An Update on Aspects of Autism for Primary Care Providers

Fred R Volkmar, M.D.
Child Study Center, Yale University

Fred.volkmar@yale.edu
Conflicts of interest

- US Grant ACE NIMH
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
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.
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
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
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
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
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

*(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
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!
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”
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
What happens to cases?

From McPartland et al. JAACAP 51:368-383, 2012
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)
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!
The culprit! $$$$$$$
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
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
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
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)
What from the surface may seem a tranquil situation...
...may mask a more basic problem!
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
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
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)
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
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
    - Guided by history and exam
    - Commercial gene panels are NOT endorsed
- **EEG if history (including regression) or exam**
- **Neuroimaging NOT routinely used**
Continued in Part 2 PDF