The Clinical Diagnosis of Autism:

Are We Over Diagnosing?

SIU MEDICINE
FORWARD. FOR YOU.

Pamela Campbell, M.D.

Associate Professor of Psychiatry

Division Chief of Child Psychiatry

SIU Neuroscience Institute

Southern Illinois University School of Medicine

Springfield, Illinois

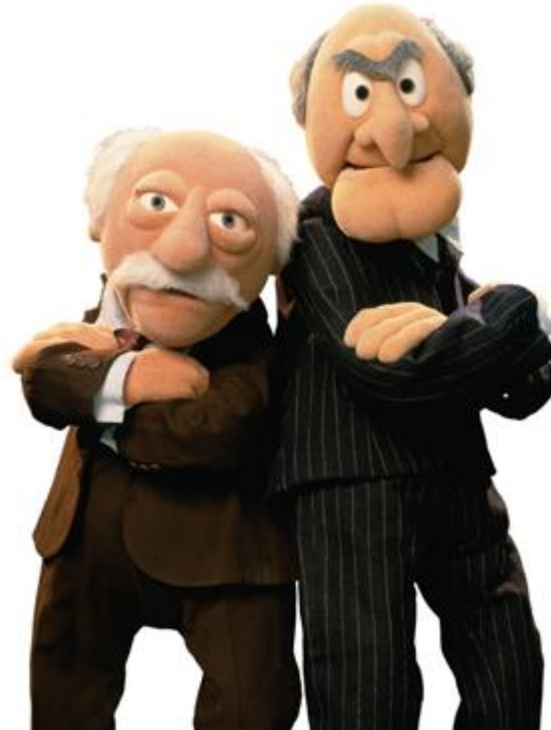
DISCLOSURES

- I have no financial disclosures that relate to this presentation.

GOALS

- Review the diagnostic history of Autism
- Discuss in detail the diagnostic criteria of autism and the possible overlap with other disorders
- Discuss pressures on clinicians in regards to diagnosing ASD.

NOT INTENDED TO BE CRITICAL



STATISTICS

- Prevalence of Autism
  - Kanner[1] estimated 150:20,000
  - 1966 estimated at 4-5:10,000 (1:1,000)
  - 1970 formal prevalence study found 4-5:10,000[2]

  - CDC Autism and Developmental Monitoring Network
    - 2002: 1:150
    - 2004: 1:125
    - 2006: 1:110
    - 2008: 1:88
    - 2010: 1:68 (14: 1,000)
    - 2012: 1:68

What Has Changed?

WHAT HAS CHANGED

1. Incidence of autism has increased.
2. Diagnosis of autism has increased.

HISTORICAL REVIEW

- Kanner[1] (1943)-early description of 11 children
  1. Inability to relate in ordinary way to people and situations from the beginning of life
  2. Failure to assume at any time an anticipatory posture preparatory to being picked up or adjust to fit the hold
  3. Lack of use of language to convey meaning to others
     a) Excellent rote memory
     b) Delayed echolalia
     c) Monotonously repetitious verbal utterances
  4. Literalness
  5. Personal pronouns are repeated just as heard
  6. Distressed by loud noises and moving objects
  7. Anxiously obsessive desire for the maintenance of sameness
  8. Limitation in the variety of spontaneous activity

HISTORICAL REVIEW

- ❖The term autism was borrowed from Bleuler who used it in the description of schizophrenics to mean withdrawn, social isolation
- ❖This suggested an overlap of the two disorders that caused confusion for many years
- ❖Until early 1970s, autism was frequently considered early onset schizophrenia

- ❖Asperger was describing a similar group of patients about a year later calling them autistic psychopathy

DIAGNOSTIC DEVELOPMENT

- Initially, the only category available in DSM I and II was childhood schizophrenia
- As usual in psychiatry, we were generally blaming the parents (refrigerator mothers)
- We finally agreed that autism was not schizophrenia, was multifactorial and was not the result of deviate parenting
- DSM III first introduced diagnostic category for autism

DSM III

- Introduced two new categories of autism:
  1. Infantile Autism (onset before age 30 months)
     a) Pervasive lack of responsiveness to people
     b) Gross deficits in language development
     c) Peculiar speech patterns if speech is present
     d) Bizarre responses to the environment
     e) Absence of hallucinations/delusions/loose associations
  2. Childhood Onset Pervasive Developmental Disorder
     a) Onset after 30 months but before 12 years of age
     b) Absence of psychotic symptoms
     c) At least 3 of the following:
        I. Resistance to change
        II. Inappropriate or constricted affect
        III. Sudden excessive anxiety
        IV. Peculiar movements
        V. Abnormal speech patterns
        VI. Under or oversensitivity to sensory stimuli
        VII. Self multilation
  3. Atypical Pervasive Developmental Disorder also described.

DSM III-R

- More complex criteria for Autism
- Requires 8 of 16 criteria in three domains: social interactions, communication and restricted interests
- Drops age limits
- Introduces PDD,NOS

DSM IV AND IV-R

- Qualitative impairment in social interaction- including shared attention, nonverbal behaviors, lack of reciprocity.
- Qualitative impairments in communication-including no speech, stereotypic or idiosyncratic speech or inability to sustain conversation.
- Restricted repetitive and stereotypic patterns of behavior, interests and activities.

DSM IV AND IV-R

- Includes 5 categories:
  - Autistic Disorder
  - Rett’s Disorder
  - Childhood Disintegrative Disorder
  - Asperger’s Disorder
  - Pervasive Developmental Disorder Not Otherwise Specified

DSM V DIAGNOSTIC CRITERIA

A. Persistent deficits in social communication and social interactions across multiple contexts: (Currently or by history)
   1. Deficits in social-emotional reciprocity
   2. Deficits in nonverbal communicative behaviors used for social interactions
   3. Deficits in developing, maintaining, and understanding relationships

DIAGNOSTIC CRITERIA

B. Restricted, repetitive patterns of behavior, interests or activities. (At Least 2)

1. Stereotyped or repetitive motor movements, use of objects, or speech
2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behavior
3. Highly restricted, fixated interests that are abnormal in intensity or focus
4. Hyper- or hyporeactivity to sensory input or unusual interest in sensory aspects of the environment.

DIAGNOSTIC CRITERIA

- 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) Cause clinically significant impairment in social, occupational or other important areas of current functioning.
- E) Behavior is not better explained by intellectual disability or global developmental delay.

REVIEW

- Kanner[1] (1943)-early description of 11 children
  1. Inability to relate in ordinary way to people and situations from the beginning of life
  2. Failure to assume at any time an anticipatory posture preparatory to being picked up or adjust to fit the hold
  3. Lack of use of language to convey meaning to others
     a) Excellent rote memory
     b) Delayed echolalia
     c) Monotonously repetitious verbal utterances
  4. Literalness
  5. Personal pronouns are repeated just as heard
  6. Distressed by loud noises and moving objects
  7. Anxiously obsessive desire for the maintenance of sameness
  8. Limitation in the variety of spontaneous activity

SO HOW DID WE GET HERE?

- Prevalence of Autism
  - ➢Kanner estimated 150:20,000[1]
  - ➢1966 estimated at 4-5:10,000 (1:1,000)
  - ➢1970 formal prevalence study found 4-5:10,000[2]

  - ➢Autism and Developmental Monitoring Network
    - ❖2002: 1:150
    - ❖2004: 1:125
    - ❖2006: 1:110
    - ❖2008: 1:88
    - ❖2012: 1:68

Are we over diagnosing autism?

If we are,why?

SOCIAL COMMUNICATION AND SOCIAL INTERACTION

- Deficits in social-emotional reciprocity, ranging, 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.

CAN OTHER DISORDERS PRESENT WITH SOCIAL DEFICITS?

- Attachment Disorders
- Mood Disorder, especially Bipolar
- Attention Deficit Hyperactivity Disorder
- Speech Apraxia
- Executive Function Disorders
- Trauma
- Children Exposed to Substances In Utero
- Social Anxiety Disorder
- Psychotic Disorders

HOW ARE SOCIAL DEFICITS IN AUTISM DIFFERENT?

- Difficulties with social reciprocity: includes abnormal social approach (smells your hand), failure of back and forth conversation (monologues), failure to initiate, respond to or maintain a conversation.
- Difficulties with speech: includes problems with pronouns, incomplete sentences, no or unique language. May be very literal and concrete. May not understand humor.
- Have to be taught pragmatic language concepts that are preprogrammed in neurotypical children

DEFINITIONS CONTINUED

- Difficulties with nonverbal communication: includes abnormal eye contact, lack of facial expression, inability to read others facial expressions.
- Difficulties developing, maintaining and understanding relationships: includes problems understanding the appropriate context for behaviors, difficulties understanding boundaries (stalking), can range from no interest in peers to insisting to be liked by everyone. Doesn't always realize when being annoying or rejected.

CRITERIA B

Restricted, repetitive patterns of behavior, interests or activities. (At Least 2)

1. Stereotyped or repetitive motor movements, use of objects, or speech
2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behavior
3. Highly restricted, fixated interests that are abnormal in intensity or focus
4. Hyper- or hyporeactivity to sensory input or unusual interest in sensory aspects of the environment.

CAN OTHER DISORDERS PRESENT WITH CRITERIA B TRAITS?

- Attention Deficit Hyperactivity Disorder
- Executive Functioning Disorders
- Obsessive Compulsive Disorders

RESTRICT, REPETITIVE PATTERNS OF BEHAVIORS, INTERESTS OR ACTIVITIES

- Stereotypies: repetitive movements such as flapping, twirling, jumping, snapping fingers, barking. Can include using objects, echolalia, or repeating phrases or scripting. Sometimes called "stimming."
- Distress at small changes, difficulties with transitions, rigid thinking, rituals, same routes, same food, same clothes.
- Limited, fixed interests, fixation on unusual objects

HYPER-OR HYPO-REACTIVITY TO THE ENVIRONMENT

- Apparent indifference to pain or temperature
- Hypersensitivity to sound/light/smell/touch
- Excessive preoccupation with a particular sense
- Visual fascination with lights or movement

- THIS IS MORE THAN BEING BOTHERED BY THE TAGS ON YOUR CLOTHES OR NOT LIKING LOUD NOISES!

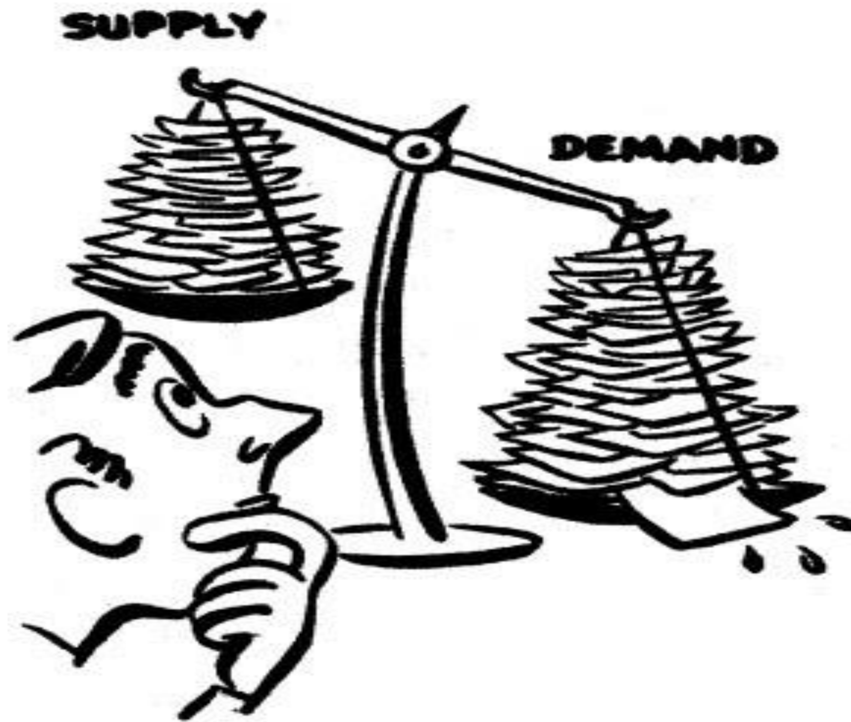
SYMPTOM CONFUSION

- Many individuals can have difficulties with sensory issues and not have autism.
- Individuals with ADHD frequently have difficulty being able to stop one activity and be able to move on to the next.
- Individuals with executive function disorders have many of the criteria including poor social skills, rigid thinking and difficulty with transitions.
- Collecting and specific interests are a normal developmental phase in children

WHERE ARE THE PROBLEMS

- Overgeneralizing symptoms
  - ✓Equating social awkwardness/shyness/anxiety with autistic impairment.
  - ✓Not considering social and behavioral impairments of other disorders such as ADHD and Bipolar/DMDD
  - ✓Too much weight on sensory issues
  - ✓Too much weight on transition issues
  - ✓Not taking trauma, attachment disorders and executive functioning disorders into account

PRESSURE



PRESSURES FROM FAMILIES

- Parents read about disorders, then present histories consistent with the disorders
- Parents want services for their children
  - ✓Many children would benefit from social skills training, yet don't have autism
  - ✓It is hard to get social skills training unless you have an ASD diagnosis
  - ✓Intensive behavioral interventions work for a variety of behavioral issues
- There can be monetary incentives to have an ASD diagnosis-SSI

PRESSURES FROM SCHOOLS AND COMMUNITIES

- Resources in schools and communities are very limited
- Services for individuals that qualify for ASD are often more intensive and expensive than other mental health services (ABA)

Providers are often caught between families wanting the diagnosis and schools resisting it.

Rock
Hard Place

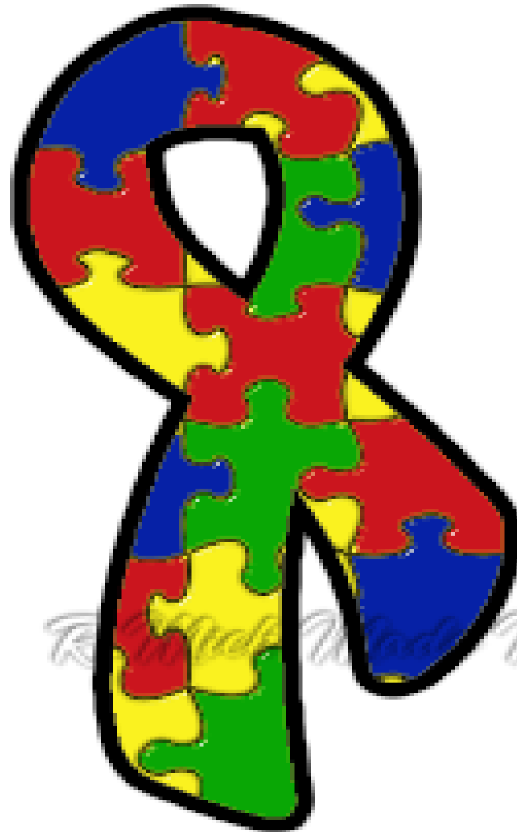
So What's A Clinician to Do?

We like to be the good guys and make everybody happy!

WHEN DIAGNOSING AUTISM

- Do a thorough evaluation, may be more than one visit
- Stick with strict diagnostic criteria
- Get speech and language, psychological and occupational assessments if possible
- Consider impairments in executive functioning, attachment disorders, trauma and substance exposure in utero
- Be cautious with parent reports of behavior, get details, videos if possible
- If standardized ADOS is available, use it.
- Expect resistance to whatever you do!

QUESTIONS?



Always
Unique
Totally
Interesting
Sometimes
Mysterious

BIBLIOGRAPHY AND RESOURCES

1. Kanner,L. (1943), Autistic disturbances of affective contact. Nerv. Child, 2: 217-250.
2. Treffert, D.A. (1970) Epidemiology of infantile autism. Arch Gen Psych, 22: 431-437.
3. Volkmar, F. et al. (2014) Practice Parameter for the Assessment and Treatment of Children and Adolescents with Autism Spectrum Disorder. J Amer Aca Child & Adol Psych, 53(2): 237-257.
4. Mandy, W., Wang, A, Lee, A, and Skuse, D.(2017) Evaluating social (pragmatic) communication disorder. J Child Psychology & Psychiatry.
5. American Psychiatric Association (2013). The diagnostic and statistical manual, fifth edition (DSM V). Washington DC APA Press.
6. CDC Autism and Developmental Disabilities Monitoring Network 2000-2012.