Autism Spectrum Disorder: An Overview

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Overview

- **WHAT:** ASD, Spectrum, Disorder, and DSM5
- **WHEN:** Brief (very) History
- **WHERE:** Prevalence
- **WHY:** What we think we know...and what we know that we don’t know about causes
- **HOW AND WHO:** Early recognition and screening
  - Diagnostic processes
  - Core features and related features
  - Interventions/treatment
  - Outcomes and the future
Why is VDH/public health involved with children with ASD and developmental concerns

- Public Health and “prevention”
- Primary, Secondary and Tertiary prevention
- Populations, data, and assurance
- Early recognition and intervention
- Maternal and Child Health and
  Children with Special Health Needs in systems of services
ASD is not a disease...

...But a neurodevelopmental *syndrome* or *disorder*: a group of symptoms that *tend* to cluster together and share a common natural history/course. A *disease* is a syndrome for which there is a known cause and/or a known pathophysiologial process.

**Autism is a spectrum disorder**

- Defined by 7 core behaviors that differ in such dimensions as onset, description, intensity
- *Boundaries* of the spectrum are defined by a committee, reviewed and revised by broad input, and finalized by consensus in the DSM5, the 2013 fifth edition of *The Diagnostic and Statistical Manual*
- A multi-dimensional spectrum disorder
ASD: The Basics: Two D’s, Delays and Differences

Two Core Areas of symptoms:

- Persistent differences (“deficits”) in social communication and social interaction across contexts, not accounted for by general developmental delays, and
- Restricted, repetitive patterns of behavior, interests, or activities

Symptoms must be present in early childhood
Symptoms together impact everyday functioning
Symptoms are not better explained by global developmental delay or other developmental or mental health disorder
3 of 3: Difficulties in Social Communication and Social Interaction:

- Social-emotional reciprocity (back and forth communication, sharing of interests, initiating and responding to social interactions), *and*
- Non-verbal communication (eye contact, body language, gestures, facial expressions), *and*
- Developing, maintaining, and understanding relationships (interest in peers, sharing imaginative play, making friends, adjusting behavior to suit social context) … *AND*
ASD DSM5 Diagnostic Criteria

2 of 4: Restricted, repetitive patterns of behavior, interests, activities:

- Stereotyped or repetitive movement, use of objects, speech
- Insistence on sameness, routines, rituals in verbal and non-verbal behaviors
- Highly restricted or fixated interests of unusual intensity or focus
- Hyper- or hypo-reactivity to sensory input, or unusual interest in sensory aspects of environment
A brief history of recognizing and defining ASD:
Leo Kanner, 1943, child psychiatrist “Autistic Disturbances of Affective Contact”
Bruno Bettelheim 1968 and “refrigerator mothers”
Diagnostic and Statistical Manual editions:
- DSM-II: A form of childhood schizophrenia
- DSM-III: Infantile Autism
- DSM-IV: Criteria: 3 areas of behavioral symptom clusters and 5 subtypes defined: Autism; PDD-NOS; Rett; Childhood Disintegrative Disorder; Asperger
- DSM5: Criteria: 2 areas of core symptoms, and single condition, ASD. Added 3 levels of severity. Added specification of with or without intellectual impairment, language impairment, known medical or genetic conditions or environmental factor, associated with another neurodevelopmental, mental or behavioral disorder.
- New to DSM5: Social Communication Disorder
What’s up with ASD Prevalence?

“Too soon to say whether autism prevalence stabilizing”
--Centers for Disease Control and Prevention, 2016

CDC’s ADDM (ASD and Developmental Disabilities Monitoring) Network

Began in 2000. 11 states. Consistent data collection methods: Medical and educational record review in 8 year olds.

Prevalence more than doubled from 2002 (0.7%) (1 in 150) to 2012 (1.5%) (1 in 68)

But, no overall change from 2010 to 2012 reported in 2016 ADDM update

National Survey on Children’s Health

Based on parent report

Prevalence nearly doubled from 2011 (1.25%) to 2014 (2.24%) (1 in 45)

Earlier recognition? Inclusion of milder forms? Diagnostic substitution? Real? Yes...
ADDM Goals & Findings:

Identify changes in ASD occurrence over time

- 2002 0.66% 1 in 150
- 2008 1.13% 1 in 88
- 2010 1.47% 1 in 68
- 2012 1.46% 1 in 68

Describe the population of children with ASD

- Cognitive: 44% average or higher; 24% borderline; 32% intellectual disability
- Gender: Boys 4.5 times more likely to be dx than girls
- Racial differences in the rate of diagnosis: White > Black > Hispanic
- Age at DX: 43% diagnosed by age 3; most dx after 4.  CAN be dx by 2.
  - Median age dx: All (50 mo); Autism (46 mo); PDDNOS (49 mo); AS (74 mo)

Compare prevalence in 11 different areas of the country

- Current prevalence: US (1:68); SC (1:81); NJ (1:41)
- Boys in MD (13.9/1000); NJ (39.1/1000)
Causes and factors in ASD: What we think we know... and know we don’t know

**ASD is NOT caused by:**

- “Refrigerator mothers” (Bettelheim)
- Vaccines and regression—Many studies, and No Evidence that:
  - MMR measles virus and “leaky gut”
  - Too many given at the same time stresses the immune system
  - Thimerosal (organic mercury antibacterial compound preservative) discontinued in 1999, but ASD prevalence rose
Causes: Genetics… and…..

Twin and sibling studies strongly suggest genetic component
Studies of younger siblings for early red flags for possible ASD are critically important for understanding the ways that ASD behavior develops and identifying it as early as possible.

“Syndromic ASD”

Increased ASD prevalence with some syndromes with known/identified genetic differences, among them: Fragile X, Rett, Tuberous Sclerosis Complex, Angelman, Smith-Lemli-Opitz, Williams, Down, diGeorge/22q
Ongoing controversies about whether the ASD symptoms are part of the syndrome and therefore “not ASD”
Genetics... and... “Epigenetics”

- No single cause likely to be found—but 90% likely to have a complex interaction of genetic factors (genes/DNA) and environment.
- Epigenetics: the study of how the environment changes the way that genes work (turning them on or off). Environment: stress, exposures, food. Epigenetic changes may also be passed onto the next generation.
- Prenatal environment and risk factors: Too early, too small, too close, maternal and paternal factors (age, infection, medications, illness).
- Whole genome microarray: small deletions and duplications (“copy number variants”). Some of these differences have been identified in others who have ASD, and but many have not..or not yet. A parent may have the same difference and have no ASD symptoms. Those which have been described in ASD “tend to cluster in genes controlling various biologic pathways important in neurotransmission.”
Towards Earlier Recognition of ASD... and Earlier Intervention

Research focus on infant siblings studies to identify early differences in behavior, images of brain development

Public outreach to increase parent and caregiver awareness of

- Normal social communication milestones in infants and recognizing delays and differences
- Red flags, and what to do: Learn the Signs. Act Early campaign. [www.CDC.gov/ActEarly](http://www.CDC.gov/ActEarly)
- Regression at any age
Early concerns recognizable in the first year

Communication:
- No babbling by 12 months
- No gestures (pointing, waving bye) by 12 months

Social: 5 behaviors that children with ASD do not do even in comparison with children who have developmental delays:
- Showing anticipation of being picked up
- Showing interest in children or peers other than siblings
- Reaching for a familiar person
- Playing simple interaction games with others (peekaboo)
- Showing affection toward familiar people
Early concerns in the second year

- Regression in social/communication, 15 – 24 months, often 18 – 21 months; may be sudden or gradual (occurs in 25%)
- No single words by 16 months
- No 2-word spontaneous by 24 months
- Limited eye contact
- Limited back and forth vocalizing
- Extremes of temperament
- Not turning to name
- Poor joint attention
- Play: limited imitation and pretend play
- Unusual language, echolalia, scripting, but little spontaneous communication
Role of developmental screening in Medical Home

- Screening for developmental delays during well-child visits, with standardized method, such as Ages & Stages Questionnaire.
- Specific screening for ASD at 18 and 24 months (up to 30 months) with M-CHAT-R (Modified Checklist for Autism in Toddlers, Revised). 20 yes/no questions probe for behaviors of concern for ASD. Score of 8 – 20 indicates high risk for ASD. Follow-up algorithm for positives.
- M-CHAT-R/F identified 67/10,000 children subsequently dx with ASD, fewer than the expected 147/10,000. Positive screens were associated with other developmental delays, however.
- In 2016 USPSTF: “There is insufficient evidence to assess the balance of benefits and harms of screening for ASD in young children for whom no concerns of ASD have been raised.”
Screening efforts in Vermont

- VDH partnership with UVM VCHIP program to promote developmental and autism screening by primary care pediatricians, following national guidelines.
- VT Child Development Clinic experience—many referrals document both developmental and ASD screens by PCP
- Expanded screening in child care, Head Start
Diagnosing ASD—Identifying Core Symptoms

- CDC: “The diagnosis of ASD at age 2 can be reliable, valid, and stable”
- Essential to begin early intervention before diagnosis.
- Collaborative, multidisciplinary diagnostic process
- Review of medical, EI, child care information, evaluations, services
- Family health and developmental history
- Extended interview with family about health and developmental history, including regression. Use of ASD-specific questions, such as ADI-R/SCQ
- Observation of child with parent/family
- Comprehensive Developmental Assessment (Bayley-3; DAS-2; others)
- Standardized play observation with ADOS-2
- Use of DSM5 diagnostic criteria
Diagnosing ASD

**Examination** to identify other health concerns or genetic signs

**Audiology** with experienced pediatric audiologist who can try many strategies to test

**Genetics Consultation:** For better anticipatory guidance, access to condition-specific family support, decrease feelings of parental guilt, specific management guidelines, if available, access to specific treatments, if applicable

**Studies not recommended as routine:**

- Imaging studies (MRI) will require sedation
- EEG
- Hair analysis, stool studies, yeast cultures, allergy testing, immunology, micronutrient assays
Diagnosing ASD—Identifying Related Concerns

- Other developmental disorders/delays/ID
- Anxiety
- ADHD
- Motor coordination/planning issues/tiptoeing
- Seizures (25 – 40 %)
- Nutrition/Eating behaviors
- Gastrointestinal symptoms/continence
- Sleep disturbances (40 – 80 %)
- Irritability/self-injury/aggression/tantrums
- Sensory/Atypical expression of pain
- Safety, Wandering (30 – 40 %)
Sleep disturbance in ASD

- Most frequent behavioral concern for parents
- May reflect ASD core symptoms, anxiety, poor self-regulation, inflexibility, sensory
- Other associations, not specific to ASD: obstructive sleep apnea; restless leg
- Sleep disorder may affect onset, maintenance, or early awakening
- Concern for safety, wandering at night
- Rule out pain, reflux.
- Treatment: Sleep “hygiene” training; consider medication, esp Melatonin

Seizures in ASD

- No one predominant type; onset in preschool, and adolescence
- More common in girls and in children with ID
- Staring spells can look like seizures—smart phone videos helpful. Ask parent if he responds to touch (he may not respond to being called)
- EEG not indicated if no seizure behavior
Diagnosing ASD in Vermont

- VT Best Practices paper: Dx by clinicians with specialized training in dx of ASD in children, using standardized tools for ASD (ADOS, ADIR)

- Resources: VDH Child Development Clinic teams include developmental pediatrician, MSW, neuropsychologist, psychologist, doctorate-level PNP and EdD/OTR, in each AHS district

- UVM VCCYF Autism Assessment Clinic, child psychiatrist, SLP, psychologist
Treatment: Start Early Intervention Early

- Begin EI as soon as ASD is suspected—do not wait for diagnosis
- Address child’s specific behaviors at a developmentally appropriate level.
- Intensive, as tolerated
- Involving parents, other caregivers.
- Strategies to apply learned skills to new environments and situations (generalization)...
- Promotion of opportunities for interaction with typically developing peers...

Treatment: Early intervention targeting core symptoms

- Emphasis on Social Communication: developing language by 5-6 portends best social outcomes, and joint attention predicts language—pointing to share, engagement, reciprocal back and forth, beginning with recognizing caregiver voice and smiling, following gaze shift (8 months), following a point (10-12 months), pointing to request (12 months), pointing to show (14 – 16 months)

- Children who cannot communicate often have challenging behaviors—give children access to communication! Visual. Basic signing. Consider augmentative and alternative communication for minimally verbal children—children who have a speech-generating device developed more spoken language.

- E.g. More Than Words Hanen Centre. Parent program, for young children, to teach reciprocal social communication, imitation, play skills
Theory of Mind

This is Sally.

Sally has a basket.

This is Anne.

Anne has a box.

Sally has a marble. She puts the marble into her basket.

Sally goes out for a walk.

Anne takes the marble out of the basket and puts it into the box.

Now Sally comes back. She wants to play with her marble.

Where will Sally look for her marble?
Intervention strategies: ABA

Applied Behavioral Analysis (ABA)

- Based on work of Skinner (1930’s); Lovaas (1980’s). A method of teaching that uses reinforcement to motivate and shape a desired behavior. Begins with a Functional Behavioral Analysis and plan. Requires skilled, certified behavior interventionist.
- A: Antecedent, such as a request that precedes the behavior.
- B: Child’s desired response.
- C: Consequence, positive reinforcer.
- Teaching by repeated trials and rewards, and practice in a variety of settings.
Other ABA-based Interventions

- Verbal Behavior Milestones Assessment and Placement Program (VBMAPP) Mark Sunderberg. Focus on language and social skills

- Pivotal Response Therapy: uses ABA approach to teach overarching, pivotal behaviors (motivation, initiation of communication)

- Early Start Denver Model (ESDM) Sally Rogers, uses ABA principles and PRT to teach communication, cognitive and language skills to children as young as 12 months—non-verbal communication, joint attention, verbal communication, social behaviors, play, imitation. 20-25 hours a week. Emotionally positive (fun!), parents as colleagues, woven into child’s entire day
Relationship-based Interventions

- Developmental, Individual-Differences, Relationship-based/Floortime. Greenspan and Wieder. Based on belief that child’s emotional development is the basis for her learning. Parent/therapist meets child at her level, follows her lead to engage her.

- RDI (Relationship Development Intervention) Parents work with an RDI-certified consultant to teach their child how to think flexibly and engage in social relationships beginning by helping children develop relationships with their parents and other family members.

- SCERTS Prizant & Wetherby Emphasizes joint attention, symbolic behavior, gestures, emotional regulation
Roles for collaboration with specific therapies

- Speech and Language Pathologist—not just “speech” but all forms of communication and pragmatic language; social skills, use of social stories, groups, video modeling
- Occupational Therapist—motor planning, fine motor, sensory aversions and sensory-seeking behaviors, often including sensory integration principles
Medical therapies

Medication management of pervasive and difficult symptoms

- anxiety, depression, irritability, impulsivity, aggression, self-harm, ADHD, repetitive behaviors, rigidity and refusals, sleep disturbance
- often diagnosed and managed initially by child psychiatrist; newer uses for medications with potential side effects and reactions
- SSRIs for anxiety, repetitive behaviors, depression
- Stimulants and non-stimulants for ADHD
- Alpha-2-agonists for aggression, explosive outbursts, self-injurious behaviors, tics, inattention, sleep
- Atypical antipsychotics for aggression and agitation
Complementary/Alternative Medicine CAM

Complementary/Alternative Medicine

- Consider evidence for benefit (is it effective?), and for harm (is it safe?)
- Know and measure target symptoms
- Consider broad scope for harm (nutritional imbalance/deficiencies; out of pocket cost; access/convenience; unnecessary and expensive tests; untested and unregulated micronutrient supplements; unknown interactions)

Avoid: Chelation; Hyperbaric Oxygen; Secretin
Early Intervention Resources in Vermont

- CIS-EI statewide begins EI evaluation and services before dx of ASD; after dx, intensifies
- Several CIS-EI areas have autism consultants
- UVM I-Team expanded eligibility to children < age 3
- Some areas have ASD-specific self-contained programs
- CIS-EI collaboration with day care programs in their region
- Early Head Start
- Availability of ABA
- Developmental and Mental Health Designated Agencies
Other supports for families and children

- TEFRA option for Medicaid coverage/Katie Beckett program
- Supplemental Security Income
- Developmental Services Designated Agencies, family supports
- Vermont Family Network
- CSHN Medical Social Worker
Care Coordination within a Statewide System of Care

- Co-located in Health Dept District offices
- Support and resource to families with CYSHN
- Member of regional CIS team
- Collaborate with regional IFS and Blueprint teams
- Liaison to Medical Homes/SW rounds
- Member of CSHN-hosted CDC team
- Liaison to health system Specialty Clinic teams
- The Vermont Blueprint for Health (Medical Home initiative) is a vision, a plan, and a statewide partnership to improve health and the health care system for Vermonters. The goals are: 1) To implement a statewide system of care that enables Vermonters with, and at risk of, chronic disease to lead healthier lives; 2) To develop a system of care that is financially sustainable; and 3) To forge a public-private partnership to develop and sustain the new system of care.

Children’s Integrated Services (CIS) is a statewide resource for pregnant or postpartum women and families with children from birth to age six. Includes: Early Intervention, Nursing & Family Support, and Children’s Mental Health.
Life course for children with ASD

- “Symptoms of ASD vary greatly from one child to the next. While it is possible for some children—studies range from 3% to 25%—to improve to the point where they no longer meet diagnostic criteria, most continue to have some degree of developmental or behavioral symptoms.”

- Symptoms that may decrease over time include stereotypies and difficulties with eye contact

- More persistent symptoms may be difficulties with social reciprocity, pragmatic language, and restrictive interests/repetitive behaviors

- Research in this area is very new and covers the years in which the definitions and prevalence of ASD expanded greatly, as well as the earlier age of diagnosis and initiation of effective interventions.