RED FLAGS

EARLY SCREENING AND DIAGNOSIS OF AUTISM

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DISCLOSURES
NONE
AUTISM 2014

• 1/42 boys and 1/189 girls

• Lifetime cost of ASD is up to 3.2 million

• US cost burden is 76 to 130 billion lifetime in birth cohort

• Early intervention may save 2/3rd of the cost

• Emotional cost as well

WHAT IS AUTISM?

Neurodevelopmental disorder with the triad of issues in communication, interaction and interests

COMMUNICATION

• Absent/delayed/disordered language without an attempt to compensate nonverbally

• Inconsistent use of words or regression

• Stereotypical or repetitive language- Examples: script usage, echolalia-not developmentally based.

SOCIAL REFERENCING

Difficulty in recognition/response of emotional significance with stimuli/emotions

Difficulty in orienting to social stimuli-ex response to name

Poor understanding of and responding to feelings of others-“theory of mind”

Decreased imitation of social behaviors

Reciprocity-back and forth of a relationship
REPEETITIVE/RESTRICTED INTERESTS

Repetitive motor movements – hand flapping, spinning, pacing etc.
Intense interests-ex letters/numbers, open close doors, lights etc
Intense interest in visual items-videos, toys with light, sound etc.
Patterns of play- repetitive themes and no pretend play.
Extreme distress with small changes

CAUSES OF AUTISM/S

- Genetics
- Risks

AUTISM/S

GENETICS/ BRAIN/ AUTISM

- Hundreds of contributing copy number variants, hundreds to thousands of pieces of genes contributing to risk
- ASD may be associated with other disorders. Example is Down Syndrome
- Evidence that genetics will give us concept of different phenotypes for autism and help clinically
- In the future, there may be specific targeted treatment medically. Ex Fragile X

RISK ASSOCIATIONS

- Older age in mothers and fathers as well as paternal/paternal grandparents
- Low folate level in mother conception/ early pregnancy, gestational diabetes, obesity…..
- Medications - Depakote (during pregnancy)…
- Extreme prematurity…..
- Environmental- pollutants, fire retardants etc..
POSSIBLE PRENATAL ORIGIN

- Changes in brains of 10 of 11 children with autism diagnosis. Similar changes were found in only 1 of 11 unaffected children.
- Patches of abnormal anatomy in parts of the brain (neocortex formed prenatally at 19 to 30 months) associated with social and communication function.
- If replicated, suggest that brain changes in autism are likely to have originated before birth.


WHY SCREEN EARLY FOR ASD?

- Common disorder 1/68 children.
- No lab test or pathognomonic sign.
- Implications for family planning-family with child with autism-3 to 20% risk of having another child with a similar disorder.
- Parent concern-18 months with usual mean age of dx-3-4 years.

EARLY INTERVENTION WORKS

Early diagnosis allows for earlier intensive intervention.
Evidence that autism cannot be cured but can have "optimal outcome."
Optimal outcome means that there are no significant functional impairments.
Easier to "rewire" brain connections when very young.

Researchers say intervention in early childhood may help the developing brain compensate by rewiring to work around the trouble spots.
WHAT DO RED FLAGS MEAN?
Red flags do not always mean a child has or will develop ASD
May have overall developmental delay
May have regulatory issues
May have communication issues
May have underlying chronic medical issues - all types including prematurity
May be normal
May have combination of above

WHAT ARE RED FLAGS?
Possible warning sign of autism in early stages.
Determined as a result of prospective studies of younger sibs of children with autism who are at higher risk of developing autism
Some of the signs can occur in normal development
Some of the signs may be related to conditions other than autism. EX developmental delay
They are related to the triad of autism issues

RED FLAGS FOR ASD
No big smile or joyful expression by 5 months
No back and forth sharing of sounds, smiles or facial expressions by 9 months or later
No babbling by 12 months
3 point gaze
No back and forth gestures, such as pointing, showing, reaching or waving by 12 months. Also eye contact.

FURTHER RED FLAGS
No words by 16 mos.
No 2 word meaningful phrases (without prompt or repeat) by 24 mos.
Any loss of speech or babbling or social skills at any age.
No back and forth sharing of sounds, smiles or facial expressions by 9 mos or later
NON VERBAL COMMUNICATION

NO BIG SMILE OR JOYFUL EXPRESSION BY 5 MONTHS

NO BACK AND FORTH SHARING OF SOUNDS, SMILES OR FACIAL EXPRESSIONS BY 9 MONTHS OR LATER

POOR IMITATION OF GESTURES
ALL KINDS OF POINTS

POINTERING TO SHOW AND TO GET & 3 POINT GAZE

RECIPROCITY

POINTING
CONCERNS BUT NOT DIAGNOSTIC IN ISOLATION

RED FLAGS, THEN WHAT

- Contact pediatrician!!
- Routine developmental surveillance is done at each visit in early childhood by pediatricians
- M CHAT R is done at 18 and 24 months - screen for autism
- MCHAT is also online as well as interactive screen from Autism Speaks
- If screen is concerning, refer for full developmental/ autism evaluation

EYE GAZE PATTERNS

Specialized research scans for a baby later diagnosed with autism (red) and a typically developing control (blue).

HEAD LAG ON PULL TO SIT

This test is recommended for babies over the age of 6 months with a family history of autism.

Place the baby on his back on the floor.

Kneel over the baby and make eye contact.

Gently tug on the baby's arm to give him a cue.

Firmly but gently pull the baby up by his arms to a sitting position.

If the baby is distracted, be sure to get his full attention before beginning the test.

The baby's head should follow his torso up and not lag backward. If the head appears to lag, be sure it's not just that the baby is trying to keep an eye on something behind him.

If the baby continues to exhibit head lag after doing the test a couple of times, have a pediatrician examine the baby for other signs of developmental delay.
EVALUATION FOR SOME RED FLAGS BUT PASS AUTISM SCREEN

Determine if symptoms are part of normal development. Ex. repetitive behaviors
Determine if there are physiologic issues causing symptoms. Ex. Premature babies may have gaze aversion
Further workup depends on symptoms and possible etiology of symptoms. Ex sleep apnea etc

EVALUATION FOR AUTISM SCREEN NON PASS

Refer for formal diagnostic evaluation by experienced clinician in development and autism
Formal audiological evaluation
Other testing may include: lead screens, thyroid tests, sleep apnea study, blood tests for metabolic errors, neurological evaluation especially with regression, Genetics evaluation or blood tests for Fragile X and chromosome micro array.

DIAGNOSIS

- Still no pathognomonic test
- Standard of care evaluation involves:
  - School Questionnaire (if appropriate)
  - Parent Questionnaire
  - Developmental Testing
  - Autism Diagnostic Observation Schedule
  - Play Observation
  - Valid at 18 months with ADOS
  - Also use home video clips

AUTISM INTERVENTIONS

- There is no one specific intervention that is considered the only absolutely proven method. ABA has statistical research and there is much proof of the efficacy in young children
- Research is ongoing in this area and is verifying efficacy especially early
- Variables- characteristics of a child, family, resources and access to services
INTERVENTION FOR ASD

Time is of the essence for early intervention

• Behaviors are less likely to become firmly established if there is early intervention
• Blooming and Pruning in brain for new pathways
• Parents are grieving re diagnosis and trying to access services at same time
• IT WORKS

AUTISM INTERVENTIONS
Programs, Agencies, Access, Logistics

• Programs- ABA(Denver Early Start, Pivotal response, discrete trial), speech therapy (Hanen, direct, group), OT, social groups, etc.
• Agencies- GGRC, school district, medical insurance, etc.
• Logistics- hours of therapy, parent work, family, finances, support
• Access- limited resources, many "hoops to go through"

PARENT SUPPORT

• Parent support is crucial-refer to Family Resource Center
• Help parents navigate the maze of systems, help each other
• Parents are grieving re the diagnosis but also re possible implications for future children
• Parents very vulnerable since there is no specific test for the diagnosis, no specific intervention absolutely proven to help their child.
• May be eligible for FMLA Family medical leave for work
• Medical home

AUTISM RESOURCES

Autism A.L.A.R.M. handout-AAP/CDC/NCHDD.
IAN site- Johns Hopkins- great reference for valid research
Autism Speaks Interactive quiz for red flags
Thinking Person Guide to Autism
10 Things Every Child with Autism Wishes You Knew-book updated 2012 Notbohn Center for Disease Control-Learn the signs Act Early