

EARLY IDENTIFICATION OF AUTISM SPECTRUM DISORDERS:

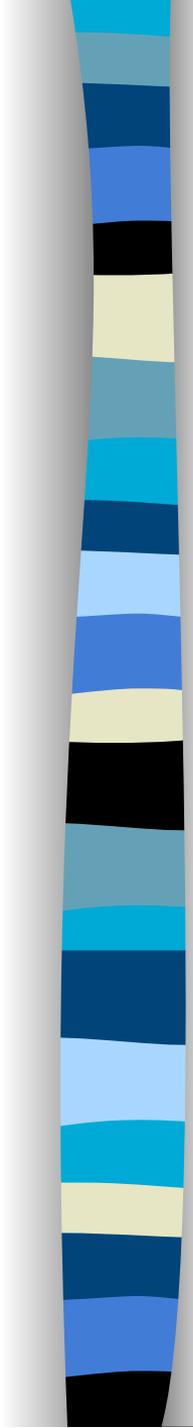
An Introduction for Those Who Work with Families

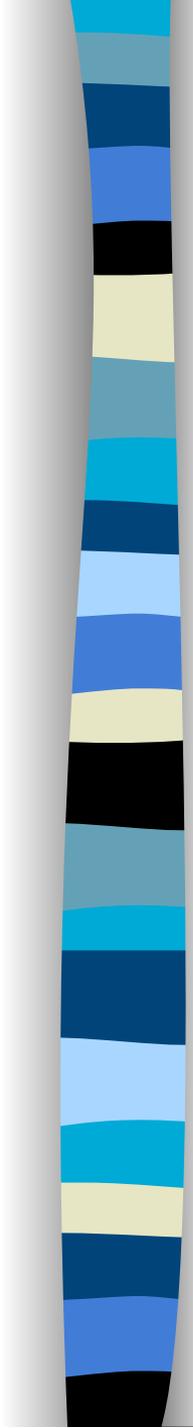


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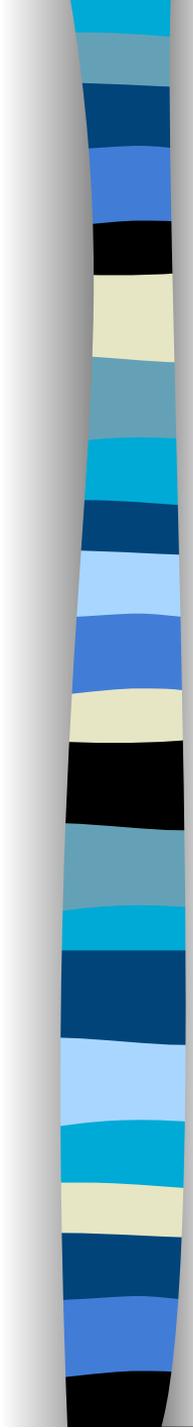
Children's Evaluation and Rehabilitation Center
Albert Einstein College of Medicine

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- The CDC contracted with the Association of University Centers on Disability (AUCD) to support regional “Act Early” Conferences and activities related to dissemination about learning the early signs of autism
 - The Rose F. Kennedy University Center for Excellence in Developmental Disabilities Education, Research and Service hosted a regional summit 3/2009 and received a mini-grant from AUCD to help support statewide “Act Early” activities



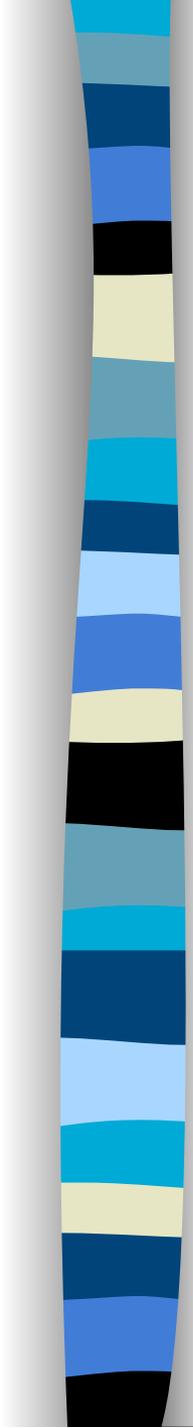
Outline for today's webinar

- What is autism?
- Why is it important to recognize the signs early?
- How can we recognize autism in young children?
- Medical and nutritional issues in autism
- Controversies
- How you can help



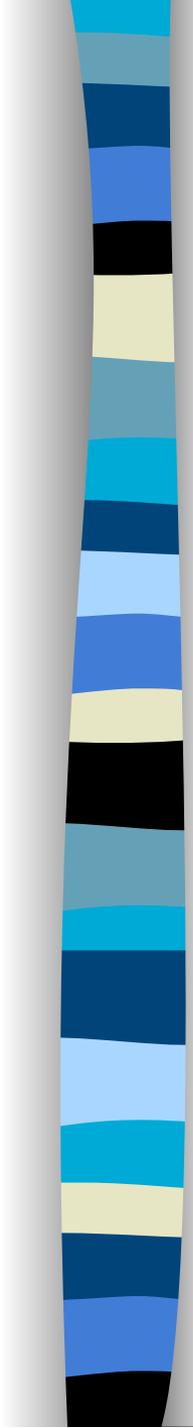
Jonathan

- A family that you work with has a 20 month old toddler about whom they express concerns: he is not yet speaking.
- At first they thought that maybe he had a hearing problem, because he doesn't seem to respond when they call his name. But a hearing test was normal.
- Relatives have assured them that boys talk later. But still they worry.....
- You observe Jonathan to be a very serious toddler whom you cannot get to smile. He does not appear interested in peek a boo or your rendition of Wheels on the bus. He has no interest in the toys in your office.



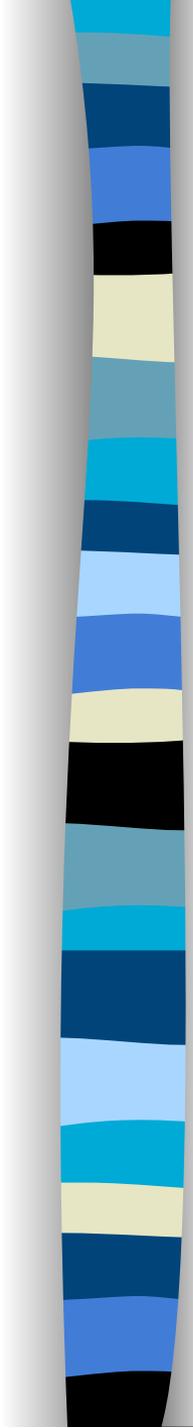
Michael

- Michael was “born talking” according to his mom, with full sentences by 18 months
- Parents have had no concerns of their own about his development
- At age 3, Michael entered a nursery program. At the parent teacher conference parents heard that the program has concerns about Michael’s social skills
- He doesn’t talk or play with the other children. In fact, the teacher realized recently that he still does not know the names of his classmates.



Michael (cont'd)

- No matter what the class is talking about, Michael steers the topic to things of interest to him: Thomas the Tank Engine and subways
- A loud sound such as a fire drill or another child crying causes him a lot of distress and he will cover his ears and try to leave the room.



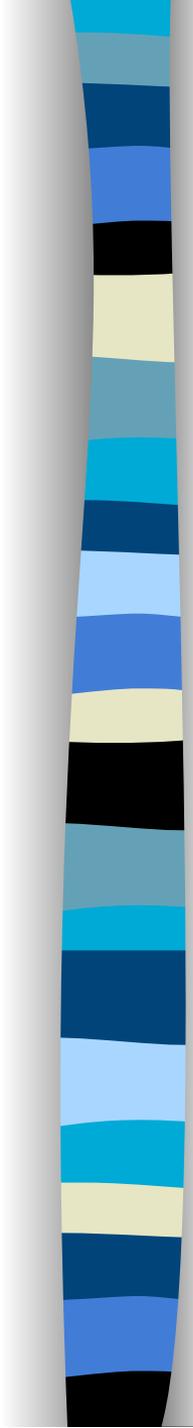
David

- David is a 23 month old toddler boy born prematurely at 31 weeks gestation.
- He had a history of reflux and motor delays and received physical therapy in the past.
- Presently, he has not developed language but is described as “an independent toddler” who climbs to get what he wants on his own.



David (cont'd)

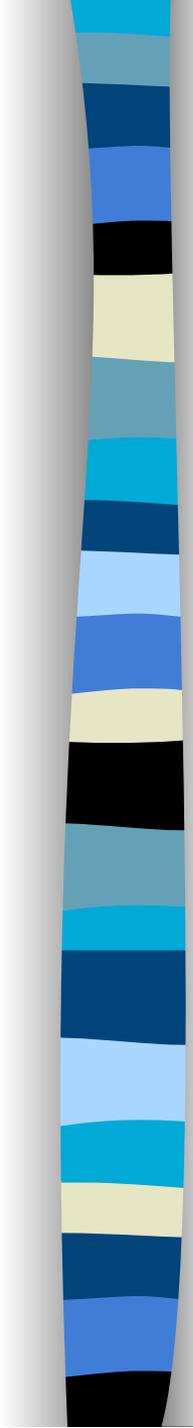
- He can entertain himself for long periods of time. He likes to line things up (blocks, toy cars, DVD cases), and repeatedly open and close doors.
- He is a very picky eater and relies heavily on a bottle and baby food. He cries and spits out food with more texture, other than pretzels and chips which he loves.



Autism Spectrum Disorders

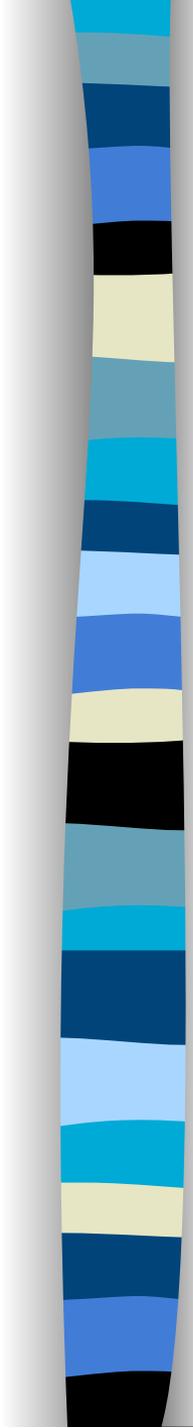
- Refer to a set of neurologically–based but behaviorally–defined disorders characterized by:
 - **Impairment in social interaction**
 - Impairment in communication
 - A restricted, repetitive repertoire of interests and activities

(DSM IV)



Demographics

- Prevalence: 1:110
- Boys 4x more affected than girls
- Previously not diagnosed until 4-5 years of age; now often diagnosed by 2 years
- Some speak fluently; others nonverbal
- 70% with cognitive impairment
- Some in regular class; others in highly restrictive settings



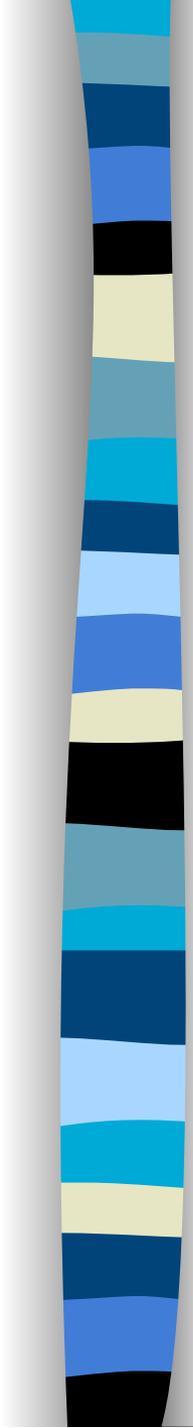
Pervasive Developmental Disorders (PDD) / Autism Spectrum Disorders (ASD)

- Autistic Disorder
- PDD.nos
- Asperger's Disorder

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- Childhood Disintegrative Disorder
 - Rett's Disorder

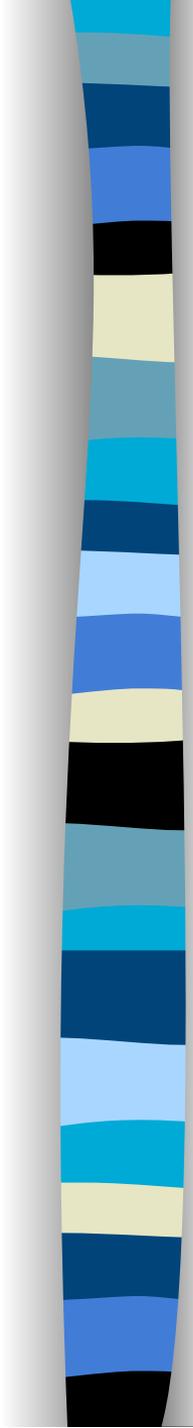
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(DSM IV)



Why the rush to diagnose autism?

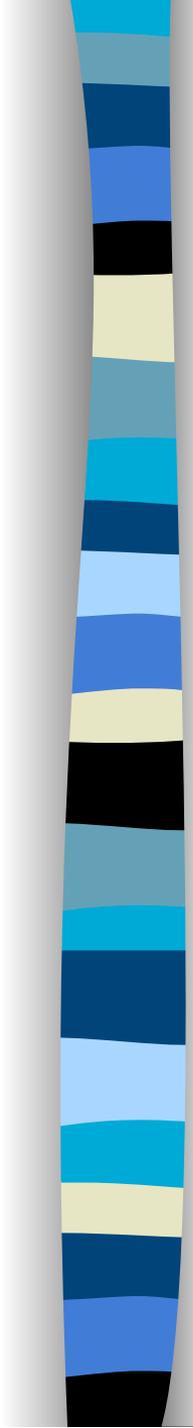
- The main research-based treatment for autism is intensive, structured teaching of skills, often called behavioral intervention. In order to help children with autism have the best outcome, it has been shown that it is critical to begin this intervention as early as possible. The only way to get this type of specialized intensive intervention is with a diagnosis of ASD.



When it comes to ASD, these are the goals:

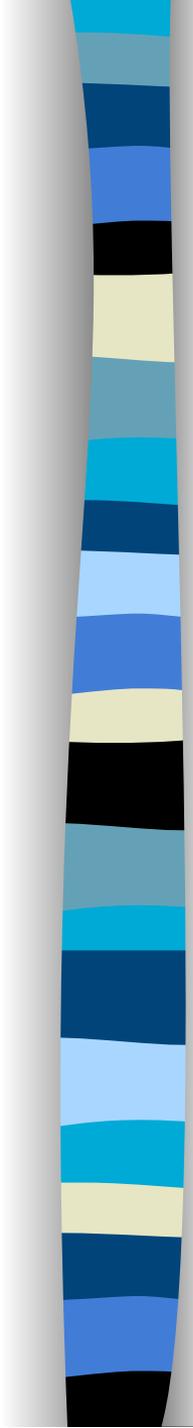
- Identification of ASD as early as possible (ideally before age 3 years)
- Initiation of intensive autism-specific intervention as soon as possible for best outcome

How can we achieve these goals for all children with ASD?



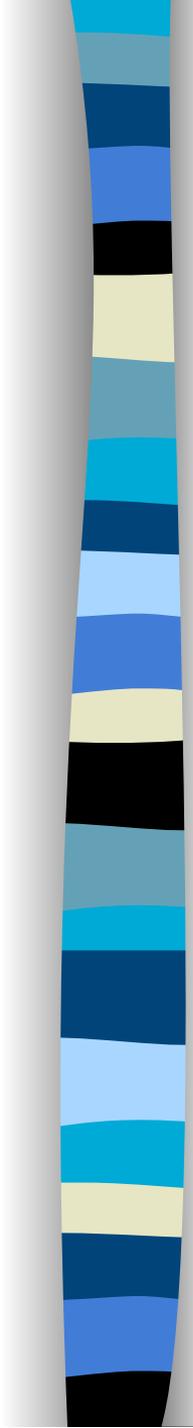
How do children with ASD come to attention?

- Parental concern
- Concern of others who spend time with the child (e.g. daycare providers, teachers, extended family members, professionals in social agencies)
- Picked up on screening by pediatrician or other healthcare providers

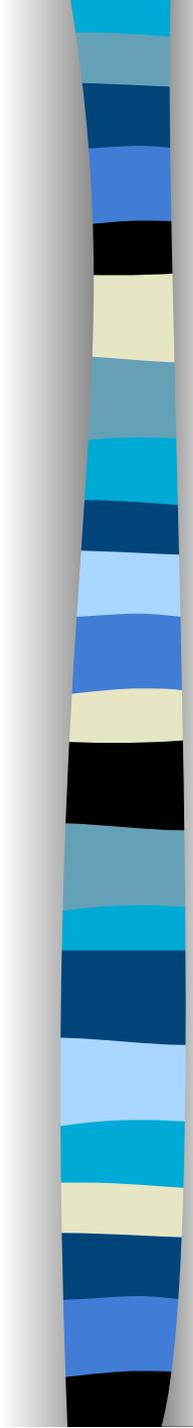


Efforts to increase early identification

- Pediatricians urged by AAP to screen for autism at 18 and 24 month visits
- Autism Speaks efforts to increase public awareness about autism
- CDC's: "Learn the Signs. Act Early" campaign targets parents, healthcare professionals, childcare providers, providing information about child development

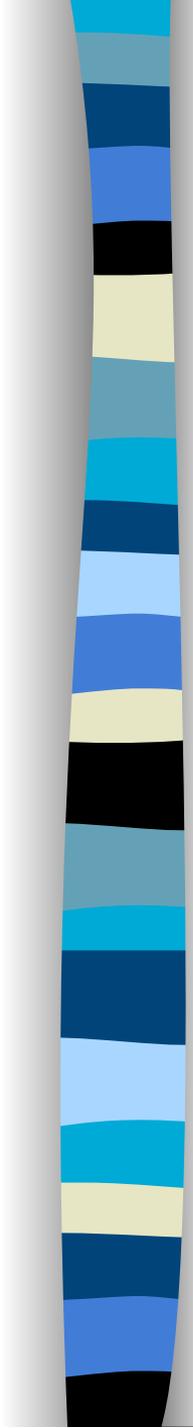


Delays in development
vs.
Atypical development



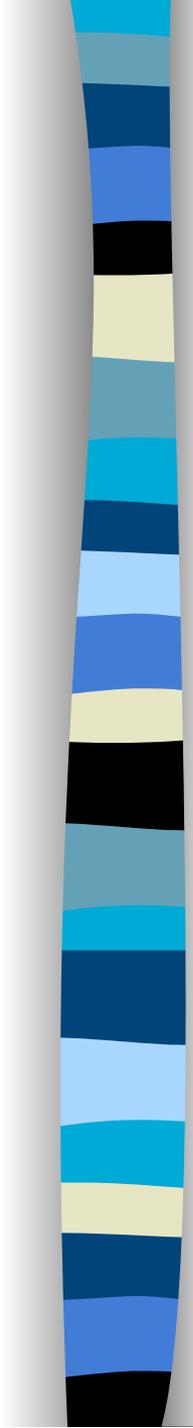
Expanded definition

- **Social impairment:**
 - Diminished social interest/motivation,
 - Lack of know-how
 - Lack of “joint attention”/”shared affect”
- **Communication impairment:**
 - Lack of language/atypical language
 - Diminished conversational ability
 - Decreased eye contact/gestural system
 - Lack of attention to language



Expanded definition (cont'd)

- Atypical interests/behaviors
 - Unusual interests
 - Perseverative interests/play
 - Lack of pretend play
 - Rigidity
 - Unusual reactions to sound, touch and smell



The spectrum of impairment:

- Social impairment

Lack of social interest → lack of social know-how

- Communication

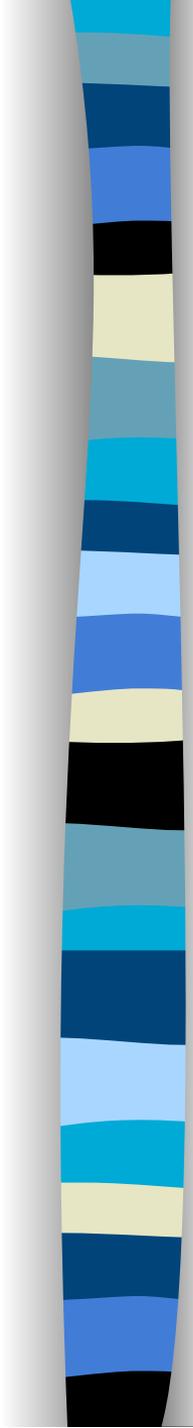
No language → highly atypical language →
“Normal language”/pragmatic difficulties

- Atypical interests

Obviously atypical → perseverative, rigid

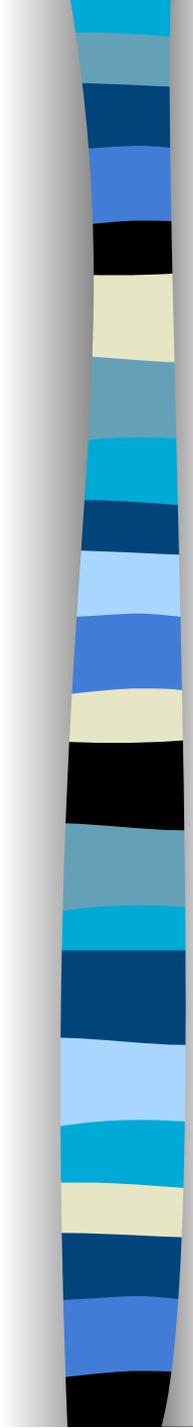
- Cognition

Very delayed → “splinter skills” → gifted



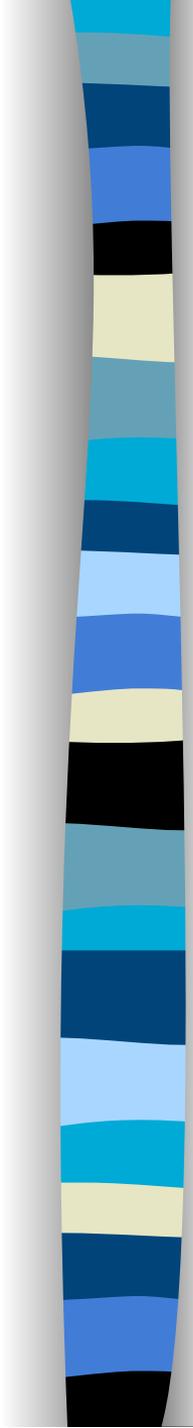
Early parental concerns in ASD

- Mean age of first parental concern is **18-19 months** (DeGiacomo & Fombonne, 1998; Harrington et al, 2006)
- **30-50%** report first concerns before 1st birthday



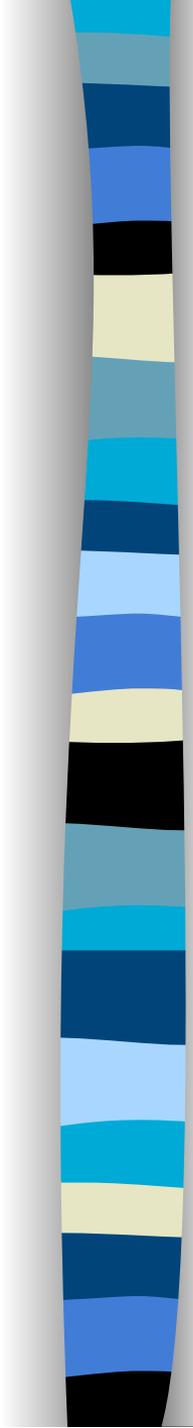
Chief Concerns: “My child...

- Doesn't talk or understand
- Doesn't listen...is he hearing?
- Is hyperactive/tantrums a lot
- Walks on tiptoes
- Doesn't play with toys
- Used to say.....
- Is he autistic?



Screening for ASD

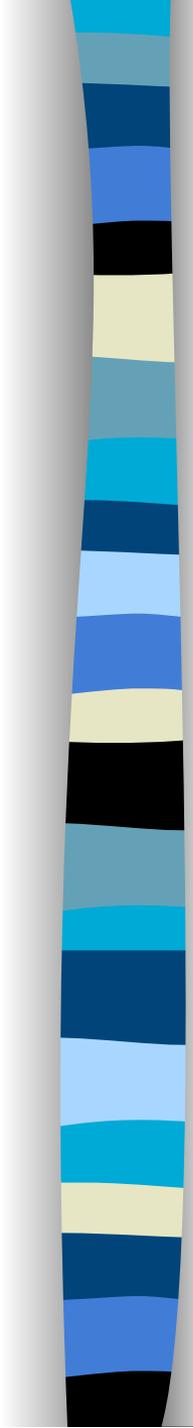
- Screening is the act of carrying out a specific instrument (e.g. questionnaire) looking for signs of a particular condition
- Screening instruments for ASD exist and make use of research findings relating to the signs of autism in infants and toddlers
- 2 streams of evidence have helped us understand those early signs



Retrospective studies

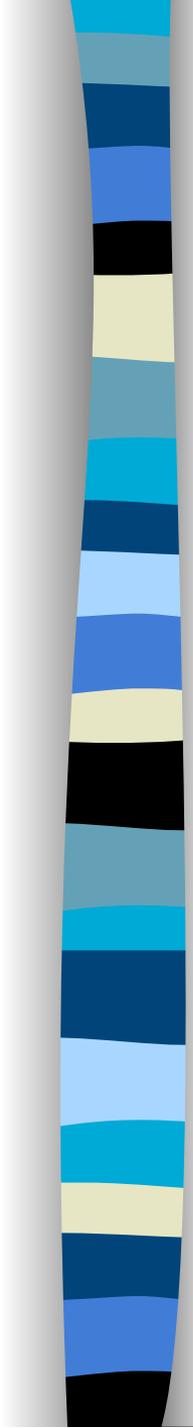
- Retrospective analysis of home videotapes of first birthday celebrations showed signs predictive of autism in 90% of children subsequently diagnosed

(Osterling and Dawson, 1994)



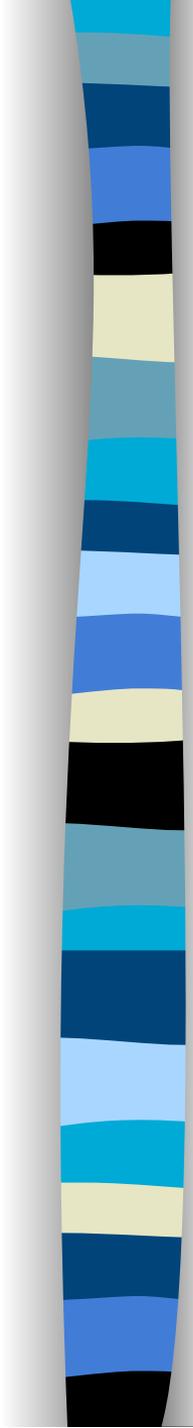
Signs Predictive of Autism in 12 month olds

- Lack of eye contact
- Failure to orient to name
- No pointing
- No showing behaviors



Prospective studies

- Looking at high risk cohorts such as “baby siblings” of children with autism:
(Bryson, Zweigenbaum, Brian, et al, 2007)
- By 12 months
 - Social/ Communication delays differentiate
 - Imitation difficulties
 - Visual fixation differences



Prospective studies (cont'd)

■ By 18 months:

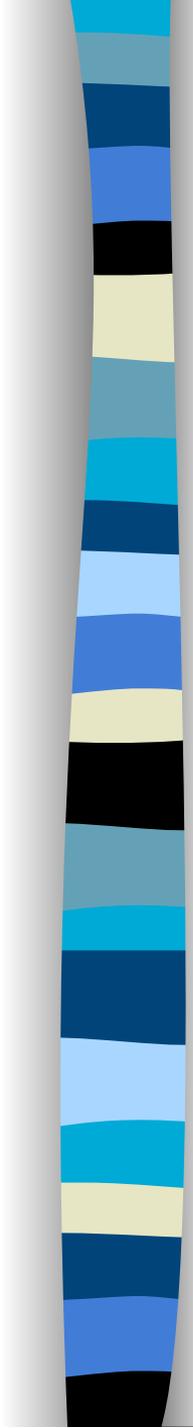
- Lack of pretend play and social play, decreased “pointing to show”, decreased social interest and joint attention

(Baron-Cohen, Allen, Gillberg, 1992)

■ By 24 months:

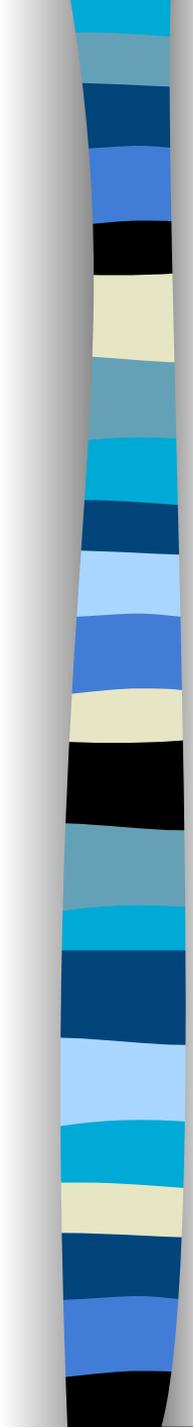
- 50% showed unusual sensory interests
- 40% hand and finger mannerisms

(Chawarska, Klin, Paul, Volkmar, 2007)



Early Signs (cont'd)

- Milder social deficits often come to light with a first organized peer exposure such as at daycare or preschool. In that context, the child's difficulties with peer interaction or unusual behaviors or interests relative to typically developing peers become evident.



Autism-specific screening instruments

- CHAT (Baron-Cohen, Wheelright, Cox, et al, 2000)
- mCHAT (Robins, Fein, & Barton, 1999)

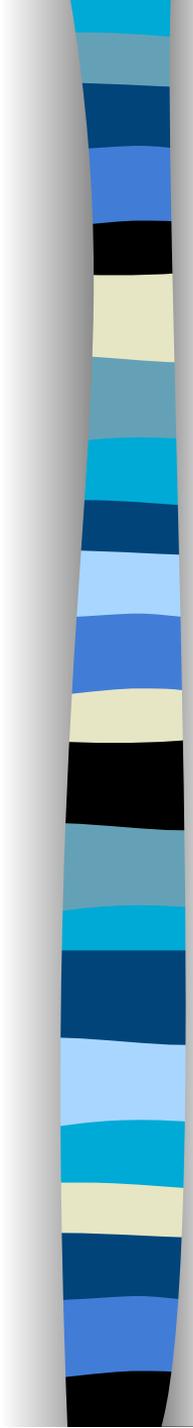
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- CAST
 - Social Responsiveness Scale
(Constantino, 2005)

M-CHAT

Please fill out the following about how your child usually is. Please try to answer every question. If the behavior is rare (e.g., you've seen it once or twice), please answer as if the child does not do it.

1. Does your child enjoy being swung, bounced on your knee, etc.? Yes No
2. Does your child take an interest in other children? Yes No
3. Does your child like climbing on things, such as up stairs? Yes No
4. Does your child enjoy playing peek-a-boo/hide-and-seek? Yes No
5. Does your child ever pretend, for example, to talk on the phone or take care of a doll or pretend other things? Yes No
6. Does your child ever use his/her index finger to point, to ask for something? Yes No
7. Does your child ever use his/her index finger to point, to indicate interest in something? Yes No
8. Can your child play properly with small toys (e.g. cars or blocks) without just mouthing, fiddling, or dropping them? Yes No
9. Does your child ever bring objects over to you (parent) to show you something? Yes No
10. Does your child look you in the eye for more than a second or two? Yes No
11. Does your child ever seem oversensitive to noise? (e.g., plugging ears) Yes No
12. Does your child smile in response to your face or your smile? Yes No
13. Does your child imitate you? (e.g., you make a face-will your child imitate it?) Yes No
14. Does your child respond to his/her name when you call? Yes No
15. If you point at a toy across the room, does your child look at it? Yes No
16. Does your child walk? Yes No
17. Does your child look at things you are looking at? Yes No
18. Does your child make unusual finger movements near his/her face? Yes No
19. Does your child try to attract your attention to his/her own activity? Yes No
20. Have you ever wondered if your child is deaf? Yes No
21. Does your child understand what people say? Yes No
22. Does your child sometimes stare at nothing or wander with no purpose? Yes No
23. Does your child look at your face to check your reaction when faced with something unfamiliar? Yes No

© 1999 Diana Robins, Deborah Fein, & Marianne Barton

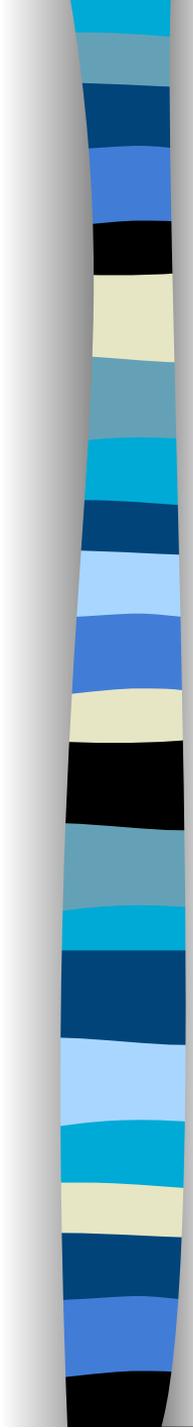


To succeed with our national goal of early identification of ASD....

- Listen to parents, elicit their concerns, provide parents with resources to follow through on their concerns
- Empower those who have contact with families with young children to support families with information/resources
- Implement ASD screening programs in healthcare settings

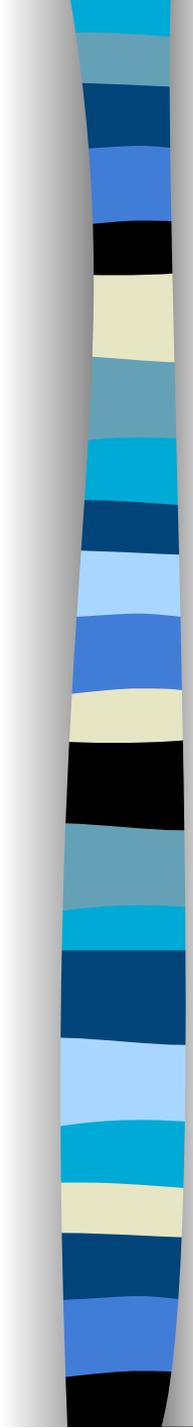
The Medical Work-up for ASD

- Diagnostic clarification
- Etiologic evaluation
- Identification and management of medical conditions seen more commonly in individuals with ASD



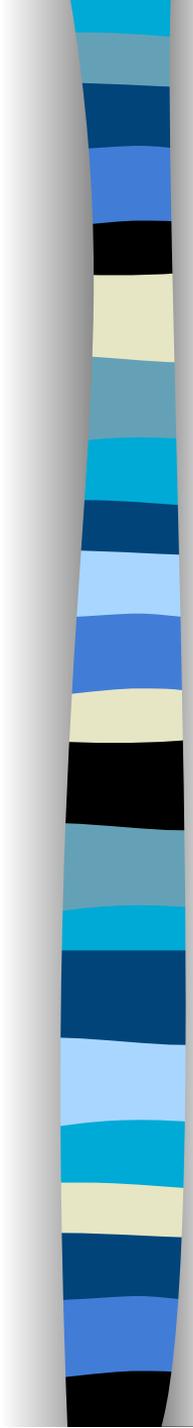
Potential active medical issues in management of ASD

- Genetics
- Regression
- Seizures
- GI/feeding issues and special diets
- Sleep problems and behavioral issues



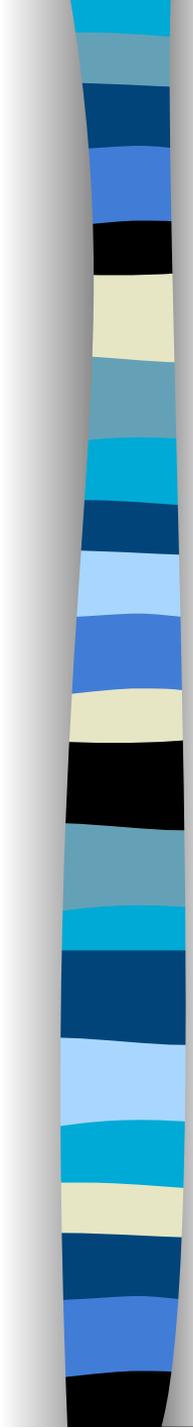
What causes ASD's?

- Conditions which result in damage to the brain: i.e. infections, metabolic processes, genetic syndromes, neurologic conditions, and teratogens
- Presently, in about 10-15% of children with ASD, doctors can identify the “cause”
- This involves looking for clues in the past medical history, family history, developmental history, physical examination, and medical work-up



What causes ASD's (cont'd)?

- Genetics is a potent factor
 - Boys more often affected suggests genetic factors
 - Twin studies (concordance rates of fraternal 5-10%; identical 90%) support a strong genetic contribution
 - No single autism gene, rather mutation of multiple gene sites can result in ASD
 - The majority of children diagnosed with autism do not have a family history of the condition
 - Overtime, we are understanding ways in which the environment and infectious exposures impact on the genome



Autistic Regression

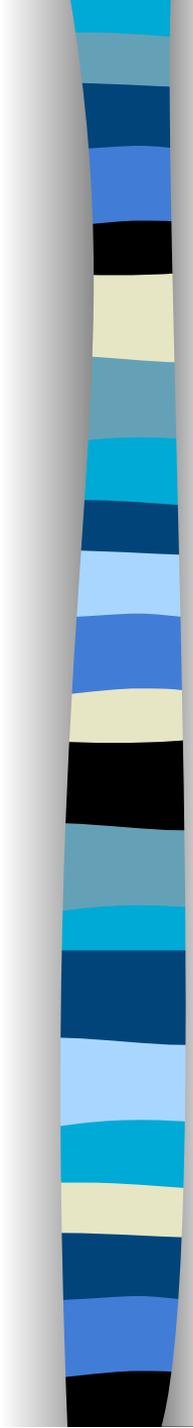
- Approximately 30% of parents of children with ASDs report a history of regression of language and/or social skills
- Most often occurring in the 15 to 24 month age range

(Kurita, 1985; Tuchman, 1991)



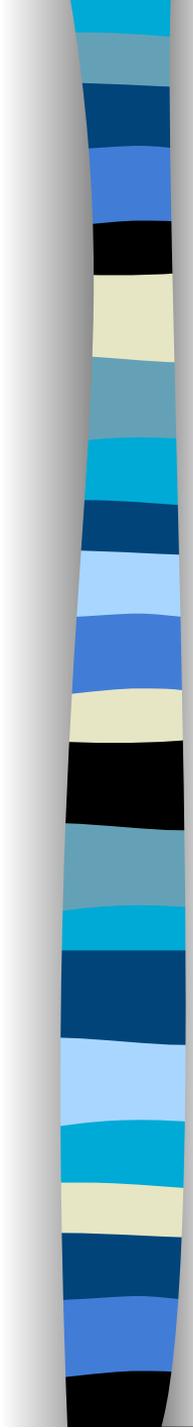
Osterling and Dawson, 1999

- Looked at phenomenon of late-onset autism by similar method
- Found increased orienting to name, eye contact in this group
- No difference in social/communication behaviors found when compared to normal controls



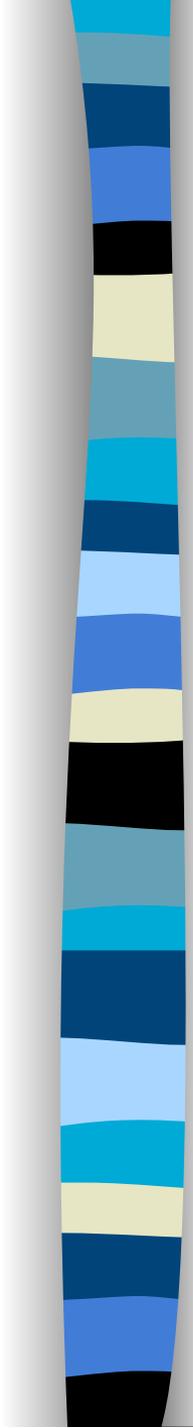
Seizures in Autism Spectrum Disorders

- The prevalence of seizures in ASD ranges from 11 – 39%
- An increased risk of seizures is associated with:
 - Severe cognitive impairment
 - Lack of language
 - A history of regression



Indications for Referral to Neurology

- History of developmental regression or stagnation
- Staring spells or involuntary movements suggestive of the possibility of seizures
- Focal neurologic examination: e.g. large or small head size, asymmetry of function or tone

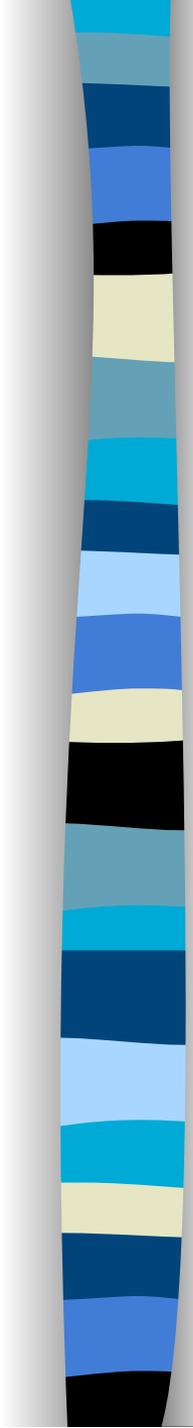


Medical Work-up

- Hearing test(s)
- Bloodwork-general Genetic studies (High resolution chromosomes or Microarray comparative genomic hybridization), Fragile X testing

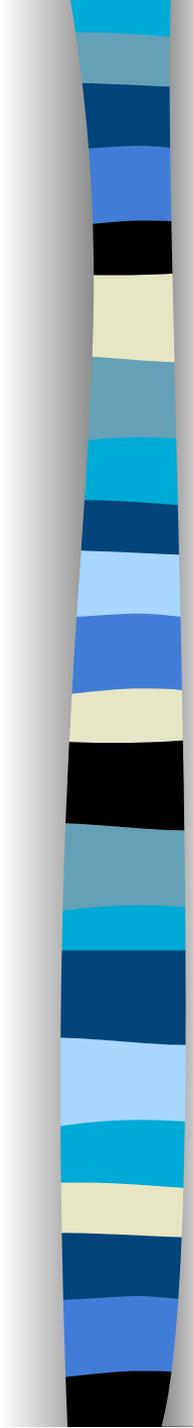
<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC1235307/>

- EEG (24 hours or more) if regression
- Brain imaging e.g. MRI for large/small head
- Targeted genetic studies for specific conditions based on abnormalities on history or exam



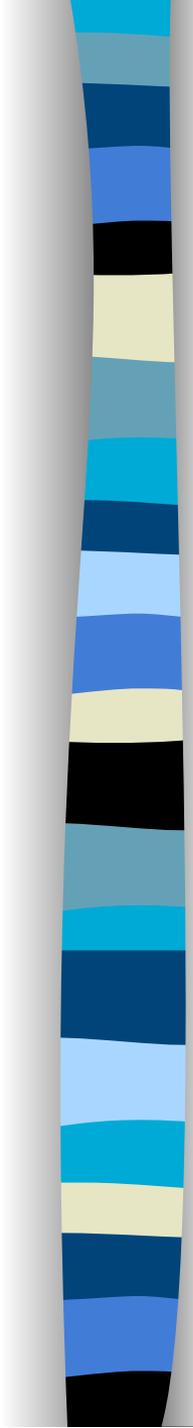
The Likelihood Of Finding Associated Medical Conditions Increases If:

- The handicap is more severe
- There are specific signs and symptoms of a particular condition
- There is a deteriorating course



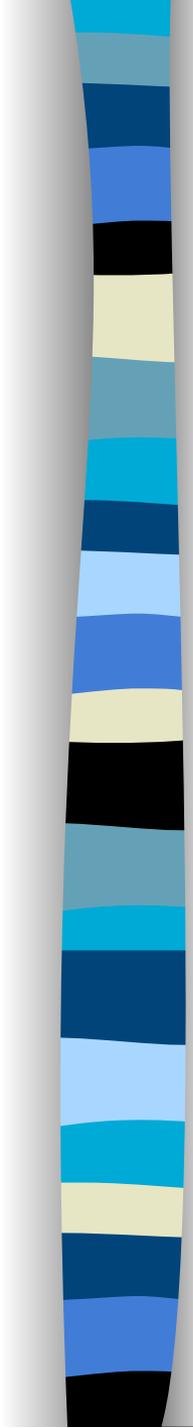
Growth and feeding issues in ASD

- The majority of children with ASD do not display physical features that set them apart from their peers
- A minority have large heads, and a smaller minority have small heads
- Height and weight are generally within normal limits



Feeding issues are common

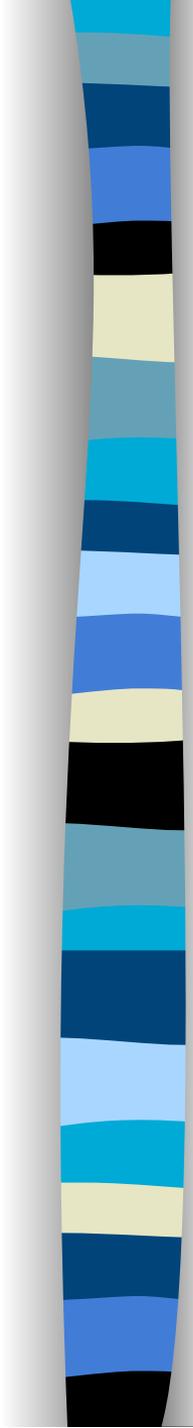
- Behavioral issues including difficulty tolerating texture, oral motor sensitivities
- Preference for certain textures or tastes
- Rigidity regarding preferred items/ brands and feeding routines
- Difficulty with self-soothing, extreme pickiness, oral-motor sensitivity may lead to reliance on bottle, immature feeding practices



Gastrointestinal symptoms

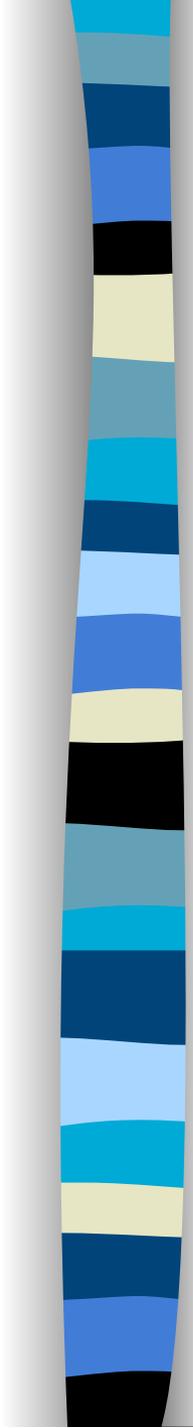
- Parents of children with ASD report more gastrointestinal symptoms than parents of children with other developmental problems
- These include: vomiting, diarrhea, constipation, abdominal pain, food selectivity, food allergies/intolerance

(Valicenti-McDermott, et al, 2006)



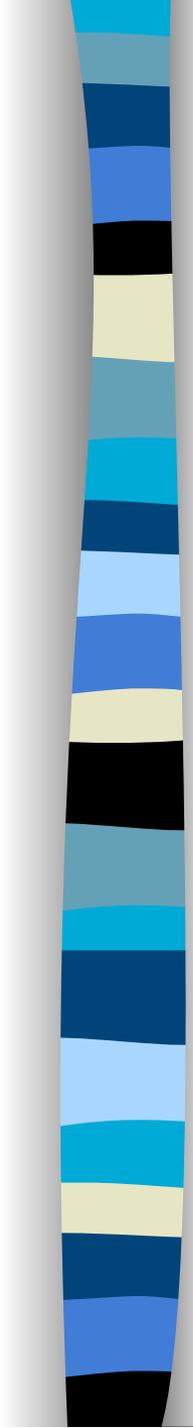
Specialized diets

- Parents of children with ASD reporting gastrointestinal symptoms are more likely to pursue specialized elimination diets
- Media portrayals and parent reports of improved symptoms on specialized diets lead parents to try diets in the hope of improvement. The most common is the gluten/casein free diet.



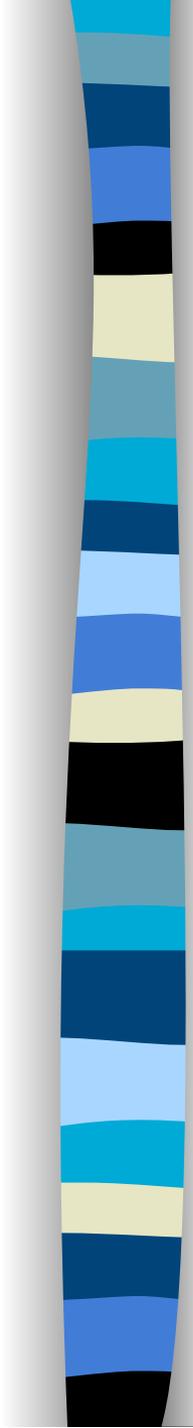
In counseling families of children with developmental issues....

- It is important to inquire about the use of specialized diets
- If children are on a casein-free diet it is important to determine the calcium and protein sources the child is receiving
- It is important to encourage the family to work with a nutritionist to make sure that **they implement a diet safely** (i.e. to reduce the risk of osteoporosis in children on long term gluten/casein-free)



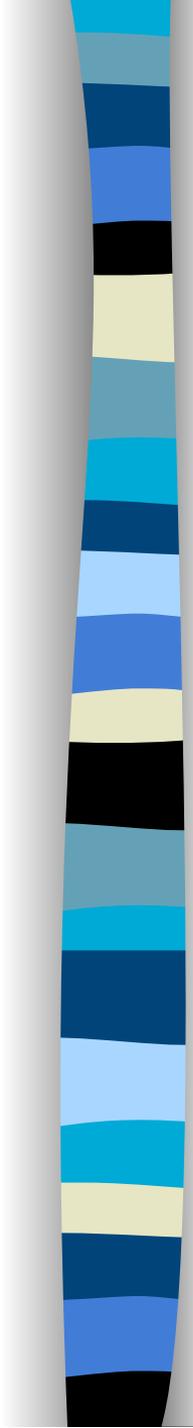
Controversies in Autism

- Non-evidence-based biomedical interventions
- Vaccinations
- Is there an autism epidemic?



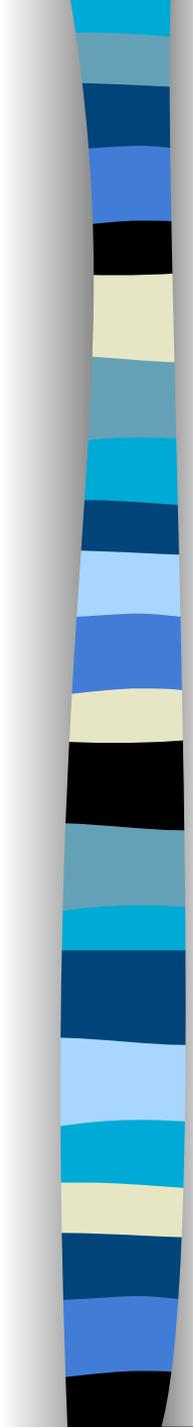
Tests Without Current Known Utility in Autism

- Allergy testing
- Yeast overgrowth
- Immune studies
- Trace elements
- Hair analysis
- Vitamin levels
- Celiac antibodies
- Intestinal permeability studies
- Stool analysis
- Urinary peptides
- Liver function tests
- Etc.



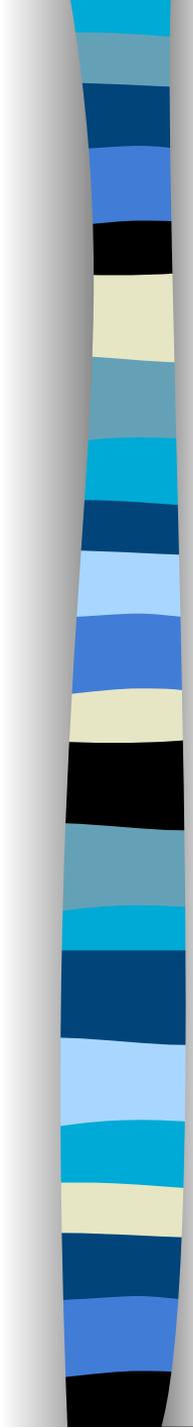
Unproven “Biological” Treatments

- Insufficient data meeting evidence-based standards
- Examples: vitamin B6, magnesium, dimethylglycine, casein/gluten restricted diets, Omega-3 fatty acids, chelation, hyperbaric oxygen
- It is important to be aware that over 50% of families of children with ASD are utilizing such treatments



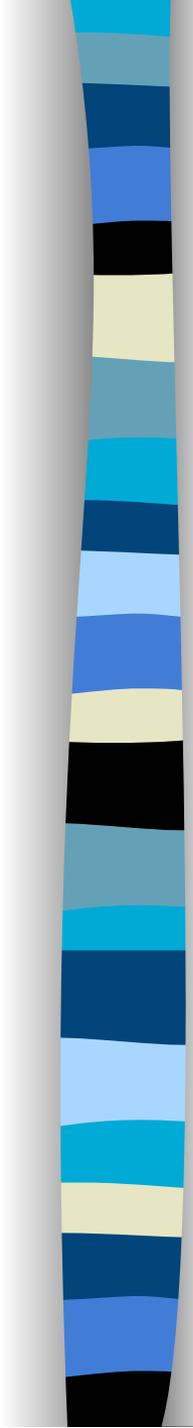
Vaccinations

- For the majority of children with ASD, the cause is unknown
- That is a ripe scenario for postulating potential causes
- In 1998, Andrew Wakefield, a British gastroenterologist claimed that he had found the cause of autism: the MMR vaccine
 - He reported data on 11 children with “regressive autism”
 - hypothesized a connection between the vaccine, the onset of autism symptoms, and a particular finding on endoscopy



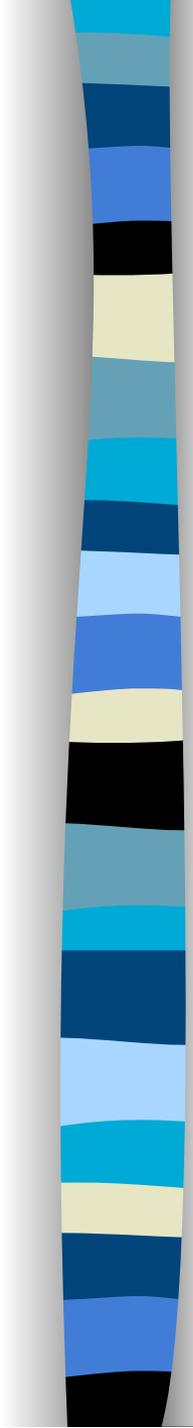
Vaccines (cont'd)

- Other researchers could not replicate these findings and it came to light that Wakefield had falsified data and committed assorted ethical violations
- In 2009, the Lancet retracted the published study and Wakefield's medical license was revoked
- But the damage was done and over the past decade the public's trust in vaccination has been eroded



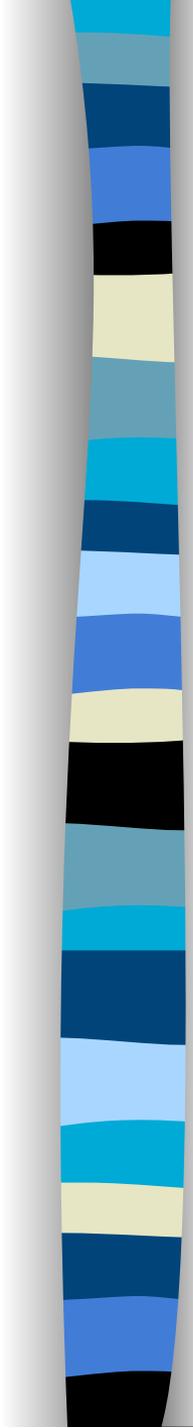
Vaccines (cont'd)

- Repeated studies do not support a relationship between MMR and autism
 - Repeated studies do not support a relationship between Thimerisol (a mercury-containing preservative used in many vaccines through 2001) and autism
- (Price et al, 2010)



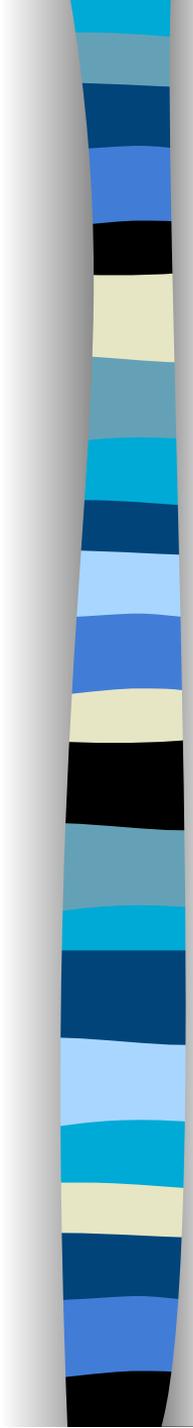
The Autism “Epidemic”

- From the mid-1960’s to the late 1980s, autism was considered a rare condition with a prevalence of 2-4/10,000 children
- Brick Township, NJ, 2001: 40/10,000
- CDC, 2003: 34/10,000
- Now, typical reported prevalence is 1/110 children



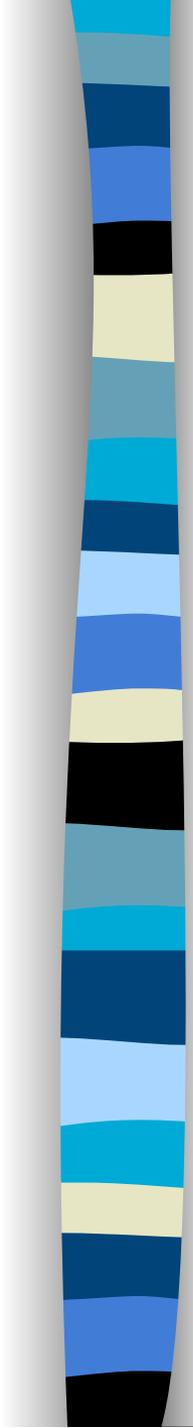
What could be responsible for this marked increase in identified cases of autism?

- Changes in diagnostic criteria to include more mildly affected children
- Diagnostic substitution for more severely affected children
- Greater public and professional awareness
- Wider availability of treatment services (the diagnosis opens the door to services and is thus desirable to parents)
- Increase in actual incidence of the disorder



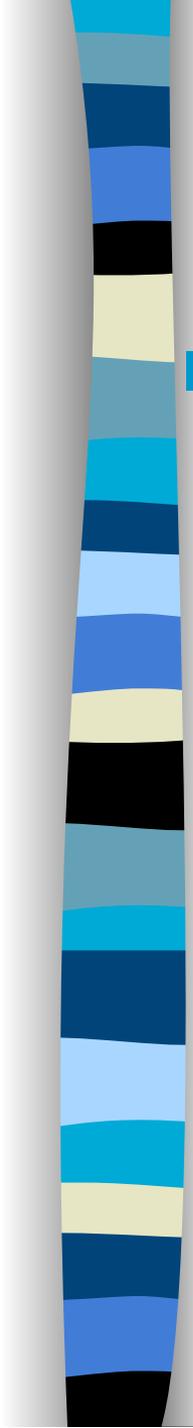
Back to Jonathan, Michael and David.....

- These 3 boys all made their way to an early diagnosis of ASD
 - Jonathan was referred by his parents
 - Michael was referred by his preschool
 - David was referred by a visiting nurse
- “It takes a village.....I mean—it takes a major metropolitan area..”



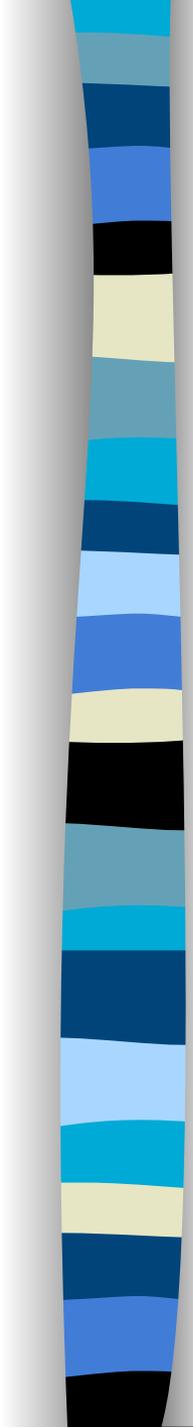
How can you help?

- Not suggesting that you screen children for ASD
- Rather, as professionals who have contact with families of young children, your awareness of the importance of early identification of autism and its early signs puts you in a position to serve as a potential resource to families
- Awareness enhances identification



How can you help? (cont'd)

- WIC's mission intersects nicely with CDC's "Learn the Signs. Act Early" campaign to improve early identification of ASD. The campaign seeks to teach parents to view a child's growth as not just physical, but as involving the attainment of developmental milestones indicative of developmental health. And you are in a unique position to disseminate that message.



Resources

- CDC “Learn the Signs. Act Early” campaign and materials:
www.cdc.gov/actearly 1-800-CDC-INFO
- American Academy of Pediatrics (AAP):
www.aap.org/healthtopics/autism
- Autism Speaks: www.autismspeaks.org
- Autism Society of America (ASA):
www.autism-society.org
- First Signs: www.firstsigns.org