

Autism Spectrum Disorder (ASD) Goals

1. Understand DSM-V criteria for ASD
2. Formulate plans to screen children for ASD
3. Construct treatment plans for children with ASD

ASD - 3 core attributes

- Impaired social interaction
- Language impairments
- Abnormal Behaviors

Prevalence rising (0.9%)

- A significant increase from 2007
 - true increase in disease?
 - increased societal awareness of ASDs?
 - changing diagnostic criteria?

CDC 2013: "Much of the prevalence increase for school-aged children was the result of diagnoses of children with previously unrecognized ASD"

Increased awareness?

- 70% with ASD had co-morbid MR in 1970s
- Only 30% have co-morbid MR in 2007 studies

Epidemiologic study in Calif concluded that early age of diagnosis & milder cases accounted for > 2/3rds of increase

Gernsbacher 2005; Fombonne 2006; Shattuck 2006; Taylor 2006; Allardotir 2007; Hertz-Picciotto 2009

Genetic Etiology ?

- Higher incidence (x10) among ASD siblings
- Genetic basis likely mirrors that of MR
 - Many syndromes, each individually genotypically rare, but phenotypically consistent with autism

NEJM 2.23.2012; NIMH/Autism-progress

Parent Age?

- Fathers aged > 50 vs < 30
 - 2.2 x more likely to have a child with ASD
- Grandfathers
 - Men who fostered a child after 50 (vs those 20-25) were 1.7 x more likely to have a grandchild with ASD

Mol Psych. 2011;16(12):1203;
JAMAPSYCH.COM;3.20.2013

Teratogens ?

- Environmental exposures
 - Unclear role, may interface with autism genes
- Some cases traced to specific exposures
 - In utero exposures to valproic acid associated with a 5-7x increased risk
 - Thalidomide & misoprostol recognized causes
- Maternal smoking

Bromley et al 2008; JAMA 2013

Rx

SSRI's?

- Single population-based, case-controlled study
 - 298 children with ASD/1507 controls
- 1 antidepressant during pregnancy
 - 3.3% (50) of controls
 - 6.3% (20) of mothers with ASD child
- Mothers with ASD child were significantly older, more educated and white than controls
- Prescription use not confirmed (from med rec review)
- Tobacco, ETOH and drug use not controlled for...

JAMA Psychiatry. 2013.

Vaccines ?

- Epidemiologic studies have not demonstrated an association between autism and exposure to *thimerosal* (which contains mercury)
- Nor the measles, mumps, and rubella vaccine
 - which never contained thimerosal

Vaccines.

- Original work by Wakefield et al
 - Anecdotal study of 12 autistic patients reporting a suspicion by their physicians about MMR
- The Lancet retracted that work in 2004!*
Accused of research fraud in 2011!

IOM 2001; Jansen 2003; DeStefano 2004; IOM 2004; Honda 2005; Taylor 2006; Schechter 2008; Offit 2008; JPeds 2013

2 y.o. Brandon....

- Brandon's Dad brings him in for a WCC and he requests a screen for Aspergers as Brandon's 6 y.o. cousin was recently diagnosed with that syndrome.....



What defines
Aspergers
syndrome?

Persuasive Developmental Disorders (DSM-IV)

Autism spectrum disorders

- Autistic disorder
- Asperger syndrome
- Pervasive developmental disorder, not otherwise specified

Other

- Rett's syndrome
- Childhood disintegrative disorder

Autism Spectrum Disorders (DSM-V)

A continuum from mild to severe, rather than specific disorders.....

Four Criteria Must be Met

- A. Deficits in social communication/interactions
- B. Restricted, repetitive patterns of behavior or activities that are abnormal in intensity or focus
- C. Symptoms present in early childhood
- D. Symptoms limit and impair everyday functioning

Severity Graded as well

Level 1 (least severe affect)

- A. Without supports, social communication deficits cause noticeable impairments. Difficulty initiating social interactions, atypical responses to social overtures.
- B. Rituals and repetitive behaviors cause significant interference with functioning. Resists attempts by others to be redirected from fixated interest.

ASDs - 3 core attributes

- Impaired social interaction
- Language impairments
- Abnormal Behaviors

Lack of social skills..

The earliest & specific sign

- Joint Attention
- Social Orienting
- Pretend Play

Social Interactions/*Joint Attention*

- The desire to share experiences with others
- 8 months follow a parents gaze when looking
- 10-12 months will follow pointing
- 12-14 months will point at things (a request)
- Receptive – smile when recognizing parent
- Concern if ignoring parent attempt to connect or poor eye contact

Social Interactions/*Orienting*

- A 12-mo will turn in response to hearing name
 - Parents may wonder about hearing
 - Hearing seems to be more attentive to environmental noises, not to voice

Social Interactions/*Pretend Play*

- An 18-month-old child will normally speak 'baby talk' into a parent's cell phone
 - a child with autism may just push the buttons repeatedly

Language Impairment..

- Delayed or odd language common
 - Less specific early sign
 - Diminished intrinsic drive to communicate

Language/Communication concerns

- At 6 months infants should babble
- By 9 months should speak jargon
- Speech delays at 18 to 24 months



Red Flags (*Am Acad Neurology*)

- No babbling, pointing by a year
- No single word by 16 months
- Lack of 2 spontaneous word by 2 years
- Any loss of language or social skills at any age

Abnormal Behaviors

1. Stereotyped or repetitive speech, movements, or use of objects
2. Excessive adherence to routines, ritualized patterns of verbal or nonverbal behavior
3. Highly restricted, fixated interests that are abnormal in intensity
4. Hyper- or hypo-reactivity to sensory input or unusual interest in sensory aspects of environment

Behaviors (less prominent)

- Stereotypic movements
 - flapping; rocking; twirling
- Difficulty with changes in routine
 - obsessive
- Repetitive use of objects

C. Symptoms present in early childhood

-but may not become fully manifest until social demands exceed limited capacities.
- Interpret speech literally
 - no understanding of idioms, jokes or lying

12 month old Danny....

- Danny's mom is concerned about Autism because although he is walking ok and feeding himself finger foods, he 'babbles a lot and only says 'Mama'; additionally he cries very easily around strangers.....

Are you concerned?
How do you screen
for Autism in your
office?

Screening

- Whenever a concern is raised
- AAP recommends screening 9, 18, 24, 30 mos
- No validated tools for < 16 months
- M-CHAT
 - Validated for 16-30 months age

M-CHAT - a good office screen

- Free: www.firstsigns.org
- A high false-positive rate
 - 85% sens/93% spec
 - PPV only about 60%

J of Autism and Developmental Disorders, 31 (2). 2001

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 dolls, 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 bricks) 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 talk?	Yes	No
18.	Does your child play independently?	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
Robins, Fein, & Barton

**Positive screen if ≥ 2 critical questions
or \geq of any 3 questions are failed**

2 y.o. Timothy

- Timothy has just failed the MCHAT at his 2 y.o. WCC



What do
you do
now??

Formal developmental evaluation

- Utilize standardized evaluation
 - Early Intervention Developmental Profile ("Michigan")
 - Battelle Developmental Inventory-2 ("BDI-2")
- A multidisciplinary autism team
 - Developmental pediatrician, audiologist, OT, psych, social worker, speech pathologist

***Learn about your community
referral sources!***

Early Intervention....

- Community programs for children <3
- Special education department of the local school district for children ≥ 3

Treatment - Behavioral Therapy

- Intensive early therapy improves cognitive, language, adaptive skills
- Sensory Tactile therapy
 - Applied Behavioral Analysis
 - www.centerforautism.com
 - Early Start Denver Program
 - SJ Rogers and G Dawson
 - TEACCH
 - www.teacch.com

AAP; Pediatrics. 2007; J Am Acad Child and Adols Psych; 2012

Medications

- Risperidone FDA approved for irritability and SIB in children with ASDs
- Fluvoxamine, fluoxetine for repetitive behaviors
- Methylphenidate for impulsivity, inattention
- Clonidine, guanfacine (centrally acting agonist) for impulsivity, outbursts, hyper-arousal

Pediatrics 9/2012; AFP 5/2012

Pets?

- Having a pet after the age of 5 demonstrated improvement in offering to share and offering comfort.....

PLoS One 2012

CAM??? (Mixed results...)

- Detoxification/chelation
- Hyperbaric O₂
- IV immune globulin
 - expensive and not recommended

Supplements ?

- No consistently proven benefit from
 - B6
 - B12
 - Omega-3s
 - Magnesium
 - Dimethylglycine
 - Secretin
 - Carnitine

Dietary restrictions ?

- Gluten-free, casein-free diet
- A randomized controlled study (35 patients) 16 outcomes measured
 - Improved ability to communicate /interact socially
 - No improvement in other measures

Poor prognosis if....

- Regression (language or other development)
 - Usually between 15-24 months age (25%)
 - Can be gradual or sudden
- Lack of social interaction by age 4
- Lack of speech by 5



Many children do improve

- Developmental gains are common
- A 2008 review of Outcome Studies concluded that between 3-25% of most cohorts lose the diagnosis
- Some have behavioral regression during adolescence



JCPP 2013;54(2):195;Neuropsych Rev 2008;18

Resources

- Parents:
 - Autism-pdd.net
 - Autism-society.org
 - [Autism speaks.org](http://Autism-speaks.org)
 - 'First 100 Days Kit' can help families arrange and advocate for effective early treatment
- Providers
 - cdc.gov/ncbddd/actearly/hcp/index.html
 - www.firstsigns.org

Summary - 3 core attributes

- Impaired social interaction
- Language impairments
- Abnormal Behaviors

Summary

- Screen all at 18 & 24-month office visits.
- Refer for early intensive behavioral therapy to improve cognitive, language & adaptive skills.
- Treat associated medical and psychiatric conditions to maximize overall functioning.