed as an explanation for the triad of ASD symptoms. Communication failures include difficulties in expressive and pragmatic abilities. Children with ASD have problems understanding rules of communicative exchange and the meaning of nonverbal
language. These deficits increase social withdrawal and atypical social and playing skills behavior. Stereotypical behaviors are observed in repetitive motor acts, ritualistic behavior and exaggerated interest in parts of objects. Children with ASD have key symptoms that may be unrecognized by primary care providers, and even specialized professionals may lack the skills and expertise to properly diagnose and help these children. Service delivery is needed in both directions, it is urgent to provide evidence-based and cost-effective interventions. The overall management implies a combined approach of medical, psychological, educational and social interventions.9 Whilst doctors may be prone to emphasize pharmacological treatments, psychologists and educators often prefer psychosocial interventions. This paper focuses on an update of evidence based recommendations for the diagnosis and treatment of ASD conditions.

**EARLY DETECTION AND DIAGNOSIS**

Parents are the first to notice early autism manifestations: 50% observe signs in the first 12 months,10 while 83.3% have concerns before age 2.11 Despite these initial observations of atypical development, there are still significant delays in seeking proper medical attention (mean delay: 22–32 months) and correct diagnosis (mean age: 53 months).12-14 Health and educational services play a crucial role in recognition and diagnosis of ASD.

In Spain, less than 38% of families receive their diagnosis through health services.12 In Bogota, up to 39% of referrals for autism is done by non-health or educational professionals mostly parents, friends or others.15 In the same line, many children (40%) are identified in school settings.16 Although the development of measures to assess the ASD in the last two decades has augmented, the cost of using these tools has increased significantly.17 Many of these instruments are very complex and are targeted to highly specialized professionals with experience in autism, so their use in the primary care setting is not feasible.18 Furthermore, some of them require training and take enormous time to administrate and rate.19,20 There are some screening and diagnostic measures available in Mexico with reported psychometric properties,21,22 but their systematic use in an autism surveillance program is still controversial, because no studies about their cost-effectiveness have been conducted.19,23

Some researchers propose that screening should be oriented to children at risk like probands’ siblings, children with low gestational age and/or weight, or children whose parents have developmental concerns.19 The use of key symptoms (red flags)24 for referral to child psychiatrist or clinical psychologist could be a more cost-effective strategy than screening instruments.25,27

Autism has a wide range of symptom presentations that can sometimes overlap with other developmental disabilities and communication disorders. Stigma associated with mental disorder bias the referral process and milder diagnosis such as expressive language delays or late language emergence are considered before autism. Therefore, it is important to raise awareness of abnormal language and social deviant development as key symptoms of autism among parents, family doctors and pediatricians. Head circumference should also be measured25,27 (table 1). There is enough evidence that early intensive intervention provides family support and improves outcome.30,31 Early detection should be followed by a rigorous assessment to confirm the diagnosis and evaluate the general functioning level of referred children and adolescents. Mexican researchers need to develop valid and reliable gold standard measures which can be used in busy settings with minimal training requirements.

Overall management includes early detection, diagnostic evaluation, psychosocial intervention and medical pharmacological treatment. This perspective is proposed in figure 1.

**TREATMENT**

According to recent guidelines,32 most of the interventions should be supported by systematic reviews or RCTs (Randomized Control Trials) when evidence is available, because many alternative medicine treatments commonly used are not evidence based.33,34

**Psychosocial treatment**

Psychosocial treatment begins with the appropriate education, which has had the greatest positive impact for ASD children in the last 50 years by reducing long stay treatments.

**Table. 1 Early Detection & Red Flags for ASD**

<table>
<thead>
<tr>
<th>Language</th>
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<tbody>
<tr>
<td>Indications for immediate evaluation</td>
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<tr>
<td>a. Language or social regression</td>
</tr>
<tr>
<td>b. Age 12 months: No babbling, pointing or gestures</td>
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<tr>
<td>c. Age 16 months: No single words</td>
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<tr>
<td>d. Age 24 months: No 2 word spontaneous phrases</td>
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<tr>
<td>Delayed speech and language skills</td>
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<tr>
<td>Confuses pronouns (e.g. You and Me)</td>
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<tr>
<td>Repeats words or phrases over and over (echolalia)</td>
</tr>
<tr>
<td>Social</td>
</tr>
<tr>
<td>• Does not respond to name by 12 months of age or older</td>
</tr>
<tr>
<td>• Avoids eye-contact</td>
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<tr>
<td>Repetitive Movements</td>
</tr>
<tr>
<td>• Flap their hands, rock their body, or spin in circles</td>
</tr>
<tr>
<td>Physical</td>
</tr>
<tr>
<td>• Increase in head circumference starting by 6 months old.44</td>
</tr>
</tbody>
</table>
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Current approaches include home and school based programs requiring parents and teachers as mediators to promote structured activities. Enrollment in these programs is recommended at an early age (2-3 years).35

The interventions and treatment methods for children with ASD include two approaches:36,37 Applied Behavioral Analysis (ABA) and the Treatment and Education of Autistic and related Communication-handicapped Children (TEACCH).

1. Applied Behavioral Analysis (ABA)

It is based on conditional operant theory to teach communication, play, social, academic, self-care, work, and community living skills, and to reduce disruptive behaviors in children with autism. This behavioral model proposed by Lovaas38,39 requires an average of 30 to 40 hours sessions per week of one-to-one instruction. The program is administered by trained certified therapists and parents at home and school settings.

The Early Start Denver Model (ESDM),40 based on ABA approach, was designed to address the needs of toddlers with ASD as young as 12 months. It is a comprehensive early behavioral intervention for infants to preschool-aged children with ASD that integrates ABA with developmental and relationship-based approaches.

A recent randomized, controlled trial study with ESDM41 intervention showed a shift in diagnosis from autism to PDD-NOS. The outcomes of this study involved an increase in IQ scores of 17 points (>1 SD) and significant gains in language and adaptive behavior.

ABA methods use discrete trial training (DTT) and incidental teaching (IT).

DTT consists of a basic four step process to teach:

a. A short verbal command or request or physical stimulus. For example: «Match colors» or «Show me blue».

b. Prompt stimulus. A prompt cue from the therapist to help the child respond correctly.

c. Response: resulting behavior, which can be a response or lack of response.

d. A consequence, which depends on the behavioral response; may include positive reinforcement of the desired behavior, or no reaction for the incorrect response.

IT focuses on teaching whenever the child shows interest in the teaching materials or activities in the natural environment. It is less structured than the DTT and allows the child to lead his/her interests and the teaching processes.42

2. The Treatment and Education of Autistic and related Communication-handicapped Children (TEACCH)43 program

It was developed by Eric Schopler and colleagues at the University of North Carolina.42 This approach is focused on modifying the environment to meet the individualized needs of children with autism. The four general components of TEACCH program are: physical organization and structure, daily schedules, work systems and task structure.

The purpose is to build a repertoire of social behaviors and communication methods through visual, verbal and tactile clues. The program aims to motivate direct learning and involvement in emotional experiences that take place in every day settings to observe the active learning and social participation of children.

Currently, a combination of ABA and TEACCH approaches is recommended;43 their use has showed significant IQ gains,44 leading children incorporation to regular classrooms.

Other interventions are:

a. Communication interventions include strategies designed by speech and language therapists.32,42 Their aim is to develop and enhance early communication skills, social communication and interaction. Modification of the communicative, social and physical environment through the use of visual prompts, routine and timetabling is recommended to minimize sensory sensitivity and complex social interactions.

b. Behavioral/psychological interventions include what was previously called behavioral management and is
now identified as Positive Behavior Support Systems.\(^4\)\(^5\)

The method requires identification of environmental contingencies to focus on the positive aspects of the environment and the child behavior. The focal interventions are designed to reduce symptom frequency and severity of specific problematic behaviors such as aggression or self-injury. This approach helps the development of adaptive skills. At present, the specific psychological intervention includes Cognitive Behavior Therapy (CBT), feasible in children with ASD who have a verbal IQ of at least 69. Children with social difficulties or with co-occurring depression and/or anxiety can benefit from this approach.\(^4\)\(^6\)

c. Interventions such as occupational\(^4\)\(^7\) and vocational support may aid children with ASD to prepare and adapt to future employment environments. Good workplace practices and proper work conduct such as timelines and understanding the concept of others privacy can be developed in adolescents with ASD.

The focus of psychosocial interventions is summarized in Table 2.

**Individualized treatment** is indicated given the heterogeneous presentation of ASD.

**Pharmacological treatment**

Family feedback regarding the etiology, diagnosis and prognosis is combined with the transient use of drugs indicated to deal with psychiatric comorbidity\(^4\)\(^8\) and/or epilepsy. Medical/pharmacological management should be used as a component of a multidisciplinary care approach.\(^4\)\(^9\)

There are some general principles regarding medication treatment assessment of the child’s home and school environment; routines in meals, daily activities, and sleep are crucial as changes in these domains are worth attempting before using medication.

In fact, there are few drugs specifically approved for use in children and adolescents with ASD. Pharmacological treatment for ASD should only be undertaken by physicians with appropriate training. It is important to define the target symptoms before medication and to measure any side effect.

Currently, there is no drug with a specific ameliorative effect on autism. Medication is a short- to medium-term intervention for symptoms such as self-aggression, irritability, hyperactivity, insomnia and anxiety; details are presented in Table 3.

At present, risperidone is indicated for symptoms such as irritability, aggression and repetitive behavior,\(^5\)\(^0\) but weight gain and metabolic syndrome must be identified as an adverse effect.\(^5\)\(^1\),\(^5\)\(^2\) Methylphenidate may be considered for attention difficulties and hyperactivity in children or young people with ASD,\(^5\)\(^3\) side effects as insomnia and weight loss should be carefully monitored.

Melatonin\(^5\)\(^4\),\(^5\)\(^5\) and clonidine\(^5\)\(^6\),\(^5\)\(^7\) had been suggested for the treatment of persistent sleep problems, resistant to behavioral interventions. Anxiety can be ameliorated with fluvoxamine\(^5\)\(^8\),\(^5\)\(^9\) or buspirone.\(^6\)\(^0\) We recommend to review the guidelines for epilepsy treatment.\(^6\)\(^1\),\(^6\)\(^2\)

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**Table 2. Recommended psychosocial interventions for ASD**

<table>
<thead>
<tr>
<th>Perspective</th>
<th>Types of Intervention</th>
<th>Recommendation</th>
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<tbody>
<tr>
<td>Education</td>
<td>PMI: Increase in reciprocal social interaction in children aged 1-6</td>
<td>As a good practice point, PMI programs should be considered as they may help families through:</td>
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<tr>
<td></td>
<td></td>
<td>• Interaction with their child</td>
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<tr>
<td></td>
<td></td>
<td>• Promote and increase development parental satisfaction, empowerment and mental health.</td>
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<tr>
<td>Communication</td>
<td>ESK/SC-I: Working in joint attention and visual support communication</td>
<td>Extrapolated evidence from well conducted case control or cohort studies indicates that interventions to support communication are recommended. The most appropriate intervention being assessed on an individual basis.</td>
</tr>
<tr>
<td>Behavioral/Psychological</td>
<td>Focal: Aberrant behavior (eg. self-injury, aggression) and daily skills.</td>
<td>Focal: A body of evidence indicates that focal interventions result in:</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• positive behavioral outcomes</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• reduce of symptom frequency and severity</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Increase of the development of adaptive skills</td>
</tr>
<tr>
<td>Specific</td>
<td>CBT</td>
<td>Specific: Requires a level of verbal and cognitive development which precludes their employment in some groups of children and young people with ASD.</td>
</tr>
<tr>
<td>Occupational</td>
<td>Social/Vocational</td>
<td>Occupational: Provides advice and support in adapting environments, activities and routines in daily life.</td>
</tr>
</tbody>
</table>

PMI = Parents Mediated Interventions; ESK = Early Communication Skills; SC-I = Social Communication and Interaction; CBT = Cognitive Behavior Therapy.
CONCLUSIONS

There are still many challenges that Mexico should meet to provide comprehensive management for children with ASD and their families, which are described as follows:

1. The exact number of people with these disabilities is unknown; epidemiological research would allow better planning of health, educational and social services for this population.

2. There is an urgent need to raise awareness of ASD disorders in the community. Previous studies have shown that at least two key symptoms (stares at nothing and abnormal language) are easily recognized by the parents as reasons for seeking medical help.63

3. It is also important to train health and educative services providers in developmental milestones with focus on language, social and communication domains in order to increase the early detection of ASD.

4. Medical assessment could be combined with the evaluation of functioning, as well as context barriers and facilitators.

5. Training in ABA and TEACCH techniques for professionals (psychologists, therapists, special education teachers, etc.) working in health and educational public institutions is highly recommended.

REFERENCES


