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**SOUTH DAKOTA**  
SANFORD SCHOOL OF MEDICINE

**Autism Spectrum Disorder:**  
**Understanding Evaluation and Diagnosis**

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# Autism Prevalence

- About 1 percent of the world population has autism spectrum disorder. ([CDC, 2014](#))
- Prevalence in the United States is estimated at 1 in 59 births. ([CDC, 2018](#))
- Prevalence of autism in U.S. children increased by 119.4 percent from 2000 (1 in 150) to 2010 (1 in 68). ([CDC, 2014](#)) Autism is the fastest-growing developmental disability. ([CDC, 2008](#))
- Prevalence has increased by 6-15 percent each year from 2002 to 2010. (Based on biennial numbers from the [CDC](#))

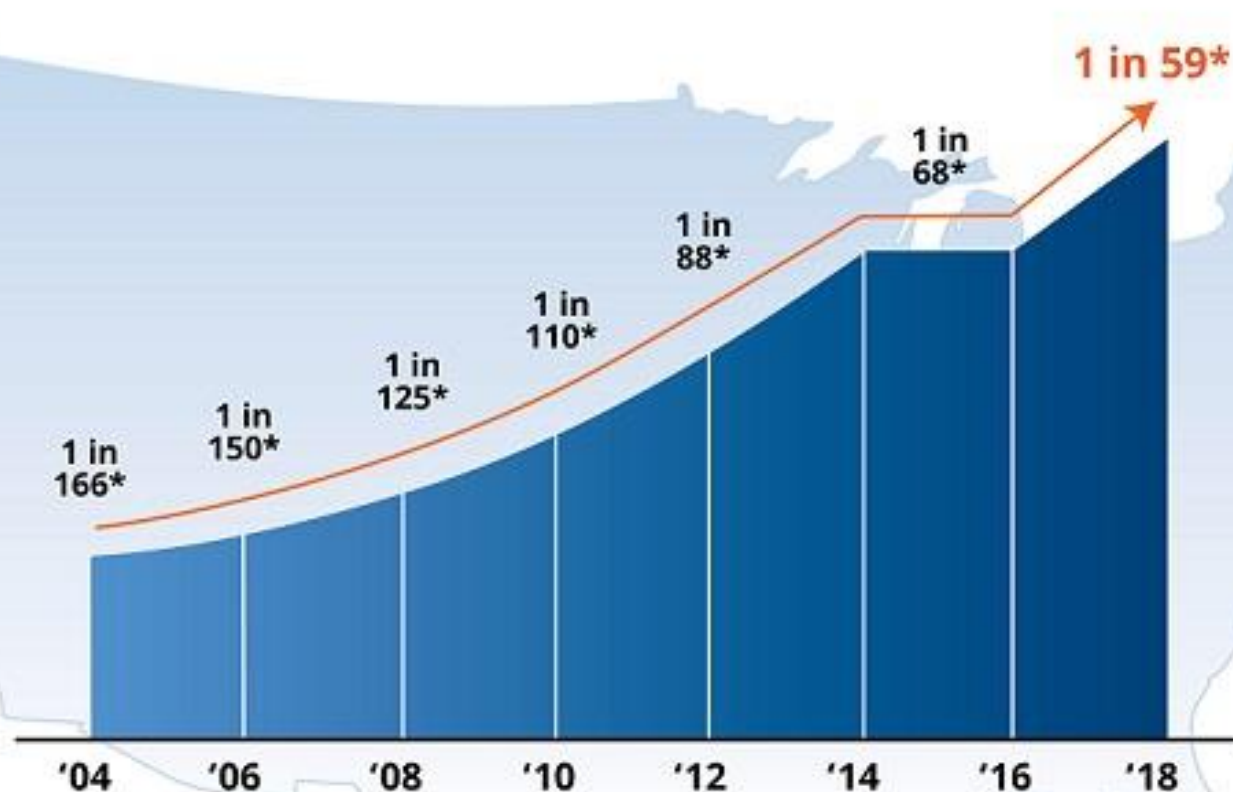


# Autism: Financial Costs

- Autism services cost U.S. citizens \$236-262 billion annually. ([Buescher et al., 2014](#))
- Most costs are in adult services – \$175-196 billion, compared to \$61-66 billion for children. ([Buescher et al., 2014](#))
- Cost of lifelong care can be reduced by 2/3 with early diagnosis and intervention. (Autism. 2007 Sep;11(5):453-63;
- The U.S. cost of autism over the lifespan is about \$2.4 million for a person with an intellectual disability, or \$1.4 million for a person without intellectual disability. ([Buescher et al., 2014](#))



# Estimated Autism Prevalence 2018



\* Centers for Disease Control and Prevention (CDC) prevalence estimates are for 4 years prior to the report date (e.g. 2018 figures are from 2014)

# What do the latest US Prevalence Rates Mean?

- Data reflect 11 sites in the United States
- Based on 8 year olds from these 11 sites in 2014
- The number of children identified with ASD varied widely by community, from 1 in 65 children in Arkansas to 1 in 34 children in New Jersey
- More children being identified with ASD
- Earlier age of diagnosis (4+ years old)
- About 50% of children with ASD have IQ's at/or above average



# Autism Spectrum Disorder

- A neurodevelopmental disorder that causes changes in the brain resulting in:
  - Severe qualitative impairment in:
    - Spontaneous, functional, and social communication (verbal and/or nonverbal communication)
  - Restricted range of activities and interests
- Onset by the age of 3 years
- Defies generalization
  - Symptom presentation severity vary greatly



# Gender and Autism

- Ratio of boys to girls: 4 to 1
  - Boys: 1 in 37
  - Girls: 1 in 151
- Why?
  - Theories, but no conclusive findings
    - Protective factors in females
    - Response to treatment
    - Different symptom presentation



# ? Causes of ASD ?

- a neurodevelopmental disorder caused by specific parts of the brain not functioning as we would expect
- Many factors are being studied to identify genetic and environmental factors
- Autism rates:
  - In the general population, about 1%
  - In a family that already has 1 child with autism, odds of having another are up to 18%
  - Among identical twins, if one has autism, odds that the other will also have autism are 70%-95%

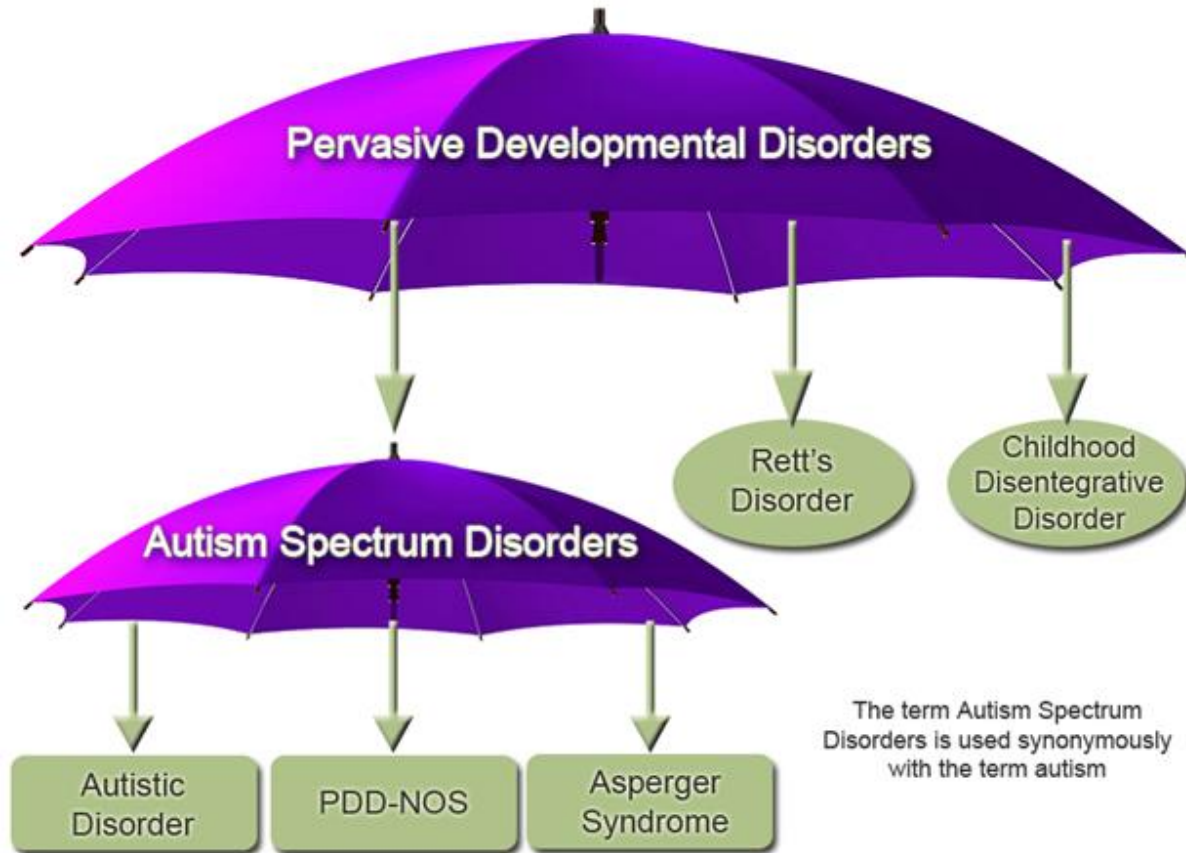




# Is there an autism epidemic?

- Some contributing factors for increased rates:
  - Definition of autism has been broadened (1994)
  - Children can now be diagnosed with more than one developmental disorder
  - In the past, children with intellectual disability were not evaluated for autism
  - We've become better at recognizing symptoms
  - Targeted efforts for screening and evaluation

# The “Spectrum”



# DSM-5: Why Change?

- Issues with consistency diagnosis applied
  - Good sensitivity
  - Poor specificity
- Better reflection of current state of knowledge about pathology and clinical presentation
- Account for variability of symptoms:
  - Onset
  - Presentation
  - Change over time



# Concerns over DSM-IV

- Validity of the Pervasive Developmental Disorders category
- Consistency of some diagnoses (e.g., high -functioning autistic disorder vs. Asperger)
- Appropriateness of the use of certain diagnoses (e.g., PDD-NOS as mild neurodevelopmental disorder, Asperger as “odd” behaviors)
- Validity of some diagnoses (e.g., childhood disintegrative disorder)



# Rett Syndrome

- Deletion of Rett Syndrome as a specific ASD
- Rett was removed as a separate disorder
- ASD behaviors are not particularly salient in Rett Syndrome except for brief period during development.
- ASD are defined by specific sets of behaviors, not etiologies (at present) so inclusion of Rett Disorder is atypical.
- Patients with Rett Syndrome who have ASD symptoms can still be described as having ASD, and clinicians should use the specifier “with known genetic or medical condition”



# Deletion of Childhood Disintegrative Disorder

- New knowledge that developmental regression in ASD is a continuous variable, with wide range in timing and nature of the loss of skills, as well as the developmental milestones that are reached prior to regression
- Rarity of CDD diagnosis makes systematic evaluation difficult, but review of accumulated world's literature shows that CDD has
- important differences from other ASD's, including the acuity and severity of regression, as well as co-occurring physical symptoms, such as loss of bowel and bladder control. (Need to look for neurological disorder)

# DSM-5 Criteria Changes

1. Merging Autistic Disorder, Asperger's Disorder, PDD-NOS into single disorder:

## Autism Spectrum Disorder

2. Combined Communication and Socialization criteria into one criterion: Social Communication

3. Symptoms must be present prior to age 3 (may not fully manifest until social demands exceed limited capacity)

4. Including a number of specifiers:

(i.e. age of onset, type of onset, level of functioning, level of support needed)

5. May be diagnosed with other disorders (ADHD, anxiety, etc.)



# Other DSM-5 Changes

- Must now have 2 in RRB
- Can be by history or current
- Added sensory issues
- New specificity





# DSM-5 Criteria Changes

Table 3

Neurodevelopmental disorders in DSM-IV and DSM-5

<b>DSM-IV</b>	<b>DSM-5</b>
Autistic disorder; Asperger disorder; childhood disintegrative disorder; and pervasive developmental disorder, not otherwise specified	Autism spectrum disorder
No equivalent DSM-IV diagnosis	Social communication disorder
Rett disorder	Not included in DSM-5
Mental retardation	Intellectual disability
Reading disorder	Dyslexia
Mathematics disorder	Dyscalculia
Disorder of written expression and learning disorder, not otherwise specified	Unspecified learning disorder
Expressive language disorder	Language impairment, late language emergence, or specific language impairment
Mixed receptive-expressive language disorder	No equivalent DSM-5 diagnosis
Phonological disorder	Speech sound disorder
Stuttering	Childhood-onset fluency disorder



# Autism Spectrum Disorders in the DSM-5

Must meet criteria A, B, C, D, & E

**A.** Persistent deficits in social communication and social interaction across multiple contexts, as manifest by the following, currently or by history (examples illustrative, not exhaustive):

-Deficits in social-emotional reciprocity (which may range, for example, from abnormal social approach and failure of normal back and forth conversation; to reduced sharing of interests, emotions, or affect; to failure to initiate or respond to social interactions

-Deficits in nonverbal communicative behaviors used for social interaction poorly integrated verbal and nonverbal communication; to abnormalities in eye contact and body-language or deficits in understanding and use of gestures; to total lack of facial expressions and nonverbal communication

-Deficits in developing, maintaining, and understanding relationships (abnormal social approach and failure of normal back and forth conversation; to reduced sharing of interests, emotions, or affect; to failure to initiate or respond to social interactions)



# Autism Spectrum Disorders in the DSM-5

**B.** Restricted, repetitive patterns of behavior, interests, or activities as manifested by at least two of the following, currently or by history (examples illustrative, not exhaustive):

- Stereotyped or repetitive motor movements, use of objects, or speech
- Insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behavior (extreme distress at small changes, difficulties with transitions, rigid thinking patterns, greeting rituals, need to take same route or eat same food every day)
- Highly restricted, fixated interests that are abnormal in intensity or focus (strong attachment to or preoccupation with unusual objects, excessively circumscribed or perseverative interests)
- Hyper- or hypo-reactivity to sensory input or unusual interest in sensory aspects of the environment (apparent indifference to pain/temperature, adverse response to specific sounds or textures, excessive smelling or touching of objects, visual fascination with lights or movement)



# Autism Spectrum Disorders in the DSM-5

C. Symptoms must be present in early developmental period (but may not become fully manifest until social demands exceed limited capacities, or may be masked by learned strategies in later life)

D. Symptoms cause clinically significant impairment in social, occupational, or other important areas of current functioning

E. These disturbances not better explained by intellectual disability or global developmental delays. Intellectual disabilities and autism spectrum disorder frequently co-occur, to make comorbid diagnoses of autism spectrum disorder and intellectual disability, social communication should be below that expected for general developmental level.

<b>Dimensional Ratings for DSM V ASD</b>	<b>Social Communication</b>	<b>Fixated Interests and Repetitive Behaviors</b>
<b>Requires very substantial support</b>	<b>Severe deficits in verbal and nonverbal. Very limited initiation of social interactions and minimal response to overtures.</b>	<b>Inflexibility of behavior, extreme difficulty coping with change, RRBs that markedly interfere in all spheres. Great Distress</b>
<b>Requires substantial support</b>	<b>Marked deficits with limited initiations and reduced or atypical responses. Impairment apparent even with supports in place.</b>	<b>Inflexible in behavior, difficulty coping with change, frequent RRBs and interfere in a variety of contexts. Some distress.</b>
<b>Requires support</b>	<b>With or without supports, noticeable impairments. Difficulty initiating social interactions and clear atypical responses. Maybe decrease social interest.</b>	<b>Behavioral inflexibility causes significant interference in one or more contexts. Trouble switching. Problems organizing and planning.</b>

# What's Missing From ASD Levels of Support?

- What type of “support”?
  - An aide; personal care assistant; 1:1 school aide; A therapist; job coach?
- In which situations do people at various levels require “support?”
- How much, and how often are supports needed?



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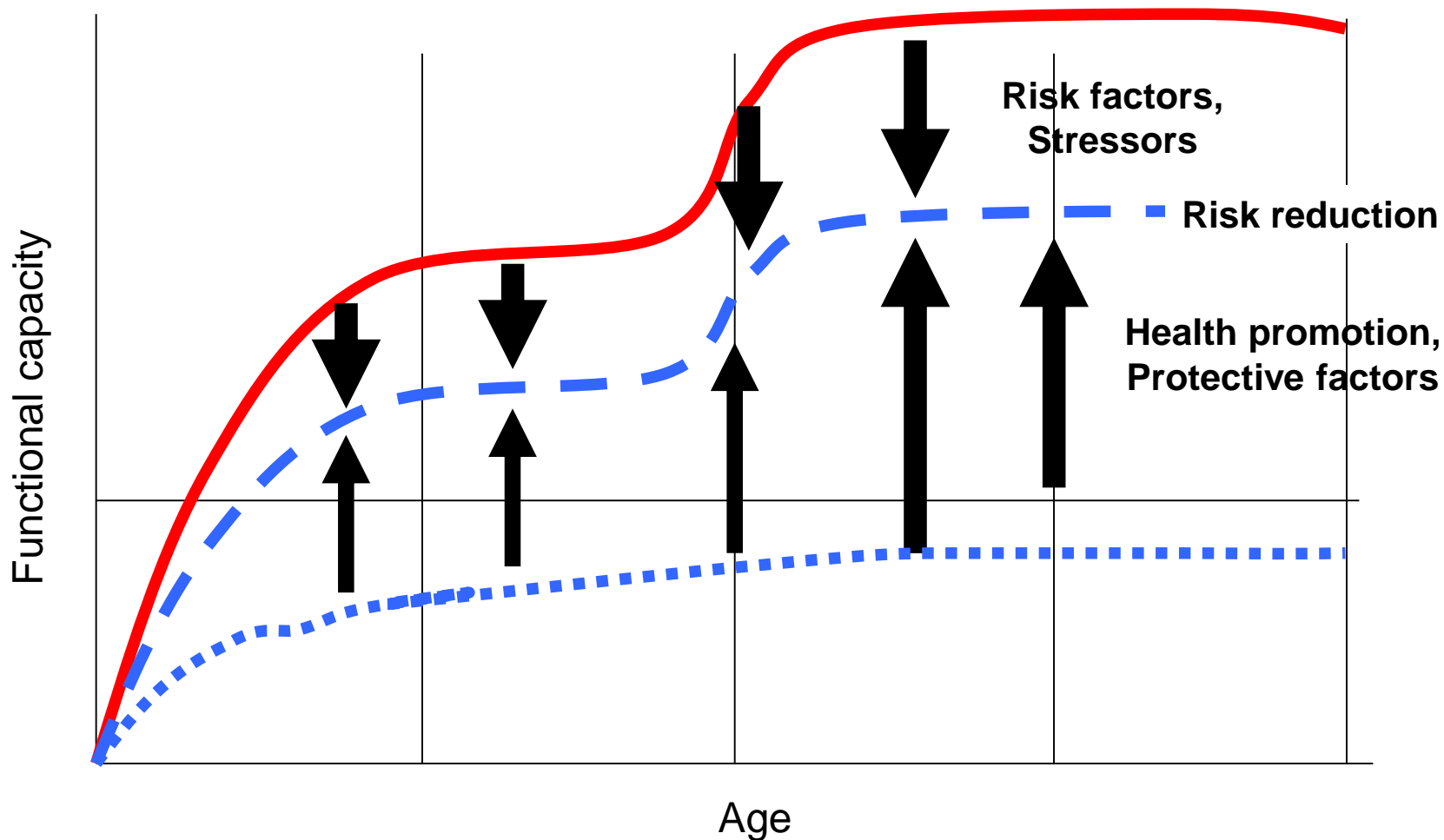
# Effective Screening Practices

**30%**: Children screened for autism and other developmental delays in the past year (percent, 10–35 months)

Data Source: National Survey of Children's Health (NSCH); Health Resources and Services Administration, Maternal and Child Health Bureau, and Centers for Disease Control and Prevention, National Center for Health Statistics (HRSA/MCHB and CDC/NCHS)



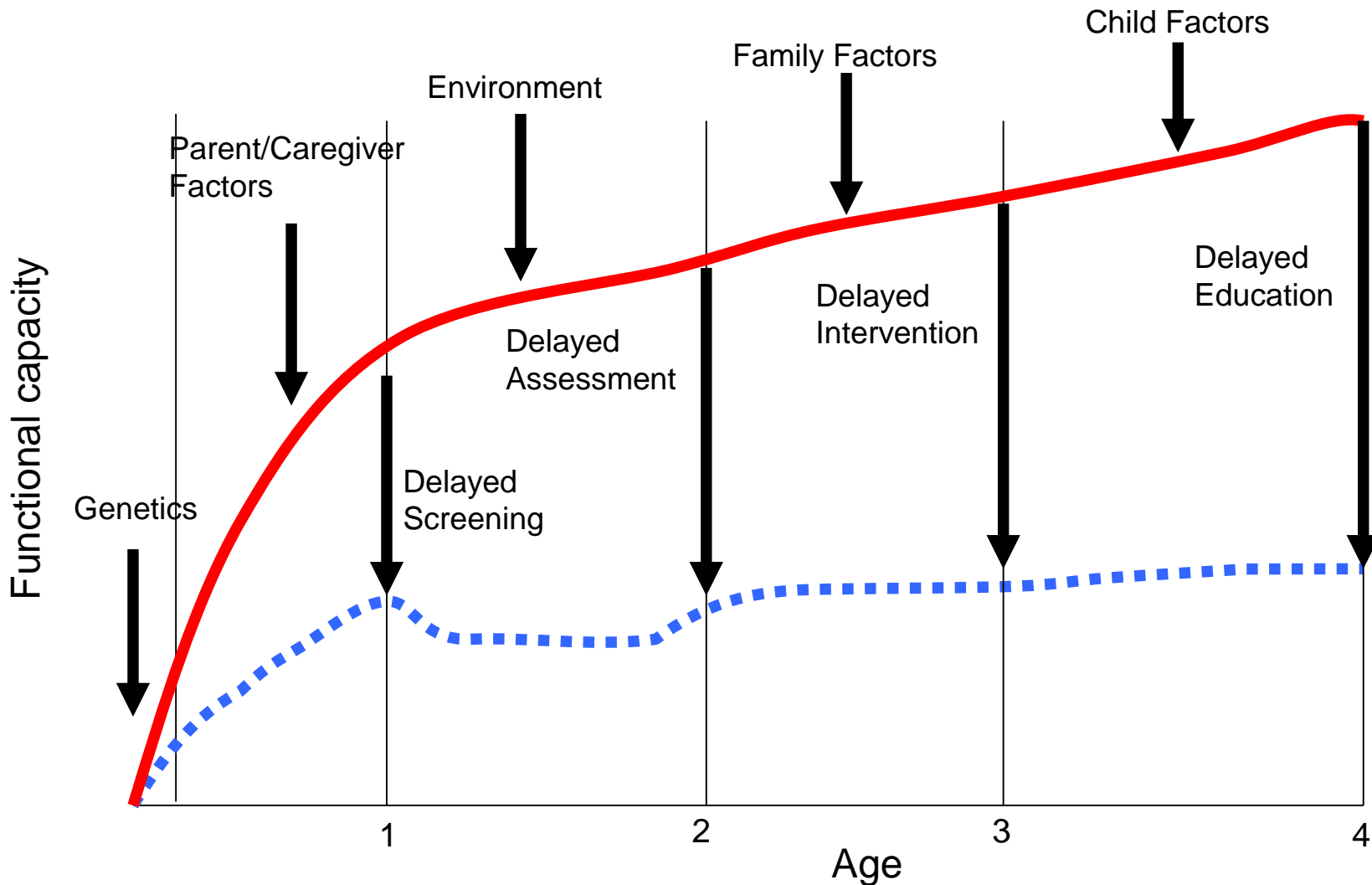
# Autism and the Life Course







# Autism, Early Childhood, Negative Factors





# Screening

- Use of a brief, objective, and validated instrument
- Goal to help differentiate children that are “probably ok” vs.. those “needing additional investigation”- those with *unsuspected* deviations from normal
- Performed at a set point in time
  - 9, 18,(24), 30 months
- Objective vs. subjective impressions
- Results always interpreted in context
  - Never in isolation
  - Aid to ongoing surveillance

# Benefits of Screening

**Assists in sorting children into 3 categories:**

- **Needs additional evaluation** - Did not pass screen
- **Needs close monitoring/surveillance**- Passed screen but has risk factors
- **Needs ongoing monitoring in the context of well-child care** - Passed screen and has no known risk factors



# Red Flags

## A person with ASD might:

- Not respond to their name by 12 months of age
- Not point at objects to show interest (point at an airplane flying over) by 14 months
- Not play "pretend" games (pretend to "feed" a doll) by 18 months
- Avoid eye contact and want to be alone
- Have trouble understanding other people's feelings or talking about their own feelings
- Have delayed speech and language skills



# Red Flags (continued)

- Repeat words or phrases over and over (echolalia)
- Give unrelated answers to questions
- Get upset by minor changes
- Have obsessive interests
- Flap their hands, rock their body, or spin in circles
- Have unusual reactions to the way things sound, smell, taste, look, or feel



# Screening Tools

- [Agnes and Stages Questionnaires \(ASQ\)](#)  
This is a general developmental screening tool. Parent-completed questionnaire; series of 19 age-specific questionnaires screening communication, gross motor, fine motor, problem-solving, and personal adaptive skills; results in a pass/fail score for domains.
- [Communication and Symbolic Behavior Scales \(CSBS\)](#)  
Standardized tool for screening of communication and symbolic abilities up to the 24-month level; the Infant Toddler Checklist is a 1-page, parent-completed screening tool.
- [Parents' Evaluation of Developmental Status \(PEDS\)](#)  
This is a general developmental screening tool. Parent-interview form; screens for developmental and behavioral problems needing further evaluation; single response form used for all ages; may be useful as a surveillance tool.
- [Modified Checklist for Autism in Toddlers \(MCHAT\)](#)  
Parent-completed questionnaire designed to identify children at risk for autism in the general population.
- [Screening Tool for Autism in Toddlers and Young Children \(STAT\)](#)  
This is an interactive screening tool designed for children when developmental concerns are suspected. It consists of 12 activities assessing play, communication, and imitation skills and takes 20 minutes to administer.

# Why does ASD need to be identified as soon as possible?

- The only proven treatment is intensive behavioral and early communication intervention.
- The neuro-developmental window for social and communication development closes very rapidly.
- Co-occurring conditions can be identified and treated.
- All forms of autism can be recognized early and reliably diagnosed by age 3.



# Co-occurring Conditions

- Epilepsy
- GI issues
- Allergies/Intolerances
- Feeding issues
- Sleep disturbances
- ADHD
- Anxiety disorders
- Depression
- Schizophrenia
- Bipolar disorder
- Genetic disorders





# Diagnosis

- diagnosis at age 2 can be reliable, valid, and stable.
- most children are not diagnosed until after age 4 years.
- Goal is to have all children diagnosed by 36 months



# Diagnosis

- By age 3 years, only 44% of children identified with concerns had received a comprehensive evaluation, despite documentation of developmental concerns among 89% of the children.
- By age 8 years, 20% of children with concerns have not received a documented ASD diagnosis or special education classification



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# Comprehensive Evaluation

- No single test can be used in determining the diagnosis or educational need.
- Consider of the child's developmental level and primary mode of communication,
- Must assess skills of everyday activities and settings (functional)
- Helps to put a context around the symptoms or problems
- Assists in “ruling in” or “ruling out” potential contributing factors
- Must be administered by trained professionals.



# Comprehensive Evaluation

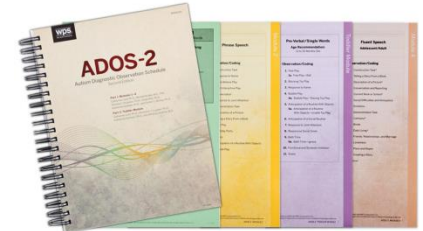
- Multi-Method:
  - Social, medical, developmental history
  - Parent/caregiver interview
  - Direct observations
  - Standardized tests
  - Informal assessments
- Multi Source:
  - parents, child, educators, other providers
- Multi-Setting:
  - Home, daycare, school, other



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# Domains to Assess

- Developmental, Health, Social History
- Cognitive Ability
- Adaptive Behavior
- Social Communication & Speech/Language
- Social Skills
- Emotional/ Behavioral status
- Sensory Processing and Motor
- Autism Specific Measures (ADOS-2, ADI-R)
- Other areas such genetic testing, neurological testing, and other medical testing, nutrition, as indicated





# Interdisciplinary Perspective

- Best practice = Interdisciplinary
- Involved disciplines:
  - FAMILY
  - psychologist
  - speech language pathologist
  - occupational therapist
  - psychiatrist, pediatrician, or primary care provider
  - others including, dietician/nutritionist, audiologist, counselor, social worker, physical therapist, neurologist, geneticist,



# Data-based Decision Making

- Recommendations made must be based from assessment data
- Appropriate inferences made from test results can only be accomplished if the instrument is both **RELIABLE** and **VALID** for the purposes and population used



# Data-based Decision Making

- Reliability – Consistency of measurement
  - Interrater reliability - degree of agreement between two observers
  - Test-retest reliability - degree to which scores are similar for a person being observed twice or taking the same test twice
  - Internal consistency reliability - examines the degree to which items on a test are related (split-half)





# Data-based Decision Making

- Validity
  - Content validity – degree that a measure adequately samples the domain of interest
  - Criterion-related validity – degree to which a measure is associated in an expected way with another measure (the criterion)
  - Construct validity – degree to which a measure of a construct is supported by other measures of that construct



# Data-based Decision Making

- **Sensitivity** measures how often a test correctly generates a positive result for people who have the condition that's being tested for (also known as the “true positive” rate).
- **Specificity** measures a test's ability to correctly generate a *negative* result for people who *don't* have the condition that's being tested for (also known as the “true negative” rate).



# Statewide Response

- Partnership with the SD Department of Education-Office of Special Education and Black Hills Special Services
- Goal: Build capacity across the state to enable specialized evaluation teams in local schools to provide comprehensive Autism Spectrum Disorder evaluations



# Statewide Response

- Level 1:
  - Initial Team Training on Comprehensive Autism Evaluations (Including Full ADOS Training)
- Level 2:
  - Phase 1: Community of Practice and Ongoing Training and Technical Assistance (establish interrater reliability)
  - Phase 2: Follow-up training for teams who've completed Level 1
- Refresher courses available



# Statewide Response

- Since 2015 over 200 professionals from over 30 local school districts and educational cooperatives have been trained.
- Ongoing technical assistance and refresher trainings provided for sustainability
- Surveys of participants report significant growth in knowledge, skills, and confidence, as well as satisfaction (at or near 100% strongly agree or agree)
- Additional survey data shows need for additional practice and community of support



# Prognosis

- Although autism is a life-long disorder, the prognosis is much better than in the past
  - For many on the autism spectrum, best outcome could include full-time job, living independently, and meaningful relationships
- No miracle cures, but intervention can greatly improve outcomes
  - Challenge: Huge increase in referrals without proportional increase in resources



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# Evidence –Based Practice for Autism Spectrum Disorders

- Expectation of recommending and implementing treatments based on sufficient research findings
- A thorough review of the treatment literature required to determine effectiveness
- National Professional Development Center on Autism:  
<http://autismpdc.fpg.unc.edu/content/briefs>
- National Autism Center  
<http://www.nationalautismcenter.org>



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