Autism Across the Life Span:
Session II: Causes of autism and the school age child
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Fred R Volkmar MD
www.autism.fm or fred.volkmar@yale.edu

Overview Session II
– Causes of autism: genetics and brain mechanisms
– School age children
  • Assessment and interventions

Autism and Related Disorders: short review
• Continuum of disabilities characterized by a triad of impairments relative to developmental level:
  • Social behavior
  • Communication
  • Repetitive or rigid behaviors and interests
• Epidemiology
  – Prevalence estimate of 1 in 110 (1:80 to 1:240)
  – 4:1 ratio of males to females for autism (IQ related!)
  – Proportionate distribution across race, ethnicity, and social class
  • If one controls for possible bias in referral
• Economics and Social Policy
  • Cost to treat: ~$3.4 million per person
  • ~$35 billion per birth cohort
  • Early identification ➔ better outcome (on average! Some children don’t do well no matter what!)
  • More children doing better and going to college!

Current Guidelines for Autism: DSM-IV
• Based on a large (nearly 1000 cases) field trial with over 100 raters at 20+ sites around the world (Volkmar, et al., 1994)
• A long process of preparation was undertaken, deliberations were very methodical (and published), and goals for the field trial included:
  – Convergence (if possible) with ICD-10 (some complexities here)
  – Good coverage over the entire range of cognitive ability
  • DSM-III was too stringent and not developmental
  • DSM-III-R overly diagnoses autism in more cognitively delayed individuals
  • In the end DSM-IV had a good balance across IQ levels
• Applicability in both clinical work and research
• Reliability

DSM-IV-TR Diagnostic Criteria
Autistic Disorder
1. Qualitative impairment in social interaction
   a) Marked impairment in the use of multiple nonverbal behaviors
   b) Failure to develop peer relationships appropriate to developmental level
   c) A lack of spontaneous seeking to share enjoyment, interests, or achievements with other people
   d) Lack of social or emotional reciprocity
2. Qualitative impairment in communication
   a) Delay in development of spoken language
   b) Marked impairment in ability to initiate or sustain a conversation with others
   c) Stereotyped and repetitive use of language or idiosyncratic language
   d) Lack of varied, spontaneous make-believe play or social imitative play
Published Research in Autism

- Intervention research has not increased as quickly!

Research Papers: 1943-1999

- # Publications
- Data presented in 5 year blocks, source: Medline
- Source: Medline, note: to keep up with everything you'd have to read about 5.25 papers each day!

Research Papers: 2000-2010

- Source: Medline

Practice guidelines

- Screening
  - Fillipack et al. 2000 Neurology 55:468-479
  - Royal College of Pediatrics: NICE guidelines
  - http://www.nice.org.uk/CG128
  - AAP Pediatrics 120:1183-1215
- Medical and Psychiatric Care
  - Volkmar et al. 1999 JACAP 38(S):32-54
  - Effective health care program (US Agency for Health Care research)
  - Royal College of Pediatrics: SIGN guidelines
    - http://www.sign.ac.uk/guidelines/fulltext/98/index.html
- Education
  - National Research Council (2001) report Educating Children with Autism
  - National Autism Center (www.nationalautismcenter.org)
    - *being revised!

EBP Resources

- In medical and mental health fields, there are pocket guides to EBP
  - Evidence-Based Medicine (3rd ed.)
    - Strauss et al.
  - How to Read a Medical Paper and related series
    - Greenhalgh et al.
  - Evidence-Based Psychotherapies for Children and Adolescents, 2nd ed.
    - Wiesz & Kazdin
  - Practitioner's Guide to Using Research for Evidence-Based Practice
    - Allen Rubin, 2008

Asperger - 1944

- Series of cases - all male
- Marked social problems
- Good cognitive/language skills
- Motor problems
- Circumscribed interests
  - + Family Hx (esp. fathers)
  - "Autistic Psychopathy"/Autistic personality disorder
Case report – Don age 8

- Birth and early development unremarkable
- Problems with peers in Nursery School
- Long standing special interests
- Testing:
  - Verbal IQ 104, Nonverbal 78
  - PPVT-R 104, EOWPVT 119
  - Vineland Socialization 46
  - (Interpersonal age equivalent 1-6)

Don’s Drawing

Case Example: “Bob”
10 ½ year old boy

- History
  - talked before he walked
  - words were his lifeline
  - Special Interests: letters/numbers, Was reading by 2 ½
  - Now Calendars, geography, foreign currency, catastrophes

- Testing Results
  - VIQ 141 PIQ 73 PPVT 138
  - Vineland Socialization 56
  - Interpersonal age 2-6

What causes autism?

- Early focus on parents
  - Partly due to Kanner’s original report
  - Emphasis on experience in psychiatry
  - Feral children approach
  - (Psychodynamic views)
- But evidence against
  - Other children in the family were fine
  - No major psychiatric problems in parents
  - No obvious deficits in parenting
- Longitudinal data began to suggest a strong role for the BRAIN and for GENETICS
Onset of Epilepsy in Autism

Cooper - data on a normative sample, Volkmar & Nelson and Dyken & McMahon large samples of children with autism

Neurobiological Findings in Autism

- High peripheral levels of serotonin
- High rates of seizure disorder
- Persistent primitive reflexes
- Increased head size (toddlers)
- Morphological changes in CNS
- Minicolumns, mirror neurons
- Fusiform gyrus and faces
- Placental abnormalities

Genetics of autism

- Early impression: NO genetics
- BUT
  - First twin study (Folstein & Rutter, 1978) Tremendous genetics
  - Increased rates for MZ (over same sex DZ) twins
- Early estimates of recurrence risk in sibs: 2% BUT
  - Stoppage rules complicate
  - Better estimates 10-15%
- 2nd Degree relatives .4-1%
- Implications:
  - Likely synergistic effects of multiple genes

Genes possibly involved in ASDs

R/Y = de novo losses/gains, G = risk ASD, P = Linkage peaks

Potential Genes involved

Environmental factors in autism

- Tremendous interest on part of parents
  - Mercury and MMR controversy
- Issues
  - Clearly strong role of genetics
  - Potential for some environmental contribution (broadly defined)
  - Need for better specification of mechanisms and subgroups
  - Data on environmental factors rather limited (Wing & Potter, 2002)
Vaccinations and Autism

- Wakefield et al. (Lancet) paper had tremendous impact
- Many problems: Paper withdrawn
- Multiple reviews have found no connection
- Parental concern → vaccination rates
- 2002-3 Italy, 5000 people hospitalized
- Sporadic outbreaks in US
- Travel related: Iowa, Chicago, San Diego
- Why the misperception
  - Role of media, regression

Dramatic rise in Measles UK

Source: myhousecallmd.com

Understanding the Social Brain in Autism

- Birth: preferential interest in face/voice, top 1/2 face
- 2-3 months: face recognition (internal features)
- 6 months: inversion effect, gender discrimination
- 9 months: strong stranger response, species effects
- Subsequent greater expertise as children age

Face Recognition in Autism

- Large literature on different approaches used by individuals with autism
- Do not do well on normed facial recognition tasks
- Do as well recognizing inverted faces as correctly oriented ones
- Rely less on salient features of face for recognition (e.g., may focus on other characteristics)
- Limitations: use of still faces

Circuitry of the Social Brain
Face Discrimination

fMRI study
- comparison to normal controls
- task: same or different:
  - people
  - objects
  - patterns
- regions of interest:
  - fusiform gyrus (face)
  - inferior temporal gyrus (objects)
- both groups equally accurate
- finding now replicated many times

Neuroanatomy of Face Recognition:
The Fusiform Gyrus

Face Recognition: Fusiform Gyrus Group Differences

A Case Study Exemplifying Expertise
Model: Digimon Expertise in an 11 year

Digimon Activations: 11 yo with Autism

Social information processing is highly dynamic

- Need for new approaches
  - Limitations of work with photographs (Ecological validity)
  - Viewing the world with new eyes
- Choice of subject – concerns and choices
  - Intensely social (small number of people)
  - Minimal action/objects (aka no terminator 2)
  - Black and white initially
  - Show short segments (not entire film)
  - Choose movie about a pleasant dinner party at a small New England college with 2 faculty members and their wives
Viewer with autism
Age: 38, FSIQ: 119,
ADOS-4 / ADI-R +,
Vineland Socialization: 69

Typical Viewer
Age: 27, FSIQ: 110


Focus on mouths vs. focus on eyes ➔ lose about 90% relevant information

Information Lost by focusing on Mouth:
All the social (nonverbal) cues of the nonspeaker
Epidemiology of Autism

- Is there an epidemic?
- More public awareness → greater concern by parents and autism
- Autism Speaks/Ad Council adtc tendency to equate autism spectrum with autism →
- ? Some increase
- DSM-V may take care of this!

Setting for intervention

- Recognize child’s difficulties in responding to complex (social/nonsocial) environments
- Balance of “pull out”, small group, classroom-based, and unstructured environments
- Classroom environment
- Continuity and consistency → across settings and across people
- Monitoring and flexibility → team approach and collaboration
- School - home communication

Social Skills

- Balance of methods
  - Adult instruction, peer, hybrid
  - Teach self-management and social skills
  - Goals[2] initiations and responses with/to peers
- All ages and levels of severity
- Most research has been done with young children

Approaches to teaching social skills

- Vary depending on age of child and level of impairment
- For younger children – focus is often on peers (with adult monitoring)
- For school age – hybrid methods (circle of friends, individual work)
- For adolescences – adult mediated with explicit teaching
Language-Communication

- Language functioning at age 5 is one of the two strong predictors of outcome
- Probably at least 75% of preschoolers with autism can develop useful speech
- Even for individuals with minimal verbal speech teaching alternative COMMUNICATION skills is critical
- Importance of augmentative approaches
  - Low and High Tech
  - Picture exchange, schedules, etc.
  - Organizational aids

Augmentative Strategies

- Augmentative forms of communication
- Manual sign, Picture Exchange, computerized communication systems, etc.
- Use of visual strategies
- No data that there is a negative result from using augmentative strategies for enhancing communication skills

Behavioral issues

- Teaching new and desired behaviors
  - Discrete trial, peer mediation, naturalistic, pivotal response
- Decrease problem behaviors
  - that interfere with learning
  - Methods:
    - Functional analysis, extinction, examination of antecedents and consequences

Organizational skills

- Social deficits \( \rightarrow \) failures in learning what to attend to
- Learning to learn skills
  - Lists, organizers, written/visual schedules
  - Software, assistive device
- Realistic, step-wise plan (goal directed)
  - Activities of daily life: homework, shopping
- Learning from experience, modifying strategies, multitasking

Adaptive skills & Generalization

- Identify appropriate targets for intervention
  - Use of Vineland and IQ levels
- Generalization across settings, people, contexts at every opportunity
- Do NOT teach skills in isolation
- Encourage functional independence and self-sufficiency
- Coordination with home/family

Occupational/Physical Therapy

- Sensory issues/sensitivities
  - Individualized program
  - Deal with sensory issues in appropriate ways
- Encourage gross and fine motor skills
  - Writing, keyboarding
- Feeding/eating and oral-motor issues
Implications for teaching – translating what we know to the classroom

- Problems in organization
  - Stepwise approach, consistency, routines, visual aids, sufficient time
- Attentional problems
  - Isolate relevant information, structure environment, support attention
- Sequencing
  - Visual cues, predictability, consistency

Implications for teaching II

- Gestalt learning style (learning in ‘chunks’)
  - Present materials across settings, encourage generalization, family involvement
- Visual learning style
  - Use visual supports, give adequate time, limit verbal language, short simple language

Implications for learning III

- Trouble with time and temporal sequences
  - Visual supports, concrete instructions, adequate time, clear expectations, clear outcome and feedback
- Trouble understanding Social Cues
  - Exaggerate, pair gestures and words, teach in context, avoid overly elaborate language, explicit teaching

Issues in mainstreaming

- Rebuttable Presumption
- Difficulties with generalization
- Use of TRAINED peers
- Effective peer mediated approaches complicated to deliver but can benefit
- The ‘holy trinity’ mainstreaming
  - (AKA the 3 WORST places to mainstream!)

Approaches to Teaching: Developmental Strategies variably emphasized in programs

- Awareness of usual developmental sequences and progressions as starting point
  - Be aware that these are sometimes violated!
- Use a child centered approach (child leads and adult follows) when possible
- Child’s preferences/motivations help guide program development
- Pay attention to the learning environment

Resources

- "more than words" – www.hanen.org
- Visual strategies - Linda Hodgdon
- Social stories – Carol Gray
- TEACCH program – Schopler and Mesibov
- Computer resources – Laureate.com, inspiration.com
- Handbook of Autism 3rd edition
MANY supports available

- Range from low to high tech
- Picture books, story boards, picture exchange
- Social stories
- Prepared books materials (esp. for medical care)
- Computers and organizational aids
  - Organizational software – inspiration.com
  - Keyboarding/laptops (esp. for more able)
  - Ipad/Iphone applications
  - Computer software and even robotics
- Have potential for many uses medically
  - Show visit plans
  - Help with communication
  - Teach relevant language

Drug Treatments

- Importance of double blind, placebo controlled studies
  - Major “placebo effect”
- Medications most frequently studied
  - Risperidone and newer 2nd generation neuroleptics – work well and several approved
  - SSRIs – used anxiety/depression, rigidity but seem to work less well in children, better in adolescents and adults not yet FDA approved
- Side effects and balance of risk and benefit
- Several good resources available
- Note the social vulnerability of autism is still NOT yet a clear target of drug treatments
  - Some new work underway on this!

RUPP Autism Network: Irritability Scale

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Other problems

- Sleep issues
  - Very common problem
- Safety issues
  - Accidents leading cause of death
- Behavioral issues
  - Interventions for problem behaviors
    - Drug and behavioral
    - Sensory issues
    - Regression

Challenges for higher functioning children

- Unawareness of the extent and impact of social disabilities & lack of appreciation of the child’s disability: e.g., “too bright”, “too verbal”
- Variability of the child’s profile and presentation across settings
- Behavioral problems may take precedence over the child’s social disability
  - “SEM”, “SED”, “ED”, “BD”
  - worst mismatch ➔ perfect mismatch
- Advocacy and services

For parents

- Providing quality and understandable information
- Use/limitations of internet
  - Type autism into google and you get 17 million hits!
  - Yale You Tube lectures!
- Importance (& downsides) of networking
- Considerable amount of miss-information available
- Evaluation of CAM particularly important
- Helping parents be informed consumers and advocates
In Summary

- Many advances and an exciting time to be in autism research/service
- Issues
  - Need for research AND integration of research with translation and communication regarding effective practices
  - Need for new models of care (screening, older individuals)
  - Developing quality resources/programs/models of care is a priority!

References (see www.autism.fm)