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Review article

# Recommendations for early diagnosis and intervention in autism spectrum disorders: An Italian–Israeli consensus conference

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ABSTRACT

On April 2013 experts in the field of autism from Italy and Israel convened in Jerusalem to discuss and finalize clinical recommendations for early diagnosis and intervention in Autism Spectrum Disorders (ASDs). In this paper, we summarize the results of this Italian–Israeli consensus conference.

ASDs constitute a class of severe and heterogeneous neurodevelopmental conditions caused by atypical brain development beginning during early prenatal life, reflecting many genetic, neurobiological and environmental influences. The first clinical signs of ASDs begin to be evident in children between 12 and 18 months of age, often after a period of relatively typical postnatal development. Recent longitudinal studies reveal substantial diversity in developmental trajectories through childhood and adolescence. Some intervention approaches have been demonstrated to be effective in improving core symptoms of ASDs, even if the heterogeneity and developmental nature of the disorder make it implausible that only one specific treatment will be best for all children with ASDs. More randomized control trials (RCTs) on early intervention are needed to identify the most effective strategies and provide the most efficient allocation of resources during the critical early intervention time period. Future research should focus on linking biological phenotypes with specific genotypes, thus establishing a foundation for the development of diagnostic screening tools and individualization of treatments.



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## Contents



## 1. Introduction

Autism spectrum disorders (ASDs) constitute a class of severe neurodevelopmental conditions caused by atypical brain development beginning during prenatal or early postnatal life and are considered to be life-long conditions, with core symptoms being permanent across the lifespan.[1] In the last years, significant progress has been made in understanding the causes of ASDs and converging links of evidence strongly point towards altered developmentally regulated brain connectivity. Studies of genetic and environmentally modulated epigenetic factors have highlighted the polygenic nature of these conditions, but etiologic causes of ASDs remain elusive in more than 80% of cases; only in 10–20% of the patients neuro-imaging and neurogenetic techniques have allowed to identify a specific medical and genetic syndromes as a cause of ASD.[2]

These disorders affect today up to one in 88–110 children, being therefore a major public health concern.[3,4] The increase in diagnosis rates is probably related to changing diagnostic criteria and to the development and use of new standardized autism-specific diagnostic tools.[5] However, taking into account the possibility of modifications in environmental or epigenetic factors, an actual increase of the prevalence of ASDs cannot be completely excluded.

In terms of clinical heterogeneity, the three symptomatological domains of ASDs and the distinction among ASD subtypes as defined by the DSM-IV have often resulted as being not congruent or not useful for studies aimed at examining the underlying structure of the ASD phenotype. Thus, the Working Group for the upcoming version of the DSM 5 has proposed a significant shift in the diagnostic conceptualization of ASDs. Proposed DSM 5 criteria may have in the future an impact on the sensitivity and the specificity of ASD diagnosis.[6,7]

The wide heterogeneity in the phenotypic presentation, regarding both configuration and severity of behavioral symptoms, reflects numerous genetic and environmental influences.[8] No biological diagnostic marker is currently available, and the diagnosis is only based on three symptomatological domains, i.e. impaired social communication, social reciprocity and repetitive/stereotypic behaviors. There are distinct subtypes of autism, which differ in terms of intellectual ability and severity of autistic symptoms, as well as in patterns of cognitive strengths or weaknesses.[9] Also the significant heterogeneity in the developmental trajectories can influence the appearance of clinical phenotypes and prospective outcomes of this population.[10]

The lack of medical tests or biological markers for identifying ASD has led researchers to focus on behavioral phenotypes, in order to detect early signs of autism. Early detection of clinical symptoms at onset and early diagnosis of ASD can improve opportunities for early intervention.[11] In recent years an increase in studies on the effectiveness of early intensive behavioral interventions has been reported; however, the current state of evidence is still unclear.[12,13]

On April 2013 experts in the field of autism from Italy and Israel convened in Jerusalem to discuss and finalize clinical recommendations for early diagnosis and intervention in ASDs. In this paper, we summarize the results of this Italian–Israeli consensus conference.