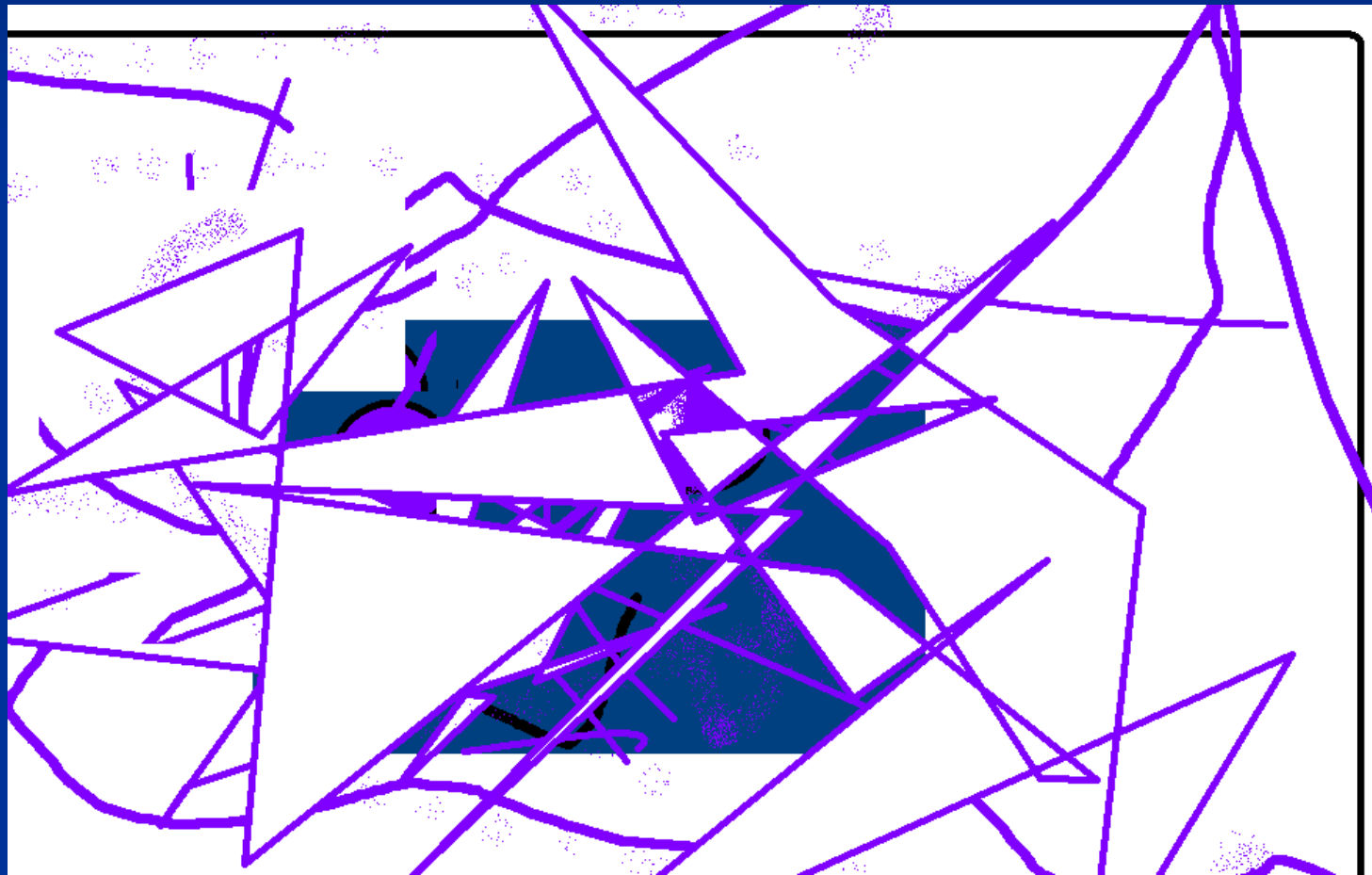


Autism

Cynthia King, MD and Lorerky Ramirez-Moya, MD



- Wing in 1980 describes two different kinds of infant with autism:
- -Placid, undemanding who rarely cries.
- -Screaming baby who is difficult to pacify.

Normal Developmental Milestones

- *Developmental milestones* are a set of functional skills or age-specific tasks that most children can do at a certain age range.
- Gross motor, Fine motor, Language, Cognitive, Social.
- <http://www.med.umich.edu/yourchild/topics/devmile.htm>



Early Interventions

- Concept: services that help a child to achieve his or her milestones from birth to 3 years old.
- The child learns and develops at the fastest rate during the first 3 years.
- The goal is helping the child to achieve the highest possible functioning and provide support and guidance to his/her family.
- 17 % kids under 18 years old are affected by Developmental, behavior or learning disability. Only 2.59 % children under 3 years old are being served through the federal funded Early Intervention Program

Early Interventions

- It is free!
- Anybody can refer to early intervention program in the community.
- They go where the child is located.
- Helps to transition to school district from IFSP(Individual Family Service Plan Basics) to I.E.P.
- They can refer or offer the child and his/her family to other programs like RESPITE, more specialized clinics, after school programs, fun activities for special needs children, support groups, education for the family and more

Early Interventions

- Needing fewer special education and other habilitative services later in life.
 - Being retained in a grade level less often
 - Being indistinguishable from classmates without special needs years after intervention.
 - Improves the functioning of the family.
- Family Infant Toddler Program, NM

What is Autism

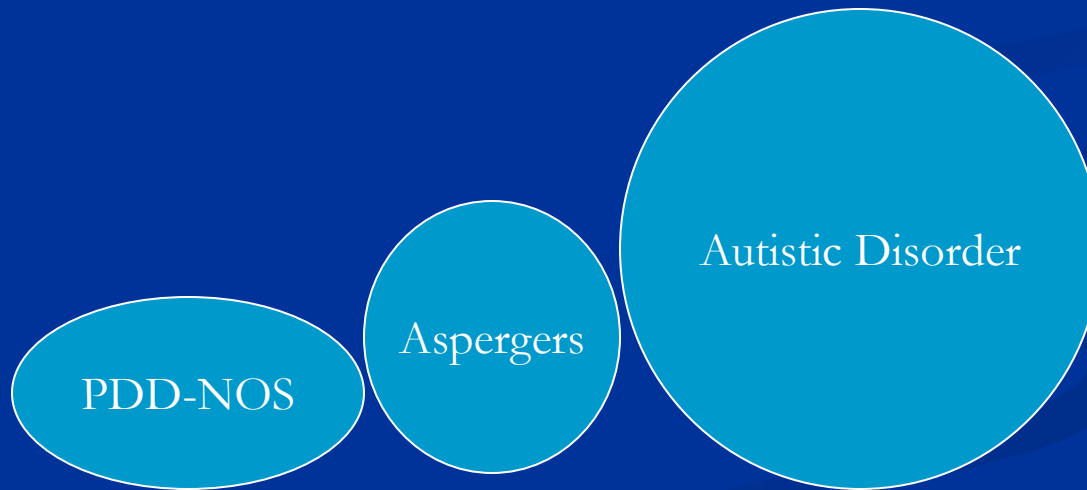
- Delay and Abnormal patterns of development before age 3 years in
- Social Development
- Communication
- Behavior and Interests

2 D's

- Delay & Deviations in Development
- Deviations = Qualitative Impairments

- Social
- Communication and Play
- Restricted Interests and Behaviors

Autism Spectrum Disorders



Types of ASDs

Diagnosis	Reciprocal Social Skills Deficits	Language Deficits	Cognitive Deficits	Restricted, Repetitive Behaviors
Autism	+	+	+/_	+
Asperger's Disorder	+	-	-	+
PDD-NOS	+	+/_	+/_	+/_

Autism

DSM-IV-TR

- A. A total of six (or more) items from (1),(2),and (3), with at least two from (1), and one each from (2) and (3)
- (1) social interaction
- (2) communication
- (3) behaviors

(1) Qualitative impairment in social interaction

- As manifested by at least two of the following:
- (a) marked impairment in use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
- (b) failure to develop peer relationships appropriate to developmental level
- (c) a lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., by lack of showing, bringing, or pointing out objects of interest)
- (d) lack of social or emotional reciprocity

(2) Qualitative impairments in communication

- As manifested by at least one of following:
- (a) delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime)
- (b) in individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others
- (c) stereotyped and repetitive use of language or idiosyncratic language
- (d) lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level

(3) Restricted repetitive and stereotyped patterns of behavior, interests, and activities

- As manifested by at least one of following:
- (a) encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus
- (b) apparently inflexible adherence to specific, nonfunctional routines or rituals
- (c) stereotyped and repetitive motor mannerisms (e.g. hand or finger flapping, or twisting, or complex whole-body movements)
- (d) persistent preoccupation with parts of objects

Social, Qualitative Impairment

- Direct gaze
- Social Smile (emerges around 2 months)
- Showing and directing attention
- Offering to share
- Seeking to share enjoyment with others
- Offer comfort
- Quality of social overtures

Social, Qualitative Impairment

- Range of facial expressions
- Inappropriate facial expression
- Imaginative Play
- Imaginative social play
- Interest in other children
- Response to approaches of other children

Communication, Qualitative Impairment

- Age of first single words (Under 24 months)
- Age of phrases (Under 33 months)
- Stereotyped utterances and delayed echolalia
- Reciprocal conversation
- Inappropriate questions or statements
- Pronominal reversals
- Neologisms/idiosyncratic language
- Verbal rituals

Communication, Qualitative Impairment

- Spontaneous imitation
- Pointing to express interest
- Nodding
- Use of another's body to communicate
- Poorly coordinated eye gaze and pointing

Restricted, Repetitive and Stereotyped Patterns of Behavior, Interests, and Activities

- Circumscribed interests
- Unusual preoccupations
- Repetitive use of objects or interest in parts of objects
- Compulsions/rituals (routines)
- Unusual sensory interests
- Hand and finger mannerisms
- Complex mannerisms or stereotyped body mvts.

Video 2



Findings

- 70% of individuals with Autistic Disorder also have an intellectual disability
- Often have strong visual and spatial skills
- 25% have a co-morbid seizure disorder
- IQ testing tends to show considerable scatter and tend to perform better on performance scales than verbal scales

Common Symptoms Not Unique to Autism

- Attentional impairment
- Hyperactivity
- Aggression
- Decreased pain sensitivity
- Sensory differences
- Sleep disturbance

Differential diagnosis

- The differential diagnosis can be challenging even for experienced clinicians

Differential Diagnosis

- Deafness
- Speech and language delays
- Intellectual Disability
- Nonverbal learning disorder
- ADHD
- Social Phobia and Selective Mutism

- Intellectual Disability, seizures, Fragile X syndrome, Down Syndrome or Tuberous Sclerosis may co-exist

Genetic Disorders Associated with ASDs

- Fragile X Syndrome
- Most common known genetic cause of Mental Retardation in males
- Fragile X in 3% of Autism cases
- 15-30% of individuals with genetically confirmed Fragile X will demonstrate characteristics of ASDs

Genetic Disorders associated with ASD

- Neurofibromatosis
- Tuberous sclerosis
- Angelman syndrome
- Prader-Willi Syndrome
- Down Syndrome (6-7% meet criteria)
- CHARGE syndrome (almost 50% meet criteria)
- Klinefelter Syndrome

Etiology

- Neurodevelopmental disorder but etiology for the neurodevelopmental abnormality is unknown
- Complex heritable disorder that probably involves multiple genes
- Increasing research into how environmental factors may play a role

MAY 15, 2006

www.time.com AOL Keyword: TIME

POP! GOES SCHOOL SODA ■ EXCLUSIVE: NINTENDO'S NEW GAME

TIME

**NEW INSIGHTS
INTO THE HIDDEN
WORLD OF
AUTISM**

BY CLAUDIA WALLIS

Nick Furth, 6, of Mine Hill, N.J.



Genetic Etiology

- Multiple Genes -2q, 7q31-35, 15q11-13, 16p, 17
- Concordance in identical twins 36% to 96%
- Fraternal twins 0% to 30 %
- Siblings 2% (vs. population rate of 0.1%)
5% have characteristics
- Male : Female 4:1
does not appear to follow any classic Mendelian patterns of transmission

Genetic Disorders Associated with Autism

- Fragile X Syndrome

Most common known genetic cause of Intellectual Disability in males

- Fragile X Syndrome is diagnosed in 3% of Autism cases

- 15-30% of individuals with genetically confirmed Fragile X will demonstrate characteristics of Autism

Possible Environmental Influences

- Prenatal or postnatal exposure to viral infections, chemicals, medications or vaccines
- Immunologic factors during prenatal development
- To date there is no published evidence linking MMR or other vaccines containing thimersol to the onset of autism

Pathophysiology

- Large brain size, rapid expansion from birth to two years

Interpreted as reflecting a failure of neuronal pruning

Increased white matter : local areas tend to be overconnected ,while links between more distant regions of brain are weak

Interconnectivity Differences

- Poor synchronization between different brain regions during thinking tasks in individuals with autism and normal IQs

Interconnectivity

- Unclear if interconnectivity problem is the result of autism or it's cause
- Because of fact that early intervention brings better results for children with ASD , it suggests that some of the odd brain anatomy and activity are secondary to lack of use
- “The neurons that fire together wire together”

Prognosis/Course

- Onset
 - Birth to 30 months (90%)
 - Regression by 24 months (35-50%)
(of language or social function)
- No substantial medically associated complications
- Normal Life Expectancy

Prognosis/Course

- Chronic and Lifelong
- Some symptoms decrease
- Some symptoms persist
 - Lack of social reciprocity
 - Language abnormalities
 - Difficulty using/understanding emotional concepts
 - Rigid and ritualistic behavior

Prognosis/Course

- 2-3% of individuals with Autistic Disorder are able to progress normally through school or live independently
- No means of prevention
- No fully effective treatment for core symptoms
- Early intervention improves prognosis

Factors that Predict Autism Outcome

- Expressive Language
 - Communicative speech by age 5
- Language Comprehension
- Intellectual Capacity
 - Non-verbal intelligence
- Adaptive Function
- Severity of Autism
 - Social
 - Restricted, Repetitive Behaviors
 - Aggression



Prevalence of ASDs

- 1 in 88 children

Possible Explanations

- Broadened criteria for diagnosis of ASDs
- Improved screening tools and more reliable evaluation instruments
- Professionals have become increasingly proficient in recognizing & diagnosing ASDs
- Heightened public awareness, parents more likely to raise a concern specifically about autism

**How can Nurses and Doctors
communicate with Patients who are
Non Verbal?**

Effective Communication

What's most important?

- The willingness of the Communicator and Communication Partner learning to communicate with each other

Tips for Communicator's Partner

- Increased general knowledge about communicator's most likely communication strengths and weaknesses

Strengths of Communicators

- Visual skills
- Like routines

Strengths of Communicators

- Often have a family member or care giver who can translate their communication attempts

Potential weaknesses of Communicators

- Verbally presented information not processed as easily as visually presented information
- Processing of information may be slowed or misunderstood

Recommendations

- Pay careful attention to how person tries to communicate
- Adjust your energy level
- Spend more time with individual
- Use any special form of communication the person needs (pointing to pictures in a communication book, high –tech speech generating device, writing, typing, iPads)

Disruptive Behaviors

Psychiatric Problems versus Medical/Physical Issues

- ? Does patient look toxic compared to baseline?
- ? Febrile?
- ? Does caregiver report an acute change in behaviors , agitation , or mental status that preceded office visit?
- ? If self injurious , does caregiver know when patient is usually self injurious?

Disruptive Behaviors

- Stay calm
- Try to be proactive and control environment as much as possible to decrease stress on patient :
lights, noise, temperature
- Notice early signs of agitation , and ask care giver what has been helpful in the past

Treatment

- 24 hours a week of active engagement

- Behavioral therapy

 - Applied Behavioral Analysis

 - TEACH model

 - Speech and Language Therapy

 - Occupational Therapy

Complementary and Alternative Medical Treatments

- Gluten/Casein free diets

 - almost 1/3 of children are treated with diet

 - initial theory of increased opiate peptides not substantiated

Many parents report improvement

Plausible that many children may have lactose intolerance or milk allergy and sleep, stool quality, and mood may be improved with less discomfort

More research needed

Treatment

- Evidence Based Psychopharmacology targetting specific symptoms
- Anxiety and obsessiveness and rigidity
 - SSRIs (fluoxetine, sertraline) helpful
- Hyperactivity, attention and concentration issues
 - can be helped with ADHD meds
- Aggression or irritability helped with risperidone
- Insomnia helped with melatonin

What information is helpful to share with physician?

- Any changes in home life (move, death of a family member, new baby)
- Any reported change in routine
- Change in sleep, appetite, energy level, decrease in enthusiasm for usual activities, decrease in self care, loss of skills
- Any suspicion of pain or loss of physical movement

What information is helpful to share with physician?

- If patient seems agitated
- What caregiver suggests helps patient to be calm
- If signs of bruises or marks