The Role of the Physician in the Assessment and Treatment of Individuals with a Spectrum Disorder

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Historical Context

- 1943 Kanner first describes Autism and describes an "inborn defect"
- Environmental explanation for psychopathology- "refrigerator mothers"
- Genetics also dismissed an inherited origin

Twin Studies

- 1980-1990's Twin Studies
- Standardized methods of diagnosis
- Concordance between monozygotic vs dizygotic pairs (60% vs 5%)
  - Heritability of 90%
  - Likely multiple genes involved
- Studies supported the heritability of a broader phenotype, ASD

Medical Conditions Associated with Autism

- 10-15% of Autism has an associated medical condition
- The rate of diagnosable medical conditions rises with increasing mental retardation
- Increasing rate of genetic abnormalities
- Diagnostic “overshadowing”

Genetic Disorders

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Table 1 | **ASD-related syndromes**

Syndrome	Gene(s) associated with the syndrome	Proportion of patients with the syndrome that have an ASD	Proportion of patients with an ASD that have the syndrome	Refs
15q duplication — Angelman syndrome	*UBE3A* (and others)	>40%	1–2%	101–103
16p11 deletion	Unknown	High	~1%	20, 35, 44
22q deletion	*SHANK3*	High	~1%	21, 22, 104
Cortical dysplasia-focal epilepsy syndrome	*CNTNAP2*	~70%	Rare	37
Fragile X syndrome	*FMR1*	25% of males; 6% of females	1–2%	105
Joubert syndrome	Several loci	25%	Rare	106
Potocki–Lupski syndrome	Chromosome position 17p11	~90%	Unknown	107
Smith–Lemli–Optiz syndrome	*DHCR7*	50%	Rare	108
Rett syndrome	*MECP2*	All individuals have Rett syndrome	~0.5%	109
Timothy syndrome	*CACNA1C*	60–80%	Unknown	24
Tuberous sclerosis	*TSC1* and *TSC2*	20%	~1%	110

Tuberous Sclerosis

- Neurocutaneous disorder affects 1/6000
- Autosomal dominant disorder, 9, 19
- Benign tumors in the brain and other organs
- Depigmented macules, UV light
- 60-80% have mental retardation and seizures
- 17-65% have Autism
- 1% of Autism have TS

Fragile X

- Most common form of inherited MR
- Characteristic facial features
- Excessive triplet repeat CGG on X
  - 55-200 premutation, 40 per 10,000
  - >200 FXS, 2 per 10,000
- Delayed processing speed, short term memory and attention
- Fragile X Tremor Ataxia Syndrome in premutation carriers
- FXS with Autism, 30%,
- Autism with FXS, 1-2%

Rett Syndrome

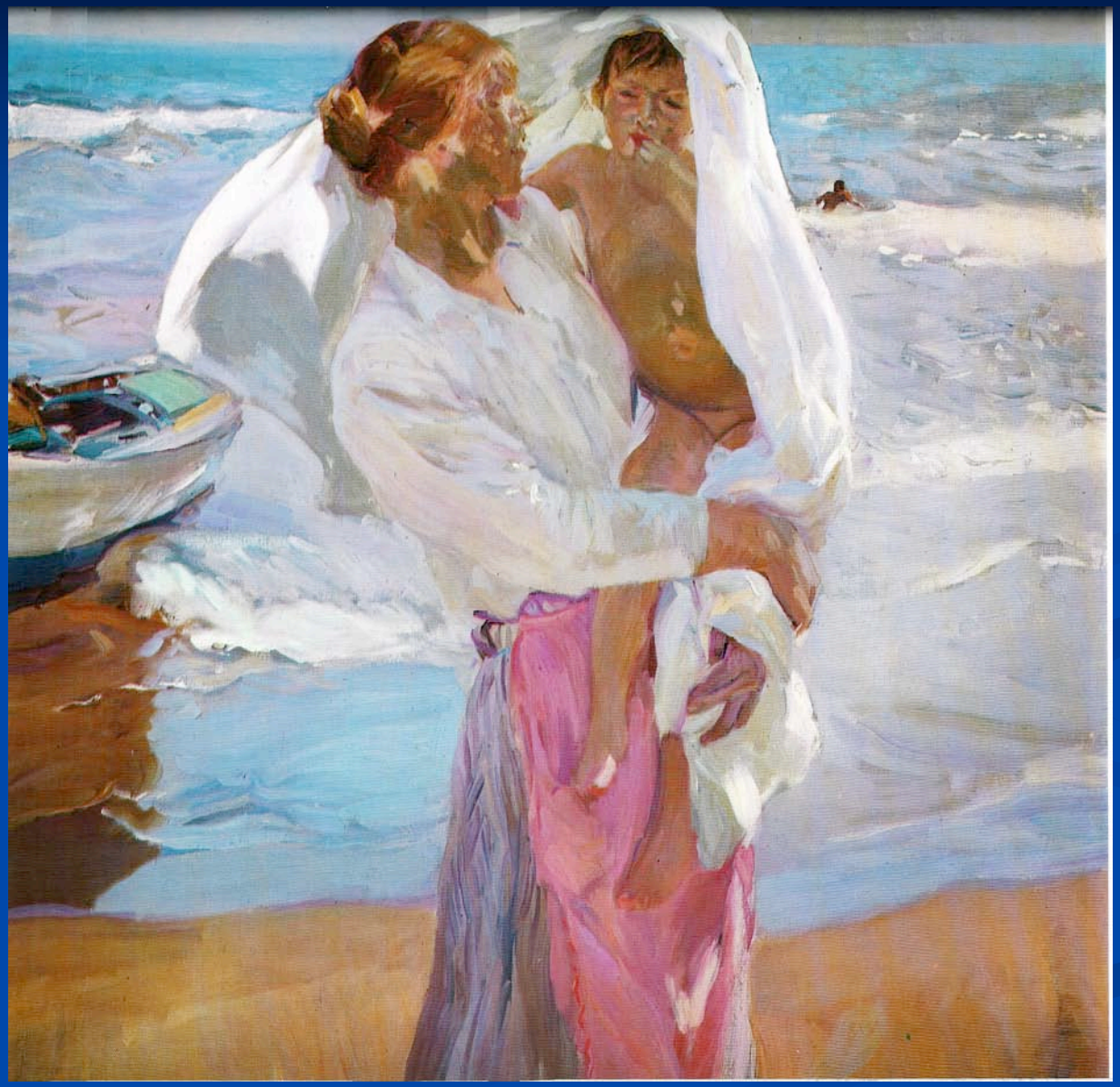
- 1/10,000 in girls
- Normal development until 6-18 months
- Loss of speech and purposeful hand use, microcephaly, seizures, social impairment, ataxia, stereotypic hand movements
- Mutation in MECP2, single gene disorder
- Although similar in appearance, it is best to consider it a separate disorder

Sleep Disorders

- The majority of children with Autism have sleep problems
- Prolonged sleep latencies
- Lengthy nighttime awakening
- Early morning awakening
- Rigid sleep rituals
- Sleep disorders

Seizure Disorders

- Occur in 30% of individuals with Autism
- Seizure onset has 2 peaks
- Risk of seizures is greater with mental retardation and language disorders
- Diagnostic overshadowing
- Consider if there is an unexplained change in behavior, or mood or a regression in function
- EEG may be normal
- Evaluation by a neurologist



Developmental Screening

- Clinician or parent rated general developmental screen
  - **P**ediatric **E**valuation **D**evelopmental **S**tatus
  - Ages and Stages Questionnaire
- M-CHAT
- Early referral for developmental services

Medical Workup

- Establish an accurate diagnosis of Autism
- Careful history and physical examination
- Auditory screening
- Ophthalmologic screening
- Newborn screening
- Specialist referral

Physical Examination

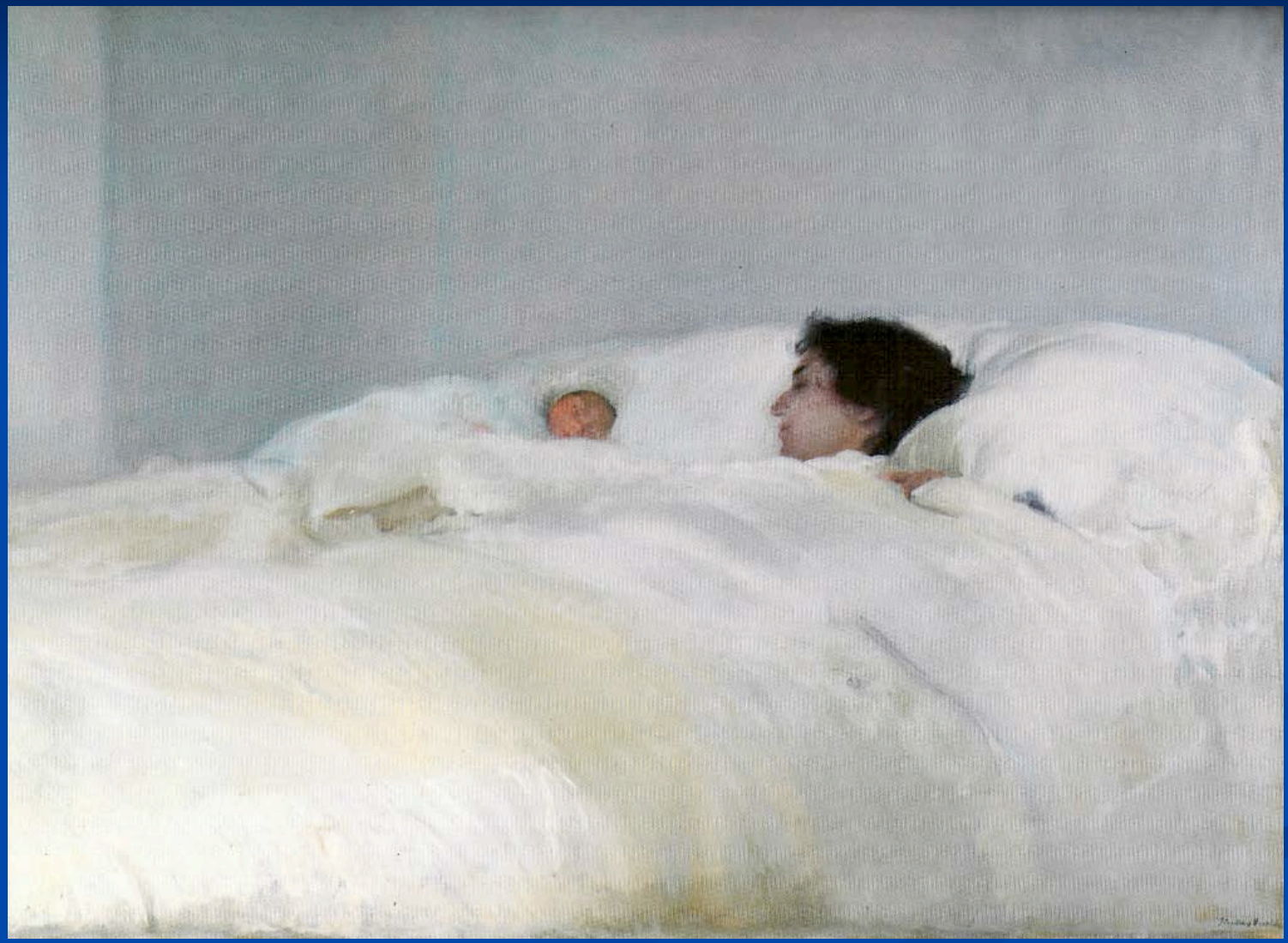
- Head circumference
- Focal neurological signs
- Cardiac examination
- Hepatomegaly
- Dysmorphic features
- Skin

Testing

- Hearing
- Vision
- Laboratory testing
- EKG, ECHO
- EEG
- Genetic Testing
- MRI

Genetic Testing

- Fragile X
- MECP2
- High Resolution Chromosomes
- Comparative Genomic Hybridization



Medical Specialists

- Family Medicine
- Pediatrics
- Developmental Pediatrics
- Neurology
- Genetics
- Psychiatry

Take Home message

- Early developmental screening and referral is essential
- Should have a reasonable not exhaustive medical evaluation
- Obtain an accurate diagnosis
- Find a physician with expertise in Autism
- Multiple disciplines bring a broader perspective of care

Co-morbid Mental Health Issues in Individuals with ASD

Scope of the problem

- Behavioral and emotional issues affect 30-40%
- Environmental and genetic factors
- Diagnostic and Statistical Manual-IV
  American Psychiatric Association
- Atypical presentations

Case presentation

- Bill is a 11 year old child who presents with aggression and noncompliance. He was failing at school both socially and academically. His family was exhausted and found that holding him more accountable had no effect on his behavior. Bill has good use of language. He worries constantly. He is repeatedly asking his parents for reassurance. He has to tap objects a certain number of times. He has many such rituals. He worries if others breath on him that he will become them. He is unable to function in most areas.

Anxiety Disorders

- Obsessive Compulsive Disorder
- Social Phobia
- Posttraumatic Stress Disorder

Obsessive Compulsive Disorder

- DSM-IV Diagnostic Criteria
  - Obsessions
    - Recurrent or persistent thoughts
    - Unable to ignore or suppress these worries
    - Intrusive and inappropriate, not real life concerns
  - Compulsions
    - Repetitive behaviors-hand washing, ordering, checking
    - Mental acts-counting, repeating
    - Compelled to perform in response to an obsession
    - Are meant to reduce distress or prevent an event, illogical

Obsessive Compulsive Disorder

- Treatment
  - Accurate diagnosis
  - Pharmacologic
  - Cognitive Behavioral
    - Education
    - Active participation
    - Response prevention

Social Anxiety

- DSM-IV Diagnostic Criteria
  - A marked and persistent fear of social or performance situations in which exposed to unfamiliar people or scrutiny by others
  - Exposure to the feared social situation provokes anxiety-panic attack, tantrums, shrinking from
  - Feared situations are avoided
  - Interferes significantly with the person's normal functioning

Social Anxiety

- Social avoidance is very common
- Exacerbates social impairment
- Lessens opportunities for social practice
- Worsens without treatment
- Can lead to profound impairment
- More commonly seen in higher functioning ASD, but…

Social Anxiety

- Treatment
  - Education
  - Support of the patient, family, school
  - Graded exposure
  - Expect an increase in behaviors
  - Treatable yet…hold on to your hat

PTSD
DSM-IV Diagnostic Criteria

- Exposed to a traumatic event that involved actual or threatened injury or death or a threat to physical integrity
- Person's response involved intense fear or helplessness
- The Traumatic event is reexperienced
  - Distressing recollections
  - Recurrent dreams
  - A sense of reliving the experience
  - Distress at exposure to cues
- Avoidance
  - Efforts to avoid thoughts, feelings or activities that arouse recollections
- Persistent symptoms of increased arousal
  - Sleep problems
  - Irritability
  - Poor concentration

PTSD

- Often associated with very aggressive behavior
- Can occur with levels of trauma that are not life threatening- but are overwhelming -domestic violence, severe bullying
- The individual with PTSD is more prone to decompensation with stress.
  - Sensory overload
  - Social overload
  - Academic overload

Post Traumatic Stress Disorder

- Treatment
  - Individual must feel safe
  - Support
  - Improve methods of coping
  - Working through the trauma
  - Pharmacology

Case presentation

- Frank is a very bright 14 year old adolescent who was isolating from his family and developing an interest in dark music and lyrics. He demonstrated increased irritability and threats towards his family members. He wanted to have a social life and yet had always struggled to make friends. His eye contact was poor, spoke in a monotone and would only answer with single words despite normal language. He was mistrustful and very hard to engage. His ADOS supported the diagnosis of PDD, NOS.

Mood Disorders

- Major Depression
- Bipolar Disorder

Major Depression
DSM-IV Diagnostic Criteria

- Symptoms present for at least 2 weeks
- Depressed mood, most of the day, almost daily
- Diminished interest or pleasure
- Significant change in appetite or weight
- Significant change in sleep
- Agitation or retardation
- Worthlessness or guilt
- Poor concentration
- Recurrent thoughts of death

Depression

- Treatment
  - Make the diagnosis
  - Understand the individual in the context of their developmental needs, ASD is not their whole story
  - Individual and family therapy
  - Review the environment
  - Find ways to connect with the world
  - A need to succeed-scaffold interactions
  - Pharmacology

Bipolar Disorder
DSM-IV Diagnostic Criteria

- Must have had the presence of a manic episode.
- Typically have also had the presence of depression at some time
- Manic symptoms include:
  - Persistently elevated, expansive or irritable mood
  - Grandiosity
  - Decreased need for sleep
  - More talkative
  - Flight of ideas
  - Increase in activity or agitation
  - Excessive involvement in pleasurable activities

Bipolar

- Atypical presentation
  - Masturbation vs promiscuity
  - Silly vs grandiose
- Family History
- Mood instability is very common in ASD but…
- Treatment
  - Pharmacologic
  - Education
  - Compliance
  - Support

Case presentation

- John is an 8 year child with Autism. He has limited eye contact although is more related with persons familiar to him. He has language although his ability to express himself is limited. He is currently in a socially and academically overwhelming school placement. Mother reports that when he returns from school he paces incessantly, talks to himself and expresses illogical and disconnected thoughts. He is laughing on his own and appears to be responding to internal stimuli. School is concerned as his behaviors there are worsening. He is increasingly erratic and unpredictable.

Psychosis

- Schizophrenia
  - DSM-IV criteria
    - Delusions
    - Hallucinations
    - Disorganized speech
    - Disorganized behavior
    - Negative symptoms-affective flattening

    *If there is a history of ASD the diagnosis is made only if symptoms are present for at least a month.

Psychosis

- The prevalence of Schizophrenia does not appear to occur with any increased frequency in ASD.
- Clinical reports like John's are very common.
- Psychosis vs Disorganization
- Disorganization typically resolves with support while true psychosis as a rule does not.

Disorganization

- Treatment
  - Rule out psychosis
  - Assess stressors
    - Social
    - Academic
    - Sensory
  - Challenge but don't overwhelm
  - Improve coping
  - Increase supports
  - Pharmacology

Attention-Deficit/Hyperactivity Disorder
DSM-IV Diagnostic Criteria

- Inattention
  - Difficulty sustaining attention
  - Does not seem to listen
  - Does not follow through on directions
  - Easily distracted
- Hyperactivity/Impulsivity
  - Often fidgets or squirms
  - Runs or climbs excessively
  - Talks incessantly
  - "On the go"

*Symptoms do not occur exclusively during the course of a Pervasive Developmental Disorder.

Attention-Deficit/Hyperactivity Disorder

- Symptoms of hyperactivity and inattention occur frequently in ASD
- Response to treatment may be atypical
- Assess inattention vs complexity of expectations
- Look for “over-focus”

Attention-Deficit/Hyperactivity Disorder

- Treatment
  - Pharmacologic
  - Work with school around appropriateness of expectations
  - Psychological testing to assess for uneven cognitive functioning
  - Skill building
  - Classroom strategies
  - Treat perseverative thinking

Take Home

- Comorbid mental health issues are very common
- Consider anxiety with avoidance, disorganization and aggression
- Consider depression particularly from mid/late childhood into adulthood with the risk of late diagnosis.
- Approach to care should be viewed broadly and include the family, mental health team, school and other community agencies.
- Balance challenge and support