



- Prevalence of ASD 1:150 children, more common than childhood cancer, juvenile diabetes, and pediatric AIDS combined
 - Boys 1 in 94*
- No established explanation for this increase-improved detection/environmental changes
- Autism is behaviorally defined, but biologically based
- Heterogeneous, can be accompanied by unusual talents and severe impairments
- Underlying biological and genetic basis still unknown, though small number of cases can be linked to other genetic disorders, or prenatal exposure to toxins or infections
- Role immune system plays within the brain and body
- How do you sort out subgroups and multiple pathways to ASD
- Ultimate goal-what is most effective prevention, intervention for these disorders
- No conflicts of interest or disclosures**

Three year old child with



Video of a boy with ASD, in a prek classroom.

By age three, what would you expect child to be doing?

What are the typical developmental milestones?

Autism Spectrum Disorders (ASD)

- Complex neurobiological disorders
- Range of developmental problems
- Lifelong condition
- Involves triad of symptoms
- ASD includes *Autism, Pervasive Developmental Disorder, Asperger's syndrome, Rett's syndrome, Childhood Disintegrative Disorder*

- ASD affects way child perceives world and makes communication and social interaction difficult
- Symptoms of ASD last throughout person's lifetime-quirky to severely disabled
- Triad includes communication, social skills, and repetitive behaviors
- Spectrum includes...

Key Definitions (DSM-IV-TR)

- Autism
 - Onset before age 3 years
 - Qualitative impairments in three core areas
 - Communication (gestures, words)
 - Social Interactions (joint attention)
 - Interests and activities (symbolic play)
 - 50-70% have MR
 - Prevalence 4 of every 1,000

- Most common of disorders in spectrum
- Evident in first two years of life-even as early as 14 months
- Symptoms range from mild to severe
- Few have “Rainman” abilities
- More boys than girls
- Increased seizure risk

Key Definitions (DSM-IV-TR)

- Pervasive Developmental Disorder, NOS
 - Unusual development in core areas of communication, or social interactions, or interests/attention
 - Does not meet criteria for any of the other ASD
 - Often occurs with cognitive impairment
 - Prevalence 1 in every 1,000

- Atypical Autism
- Severe, pervasive impairment in social, communication or behavior
- More commonly DX in lower functioning children

Key Definitions (DSM-IV-TR)

- Asperger's syndrome
 - Qualitative impairment in social interaction
 - Restricted, repetitive patterns of behavior, interest and activities
 - No obvious delays in language (single words by 12 months, 2-words by age 2, phrases by age 3)
 - No clinically significant delays in cognitive or adaptive functioning
 - Appear clumsy
 - Prevalence 2.5 in every 1,000 individuals

- No clinically significant delay in language: single words by 2, phrases by 3
- No clinically significant delay in cognitive, self help skills, adaptive behavior, or curiosity
- Poor motor coordination
- Clumsy gestures
- Do not understand personal space
- Difficulty picking up social cues
- Approaches others only to have needs met
- Expert on things, not people
- Make better adults than children

Leonard & Sheldon *The Big Bang Theory*



- Monday night on CBS
- Sitcom around 4 brilliant young men, with Ph.D.'s in Physics, and a waitress at Cheesecake factory
- Clumsy social approach, awkwardness in social relationships

Key Definitions (DSM-IV-TR)

- Rett's Disorder
 - Normal development first 5-18 months
 - Normal head circumference at birth, deceleration between 5 and 48 months
 - Loss of hand skills, then stereotypical hand movements
 - Loss of social engagement, language, motor skills
 - X-linked MeCP2 gene
 - Prevalence 1:15,000 live female births

- Males die in utero (X-linked gene)
- Progressive disease-in early stages look like ASD, but deteriorate
- Requires ongoing medical support/therapies
- May live into 40's
- Burden to families
- Occurs randomly in all populations

Key Definitions (DSM-IV-TR)

- Childhood Disintegrative Disorder
 - Normal development first 2 years
 - Loss of acquired skills by age 10
 - Language/IQ
 - Bowel and bladder
 - Play
 - Motor skills
 - IQ moderate to profound MR
 - Emergence of stereotypic behaviors
 - Prevalence 1: 100,000 individuals

- Heller's syndrome, dementia infantilis, disintegrative psychosis
- Unknown etiology
- Linked with tuberous sclerosis, neurolipidoses, pertusis, measles
 - Fearfulness/anxiety symptoms
 - Seizures after onset
- Treatment: antipsychotic, antiepileptic
- Behavior management
- Educational services
- Poor prognosis
- Normal life span

Features of ASD

- Prevalence ASD 1:150 children
 - Autism 4 per 1,000
 - PDD and Asperger's 1-2 per 1,000
- 3 to 4 times more common in boys
- Genetic factors thought to underlie ASD's
- Severity of symptoms varies
- Prognosis varies
- No known cures

- Vast majority of cases are idiopathic, or cause is unknown
- Levels of severity and combinations of symptoms vary
- Best scientific evidence points to a combination of genetic factors and environmental conditions, yet unknown
- Much more common in males, (with exception of Rett)
- Sleep problems-some evidence of a melatonin deficiency
- Wide range of treatments available-some are supported by research-no single therapy works for every child

ASD Signs and Symptoms

- Symptoms present in three crucial areas
 - Communication
 - Social skills
 - Behavior/Play
- Development may seem normal first few months or years of life
- Symptoms and skills vary among children with same diagnosis

•Communication

- Delays in first three years (not Asperger's), but most learn to communicate in some way
- May parrot what they hear-long after age 3
- Speak without inflection, or in sing-song, high pitched, robot like
- Do not match body language, facial expressions to speech
- Inability to interpret body language, tone of voice, sarcasm
- Have difficult time expressing emotions, and are at risk for depression/anxiety

•Social Skills

- Difficulty learning to engage, share interactions with others
- Avoid eye contact
- Resist attention or are passive
- Fail to seek comfort
- Do not understand social cues
- Difficulty regulating emotions

•Behaviors/Play

- Repetitive behaviors, such as flapping, walking on toes
- Spend hours lining up toys
- No pretend play
- Upset by changes in routines
- Preoccupation-Thomas the Tank engine, vacuumed cleaners, plane schedules

Red Flags: Communication

- No babbling by 12 months
- No pointing by 12 months
- No single words by 16 months
- No 2-word spontaneous phrases by 24 months
- Speaks with abnormal rhythm or tone
- Can't start a conversation or keep it going
- May repeat certain words or phrases but doesn't use them appropriately
- Loses ability to say words

Red flags for identification-Developmental Milestones

Three Year Old, with ASD



Three year old boy-mother is engaging with him in an pleasurable activity, and trying to stretch his speech. What would you expect a typical three year old to be doing at this age? What behaviors do you see?

Red Flags: Social Skills

- No smiling by 6 months
- No imitation facial expressions by 9 months
- Fails to respond to own name at 12 months
- Has poor eye contact
- Appears not to hear you
- Resists cuddling and holding
- Appears unaware of other's feelings
- Seems to prefer to play alone
- Retreats into "own" world

Red flags for early identification-Social and Emotional Development

3 Year Old Child, with ASD



Brothers making pancakes. Brother facing away is a three year old with ASD. What behaviors do you expect for that age? What do you see?

Red Flags: Behavior

- Performs repetitive movements: rocking, spinning, hand flapping
- Develops specific routines or rituals
- Becomes disturbed with slight changes in routines or rituals
- Moves constantly
- Fascinated with parts of objects
- May be unusually sensitive to light, noise, or touching

- Red flags, beginning around 12-14 months-which become more entrenched as child matures
- Challenging behavior when these routines are disrupted or disturbed

Preschool Child, with ASD



Three year old at daycare. Note his behavior, attempts of teacher to draw him into activity, and typical peers

ASD and Learning Problems

- Majority have academic deficits
- Poor coping skills and problem-solving
- Communication problems
- Concrete thoughts
- Attention problems
- Challenging behaviors
- Most will need some assistance as adults
- Less than 5% have genius-like abilities

- All children can learn
- Children with ASD have learning styles quite different from peers
- Many skills have to be explicitly taught and reinforced
- Rigid thinking-once they learn to do something, have difficulty switching
- Family and intervention team need to be on same page
- Many individuals will need ongoing assistance in adulthood

ASD and Mental Health Disorders

- Shared symptoms
 - ADHD
 - Anxiety
 - Bipolar
 - Depression
 - OCD
 - Mental Retardation

High rates of co morbidity (Bruin, Ferdinand, et. Al, 2007)

Children 6-12 years with PDD

- 80% one co morbidity
- 61.7% disruptive behavior disorder
- 55.3% anxiety disorder

Lubetsky (2007)-children with ASD

60% MR

*Because of high co morbidity, clinical assessment of DSM-IV disorders should be completed

ASD and Medical Disorders

- Shared symptoms and genetics
 - Epilepsy
 - Fragile X
 - Tuberous sclerosis
 - Tourette's syndrome
 - Prader-Willie
 - Angelman's syndrome

National Institute of Child Health and Human Development (NICHD) doing research on autism to understand which genes may play role in the condition.

In 5% of ASD, another single gene or chromosome disorder is present

Scientists are studying known genetic disorders, with symptoms common to individuals with ASD

- Many people with ASD have seizures, as do individuals with Epilepsy
- Fragile X and Rett share symptoms with Autism, caused by genes on X chromosome
- Tuberous sclerosis affects nervous system and can result in combination of symptoms including seizures, developmental delays, and behavioral problems
- Prader Willi and Angelman both share behavioral symptoms with ASD and are caused by errors on chromosome 15. About 4% of individuals with ASD also show this error

Causes of ASD

- No single, identifiable cause
- Seems to be related to abnormalities in several areas of brain
- Environmental factors, e.g. viruses may trigger symptoms
- Structural (anatomic, cellular)
- Genetic component
 - Identical twins 60%
 - Siblings 6-8%
 - Other family members 2%

- No known biological markers at this time
- Best scientific evidence points to combinations of factors causing ASD
- Genetic components on their own or in combination with exposure to environmental toxins
- Timing or exposure unknown (pre-post natally)
- Small number of cases linked to other genetic disorders

Genetic Investigation

- **As many as 12 genes may be involved (NIH, 2005)**
 - HOX genes (brain stem and cerebellum)
 - Chromosome 7 (AUTS1-speech and language disorders)
 - Chromosome 13 (families with ASD)
 - Chromosome 15 (Angelman and Prader-Willie)
 - Chromosome 16 (seizures and tuberous sclerosis)
 - Chromosome 17 (problems with serotonin, OCD)
 - X Chromosome (46 chromosomes; XX, XY)

ASD Myths

- Parents/parenting styles cause ASD
- Vaccines containing thimerosal (mercury) cause ASD
- More white children affected
- Children grow out of ASD
- One proven approach (e.g. ABA, Floor Time)
- Dietary changes will cure core symptoms

- Kanner, then Bettleheim perpetuated myth that ASD caused by cold, rejecting mothers
- In 1970, Dr. Rimland, founder of Autism Society of America helped medical community understand that ASD was a biological disorder
- Autism effects all children equally
- In some children, symptoms improve. Most will carry a degree of disability for their lifetime
- One size does not fit all-intervention must be individualized to child and family
- 70% of ASD children report GI symptoms, eg pain, vomiting, diarrhea, or constipation, thus thought to be related to food sensitivities-diets that avoid refined carbs, gluten free diet, yeast free diet, low oxalate diet used/no evidence that they improve core symptoms, and children may not receive proper nutrients
- Some speculation that children with ASD are the “yellow canaries”-to toxins in foods, environment, etc. yet to be proven

Components of ASD Diagnosis

- Hearing evaluation
- Developmental assessment
 - Levels of performance in 5 domains
 - ASD specific tools
- Developmental history
 - Address core features of ASD
 - Health history
- Speech and language
 - Form, content, and pragmatics

Dx through a multidisciplinary team eva

- ABR hearing
- Standardized assessment tools for development, adaptive behavior, and ASD symptoms
- Information gathered from caregiver
- In depth speech and language assessment

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Specialized ASD Tools

- Caregiver report and observational measures
 - Autism Diagnostic Observation Schedule (ADOS)
 - Child Autism Rating Scale (CARS)
 - Child Behavior Checklist (CBCL)
 - Gilliam Autism Rating Scale (GARS-2)
 - Gilliam Asperger's Disorders Scale (GADS)

Caveat: Tools may not be useful for children under age 3 or children with no language

Medical Tests

- Electroencephalogram (EEG)
- Metabolic Screening (blood & urine)
 - Lead screening
- Magnetic Resonance Imaging (MRI)
- Computer Assisted Axial Tomography (CAT Scan)
- Genetics (FISH)

American Academy of Neurology Guidelines

EEG-inadequate evidence for ASD-indicated for seizures, or history of skill regression

Metabolic screening-presence of clinical or physical findings, such as lethargy, vomiting, seizures, global developmental delays, questionable adequacy of newborn screening

Lead screening-developmental delays or pica

MRI/CAT scan -no clinical evidence to support routine neuroimaging

Genetics-strong evidence supports: high resolution chromosome study and analysis for Fragile X, especially if dysmorphic features, family history, MR

Evidence-Based

- Educational interventions
 - Behavioral Approaches
 - Positive Behavior Support (e.g. HOT DOCS©)
 - Discrete Trials (Applied behavior analysis)
 - TEACCH Program
 - Speech and language (e.g. *Hanen*)
 - Social skills training (e.g. Social Stories)
 - Early relationships (e.g. Greenspan's *Floor time*)

- Interventions which address core features of ASD
- Educational approaches
- Children learn what you teach them

Pharmacological Interventions

- No medications address core features of ASD
 - Anti-infection medication (sinuses, fungal, bacteria)
 - Antipsychotics (Risperidone*)
 - Stimulants-off label only
 - Sleep aides-off label only
 - SSRI's-off label only
 - Anticonvulsants-off label only

- Treat underlying medical conditions
- Risperidone FDA approved for children ages 5-16 for symptoms of irritability
- Other medications are off label only
- Careful diagnosis of co-morbidity-medications for problem symptoms such as self injurious behavior, inattention, anxiety, depression, sleep problems



Non-Evidenced Based, Complementary Approaches

- Detoxification
- Dietary modifications
- Nutritional supplements
- Herbal medications
- Hyperbaric Oxygen (HBOT)
- Sensory Integration Therapy
- Auditory Integration Therapy
- Music Therapy
- Facilitated communication
- Others...

Effectiveness of Intervention for Children with ASD

- What works more often opinion than evidence
- Belief that particular intervention can change outcomes
- Many individuals already receiving multiple treatments (eclectic approach)
- Difficulty in transferring intervention from research to practice setting

Child factors

- Developmental issues
- Challenging behavior
- Adolescence

Intensity

- How much is needed?

Duration

- How often?

Setting

- Where?

Evidence-base for ASD Interventions

- Interventions work best for:
 - Higher functioning children
 - Children with less severe behavioral symptoms
 - Children who begin intervention early (<60 months)
 - 25 hours per week of engagement
 - Intervention across natural settings
 - Multiple methods used

- Very little research with toddlers
- About half children show improvement, regardless of approach
- Studies do not have comparison groups
- Cannot extend preschool approaches to toddlers/families
- Home/childcare settings versus laboratory/clinical applications

Issues Related to Early Intervention

- Dilemma in early identification
 - Children identified early show less severe symptoms compared with those identified later
 - Children flagged early for ASD may not meet criteria later on
 - Changes in natural course?
 - Result of early intervention?
 - Misidentified?

Landa (2008) Kennedy Krieger studied high risk for ASD infants (107 siblings of children with ASD), found that 50% could be reliably dx with ASD as early as 14 months

Pediatrician's Role

- Pediatric well-child screenings at 9, 18, 24 and 30-months (APA Guidelines, 2008)
 - Ages and Stages
 - M-CHAT (18 month & 24 month visit)*
- Referral for formal evaluation (IDEA, 2004)
 - **Early Steps** (0-3)
 - **Child Find Public Schools** (3-21)
- Ongoing care management

•Increased risk for other medical conditions, including behavioral conduct problems, ADHD, stuttering, depression/anxiety, hearing and vision problems, food allergies, frequent headaches,

•Florida House and Senate Bill 1291 “Window of Opportunity Act” will require training for health and childcare professionals to insure timely screening, assessment, and intervention for children with ASD; health insurance coverage for well baby/well child screenings, diagnosis, and for intervention and treatment of ASD

•Ongoing care management-Children with ASD have more chronic medical concerns than typical peers, often require services from multiple providers & systems of care, providers change, family functioning may vary, children will transition into adult care system. Concept of the “Medical Home” for children with disabilities, including Autism

Helpful Resources

- American Academy of Pediatrics <http://www.dbpeds.org>
 - AAP Tool kit (2008) Caring for Children with ASD
- Autism Speaks <http://www.autismspeaks.org>
 - First 100 Days Tool Kit (2008)
- Autism Society of Florida <http://www.autismfl.com/>
- Centers for Autism and Related Disabilities (CARD)
- Center on the Social and Emotional Foundations for Early Learning <http://csefel.uiuc.edu>
- HOT DOCS© <http://www.usfpeds.hsc.usf.edu>
- Genes and Autism <http://www.nichd.nih.gov/autism>
- IDEA, 2004 <http://idea.ed.gov>
- National Institute of Mental Health <http://www.nimh.nih.gov/publicat/autism.cfm>
- Partnership for Effective Programs for Students with Autism <http://www.autism-society.org/site/PageServer>

AAP Tool Kit (2008) Caring for Children with ASD

Contains screening, surveillance guidance using validated tools

Information re ASD screening at 18 & 24 months

Physician fact sheets

Family Handouts

English & Spanish

Autism Speaks: First 100 Days Tool Kit-Kit to assist families in getting critical information needed in the first 100 days after DX



Thank you!!!!

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