Autism Spectrum Disorders

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Objectives

- To address the most common questions asked at autism evaluations
- To answer questions you may have
- Provide my contact information for future questions or consults as needed

Most Common Questions

- 1) Is autism new?
- 2)What is autism?
- 3) How do you diagnose a child with autism?
- 4)What questions do I ask parents if they are concerned about autism?
- 5)When should I be concerned or need to refer for more evaluation?

Most Common Questions

- 6) Why do we see so much Asperger’s children in the Treasure Valley?
- 7) How do I screen these kids quickly?
- 8)Why is autism increasing?
- 9) How often do you find a cause for autism?
- 10) What is the typical workup?
- 11) What role do vaccines and mercury play in autism?

Most Common Questions

- 12) What is the prognosis?
- 13) Is there a cure? Treatments?

Facts and Statistics

- 1% of population of children in US age 3-17 have autism (2-6/1,000 children)
- Prevalence estimated 1 in 110 births
- 4 million US births/yr (24,000 will be dx autism)
- More common than childhood cancer(1.5/10,000)
- 1- 1.5 million Americans have Autism
- Fastest growing developmental disability 1,148% growth rate
- 10-17% annual growth
- $60 billion annual costs

Facts and Statistics

- 60% costs in adult services
- Cost of lifetime services can be reduced 2/3 with Early Intervention Services
- In 10 years , annual costs will be $200-400 billion

Autism Myths

- 1) All autistic children are alike.
- Not all autistic people are like rain man. It is a spectrum. The only thing they all seem to have in common is the their difficulty with social communication.
- 2)Autistic children don't have feelings.
- Most are highly empathic and capable of expressing love…though often in idiosyncratic ways.

Autism Myths

- 3) Autistic children can’t build relationships.
- Yes they may not be the cheerleader but they often are able to have solid relationships at the least with their closest family members.
- 4) Autistic people are a danger to society.
- While they may show aggression it is often out of frustration or sensory overload not out of malice

Autism Myths

- 5) All autistic persons are savants.
- While some do have “splinter skills” the majority do not.
- 6) Autistic people have no language skills.
- Children with classic autism can be non-verbal or close to non-verbal but autism is a spectrum. Some are very verbal and even have hyperlexia.

Autism Myths

- 7) Autistic people can’t do or achieve most of anything
- This is the most damaging of the myths.
- Often they are the creative innovators in our midst.
- They see the world through a different lens and if respected can sometimes change the world

Is Autism New?

Famous People with Autism

- Mozart and Bach- Composer
- Charles Darwin-Naturalist/Evolutionist
- Emily Dickenson-Poet
- Albert Einstein- Physicist
- Thomas Edison-Inventor
- Thomas Jefferson-President
- James Joyce- Writer Ulysses
- Mark Twain-Writer
- Michelangelo- Artist
- Isaac Newton-Scientist
- Van Gogh-Artist
- Andy Warhol-Artist
- George Orwell- Writer
- WB Yeats-Poet
- Stanley Kubrick-Film Maker
- Bill Gates- Microsoft Billionaire

Famous People with Autism

Albert Einstein

Asperger’s syndrome

What is Autism?

Autism Definition

Autism is a neurodevelopmental disorder with a strong genetic basis but of unknown etiology.

- It is not a disease.
- It is a disorder characterized by a cluster of behavioral and neurodevelopmental symptoms rather than any physical or lab abnormality.
- Onset occurs before age three.

Triad of Abnormal Behaviors define Autism

- 1) Qualitative impairment of social relatedness significantly below child's level of developmental functioning.
- 2) Qualitative impairment in communication characterized by the lack of desire to communicate(pragmatic deficit- dissociation between form and function of language)
- 3) Restrictive, repetitive, stereotypic patterns of behavior, interests, and activities(may occur after 3 years old)

There is no pathognmonic physical sign, biologic test, developmental profile or behavior phenotype that entirely discriminates autism from other disorders.

Most distinguishing characteristic of lack of joint attention and mind-blindness.

"Lack of capacity to share attention& emotion with others is specifically and universally impaired." (Bristol 1996)

Historical Perspective

- 1943 Kanner describes 12 kids with autism.
- “Refrigerator Moms”- coined by Kanner. Term to describe a parent who was cold and uncaring cased trauma to child that they became autistic. This idea persisted until the 1960s!
- 1944 Asperger describes Asperger syndrome.
- DSM-1(1952) Autism included in “schizophrenia-childhood type”
- DSM-11(1968) Childhood schizophrenia-psychotic reaction in childhood manifesting primarily as autism.

Dr. Leo Kanner

- “Autistic Disturbance of the Affective Child” in Journal of the Nervous Child (1943)
- N= 12(9 males/3 females)
- Description: “He seems to be self satisfied. He has no apparent affection when petted. He does not observe when anyone comes or goes. Never seems glad to see mother, father, or any playmate. He seems to draw into his shell and live within himself.”
- Autism from greek “auto” meaning self. Term autism had been used before in describing schizophrenia. Felt until 1970’s they were the same disorder.

Hans Asperger

Hans Asperger, an Austrian pediatrician who only published in German was working at the same time period as Dr. Kanner on opposite sides of the Atlantic. He worked with children who although bright spoke in one sided lecturing fashion like "little professors" on topics in which they had an absorbing interest. The children were socially unaware with little ability to form friendships or read others emotional states. They had difficulty with non-verbal communication, prone to behavioral problems, and appeared clumsy.

His work remained unknown until 1991 when it was translated into English. In 1994 Asperger Syndrome label in DSM IV.

Hans Asperger with Young Boy

Historical Perspective

- DSM III (1980)-No longer viewed as same disorder as schizophrenia. Three types:
- Infantile autism
- Childhood Onset PDD- more flexible and inclusive
- Atypical Onset PDD-sub-threshold condition for those who failed to meet criteria.

Historical Perspective

- DSM-IIIR(1987)significant changes.
- British psychiatrist Wing took a broader view and coined term "Autism Spectrum Disorder"(he had a autistic child)
- 16 criteria- 8 needed for diagnosis(2-1-1)
- At least two impaired social interaction
- At least one impaired communication
- At least one restricted behavior and interests
- Autism disorder replaced Infantile Autism
- PDD-NOS –sub-threshold condition replaced atypical autism.
- No age requirement for initial onset of symptoms

How do you make the diagnosis of autism?

Clinical Diagnosis of Classical Autism (DSM-IV) 1994

- 1. Socialization (4 items)
- 2. Communication (4 items)
- 3. Behavioral/ Interests (4 items)

- Needs six or more items with:
- At least two from 1
- At least one from 2
- At least one from 3

1. Socialization (need 2)

- ! Non-verbal communication
- ! Peer relationships
- ! Sharing interests
- ! Social reciprocity

2. Communication (need 1)

- ! Spoken language
- ! Sustained conversation
- Idiosyncratic language
- ! Make believe play

Behaviors/Interests (need 1)

- Restricted interests
- Preoccupation with parts
- Inflexible routines
- Stereotypical Behaviors

Clinical Diagnosis of Autism (DSM-IV)

- B. Delay or dysfunction before age 3
- C. Not better accounted for by Childhood Disintegrative Disorder or Rett Syndrome

Historical Perspective

- DSM-IV(1994) Age criterion back to 3 years
- Autism one of five Pervasive Developmental Disorders:
- 1)Autistic Disorder
- 2)PDD-NOS- some but not all criteria met for autistic disorder
- 3)Asperger’s Disorder
- 4)Rett Syndrome
- 5)Childhood Disintegrative Disorder
- DSM-IV TR(2000) Autism spectrum Disorder
- ASD is Autism + PDD-NOS+ Asperger’s

PDD Umbrella
DSM-IV(1994)

Autistic disorder (classic autism)	Onset by age 3 years impairments in social interaction and communication, repetitive behaviors, and inability to engage in imaginative play; in some cases, severe regression in social interaction and language skills between 18 and 24 mo
Asperger's syndrome	Characteristics similar to classic autism; exceptions include no delays in language skills or cognitive skills; individuals have pragmatic deficits; sometimes referred to as "high-functioning autistic"
Pervasive developmental disorder–not otherwise specified	No specific autistic signs but severe and pervasive impairments in specified behaviors
Rett syndrome	A neurologic degenerative condition that affects only females; a period of normal development followed by loss of previously acquired skills; usually diagnosed around 18 mo; patients begin to exhibit reduced muscle tone (hypotonia); gene discovered in 1999
Childhood disintegrative disorder	Marked regression after 2 years of normal growth; regression may occur in motor skills, social skills, language, and bowel and bladder control; typically occurs between ages 3 and 4 years but

Rett Syndrome

Normal development and HC in first 5 months with onset deceleration in head growth and loss language and social skills between 5-30 months. Most notably loss of purposeful hand skills and stereotypic handwringing. Mutation of MECP2 gene on X chromosome >80% females. Now rare males identified.

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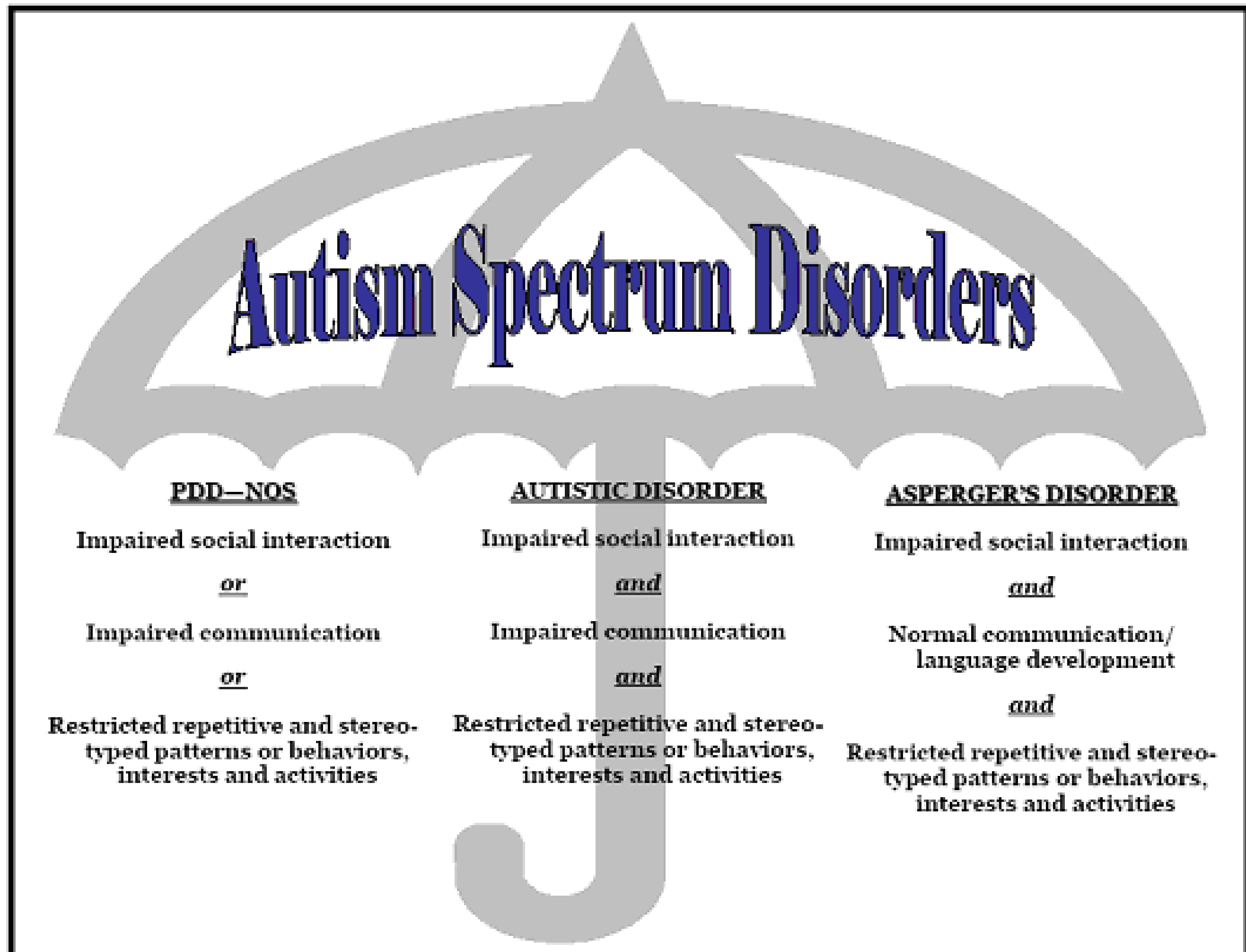
CDD

These individuals present with behavioral features seen in autistic disorder but critical difference is delayed onset after 2 years NORMAL development. Very rare, strong male preponderance. Global regression including loss bowel and bladder control. Most develop seizures and have very low IQ. DDx: Mitochondrial Disorders

ASD
DSM-IV-TR (2000)



Disorder

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. Qualitative impairment in social interaction, as manifested by at least two of the following:
      a. Marked impairment in the 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 a 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 the 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 the 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

B. Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years: (1) social interaction, (2) language as used in social communication, or (3) symbolic or imaginative play.

C. The disturbance is not better accounted for by Rett's Disorder or Childhood Disintegrative Disorder.

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What questions do I ask in evaluating a child for possible autism?

Assessing For Classic Autism: Social Interaction

- Does your child:
- 1)Pay attention or look toward you when you speak or enter the room? Eye contact
- 2)Enjoy cuddling like other children?
- 3)Enjoy playing imitation games(peek a boo)?
- 4)Show interest in other children his or her age?

Assessing for Autism: Communication

- Does your child:
- 1. Point to items of interest that he/she wants?
- 2.Respond yes or no with a head nod or shake?
- 3.Lead you by the hand to a desired activity or place?
- 4. Give consistent responses when he/she hears their name or simple commands?

Development of Social Communication

- 1. Under three months of age
- The child develops “eye contact” with the caregiver along with an exchange of voice or facial expressions (coo and social smile) in the form of rhythmic turn taking
- Autistic children have a deficit in eye contact- stare through you, look at mouth instead of eyes, or no eye contact at all.

Development of Social Communication

- 2. Over nine months- Joint attention
- An activity in which both child and caregiver look at the same target and share information through pointing and gazing.
- Example: A one year old looks at her favorite snack on a high countertop. Knowing that Mom is in the room with her she points at the goodies and turns to look at her mother. The child alternates her gaze between the snack and her mother until she knows they have ”connected.”
- Triad of actions: child focusing on something, pointing, and looking back at parent.
- Lack of reaction to pointing(joint attention) may be a sign of autism.

Assessing for autism: Behavior

- Does your child:
- 1. Have repetitive behavior that you find unusual or odd?
- 2. Show any interest in pretend play?
- 3.Uses toys in appropriate imaginative ways(such as making motor noises when rolling a car or train, using dolls or action figures to act out scenes ,etc.)?
- 4. Show especially strong attachment to a specific object and if removed suffers meltdowns?
- 5. Shows preoccupation with a few select items or interests?

Autism: Age of Recognition

FIRST YEAR	25%
1-2 YEARS	50%
BEYOND 2 YEARS	25%

MUCH TOO LATE!!!

RECOGNIZING EARLIER IS THE KEY!!

FIRST BIRTHDAY PARTY

When does autism start?

Dr. Susan Bryson- Canadian researcher

Preliminary data of 6 month olds children who were later diagnosed with autism generally show good eye contact but are quieter and more passive than matched peers. They may lag behind motor development in sitting up and reaching for objects.

Her research shows are signs autism more obvious by first birthday.

Extreme reactivity-overexcited or barely notice at all environment

Repetitive behaviors- rocking back and forth or fixating on an object (candle)

Less responsive to playful interactions of others

Typical 1 yo: Face lights up, makes eye contact, makes sounds/laughs

Autistic- little facial expression, little/no eye contact, takes enormous energy to get a response

When do I need to refer a child?

Absolute Indication for Immediate Evaluation for Autism

- By 6 months- no big smiles or warm joyful expressions
- By 9 months- no reciprocal sharing of smiles, sounds, or facial expressions
- By 12 months- lack response to name
- By 12 months- no babbling(baby talk)
- By 12 months- no back and forth gestures- pointing , showing , reaching , waving
- By 16 months- no spoken words
- By 24 months-no two word spontaneous(not echolalia) phrases
- Any loss of language or social skills at any age

Other “red flags” Autism

- Poor eye contact
- Does not respond to name
- Lack interactive play(indifferent to other kids)
- Lack joint attention
- “very good baby” “never cries”
- Not know how to play with toys
- Spins objects or self
- Stereotypies(hand flapping)

Why do we see so much Asperger's in the Treasure Valley?

Asperger's Syndrome

Asperger's Syndrome

Children and adults with Asperger syndrome might:

- Have trouble understanding other people's feelings or talking about their own feelings.
- Have a hard time understanding body language.
- Avoid eye contact.
- Want to be alone; or want to interact, but not know how.
- Have narrow, sometimes obsessive, interests.
- Talk only about themselves and their interests.
- Speak in unusual ways or with an odd tone of voice.
- Have a hard time making friends.
- Seem nervous in large social groups.
- Be clumsy or awkward.
- Have rituals that they refuse to change, such as a very rigid bedtime routine.
- Develop odd or repetitive movements.
- Have unusual sensory reactions.

Source: Centers for Disease Control

Classic Autism vs. Asperger's

- Speech delays
- Failure to make friendships
- Often lower IQ

- No clinically significant delay in language
- Difficulty making friends- want friends just don't know how
- Normal to high IQ
- "little professors" or "odd genius"

Asperger's

Engineers
Artists
Doctors
Lawyers
Accountants
Writers
Computer Programmers
Professors

Highly genetic-
Often see many in a family!

Very verbal but poor pragmatic language
Can't read social cues
Want to be alone or make friends but don't know how

How do you screen for autism quickly in a busy practice?

Identifying Infants and Young Children in Developmental Disorders

- Developmental Surveillance- every well child visit
- Developmental screening- 9, 18, 24 months at a minimum
- Ages and Stages Questionaire (ASQ)
- Screening with autism specific tool (M-CHAT) at 18 months and 2 years (AAP,2001)
- Early diagnosis of autism immensely improves outcomes!!
- ASQ and M-Chat available on our website: meridianpediatrics.com under forms and policies.

M-Chat
Autism Specific Screener

- Free screener available on firstsigns.org
- 23 questions- can fill out in 10 minutes by parent while waiting in office
- Easy to score
- If fail 2 or more critical items or any three should be evaluated in more depth by a specialist
- Critical items- question 2,7,9,13,14,15

M-CHAT Critical Questions

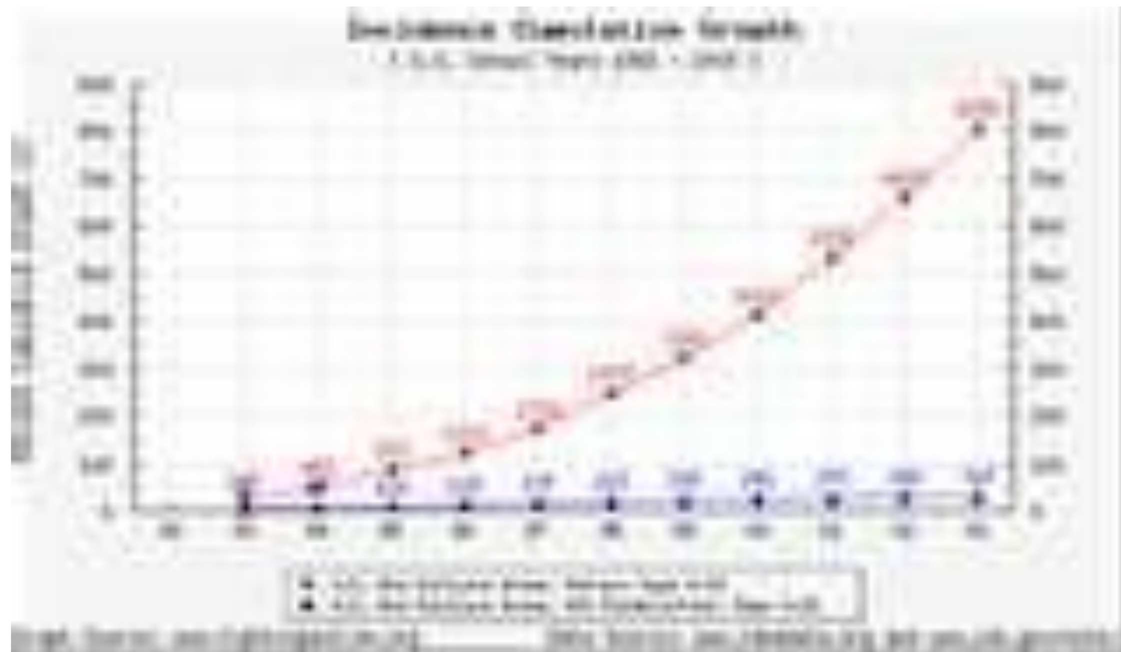
- 2. Does your child take an interest in other children?
- 7.Does your child use his index finger to point to indicate interest in something?
- 9.Does your child ever bring objects over to you(parent) to show you something?

M-Chat Critical Questions

- 13. Does your child imitate you?(you make a face and child imitates it)
- 14.Does your child respond to their name when you call?
- 15. If you point to a toy across the room does your child look at it?

- If fails screen refer or do more investigation!
- Failing the M-Chat does not mean the child has autism.

Why is Autism increasing?



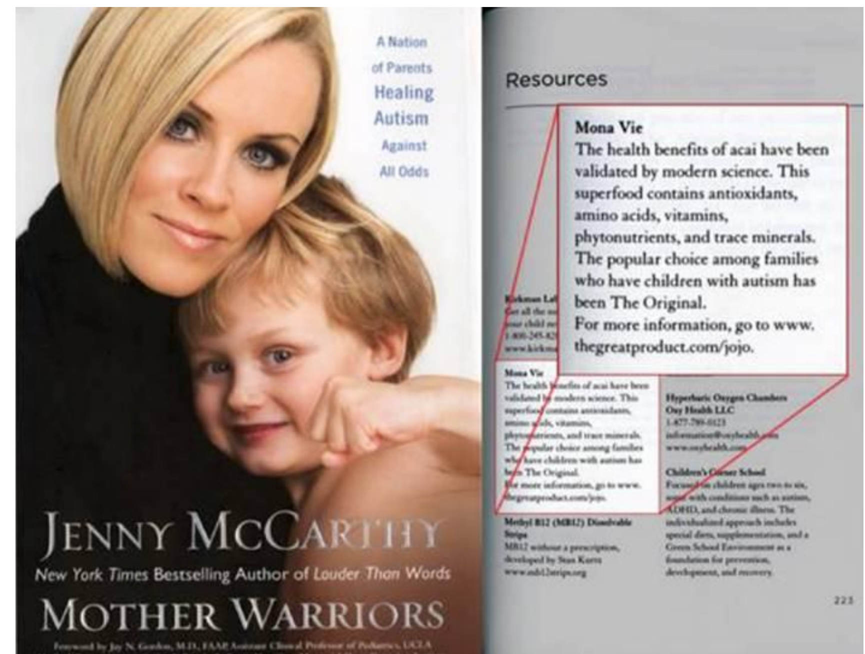
Autism
Possible reasons for the rise

- 1. Broadened diagnostic criteria, concept of spectrum(including Asperger and PDD-NOS)
- 2. Decreasing number of children being diagnosed without MR- relabeling
- Autism diagnosis with MR:10% in 1980’s, 25% in 1990’s, 40-60% in 2001
- MR/LD with autistic behavior is now called Autism with MR/LD

Autism
Possible Reasons for the Rise

- 3.Increased Public recognition-less stigma

- Increased professional awareness- availability of screening tests and screening guidelines
- No screening tools until 1992(CHAT)
- No formal evaluation tools until late 1980s
- CARS 1988,ADOS 1989,ADI 1994
- Increase in screening young children (MCAT)
- MCHAT free screener on firstsigns.org and now standard of care by AAP to screen at 18mo and 2 years.
- AAP recommends developmental screener at each well child visit- Denver out, ASQ in.



Autism
Possible Reason for Rise

- 4. Autism as a Diagnostic category in Special Education added in 1991
- In 1986 PL 99-457 expanded educational services for young children through Early Intervention Services (0-3 yrs) and school district developmental preschool programs(3-5 yrs).
- In 1987- mean age autistic child enter education system 6.9 yrs(1st grade) and in 1994 3.3 yrs(PK).
- Greatest rise in prevalence of autism is younger age (2-4 yrs).
- Diagnosis provides access to increased services(early intervention, DT, IBI,ABA)

Autism
Possible Reason for Rise

- 5. De-institutionalization
- Prior to 1990, 55% children with autism lived in an institution- not counted in education agency counts and not cared for by community doctors.
- 30-40% persons in institutions diagnosed with MR also had autism.
- 1990 ADA law unconstitutional to segregate persons with disabilities into large institutions
- 1990s-100,000 persons with MR moved into community. If 40% also have ASD-40,000 “new” children entered special education for the first time.

A true rise in prevalence??

- Unclear whether certain environmental or immunologic or yet unidentified factors may be playing a role.
- Etiology- Unknown
- What do we know? Strong genetic predisposition with a trigger
- Heritability estimates >90% (multifactorial)
- Monozygotic Twins(Identical and share 100% genes)- concordance autism 60%, broader phenotype 92%
- Dizygotic Twins(share 50% genes)- concordance autism 0-3%, broader phenotype 10-30%)
- Recurrence rate in children with ASD sibling is 3-7%
- Twin and multiplex family studies support “multiple gene” theory and a possible trigger effect of regulator gene or environment
- Idea of “perfect storm”

How often do you find a cause?

Etiology of autism

- Syndromic 10-20%
- Unknown 80-90%

Syndromic Autism
Medical/Genetic Cause

Syndromic ASD

- 10-20% ASD is associated with(and/or caused by) a known genetic or medical disorder
- Meta-analysis of 23 epidemiological studies revealed known cause only 6% (JAMA 2001)
- One study detailed workup including telomeric FISH(FISH for 22q11,15q11-13,mecp2), Metabolic studies, MRI, and EEG found a cause in 20%
- Typical workup: All kids need Hearing and Vision, Growth parameters, High resolution Chromosomes, Fragile X, and Microarray CGH (Comparative Genomic Hybridization)- replaces FISH.
- Thorough skin exam using a Wood’s lamp all kids
- CGH tests for deletions, microdeletions, rearrangements, and duplications in the human genome. It does not detect balanced alterations.
- EEG, MRI, Metabolic studies, lead levels based on history

What is your typical workup?

Work up Autism

- Hearing and Vision and growth parameter screening in all kids
- Lead testing if history of pica
- Consider metabolic tests: Urine OAs, and serum AA, blood lactate and pyruvate levels especially if history of regression in developmental milestones.
- Review newborn screen in all kids
- MECP2 mutation(esp female)- Rett Syndrome
- DNA analysis for fragile X all kids- more commonly seen in males
- High Resolution chromosomes and microarray CGH all kids.

Growth Parameters
Head Growth in Autism

- HC slightly small or normal at birth
- Very rapid head growth in the first 2-4 years then gradually slows, max size by 3-6 years age
- Adolescent/ adult head size usually normal
- Increased head size reflective of increased brain size- lack of pruning especially in frontal/temporal lobe

Fragile X (FXS) Testing

Most common cause of inherited mental impairment.

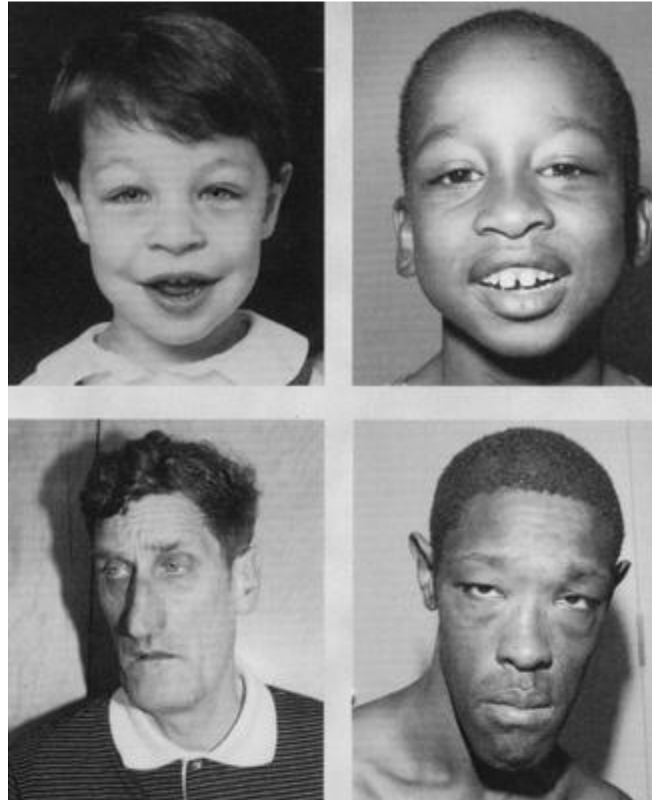
5-10% persons with autism have Fragile X.

15-25% persons with FXS have autism.

50% FXS have some autistic features.

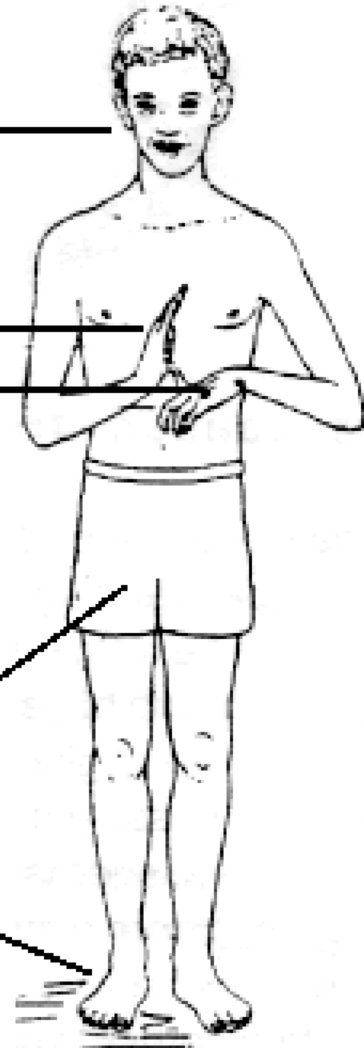
Co-occurrence both suggests worse prognosis but both are a spectrum in terms of severity.

Important to screen all because physical features develop over time .



Fragile X

- normal structure
- broad forehead
- elongated face
- large prominent ears
- strabismus (crossed eyes)
- highly arched palate
- hyperextensible joints
- hand calluses (from self-abuse)
- pectus excavatum (indentation of chest)
- mitral valve prolapse (benign heart condition)
- enlarged testicles
- hypotonia (low muscle tone)
- soft, fleshy skin
- flat feet
- seizures (in about 10 percent)



Wood's Lamp
Neurocutaneous Syndromes

- Neurofibromatosis
- Café o lait spots, axillary freckling, neurofibromas
- MR, LD
- Can have comorbid autism- 0.2-14% kids with autism have NF in 3 studies

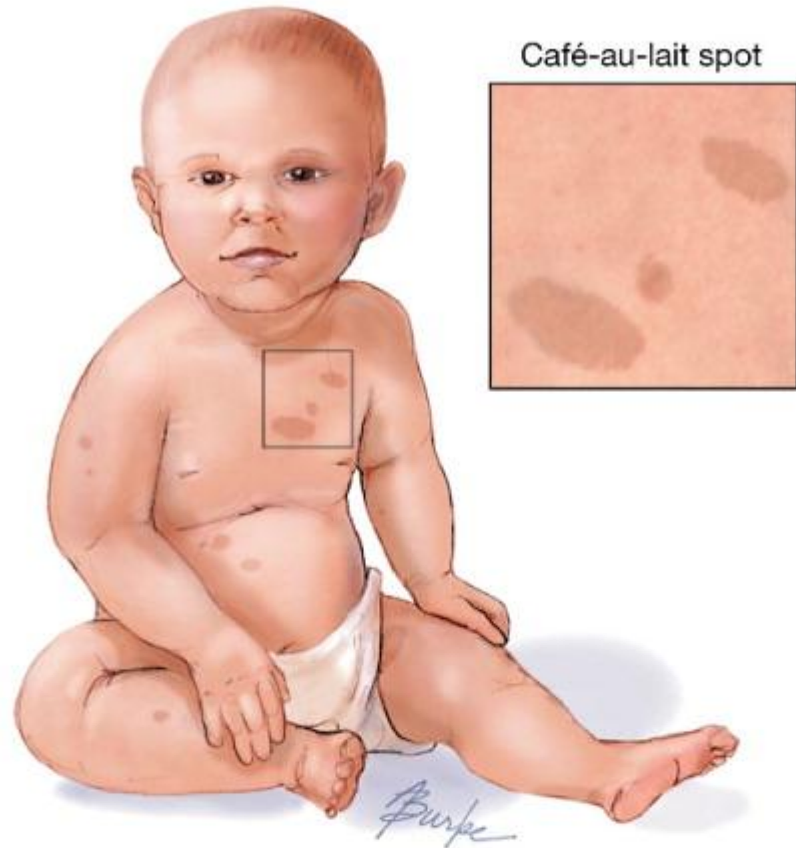
- Tuberous Sclerosis
- Phenotype-seizures, MR, skin and brain abnormalities
- Facial angiomas
- Ungal Fibromas
- 0.5-3% of kids with autism have TS
- 14% kids ASD and seizures have TS.
- 25-60% with TS have autism

NF-1

Autosomal Dominant inheritance

Can be a normal birthmark

Multiple birthmarks in the setting of LD or MR need to consider NF



Tuberous Sclerosis

Ash Leaf spot- hypomelanic spot

May be only sign at birth

Need woods lamp to visualize

95% kids with TS have a skin spot

Over time develop angiofibromas

50% LD

25-60% will meet criteria for autism

High Resolution Chromosomes Microarray CGH

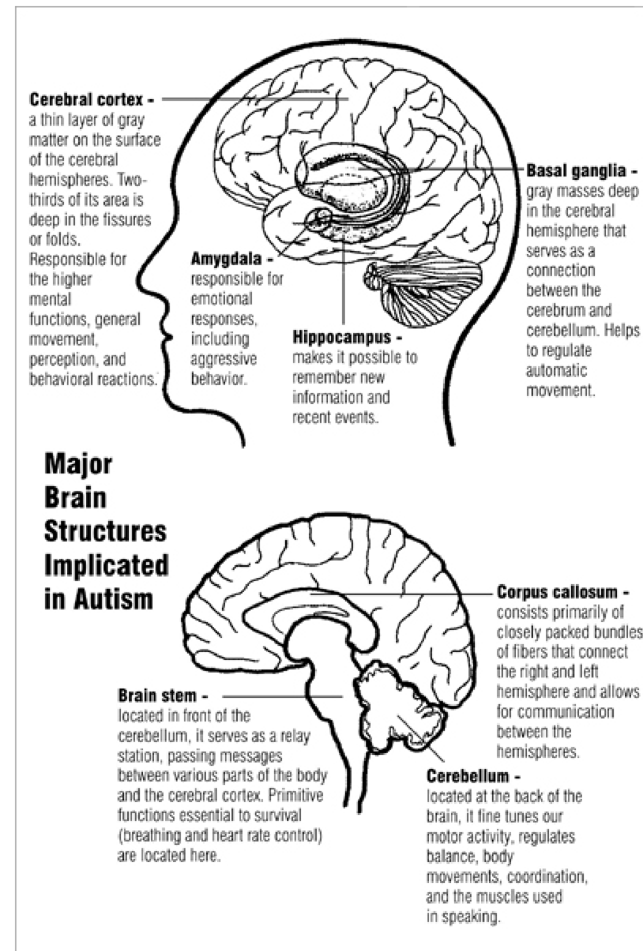
- Associated Syndromes with Autism:
- PKU
- Charge
- Prenatal Rubella
- Cornelia De lange
- Angelman
- Smith Magenis
- FAS
- Smith-Lemli-Opitz
- Down Syndrome (7% meet criteria for ASD)

Autism, Seizures, and EEG abnormalities

- Seizures seen in 5-40% of autistic children
- Peaks 0-5 yrs and adolescence
- With severe MR, 25% have seizures
- With severe MR and motor abnormalities(ataxia, hemiparesis) 65% have seizures
- Abnormal EEG in 10-80% autistic especially focal spikes in bilateral temporal lobes
- Almost all kids on the spectrum will show diffuse background slowing on an EEG.
- Thus, EEG if history of moderate to severe MR, motor abnormalities, or any abnormal neurological signs.

Autism- MRI

- Variety of abnormalities been reported involving the cerebral cortex, temporal lobes, basal ganglia, corpus callosum, and brain stem
- Most often reported;
- ! Size lobules VI and VII cerebellar vermis
- ! Size mesial temporal lobes
- ! Neuronal size and ! numbers of cells in limbic system- emotional area
- MRI kids with regression seizures, or abnormal neuro exam



Autism and Mirror Neurons Dapretto et al.

- These are neurons in the premotor frontal lobe that become active in response to emotional stimuli
- These neurons appear to be hypoactive in children with autism
- Such hypoactivity may explain the autistic child's inability to " read" the emotions of others and may underlie their social isolation
- (DaPretto)- "For an autistic child, a social situation is like a square dance where the caller is speaking Swahili"
- Research looking at what is the regulator to activate or deactivate the activity of these mirror neurons.
- For now kids with all kids autism need social skills training

Idiopathic autism

- 80-90% without an underlying disorder- workup is normal.
- Multifactorial- both genetic and environmental influences
- Polygenetic-estimated 3-10(some feel as high as 20) genes involved.
- “gene causing” now “gene susceptibility”
- Combination of factors helps to explain spectrum of features but makes finding cause and best treatments difficult!
- Autism is a spectrum:
- MR/nonMR, verbal/nonverbal, typical/savant, GI and immune sx/nonsx, mild/severe ASD, aloof and passive/interactive but odd

Polygenetic:
Autism Hotspots

7p- regulator gene that codes for social behavior in knockout mice

7q: 7q11,7q22-31.2,7q31-35,7q22

13q- Serotonin receptor gene

15

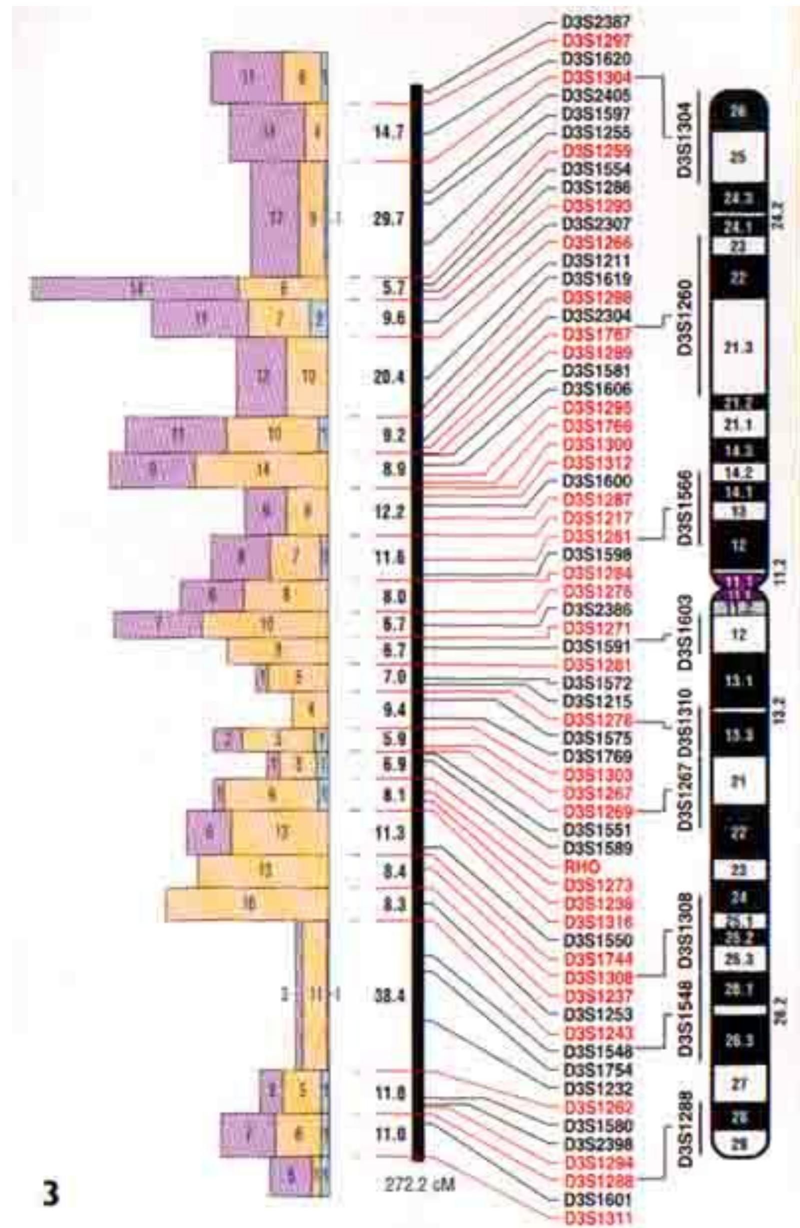
15q11-13 duplication found in 5-10% Persons with ASD

16- assoc site for Tuberous Sclerosis

17- Serotonin transport gene seen in Smith Magenis and ASD

22

22q11-Velocardiofacial syn and autism



What roles do vaccines and mercury play in autism?

Environmental Influences
Viruses and Vaccines

Prior to MMR vaccine viruses known to cause autistic symptoms in children- CMV and intrauterine rubella

- Vaccines:
- MMR controversy
- Thimerisol(Mercury) controversy

MMR and Wakefield vaccine study

"Ileal lymphoid-nodular hyperplasia, nonspecific colitis and pervasive developmental disorder in children"

Wakefield AJ et al Lancet 1998 351:637-41

Study Hypothesis: MMR vaccine causes a series of events that include intestinal inflammation, loss of intestinal barrier function, encephalopathic proteins enter the bloodstream with consequential development of autism

Dr. Wakefield described 12 children with neurodevelopmental delay, 8 with autism. All had GI complaints and developed autism within one month of receiving the MMR vaccine.

Wakefield MMR study Fraud

- Four years after the study no investigator able to reproduce his results.
- In 1995, Wakefield while investigating Crohn's disease was approached by Rosemary Kessick, a parent of a child with autism who had bowel problems. She ran a group named Allergy induced Autism.
- In 1996 Wakefield stated to investigate possible link MMR and Autism.
- In 1998, he published results in Lancet claiming MMR caused autism in 12 children.
- Immediately following paper BBC news conference Wakefield stated we should stop the three in one jab of measles, mumps, and rubella vaccine.
- In 2002, meta analysis show no researchers able to duplicate his results.
- In 1993 Japan stopped vaccinating with the MMR vaccine and the rate of autism continued to rise.
- In England 1998 they also stopped MMR vaccine- first time in over 12 years rise in measles deaths but incidence autism still rose.
- In 2004, Brian Deer a reporter uncovered undisclosed financial conflicts of interest by Wakefield.

Wakefield MMR Study Fraud

- Some of the 12 parents in the study were recruited by a UK lawyer preparing a lawsuit against the MMR vaccine manufacturer
- Royal Free Hospital where Wakefield worked received 55,000 pounds from UK Legal Aid Board to do research on MMR vaccine.
- Wakefield personally received 435,000 pounds($750 K) from the lawyers suing the vaccine company which he did not report to the study review board. He started to receive payments two years before the Lancet paper was published to build a case against the MMR vaccine.
- Wakefield also applied for a single jab measles vaccine patent one year before his Lancet paper- if MMR was discredited he stood to gain an additional huge financial benefit. This was also not disclosed either.
- Later records showed that 5 of12 kids were not previously “normal” prior to the MMR. Only 1 of 12 of the kids had regressive autism. Three of 12 never diagnosed with autism at all.
- BMJ 2001 Other authors retracted their claim and conclusion” patients were recruited through anti-MMR campaigners, and the study was commissioned and funded for planned litigation.”
- 2010 Wakefield UK Medical license revoked.

Result Wakefield Study

- In Britain, measles and mumps cases soared in the thousands.
- In the US, outbreaks of measles, pertussis, and Haeophilus influenza type b involved kids whose parents opted out of immunizations for fear cause autism.
- Dr. Wakefield still has defenders especially parents of autistic kids.
- JB Handley founded Generation Rescue, now headed by Jenny McCarthy, told CNN that "BMJ did not remotely discredit Dr. Wakefield's study."

Before Vaccines each year in the US…

- Measles would infect 4 million children and kill 3,000
- Diphtheria would kill 14,000 mostly teens
- Rubella would cause 20,000 babies to be born blind, deaf, and mentally retarted
- Pertussis would kill 8,000 children
- Polio would paralyze 15,000 children and kill 1,000
- No single medical advance has had a greater impact on human health than vaccines!
- As evidence against MMR virus as causative factor mounted, anti-vaccine groups turned their focus towards thimerisol and mercury.

What is Thimerosal?

- Thimerosal is 49.6% mercury by weight as the sodium salt of ethylmercurithiosalicylate
- Thimerisol (composed in part of ETHYLmercury) was used as a preservative in some vaccines(but not MMR) since the 1930’s.

Mercury Concerns

- 1n 1999 the FDA concluded that the current vaccination schedule which recommended an increased number of vaccines, some infants might receive cumulative doses of ETHYLmercury that exceeded EPA safety guidelines for Methylmercury.
- 1960- 5 vaccines diptheria, pertussis,polio, and smallpox required 8 shots by the age of 2.
- 1999-children received 11 vaccines routinely and as many as 20 shots by age 2.
- The safety guidelines were based on oral exposures to MeHg not parenternally doses of EtHg.
- AAP and US Public Health recommended removing thimerisol from vaccines since no data on EtHg
- 2001- thimerisol removed from all vaccines except some influenza vaccines.

Role of thimerisol and autism

- No published studies of known ethylmercury poisoning.
- Ataxia, tremor, visual disturbances, renal tubular acidosis, and skin rashes known complications of methylmercury exposure DO NOT characterize the symptoms of autism!
- These symptoms are more compatible with metabolic disorders and extrapyramidal cerebral palsy.
- Males with autism far outnumber females with autism, yet they are equally immunized.
- Theoretically premies should experience a higher toxicity risk associated with neonatal HepB(more Hg load per BW) and immature liver to metabolize Hg to a inorganic(safe) form.
- Yet prematurity is not a risk factor for autism. (Peds 2001 and Bristol 1996.)

IOM MMR and Thimerosol Feb. 2004

- CDC and NIH asked the IOM(institute of medicine) to establish an independent expert committee to evaluate the evidence on possible causal associations between vaccines and certain negative health outcomes.
- Final report Feb 2004- meta-analysis concluded: There is currently no scientific evidence of causal relationship between MMR and Thimerisol containing vaccines and autism.
- Despite this DAN doctors still chelate for mercury- can cause seizures and death.

Costs of Autism

- Average lifetime cost for a person with Autism is $3.2 million
- Annual cost to provide services for all persons in the US is $60 billion
- Other costs:
- “ Parents of autism are exhausted and broke. Too many of them go to bed at night and pray that one day their child will look at them smile and say Mommy.”

Prognosis of Autism

- Independent as adults 5-10%
- Some degree of independence 25%
- Highly dependent 60-65%

- Acquisition of meaningful speech by 5 years is a key predictor with none the outlook is very poor.

Autism

- The Ten Pathways to Cure

Is there a cure for autism?

“ Curing” Autism

- 1. Screen at all well visits for developmental red flags especially at 9,18,24, and 30 months per the AAP.
- 2. If uncertain about diagnosis, quickly seek a second opinion
- 3. Seek and treat any underlying treatable cause

“Curing Autism”

- 4. Treat any accompanying medical and neurological disorders

- 5. Treat behavioral dyscontrol
- A. non-pharmacologically
- B. pharmacologically

Behavioral Disorders in Autism

- ADHD
  Stimulants,Stattera, Intuniv
- Tantrums/Aggression
  Risperdal,Intuniv
- Self-Injury
  Risperdal
- Anxiety
  Prozac,Zoloft

Behavioral Disorders in Autism

- OCD/Stereotypies
  Prozac,Zoloft
- Depression
  Prozac,Zoloft
- Mood Swings

  Risperdal, Depakote, Atypical antipsycotics

Sleep Disturbance

  Mellatonin, Clonodine,Trazadone

“Curing” Autism

- 6. Intensive Early Intervention, Developmental therapy, and or IBI

- 7. Speech and Language therapy, physical therapy, occupational therapy, sensory integration therapy, social skills training and groups, augmented/alternative communication

“ Curing” Autism

- 8. Parent education, training, and support

- 9. Complementary/alternative therapies- consider if no apparent risk and some scientific support- GFCF diet, Vitamins and fish oil
- ASATonline.org

“Curing” Autism

- 10. Don’t give up…..

  KEEP PADDLING!!