Special Topic:
Autism Spectrum Disorder
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OUTLINE:

- Characterization and Prevalence
- Symptoms
- Diagnosis
- Possible Causes
- Treatment and Therapy
- Fear That Vaccines Cause Autism
Characterization and Prevalence

- Autism spectrum disorders (ASDs)
  - Autism
  - Asperger syndrome
  - Pervasive developmental disorder not otherwise specified (PDD-NOS)
Characterization and Prevalence

- General characteristics of ASDs
  - Neurodevelopmental disorders
    - Impaired communication skills
    - Poor social skills
    - Unusual behaviors
  - Symptoms begin in early childhood and last a lifetime
Characterization and Prevalence

- Prevalence
  - CDC estimates 1 in 110 children in the U.S. has an ASD
  - Four times more common in males
  - Estimates of prevalence have increased in recent decades
    - True increase in number of cases?
    - Increased awareness and recognition?
    - Improved and expanding diagnostic criteria?
Characterization and Prevalence

- History
  - Autism first described as a disorder in 1943 by Leo Kanner
  - Asperger syndrome first described by Hans Asperger in 1944
  - PDD-NOS formally defined in 1987
Characterization and Prevalence

- *Diagnostic and Statistical Manual of Mental Disorders (DSM)*
  - Handbook used by psychiatrists and psychologists to classify mental disorders
  - Published and regularly updated by the American Psychiatric Association
Characterization and Prevalence

- ASD symptoms
  - Marked deficits in communication
  - Poor social skills
  - Performance of unusual and repetitive motor patterns
- Within autism, Asperger syndrome, and PDD-NOS, there is tremendous variation in the presence and intensity of symptoms
Figure 18a.1 Autism spectrum disorder (ASD) is characterized by deficits in social communication and interaction and by the performance of unusual behaviors.

(a) Failure to make eye contact is an example of an impairment in nonverbal communication that may occur in someone with ASD.

(b) Social withdrawal occurs in some people with ASD.

(c) Restricted and repetitive behaviors are also common in people with ASD. Here, an autistic teen continually touches his ears.
Characterization and Prevalence

- There is considerable overlap in diagnostic criteria for autism and Asperger syndrome, with some differences
  - Some with autism are intellectually challenged
  - Some with Asperger syndrome are intellectually gifted
  - Some with autism never speak
  - Some with Asperger syndrome speak at length
Characterization and Prevalence

- Some diagnostic criteria differ
  - Autism
    - Clinically significant delay in language or no speech at all
  - Asperger syndrome
    - No clinical delay in language or cognitive development
Table 18a.1 Diagnostic Criteria for Autism Spectrum Disorder Proposed for *DSM-5*

<table>
<thead>
<tr>
<th>Criteria</th>
<th>Deficits and Symptoms</th>
</tr>
</thead>
</table>
| A. Persistent deficits in social communication and social interaction across contexts, not accounted for by general developmental delays | 1. Deficits in social-emotional reciprocity  
2. Deficits in nonverbal communicative behaviors used for social interaction  
3. Deficits in developing and maintaining relationships appropriate to developmental level (beyond those with caregivers) |
| B. Restricted, repetitive patterns of behavior, interests, or activities | 1. Stereotyped or repetitive speech, motor movements, or use of objects  
2. Excessive adherence to routines, ritualized patterns of verbal or nonverbal behavior, or excessive resistance to change  
3. Highly restricted, fixated interests that are abnormal in intensity or focus  
4. Hyper- or hypo-reactivity to sensory input or unusual interest in sensory aspects of the environment |
| C. Symptoms must be present in early childhood (but may not become fully manifest until social demands exceed limited capacities) | |
| D. Symptoms together limit and impair everyday functioning | |

*Modified from Proposed Revision to *DSM-5*, January 26, 2011.*
Verbal communication

- Skills vary dramatically
  - 40% do not speak at all
  - 25% use some words and then lose them between 15 to 24 months of age
- Some exhibit delays in speaking
- Some develop speech at the appropriate time but speak with impaired use of language and poor social skills
Characterization and Prevalence

- Nonverbal communication
  - Using and understanding nonverbal communication is problematic
    - Facial expression disconnected from words
    - Limited or no eye contact
    - Poor understanding of body language of others
Characterization and Prevalence

- Social skills
  - Impairments in communication often associated with impairments in social skills
  - Inability to imitate the actions of others
    - Major disadvantage because this is how young children learn to speak and act in social situations
  - Inability to cope with change
Characterization and Prevalence

- Other behavioral conditions frequently found in children with ASDs
  - Impulsivity
  - Hyperactivity
  - Aggressiveness
  - Self-injurious behavior
  - Atypical responses to sensory stimuli
    - May overreact or underreact
Characterization and Prevalence

- Motor patterns
  - Repetitive movements (self-stimulatory activities)
    - Hand flapping
    - Rocking
    - Head banging
  - Unusual rituals
  - All-consuming interests
Characterization and Prevalence

- **Timing of symptom development**
  - Underlying neurological defects are thought to occur early in embryonic development
  - Symptoms usually become apparent in first few years after birth
    - 30% to 50% of parents notice problems during the first year
    - 80% to 90% of parents notice problems by 2 years
Diagnosis

- Diagnostic criteria have changed several times in recent decades
  - Changes reflected in multiple editions of the DSM
  - Example: in 1980, autism was formally differentiated from schizophrenia as a developmental disorder
Diagnosis

- Routine developmental screening of infants and young children occurs at their periodic physical examinations
  - Failure to meet age-specific developmental milestones may lead to further screening
    - Several diagnostic checklists and rating scales are available for diagnosing ASDs
  - Identifying developmental delays is important
    - Early diagnosis can lead to early intervention
Figure 18a.2 The pace at which a young child develops can be viewed in the context of age-specific developmental milestones.

(a) One milestone, typically attained by the end of the first year, is the ability to pull up to a standing position.

(b) The motor skills needed to build towers of blocks or other toys are usually attained after the first year.

(c) Some milestones concern social skills and behavior; these include showing independence and defiance.
Figure 18a.3 Routine developmental screening at periodic visits to the pediatrician can help identify developmental delays or losses.
Table 18a.2 Some Developmental Milestones Used to Assess a Young Child’s Progress in the Areas of Communication, Social Behavior, and Motor Skills

<table>
<thead>
<tr>
<th>Age</th>
<th>Developmental Milestones</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Communication</td>
</tr>
<tr>
<td>By the end of the</td>
<td>Babbles with changes in tone</td>
</tr>
<tr>
<td>first year</td>
<td>Tries to imitate words</td>
</tr>
<tr>
<td></td>
<td>Uses simple gestures and single words</td>
</tr>
<tr>
<td>By the end of the</td>
<td>Uses simple phrases and several single words</td>
</tr>
<tr>
<td>second year</td>
<td>Points to named objects</td>
</tr>
<tr>
<td></td>
<td>Repeats words heard in conversation</td>
</tr>
<tr>
<td>By the end of the</td>
<td>Recognizes and identifies common objects</td>
</tr>
<tr>
<td>third year</td>
<td>Uses sentences with four or five words</td>
</tr>
<tr>
<td></td>
<td>Speaks such that strangers can understand</td>
</tr>
<tr>
<td></td>
<td>most words</td>
</tr>
</tbody>
</table>

*More complete lists of developmental milestones can be found at www.cdc.gov/ncbddd/autism/actearly/screening.
Possible Causes

- Theories
  - Autism is caused by aberrant parenting
    - “Refrigerator Mother” hypothesis
    - Dispelled in 1960s
  - Autism is a neurodevelopmental disorder with a genetic basis
    - Probably multiple causes
    - Genes and environment both play a role
Possible Causes

- Two main types of studies indicate that autism has a strong genetic component
  - Family studies
  - Twin studies
Possible Causes

- Family studies
  - Higher than expected rates of autism among family members
    - Rate of recurrence of autism in siblings of autistic individuals is 2% to 6%
      - This rate is 10 to 60 times that in the general population
Possible Causes

- Twin studies
  - Monozygotic twins
    - Nearly genetically identical
    - If one monozygotic twin has autism, then there is a 75% chance that the other twin will have autism
  - Dizygotic twins
    - No more genetically similar than siblings who are not twins
    - If one dizygotic twin has autism, then there is a 3% chance that the other twin will have autism
Possible Causes

- Environmental risk factors
  - Prenatal exposure to certain drugs (thalidomide, valproic acid)
  - Maternal illness (rubella) during pregnancy
  - Drugs used to induce ovulation during infertility treatments?
Treatment and Therapy

- No cures for ASDs
- No single therapeutic drug is known to improve core symptoms
  - Oxytocin is currently being tested
- Educational interventions can help children reach their full potential
  - Individualized educational plan (IEP)
    - May include speech therapy, occupational therapy, sensory integration therapy
Fear That Vaccines Cause Autism Spectrum Disorder

- Vaccines
  - Prevent potentially deadly diseases (measles, mumps, rubella, polio, etc.)
- Immunization programs
  - Reduced or eradicated serious infectious diseases
  - Saved innumerable lives
Fear That Vaccines Cause Autism Spectrum Disorder

- Concerns about a possible link between certain vaccines and autism
  - MMR vaccine
  - Thimerosal-containing vaccines
    - Thimerosal
      - Mercury-containing compound used to prevent growth of microorganisms in certain vaccines
      - Reduced or eliminated from vaccines as a precautionary measure
Fear That Vaccines Cause Autism Spectrum Disorder

- Current scientific evidence does not support a causal association between:
  - The MMR vaccine and autism
  - Thimerosal-containing vaccines and autism