An Update on Aspects of Autism for Primary Care Providers

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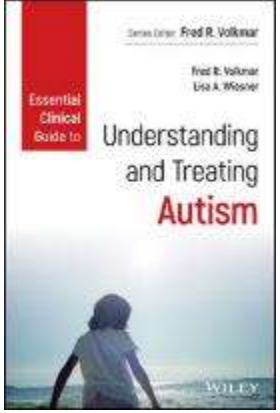
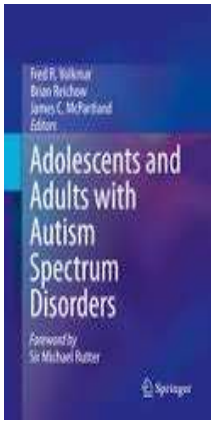
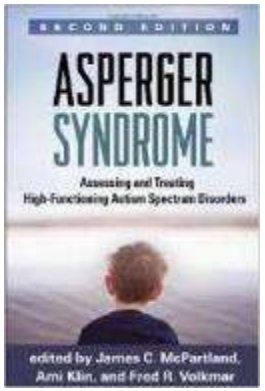
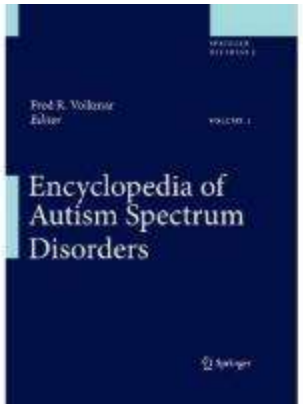
Conflicts of interest

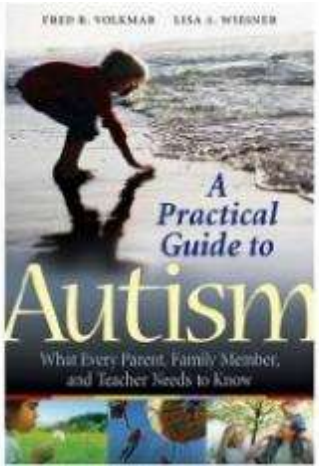
- **US Grant ACE NIMH**

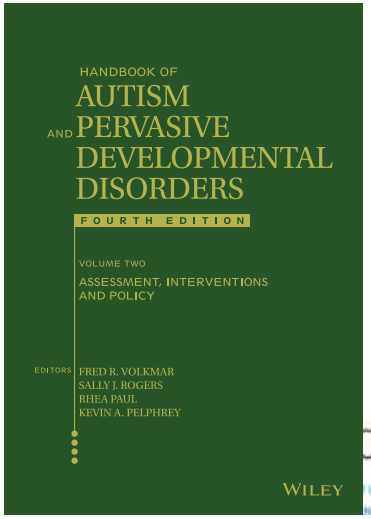


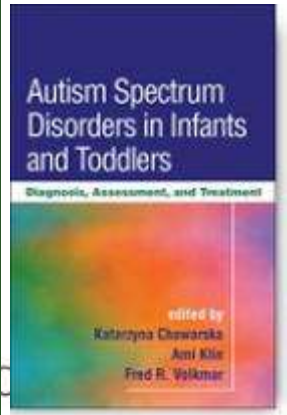












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Overview

- **What is autism?**
  - Autism as a social learning disability
- **Screening and Diagnosis**
- **Medical Issues**
  - Medical Home, approaches to patient
  - Common Problems
- **Treatment Updates**
- **Outcome**
- **References/Resources**

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What is autism?

- **Autism is first and foremost a**
  - SOCIAL LEARNING DISABILITY
- **It shares many features with other disabilities**
  - In general people don't 'outgrow' it
  - It can be tremendously helped (often)
    - With appropriate supports and realization of what needs and vulnerabilities are
- **It differs from other disabilities given its early onset and pervasive effects**

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A (quick) discussion of terms

- **Autism, Autism Spectrum Disorder(s), Asperger's, PDD, etc. -need better term**
- **Keep in mind that there is a BROAD range of syndrome expression**
  - If you meet one person with autism you have met one person with autism
- **Disorder vs. Differences is indeed a discussion**
  - One of the major findings of past decade
- **Normative = neurotypical = some hypothetical population average**

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Screening and Diagnostic Instruments- Good and Bad News!

- **Large number of screeners available**
  - At least 37 now available
    - Some for young children, other for school age
  - Several good Diagnostic instruments
    - ADI-R: parent report
    - ADOS: Child Assessment
    - CARS-2: Child assessment

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Warning signs: birth to 1 year

- **Social Symptoms**
  - Limited ability to anticipate being picked up
  - Low frequency of looking at people
  - Limited interest in interactional games
  - Limited affection toward familiar people
  - Content to be left alone
- **Communication Symptoms**
  - Poor response to name (doesn't respond when called)
  - Does not frequently look at objects held by others
- **Restricted interests and stereotyped behaviors**
  - Mouths objects excessively
  - Does not like to be touched

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Warning signs: 12-36 months

- **Restricted interests and stereotyped behaviors**
- **Hand or finger mannerisms**
- **Inappropriate use of objects**
- **Repetitive interest/play**
- **Unusual sensory behaviors**
- **Hyper/hyposensitivity to sounds, texture, tastes, visual stimuli**

- **Regression**
  - Loss of words
  - Loss of social engagement
  - Reported 20% of cases
    - Different patterns
    - Reason for continued vigilance
    - May represent different subtype of autism

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Early Screening Guidelines

- **Level One: Routine developmental surveillance**
  - Performed on all children at all well-child visits
  - Identifies children at risk for atypical development
  - Red flags indicate additional screening
- **Level Two: Diagnosis and evaluation of autism**
  - In-depth evaluation of children identified as at-risk
  - Differentiates autism from other developmental disorders
- AAP, AACAP have recommended early screening
- BUT several groups now recommend against
  - NICE
  - US Health Policy Guidelines

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(Johnson & Myers, 2007)

Problems for screeners

- **Level I and II screeners**
  - In reality mostly level I
  - Validity studies tend to be somewhat limited
  - More population based studies needed
    - Note in Norway M-Chat
  - Controversy regarding screening
    - Conflicting recommendations
  - Need for more mobile based app type approaches

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Problems for diagnostic instruments

- **These do NOT replace clinical judgement**
- **Originally focused: school age BOYS (US/UK) children of mild-borderline ID**
- **Growing body of work on potential biases**
  - Social class, cultural issues
  - Work less well for more intellectually disabled and for the higher cognitive functioning
  - And in girls!

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Problems with DSM-5

- **For autism**
  - Marked reduction in criteria, flexibility
  - Problems for
    - Higher functioning
    - Asperger's
    - PDD-NOS
  - Given the increased awareness of the broader autism spectrum this is unfortunate
  - "social communication disorder"

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McPartland et al 2012

– JAACAP 2012 Apr;51(4):368-83.

- Re-analyzes data from 933 cases in DSM-IV field trial
- 657 clinician diagnosed ASD, 276 non-ASD
- Cross -alked criteria from field trial to DSM-5
- 60.6% ASD retained DSM-5 diagnosis
- Specificity high (94.9%)
- Se varied in several ways
  - by dx: Autism =.76, Asp= .25, PDD-NOS= .28
  - And by IQ <70 Se=.70, >70=.46

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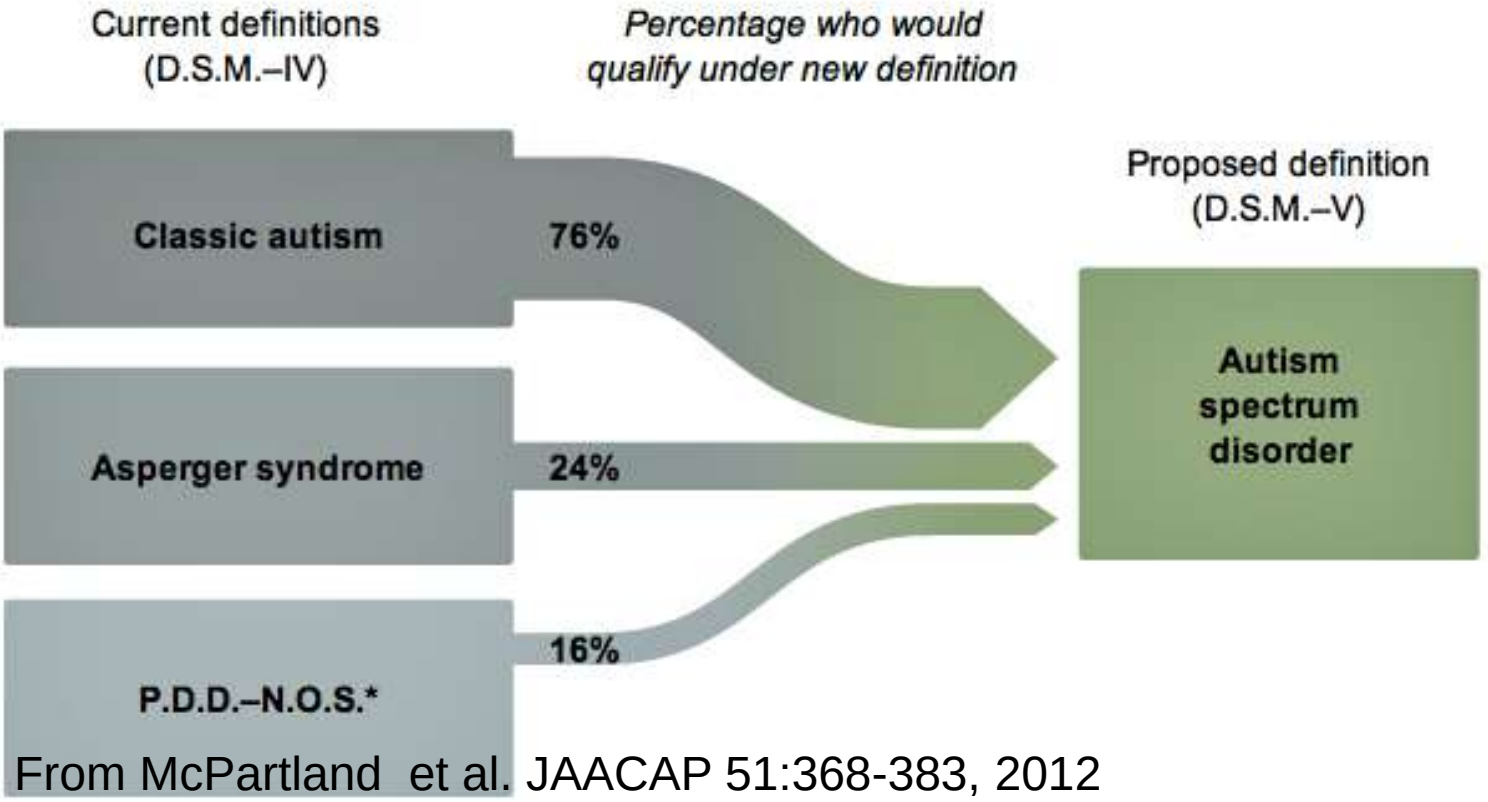
What happens to cases?

From NY Times

Redefining Autism

In a preliminary analysis, three researchers estimate that far fewer people with autism or a related disorder would meet the criteria for autism spectrum disorder after a change proposed for the fifth edition of the Diagnostic and Statistical Manual of Mental Disorders, or D.S.M.

Related Article »



From McPartland et al. JAACAP 51:368-383, 2012

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DSM-5 – 5 years late

- Smith et al JADD 45(7)3541-2552
  - 25 studies of DSM-5
  - Compared to DSM-IV
    - 25-50% of cases LOST diagnosis
    - Esp. higher functioning, Asperger's, PDD-NOS
  - Issues for young children as well
    - Barton et al 2013 (similar to problems with screeners)

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How did this happen?

- **A well meaning and informed group of individuals but what were problems**
  - In house
    - Not at academic center
  - Use of existing data sets (large but collected in highly standardized way)
    Disraeli
  - Lack of field trials
  - And a very real problem in addition!

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The culprit! $$$$$$$

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DSM-5 Background

- **Nearly 2 decades since DSM-IV**
- **Some basic decisions**
  - Eliminate subthreshold concepts (all of DSM 5)
  - Look at new approaches
    - Reliance on data from diagnostic instruments (ADOS/ADI)
    - CAUTIONARY NOTE!
  - "field trials" and process issues
  - "new" social communication disorder

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Potentially problematic or beneficial decisions!

- **Overall decisions**
  - Eliminate "subthreshold"
  - Rely on research diagnostic instruments
    - Rather than field trials
- **Autism specific issues**
  - Autism spectrum disorder
    - Levels of symptoms severity
  - Move from 3 categories to 2
  - From polythetic to mixed decision

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Use of factor analysis

- **2 or 3 factors?**
  - Kanner (2) ➔ Rutter 3 (till now)
  - In DSM-IV field trial
    - 3, 2, or 5-factor solutions worked
    - BUT 3-factor was consistent with older work AND gave much greater flexibility of combinations (>2000 for DSM-IV)
    - For DSM 5 12 combinations
    - General problem with factor analysis
      - See Gould Mis-measure of Man book
      - Paper in Press in JADD

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Not everything can (should) be combined!

Use of research instruments

- **Excellent instruments available but**
  - Need considerable training
  - BOTH history AND current assessment
  - Which instruments to use?
  - Criteria/items may be less useful in clinical practice settings
  - Diagnostic instruments work best at 'midrange' functioning and age (school age children with borderline to moderate Intellectual deficiency)

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What from the surface may seem a tranquil situation…

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…may mask a more basic problem!

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Screening and Diagnosis

- **Why is early diagnosis important?**
  - Children <5 have most potential for major gains
  - Presumption that for many (not all) early intervention may make a MAJOR difference
  - Issues in diagnosis under 3 years
    - Child may have social and communication problems but NOT yet the repetitive behaviors
    - The latter emerge by 3

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New approaches to screening

- **Less pencil and paper based**
- **MORE focused on tasks that the child engages in**
  - EEG, eye tracking, listening, etc.
  - MANY potential advantages
  - BUT what are the problems?
    - Avoid fishing in stocked ponds!
    - Population based studies needed

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Clinical Evaluation: History

- **Pregnancy, labor, delivery**
- **Developmental milestones**
  - (sometimes baby diaries/videos help)
- **Family History**
- **With age/TIME**
  - Educational interventions
  - Medical interventions
  - CAM
  - Course (major changes, regression or moves to better)

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Initial assessments

- **Practice guidelines available**
- **Medical evaluations**
  - Hearing & vision
  - Dysmorphic features or + family history
    ➔ genetic assessments
    - ACHG has online guidelines
  - Most frequent problems
    - Seizures
    - Associated medical conditions
      - Fragile X, tuberous sclerosis

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Physical Exam

- **Look for**
  - Any unusual dysmorphic features
    - Genetic conditions esp.
      - Fragile X, Tuberous sclerosis
    - Any suggestion (exam/history) of seizure disorder
  - Head size
    - Macrocephaly (and body size!)

Laboratory Studies

- **Lead level**
- **Genetic testing**
  - An area where technology evolving rapidly
    - Recommendations from ACMG
      - Genet Med 15:5:399-407
      - Guided by history and exam
      - Commercial gene panels are NOT endorsed
- **EEG if history (including regression) or exam**
- **Neuroimaging NOT routinely used**

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Continued in Part 2 PDF