

Research in Autism Spectrum Disorders: What Do We Know and What Do We Need to Know?

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Heterogeneity in ASD

- ❖ About half of children with ASD learn to talk and about half are nonverbal; a larger proportion are verbal with early intervention.
- ❖ Lower functioning children may display severe behavior problems (aggression, self-injury) and require special education throughout school; some require 1:1 instruction or care.
- ❖ Higher functioning children may learn to read and write and can be taught in a regular classroom.

DSM IV Diagnostic Criteria for Pervasive Developmental Disorder (PDD)

Impairment in Social Interaction

- ❖ *Impairment in the use of nonverbal behavior; Lack of spontaneous sharing; Lack of social/emotional reciprocity; Failure to develop peer relationships*

Impairment in Communication

- ❖ *Delay in or lack of development of spoken language & gestures; Impairment in the ability to initiate or maintain conversation; Repetitive and idiosyncratic use of language; Lack of pretend play*

Restricted Repertoire of Activity and Interests

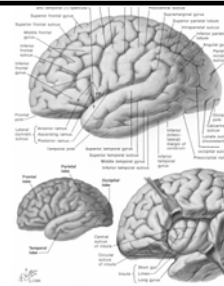
- ❖ *Preoccupation with restricted patterns of interest; Inflexible adherence to routines; Repetitive movements; Preoccupation with parts of objects*

Diagnostic Tools for ASD

- Interview with family:
 - ❖ Autism Diagnostic Interview (ADI)
- Assessment of child:
 - ❖ Autism Diagnostic Observation Schedule (ADOS)
- Observation of child:
 - ❖ Childhood Autism Rating Scales (CARS)
- Clinical diagnosis based on the DSM IV

Neuropathology Findings

- ❖ cerebellum
- ❖ brain stem
- ❖ basal ganglia
- ❖ limbic system
- ❖ cortex



Multifactorial Etiology

- ❖ Genetics: 90 to 95%
 - No "autism" gene yet
 - Polygenetic: 5-20 genes
 - Regulator genes: impact early in development and have a cascading effect of abnormalities
- ❖ Environmental: 5 to 10%
 - Brain pathology within first trimester of pregnancy
 - Primary versus Secondary impact
 - Timing: prenatal versus postnatal

Epidemiology

- ❖ Prevalence Rate
 - ASD in 1970: 3 to 4 per 10,000
 - ASD in 2000 in US: 3.4 per 1,000
 - ASD in 2000 in Europe: 6 to 7 per 1,000
- ❖ Recurrence Rate
 - 5% among siblings (3-9%)
 - 25% if there are 2 sibs with ASD
- ❖ Gender Ratio
 - 4 males: 1 female
 - Impact of IQ: 1:1 → 4:1 → 15:1
Low IQ High IQ

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National Academy of Sciences

Committee on Educational Interventions for Children with Autism

- | | |
|-------------------------|----------------|
| ❖ Catherine Lord, Chair | ❖ Alan Leslie |
| ❖ Marie Bristol-Power | ❖ Gail McGee |
| ❖ Joanne Cafiero | ❖ Samuel Odum |
| ❖ Pauline Filipek | ❖ Sally Rogers |
| ❖ James Gallagher | ❖ Fred Volkmar |
| ❖ Sandra Harris | ❖ Amy Wetherby |

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Committee's Charge

- ❖ To evaluate the scientific evidence on educational interventions for children with autism from birth to age 8 and address:
 - Diagnosis, Assessment, and Prevalence
 - Role of Families
 - Goals for Educational Services
 - Characteristics of Effective Interventions
 - Public Policies
 - Personnel Preparation
 - Needed Research

Committee's Process

- ❖ Conducted a systematic and rigorous assessment of research with an eye toward convergence of evidence from independent sources and different methodologies.
- ❖ Established guidelines for evaluating scientific evidence based on:
 - Internal Validity: Control for nonspecific factors such as maturation, expectancy, and experimenter artifacts
 - External Validity: Selection biases addressed in random assignment, sample size, and well-defined populations
 - Generalization: Documented in a natural setting outside of experimental intervention or with functional outcomes

What Do We Know About the Effectiveness of Intervention for Children with ASD?

(National Research Council, 2001: www.nap.edu)

- ❖ Studies have reported substantial changes in large numbers of children receiving a variety of intervention approaches, ranging from behavioral to developmental.
- ❖ The most common reported outcome measures are changes in IQ scores and post-intervention placement.
- ❖ Many single-subject design studies have demonstrated progress in individual responses to specific intervention techniques in a short time.
- ❖ Even in treatment studies with the strongest gains, children's outcomes are variable.

Characteristics of Effective Interventions: Recommendations

Six kinds of instruction should have priority:

- ❖ Functional, spontaneous communication
- ❖ Social instruction in various settings
- ❖ Teaching of play skills focusing on appropriate use of toys and play with peers.
- ❖ Instruction leading to generalization and maintenance of cognitive goals in natural contexts
- ❖ Positive approaches to address problem behaviors
- ❖ Functional academic skills when appropriate

What are the **Active Ingredients** of Effective Programs for Children with ASD?

(National Research Council, 2001: www.nap.edu)

- ❖ Early is better
 - Intervention by 3 ½ years is more effective than after 5 years
- ❖ Intensity matters
 - Active engagement for 25 hour per week
 - Low student/teacher ratio (no more than 2:1)
- ❖ Family participation is essential
 - Families were a critical component in effective programs
- ❖ Goals need to be individualized and monitored regularly
 - Goals and supports need to target behavior, social, and academic challenges
 - Progress should be documented within 3 months

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Can ASD be diagnosed early?

- ❖ There is no biological marker of ASD.
- ❖ ASD can be diagnosed reliably by experienced professionals down to 24 months of age.
- ❖ Over 85% of children diagnosed by experienced professionals at 2 years of age are still on the autism spectrum at 4 years of age.
 - *Social Impairments and Communication Impairments* were prominent at 2 years of age.
 - *Restricted Repertoire of Activities and Interests* were not prominent in some children until 3 years of age.

(Lord, 1995; Stone, Lee, Ashford, Brissie, Hepburn, Coonrod, & Weiss, 1999)

Core Social Communication Deficits of ASD: Clues for Earlier Identification

- ❖ Limited gaze shifts and lack of shared positive affect
- ❖ Lack of communicating to share attention and interests
- ❖ Lack of conventional and symbolic gestures
- ❖ Unconventional means of communicating (e.g., using person's hand as a tool; self-injurious behavior; echolalia)
- ❖ Poor coordination of gestures, sounds, and eye gaze
- ❖ Limited use of sounds and inventory of consonants
- ❖ Limited use of speech
- ❖ Poor language comprehension
- ❖ Limited conventional use of objects and lack of pretend play but good constructive play

(Stone, Ousley, Yoder, Hogan, & Hepburn, 1997; Wetherby, Prizant, & Hutchinson, 1998)

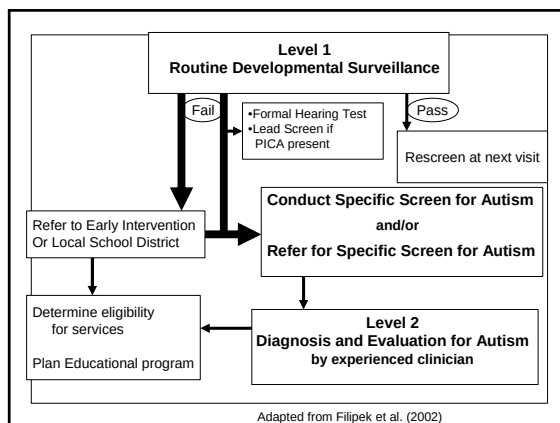
Practice Parameters for Screening and Diagnosis of Autism Spectrum

Absolute Indications for Immediate Further Evaluation:



- No babbling by 12 months
- No gesturing (pointing, waving, bye-bye) by 12 months
- No single words by 16 months
- No 2-word spontaneous (not just echolalic) phrases by 24 months
- ANY loss of ANY language or social skills at ANY age

Filipek, P., Accardo, P., Baranek, G., Cook, E., Dawson, G., Gordon, B., Gravel, J., Johnson, C., Kallen, R., Levy, S., Minshew, N., Prizant, B., Rapin, I., Rogers, S., Stone, W., Topley, S., Tuchman, R., & Volkmar, F. (1999). The screening and diagnosis of autistic spectrum disorders. *Journal of Autism and Developmental Disorders*, 29, 439-484.



First Signs

A national program to improve
developmental screening and referral
practices for young children

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Red Flags of Autism Spectrum Disorders and Developmental Delays in the Second Year of Life

ASD Red Flags

- Lack of showing
- Lack of coordination of nonverbal communication
- Lack of sharing interest or enjoyment
- Repetitive movements with objects
- Lack of appropriate gaze
- Lack of response to name
- Lack of warm, joyful expressions
- Unusual prosody
- Repetitive movements or posturing of body

ASD & DD Red Flags

- Lack of pointing
- Lack of playing with a variety of toys
- Lack of response to contextual cues
- Lack of communicative vocalizations with consonants

Wetherby and Woods (2003)



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Early Brain Overgrowth in ASD

- ❖ Most children with ASD have normal head circumferences (HC) at birth.
- ❖ HC studies indicate abnormal acceleration of brain growth from birth to 2 years of age.
- ❖ MRI studies indicate accelerated brain volume between 2 and 4 yrs of age and then brain volume declines over childhood and reaches normal levels in adolescents and adults.
- ❖ Abnormal brain overgrowth appears to be due to enlargement of gray and white matter in cerebral, cerebellar, and limbic structures.

(Courchesne et al., 2001; 2003; Piven et al., 1996; Sparks et al., 2002)

Meaningful Communication Outcome Measures: Recommendations

- ❖ Gains in initiation of spontaneous communication in functional activities
- ❖ Generalization of gains across activities, interactants (adults and peers), and environments

Empirically Supported Strategies for Initiation and Generalization

- ❖ Environmental Arrangement—modify the environment to prompt or cue a child to initiate social interaction
- ❖ Natural Reinforcers—provide access to objects or events that the child desires or removing undesired objects or events
- ❖ Time Delay—provide a stimulus and wait briefly before giving a verbal prompt for a child to respond
- ❖ Contingent Imitation—imitate a child's actions immediately following the child's actions

(Hwang & Hughes, 2000; Koegel, 1995; McGee, 1999)

Core deficits in joint attention...

- Difficulty orienting and attending to a social partner
- Difficulty coordinating attention between people and objects/ events
- Difficulty drawing another's attention to objects or events for the purpose of sharing experiences
- Difficulty reading / sharing affect, emotional states, or perspectives

What are child predictors of later language outcomes for children with ASD?

- ❖ Joint attention skills in preschoolers predicted gains in language 1 to 10 years later (Mundy Sigman, & Kasari, 1990; Sigman & Ruskin, 1999)
- ❖ Social communication skills in the second year predicted language outcomes 1 to 2 years later; understanding was the strongest predictor but initiating joint attention predicted the most unique variance in language outcome (Charman et al., 2003; Wetherby, Watt, Morgan, & Shumway, 2005)
- ❖ Level of repetitive behaviors and restricted interests in the second year of life was a significant predictor of similar behaviors on the ADOS as well as verbal and nonverbal outcomes 2 years later (Watt, Wetherby, & Woods, 2005)

What are caregiver predictors of later language outcomes for children with ASD?

- ❖ Children with ASD whose caregivers showed higher levels of synchronization during play at preschool age developed better joint attention skills 1 year later and better language outcomes 10 and 16 years later
- ❖ The strongest predictor of language gain was the proportion of caregiver utterances that follow the child's attentional focus and allow the child to continue the ongoing toy engagement

(Siller & Sigman, 2002)