



Autism Spectrum Disorders

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Autism Spectrum Disorders

- Autism Spectrum Disorders (ASDs) are lifelong, neurobehavioral disorders
- Impact child's behavioral, social and communication skills
- Five disorders – with a spectrum – from severe to high-functioning
- “You meet a child with Autism and you have met a child with Autism” each child is unique with different strengths and weaknesses

Autism Spectrum Disorders

- Fastest growing serious developmental disability in the U.S.
- More children will be diagnosed with an ASD this year than with AIDS, diabetes & cancer combined
- 1 out of 110 children born today
- 1 out of 70 boys – boys four times more likely
- Prevalence rate is increasing at an estimated 10-17% per year
- ASDs affect all genders, races, ethnicities and socio-economic levels
- No medical cure
- Early intervention is key

Common Co-morbidities

- Speech/Communication delay
- Seizures - As many as 39% of people with autism may have a seizure disorder
- Gastrointestinal symptoms - Surveys suggest that 46-85% of children with autism also have GI problems
- Sleep Dysfunction
- Sensory Integration Dysfunction
- Eating Disorders – refusal of food or Pica
- Depression

Red Flags – Developmental Milestones

- No big smiles or other warm, joyful expressions by six months or thereafter
- No back-and-forth sharing of sounds, smiles, or other facial expressions by nine months or thereafter
- No babbling by 12 months
- No back-and-forth gestures, such as pointing, showing, reaching, or waving by 12 months
- No words by 16 months
- No two-word meaningful phrases (without imitating or repeating) by 24 months
- Any loss of speech or babbling or social skills at any age

*This information has been provided by First Signs, Inc. ©2001-2005. For more information about recognizing the early signs of developmental and behavioral disorders, please visit <http://www.firstsigns.org> or the Centers for Disease Control at www.cdc.gov/ncbddd/autism/.

Diagnosis

- Complex disorder
- No single test
- Child can be seen by multiple specialists including: Developmental Pediatrician, Psychologist, Neurologist, Speech Pathologist
- DSM-IV Criteria
- Early intervention is key

Multidisciplinary Approach

- New program
- Initial screen directs child to appropriate clinic or Multidisciplinary ASD clinic
- Each child is seen for 30 min by each specialty – Developmental Pediatrics, Neurology & Psychology
- Child stays in room – minimizes stress and waiting time
- Team discusses each child and need for follow-up testing
- After testing, physician and clinical social worker have follow-up meeting with parents to explain diagnosis, answer questions, provide information, support and recommended services
- Children are seen at Amherst General and WCHOB

Neurological Perspective

- Neurological exam
- Etiology - possible causes
- Other neurological conditions such as seizures or Tuberous Sclerosis
- MRI or EEG tests
- Medication management (if necessary)

Clinical Evaluation - History

Perinatal and developmental history should include:

- milestones
- regression in early childhood or later in life
- encephalopathic events
- attentional deficits
- seizure disorder (absence or generalized)
- depression or mania
- behaviors such as irritability, self-injury, sleep and eating disturbances, and pica

Neurology 2000; 55:468-479

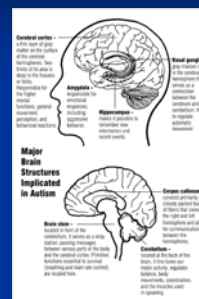
Clinical Evaluation

The physical and neurologic examination should include:

- longitudinal measurements of head circumference
- examination for unusual features (facial, limb, stature, etc.) suggesting the need for genetic evaluation
- neurocutaneous abnormalities
- gait
- tone
- reflexes
- cranial nerves
- determination of mental status, including verbal and nonverbal language and play

Neurology 2000; 55:468-479

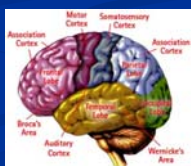
Neurological Basis



www.nimh.nih.gov/health/publications/autism/complete-publication.shtml

Neurological Basis

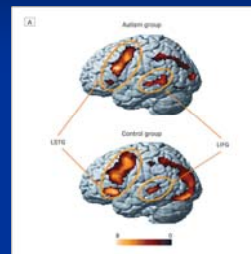
- Current evidence supports autism as a disorder of the association cortex, both its neurons and their projections
- Essentially it is a disorder of connectivity, which appears to primarily involve intrahemispheric connectivity



Minshew, N. J. et al. Arch Neurol 2007;64:945-950.

Functional MRI abnormalities observed in ASD

- Fig.1 Brain activation of autism and control groups during sentence comprehension (sentence vs fixation contrast). Participants with autism show less activation in the left inferior frontal gyrus (LIFG) than the control group, but more activation in the left posterior superior temporal gyrus (LSTG) than the control group.



Minshew, N. J. et al. Arch Neurol 2007;64:945-950.

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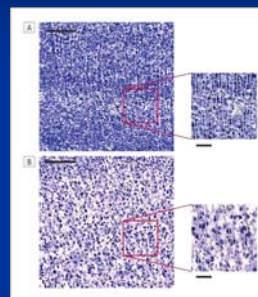
Neurological Basis

- Minicolumns are composed of radially oriented arrays of pyramidal neurons (layers II-VI), interneurons (layers I-VI), axons, and dendrites
- Minicolumns have been hypothesized to be the smallest radial unit of information processing in the cortex, but this function has not been confirmed
- In autism, minicolumns have been reported to be increased in number and narrower in width, with reduced neuropil space, with smaller neuron cell bodies and nucleoli
- These abnormalities have been observed bilaterally in cortical areas 3, 4, 9, 17, 21, and 22.

Minshew, N. J. et al. Arch Neurol 2007;64:945-950.

Micrographs of Brodmann area 4, lamina III, from a patient with autism (A) and from an age-matched control (B)

- Fig 3. Micrographs of Brodmann area 4, lamina III, from a patient with autism (A) and from an age-matched control (B).
- Insets highlight the cores of minicolumn fragments identified by a software program, illustrating the reduction in minicolumnar width in autism. Scale bars measure 200 μ m in the full images and 50 μ m in the insets. Image courtesy of Manuel Casanova, MD



Minshew, N. J. et al. Arch Neurol 2007;64:945-950.

Neurological Basis

- Because the narrowing of the minicolumns was largely related to a reduction in the neuropil space occupied by unmyelinated projections of gamma-aminobutyric acid inhibitory interneurons, a deficit in cortical inhibition was hypothesized and proposed to explain the 30% prevalence of seizures, the sensory sensitivities, and the bias in information processing toward low-level perceptual processing

Minshew, N. J. et al. Arch Neurol 2007;64:945-950.

Genetic Testing

- Family studies have shown that there is a 50-to-100-fold increase in the rate of autism in first-degree relatives of autistic children
- Although at least one autism linked abnormality has been found on almost every chromosome, X, 2,3,7,15,17 and 22 seem to have the most correlation

Pediatrics, 2007 Vol 120(5):1183-1215

Genetic Testing

- A chromosomal abnormality reported in possibly more than 1% of autistic individuals involves the proximal long arm of chromosome 15 (15q11-q13), which is a greater frequency than other currently identifiable chromosomal disorders



Pediatrics, 2007 Vol 120(5)1183-1215

Fragile X



Fragile X

- The most common known inherited genetic cause of ASD and of MR.
- Accounts for about one-half of cases of X-linked mental retardation and is the second most common cause of mental impairment after trisomy 21
- It is caused by mutation in the FMR1 gene. The vast majority of cases are caused by a trinucleotide (CGG)_n repeat expansion of greater than 200 repeats.
- Phenotype includes MR, macrocephaly, large pinnae, large testicles, hypotonia and joint hyperextensibility
- Yield of DNA testing has ranged from 0-8% with a mean of 3-4%
- Yet 30-50% of individuals with Fragile X will demonstrate characteristics of ASDs

J Dev Behav Pediatr. 2001; 22:409-417

Development & Behavior

- According to DSM-IV diagnosis criteria – child with Autism must have impairments in: 1) social interaction, 2) communication and 3) repetitive patterns of behavior, interests and activities
- Developmental history
- Observe how child moves, talks, interacts, plays
- Activities may include: stacking blocks, doing puzzles, writing and drawing pictures

Development cont.



Pictures & Story by 13 y.o. male with Asperger's

The cat is happily screaming because he just remembered that he is going to get a big fish from his friend and then he is going to get rich so then he can buy all the fish in the world.



DSM V - Ideas

- How will Autism and related Spectrum disorders be diagnosed in 3 years?
- What providers/evaluations will be useful?

Autism – Developmental Aspects

- Setting: DSM V – which proposes to combine the Asperger's group with Autism and PDD- NOS
- How do you then decide on therapeutic needs, therapy and educational methods, and intensity of remediation ?
- Need better tools to assess the skills of:
 - Language and communication
 - Social interaction
 - Intelligence or achievement
 - Emotional development

Language Assessment

Current methods in clinic:

- CLAMS (0-36 mo levels) from Capute scales
 - Receptive skills on Bracken (3 yr – 7 yr)
 - Conversational tasks (all)
- Currently multiple language scales used by Early Intervention, CPSE and CSE evaluators as well as tests for apraxia

Cognitive Tasks

Current methods in clinic

- CAT (0-36 mo levels) from Capute scales
- Educational tasks, drawing tasks, digit memory, picture vocabulary tasks

Classic Intelligence and achievement tests

- WPPSI or Stanford Binet IV (younger children)
- WISC 6-16 yrs
- WAIS >16 yrs
- WIAT and curriculum based levels

Social Assessment

■ Physicians

- History of changes – feeding, eye contact, crying and calming skills, attempts at language interaction from social smile to words – with attention to regression

■ Psychology

- Checklists and structured testing
 - SRS, TRF, CBCL, Greenspan, ADOS

Autism assessments w/ scores

- MCHAT
- ADOS
- CARS & GARS 2
- Asperger's scales
 - PDDST
 - CAST
 - AQ
 - GADS – Gilliam Asperger's Disorder Scale

MCHAT comments

- What is a “failed MCHAT”?
- What does it mean?
- Why do premature babies fail the MCHAT a little too much (when it is not Autism)?
- Should we keep asking practices to screen toddlers for Autism in some way?

Emotional & Psychological Well-Being of Child & Family

- Autism “happens” to the whole family
- In a survey of over 8,000 Mothers of children with Autism, 48% reported being currently or previously treated for depression
- Depression fairly common among older, higher-functioning children with autism
- Care Coordinator and Education Plan will:
 - Empower families with information and support
 - Minimize frustration in navigating a complex system
 - Help improve emotional well-being of child & family

Psychological Perspective

Review information before clinic

- Reports: IEP, EI, CPSE, CSE
- Checklists: SEGC, SCQ, CBCL, TRF
- Current programs and services
- Parent questionnaire, current concerns & stressors

Psychological Evaluation

Evaluation Interview with parent and child

- Further clarification of concerns and what help family is looking for
- Diagnostic engagement of child in activity or conversation depending on developmental level of child

Observation – frequency and quality of social interaction:

Eye contact	Use of pointing and other gestures
Joint attention	Speech tone and rhythm
Enjoyment of joint attention	Facial expressiveness and congruence with topic
Response to name	Functional and symbolic play
Social smile	Imagination in play
Social initiation and responsiveness	Any evidence for taking another's point of view
Back and forth conversation	Mannerisms that are unusual or repetitive

Psychological eval. cont.

Additional Information:

- Gilliam Autism Rating Scale, 2nd edition (GARS2)
- Gilliam Asperger's Disorder Scale (GADS)
- Social Responsiveness Scale (SRS)
- Autism Diagnostic Observation Schedule (ADOS)
 - Teacher/Caregiver Report Form
 - Observation of child at school (rare)

Complications in Diagnosis

- Significant medical histories
- Significant disruptions and/or stress in the child's environment
- Other diagnostic considerations:
 - ADHD
 - Anxiety Disorder (e.g., OCD, social, PTSD)
 - Reactive Attachment Disorder
 - Sensory processing disturbances
 - Nonverbal Learning Disorder
 - Oppositional and other behavioral disturbances
 - Mood disturbances

Patient Demographics

As of April 30, 2010 – 79 children have had initial evaluations in the center

Data on the first patients show

Gender: Male 84%
Female 16%

Ages: Youngest 2.3 years old
Oldest 17 years old
Median 5.7 years old

Patient Demographics – cont.

Race Description:	White	86%
	African-American	6%
	Biracial	4%
	Other	4%
Financial Mix:	Private	59%
	Managed Medicaid	23%
	Medicaid	16%
	Self	1%

Patient Demographics cont.

First 79 patients came from 48 different zip codes
All in New York



Patient Diagnosis

Of children who have been diagnosed thus far:

Autism Spectrum Disorders	59%
Behavioral	20%
Anxiety	9%
ADHD	9%
Other	3%

Family Support

- Parents need support
- Excellent phone access
- Meetings with families
- Follow-up process – outreach calls after appt and after diagnosis
- Family resources – information package
- Support Groups - monthly

Resources

- Autism Speaks – www.autismspeaks.org
- Autism Society of America www.autism-society.org
- AAP – Autism Page
www.aap.org/healthtopics/autism.cfm
- CDC Autism
www.cdc.gov/ncbddd/autism/index.html
- National Institute of Neurological Disorders & Stroke (NIH)
www.ninds.nih.gov/disorders/autism/autism.htm
- Interactive Autism Network (Kennedy Krieger research)
www.ianproject.org

Community Resources

- Autism Society of America – Western New York Chapter - www.autismwny.org
- NY ACTs - www.omr.state.ny.us/nyacts
- CASE – Center for Autism Support & Education
www.parentnetworkwny.org/Home/Programs/CASE
- Parent Network of WNY - www.parentnetworkwny.org
- Early Childhood Direction Center www.wchob.org/ecdc
- NYS OMRDD autism website
www.omr.state.ny.us/autism/index.jsp
- A Few Services: Autistic Services, Inc., Buffalo Hearing & Speech, Canisius Connections Program, Summit Educational Resources

Referrals

(716) 878-7600

Thank You!