

## Seeing the Forest AND the Trees: Diagnostic Complexities in the Age of DSM-5

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**Structure of today's talk**  
Future talks in this series



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## Overview

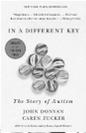
- I. The 'Discovery' of Autism and Origins of the Broad vs. Narrow Concept
- II. From Kanner to DSM-III (1980)
- III. The Origins of DSM
- IV. From DSM-III to DSM-5
- V. DSM-5 and its Impact
- VI. Current Controversies
  - I. Broader Autism Phenotype
  - II. Asperger's
  - III. Age and IQ related issues: adults, infants
  - IV. Gender
  - V. Culture
- VII. Summary Where to go from here!



Could we infer the forest from a leaf?

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## The 'Discovery' of Autism



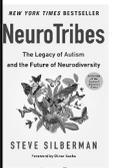
- Was there autism before Kanner?
  - Early reports of feral children – date from Roman times if not before
  - Victor – the Wild Boy – reported on by Itard may be first reported case
  - Donvan and Zuker – In a Different Key – found reports from state training schools in the 1800s
- Leo Kanner (1943) gave the classic description of "early infantile autism"
  - Two essential features:
    - Autism
    - Insistence on sameness/resistance to change
    - Also noted the language/communication issues
    - Believed it was inborn
  - Some false leads:
    - Impression of normal levels of intelligence – did well with SOME tasks on IQ tests
    - Not related to medical conditions
    - In early cases parents were all very successful and well educated





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### Asperger and the Origins of the Broader Autism Phenotype




- Hans Asperger (1944) – Autistic Personality Disorder
  - Used same word autism to denote severe problems in social interaction
  - Saw this more as a personality rather than developmental disorder
  - All boys, special interests that interfered with functioning, ran in families (first to really highlight genetics)
- Little interest until 1980 – Wing published a description in English with case reports
  - Origins of differences in conceptualization start with Wing
  - Everyone cited Wing's criteria but she really didn't have any AND
  - She saw this as part of broader spectrum
- Other concepts as well: atypical personality development (B. Runk), schizoid children, and others
- Beginning of the debate about Broader Autism Phenotype (BAP)

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### What is in a name?

αὐτός

The intended sense of αὐτός is generally defined by its grammatical context. When used as a lone nominal without an article, it is generally the third person personal pronoun. When appended to a nominal and not possessing the definite article it is "self". When combined with the definite article, either appended to a nominal or on its own, it is "same".

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### Early Development of Autism as a Diagnostic Concept

- Evolution of concept
  - ? Continuity with Childhood Schizophrenia (official term for autism before 1980)
  - Some aspects of Kanner's report
- Research in the 1970's helped establish the validity of autism as a diagnostic concept
  - Autism was **BRAIN BASED** – high rates of epilepsy, often in teenage years
  - Autism was **STRONGLY GENETIC** – much higher rates in identical vs fraternal twin
  - Autism responded best to **STRUCTURED TEACHING** rather than unstructured psychotherapy
- Work with childhood schizophrenia began to show that it was:
  - Extremely rare (esp. before puberty)
  - Onset much later than autism
  - Different in clinical features and family history as compared to autism
- Attempts in the 1970's to develop explicit guidelines for diagnosis
  - Rutter (1978) Social and language features (not due to MR), restricted interests and early onset
  - Ritvo/INSAC (1978) unusual rates & sequences of development, sensitivities to environment

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### A very short review of classification





- Several origins:
  - General interest in classification – e.g. Linnaeus
  - In Medicine -Origins in late 1800's work on causes of death and infection control
- Purposes for classification
  - Enhance communication and research, epidemiology, service planning
  - Tensions between clinical and research purposes
  - Different approaches: categorical, dimensional, ideograph
- In Medicine the International Classification of Diseases (ICD) is the world wide manual
  - The DSM is a "local variant" of the ICD but in fact dominates the world
- Common misconceptions
  - Disorders NOT people are classified
  - Labels may give entitlements to services
  - Assigning a label is NOT the same thing as having an explanation
- Important constraints on systems of classification

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### Evolution of DSM: I → III

DSM-I and II – arose in context of growing awareness of mental illness and esp. needed with WW II

Heavily theoretical

Only a few categories for children

MR, childhood schizophrenia, adjustment and other 'reactions' (hyperkinetic, withdrawing, overanxious, runaway, unsocialized aggressive, group delinquent and 'other')

DSM-III

Wash U school of psychiatry approach

Research Diagnostic criteria

Multiaxial framework

Included 'subthreshold categories'

Important note: DSM III and beyond intended BOTH for clinical and research work – ICD has adopted the two book approach!

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### DSM-III – Recognition of Autism

DSM-III

- First recognition of "Infantile Autism"
- New overarching class of disorder to which this belonged: the
  - Pervasive Developmental Disorder
    - Infantile Autism/Residual Infantile Autism
    - Childhood Onset PDD/ Residual COPDD
    - Atypical PDD (subthreshold – but essential **Broader Autism Phenotype- BAP**)

Problems:

- Great to have recognition BUT lack of developmental orientation and complexities of diagnosis

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### From DSM-III to DSM-IV

- Problems in DSM-III quickly recognized
- DSM-III-R (1987) - Autistic Disorder
  - Better name, more criteria (3 areas), more flexible, more developmental
  - Problems – a bit overly broad – esp. at ends
- DSM-IV – (1991) major redo of whole book
  - For autism several steps: invited reviews, data reanalyses, working WITH ICD-10 revision
  - → Field Trial

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### DSM-IV Field Trial

- International effort
  - 21 sites, 125 rates, almost 1000 cases
- Info on case and raters
  - Clinical diagnosis
  - Rater info/experience
  - Case information
- Ratings of various criteria sets
- Final set: 12 items (3 areas: social, communication, restricted behaviors), at least 6 (2 from social)
  - Note: good reliability with experienced clinicians, good interrater reliability, and results of factor analyses

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### DSM-IV Continued

- For autistic disorder > 2200 ways to get a diagnosis of autistic disorder
- New Categories (compatible with ICD-10)
  - Asperger's
  - Childhood Disintegrative Disorder
  - Rett's
  - Pervasive Developmental Disorder Not Otherwise specified
  - (Broader autism phenotype)

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### Asperger's Disorder: 1944 –1994

- Little interest until Wing's 1981 paper
  - Suggested modifications, case reports, issue of continuity with autism – several views arose
- Subsequent research limited in several ways
  - Small samples, tendency towards circularity in findings
  - Inconsistent approaches and terminology
  - Convergence with other concepts:
    - NLD, Right Hemisphere Learning Disability, Semantic-Pragmatic Processing Disorder, Schizoid personality
- DSM- IV Field Trial
  - About 50 cases, significant differences from autism and PDD-NOS

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### Asperger's Disorder

- Considerable resistance to inclusion
- DSM-IV definition somewhat problematic
- Complexities with several views of best approaches to diagnosis: "verbal" autism, PDD-NOS, something more unique and different from autism
  - At least 6 different approaches!
- But it was included and research and clinical interest as well as public awareness started to increase dramatically

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### PDD-NOS

- Early 'back story'
- Relation to autism?
  - Importance of genetic work
- Subtypes
  - Critically important question
  - Need for larger samples and research
  - Various subtypes proposed
    - Some cases with more attentional problems
    - Others with more affective lability
    - Others with more language/thought problems

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### Work on Genetics – Complexities

- MANY genes involved
- Some single genes
- Extremely complex

Single gene, Mendelian

“Common autism” (multiple genes, environment)

Cytogenetic

15q mat dup

22q11 del

22q tel del

2q tel del

Genome-wide Arrays  
~8% diagnostic yield in ASD at EGL

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### Broader Autism Phenotype

- Several lines of work began to converge
  - Studies from psychology began to support notion that some individuals had some features of autism without the whole syndrome
  - Other studies shows some traits in family members who didn't have the full syndrome
  - Neurobiological research (e.g. MRI) also showed some family members with features similar to autism in sibling
  - Epidemiology – MANY more children had some features without the whole autism clinical pictures
  - And the complicated genetics

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### A Momentary Pause

- Nearly two decades pass
- Both DSM and ICD start to think about revisions
- Good reasons for this
  - More knowledge, better understanding, tweak disorders
- Less Good reasons for this
  - The DSM is a (if not THE) major source of income for the APA and DSM-IV was losing steam

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### DSM-5

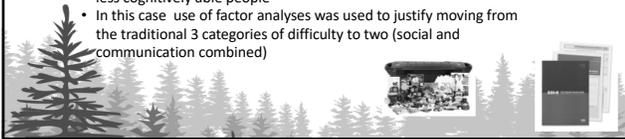
Nearly 2 decades since DSM-IV appeared

- Some basic decisions
  - Eliminate subthreshold concepts (all of DSM 5)
  - Look at new approaches
    - Reliance on data from diagnostic instruments (ADOS/ADI)
  - ‘field trials’ and process issues
  - Now based at APA headquarters rather than an academic institution
  - Note in contrast to previous versions several members resigned during the DSM-5 development process

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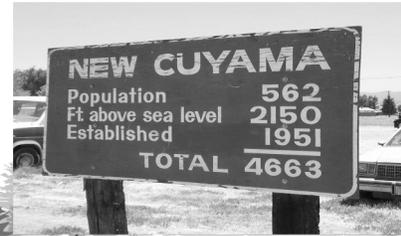
### Use of research instruments to derive criteria

- These are excellent instruments BUT require extensive training AND they come from DSM-IV and ICD-10 in first place (dog chases its tail)
- They are NOT designed for routine use
- Their background is that they were first developed for school age children (mostly boys) of borderline to mild intellectual disability – and do NOT work so well for older/younger people NOR for more and less cognitively able people
- In this case use of factor analyses was used to justify moving from the traditional 3 categories of difficulty to two (social and communication combined)



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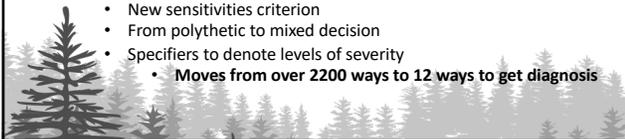
Not everything can (should)  
be combine!



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### DSM-5 Decisions

- Drop PDD in favor of Autism Spectrum Disorder
  - A welcome change but not the singular rather the plural
- Overall decision
  - ONLY autism spectrum disorder
    - Drop Asperger's, drop PDD-NOS
    - New social communication disorder diagnosis
- For Autism Spectrum Disorder
  - Changes in approaches - Move from 3 categories to 2
  - New sensitivities criterion
  - From polythetic to mixed decision
  - Specifiers to denote levels of severity
    - Moves from over 2200 ways to 12 ways to get diagnosis

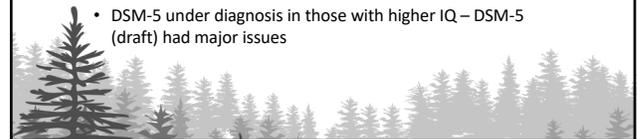


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### Impact of DSM-5

Mattilla et al 2011 (JAACAP 2011 583-592)

- First study of DSM-5 but DID not use final criteria set
- epidemiological study 5,484 8 year old (Finland)
- Large group had ASSO, 110 seen for assessment,
- Looked at DSM-IV and DSM-5, separate analysis IQ < > 50
- DSM-5 under diagnosis in those with higher IQ – DSM-5 (draft) had major issues

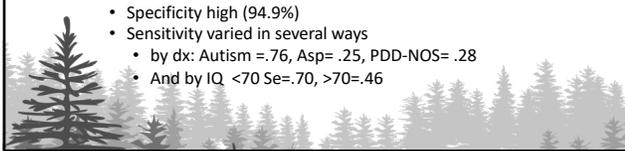


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### Impact of DSM- 5. McPartland, Reichow & Volkmar

JAACAP 2012 Apr;51(4):368-83.

- Reanalysis data from 933 cases in DSM-IV field trial
- 657 clinician dx'd asd, 276 non asd
- Cross walked criteria from field trial to DSM-5 – used FINAL criteria set
- 60.6% ASD retained DSM-5 diagnosis
- Specificity high (94.9%)
- Sensitivity 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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Published: January 19, 2012

### Redefining Autism

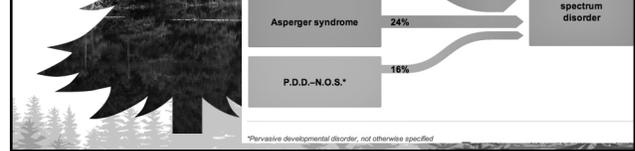
In a preliminary analysis, three researchers estimate that far fewer people with autism or a related disorder would meet the criteria for autism spectrum disorder after a change proposed for the fifth edition of the Diagnostic and Statistical Manual of Mental Disorders, or D.S.M. Related Article »



Current definitions (D.S.M.-IV)	Percentage who would qualify under new definition
Classic autism	76%
Asperger syndrome	24%
P.D.D.-N.O.S.*	16%

Proposed definition (D.S.M.-V): Autism spectrum disorder

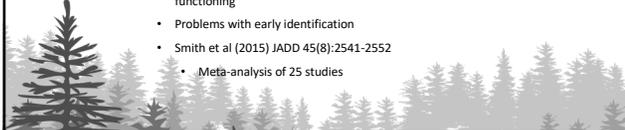
\*Pervasive developmental disorder, not otherwise specified



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### Other studies and response

- Response of DSM-5 work group
- "Grandfather in" cases with "well established" diagnosis
- Two major problems quickly identified
  - Marked drop in diagnosis in higher functioning
  - Problems with early identification
- Smith et al (2015) JADD 45(8):2541-2552
  - Meta-analysis of 25 studies

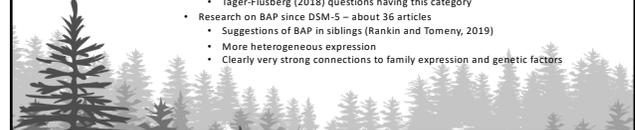



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### Continued and New Controversies

Broader Autism Phenotype

- As noted previously about 85% no longer qualify for ASD diagnosis
- Does the new Social Communication Disorder category work for them?
  - In first place this is a COMMUNICATION disorder
    - Pragmatic language deficits, NO Restricted Repetitive Behaviors
  - It is more restrictive than the old PDD-NOS
  - Specifically excludes cases with ASD
  - Does not simply equate with broader autism phenotype
  - Only about 12 papers since DSM-5 appeared. (2013)
  - Tager-Flusberg (2018) questions having this category
- Research on BAP since DSM-5 – about 36 articles
  - Suggestions of BAP in siblings (Rankin and Tomeny, 2019)
  - More heterogeneous expression
  - Clearly very strong connections to family expression and genetic factors



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### Continued and New Controversies

Whither Asperger's Disorder

- Before DSM-IV there were various differing approaches to diagnosis
- Most claimed to be from Lorna Wing's 1981 "criteria" but she didn't really give any
- Asperger's included only reluctantly in DSM-IV
  - Reflect in criteria and exclusionary rules
  - Problems quickly noted (Miller and Ozonoff, 1997)
  - Major changes made in text NOT criteria in DSM-IV TR

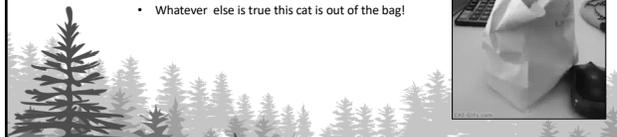


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### Continued and New Controversies

Whither Asperger's Disorder

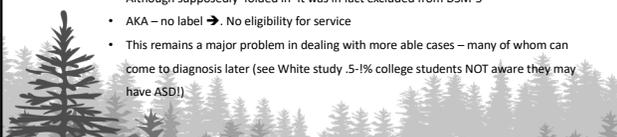
- As noted previously about 75% of cases with AS lose ASD as a label
- From a research viewpoint:
  - 1944-1981. 1982-1991. 1991- Present
  - 8 papers. 15 Papers. 2284 PAPER
  - Whatever else is true this cat is out of the bag!

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### Continued and New Controversies

- Why delete it?
  - There clearly were differing views (esp. from Center to Center)
  - Not yet a consensus on its validity apart from autism
  - Nonpeer reviewed paper by Happe (see file to right)
- Why not delete it?
  - MANY people identified with it as something similar to but not quite the same as autism!
    - Although supposedly "folded in" It was in fact excluded from DSM-5
    - AKA – no label →. No eligibility for service
    - This remains a major problem in dealing with more able cases – many of whom can come to diagnosis later (see White study .5-1% college students NOT aware they may have ASDI)

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### Continued and New Controversies

- Asperger's POST DSM-5 – Data continues to emerge
  - E.g. Chiang et al. 2014 (JADD 44(7): 1577-86) Meta-analysis
  - 52 STUDIES >1000 HFA >1000 ASP
  - RESULTS
    - A) individuals with ASP had significantly higher full-scale IQ
    - B) within individuals with ASP had significantly higher VIQ than PIQ
    - C) within those with HFA the VIQ was similar to PIQ



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### Continued and New Controversies

Age-Related Issues - ADULTS

- Many more able individuals may come to diagnosis later
  - Asperger mentioned this originally (1944)
  - White et al. (2011). 7-1.9% of college freshmen met either of two approaches to diagnosis
  - Differences in presentation – complex psych histories
    - Lai & Baron-Chen, 2015; Huang et al., 2020
- DSM – 5 criteria may work less well
  - remember where instruments come from!
    - Jackson & Volkmar, 2019; Magiati and Howlin 2019
  - Failure to have problems picked up on early in life may be a disqualifier

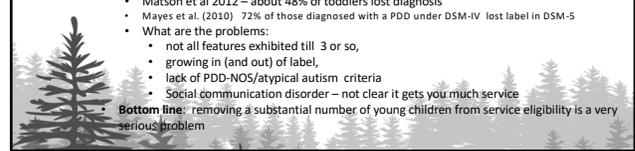


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### Continued and New Controversies

Age-Related Issues - Toddlers

- Increased interest in very young children
  - At risk groups (siblings),
  - Importance of early diagnosis and treatment
  - But also aware of broad range and screening issues AND developmental course
- DSM-5
  - A series of studies point to problem
    - Matson et al 2012 – about 48% of toddlers lost diagnosis
    - Mayes et al. (2010) 72% of those diagnosed with a PDD under DSM-IV lost label in DSM-5
    - What are the problems:
      - not all features exhibited till 3 or so,
      - growing in (and out) of label,
      - lack of PDD-NOS/atypical autism criteria
      - Social communication disorder – not clear it gets you much service
  - **Bottom line:** removing a substantial number of young children from service eligibility is a very serious problem

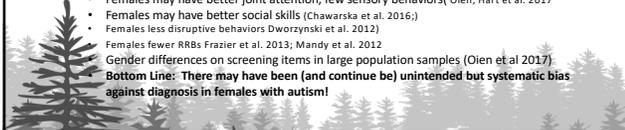


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### Continued and New Controversies

Gender Related Issues

- Implicit assumption that criteria are agnostic to gender BUT is this true?
- Dworzynski et al., 2012 Females with ASD require more symptom severity and greater impairment BUT
  - Females more likely to exhibit internalizing behaviors than boys(Mandy et al., 2012
  - Females may be better at 'camouflaging' than males (Bargiela et al., 2016).
- Prevalence studies consistently show male predominance but
  - Females less likely to engage in RRBs than males(Charman et al. 2017)
  - Males show more externalizing behaviors (Mandy et al. 2012; Solomon, Miller et al. 2012 )
  - Females may have better joint attention, few sensory behaviors(Oien, Hart et al. 2017
  - Females may have better social skills (Chawarska et al. 2016.)
  - Females less disruptive behaviors Dworzynski et al. 2012)
  - Females fewer RRBs Frazier et al. 2013; Mandy et al. 2012
- Gender differences on screening items in large population samples (Oien et al 2017)
- **Bottom Line: There may have been (and continue be) unintended but systematic bias against diagnosis in females with autism!**



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### Continued and New Controversies

Cultural Issues

- Interest from two sources
  - Growing interest in developing countries in ASD –
  - Growing awareness WITHIN THE US of potential cultural bias in diagnosis
    - (notes rates of autism in Los Angeles!)



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### • Continued and New Controversies

- Autism across cultures has a small but growing literature (see Frith et al. 2014)
- Some obvious areas of cultural difference – e.g. eye contact
- The VAST majority of work done in Western countries
- Potentially very significant cultural issues in understanding both diagnosis and treatment
  - Grinker et al. 2015
- Problems noted at several levels
  - Screening instruments
  - Diagnostic instruments
  - Applicability of usual diagnostic criteria
  - Need for coordination and involvement of various stakeholders and respect for cultural variations and practice



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### Continued and New Controversies

Cultural Issues

- Within the U.S. – **UNDER** diagnosis in minority groups esp. inner city but also potentially others
  - Social communicative features seem more problematic Vanegas et al. 2016
  - Tek & Landa (2012) lower levels of communication skills in minority groups even when parent education controlled for
  - Screening tools may operate differently in minority groups
  - Several studies suggest higher positive reports from Hispanic families using M-CHAT-R
  - Some apparent differences in item response based on maternal education (Scarpa et al., 2013)
- Various factors appear to impact lower diagnostic rates in inner city populations
  - See Mandel et al. 2007, 2009

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### Yet one more complexity Co-morbidity

- Having a problem like ASD only makes one MORE vulnerable to other problems
- And, esp. with age further complicating assessment and diagnosis
  - School age: ADHD, anxiety
  - Adolescence: Anxiety and depression
  - Many others as well



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### Summary

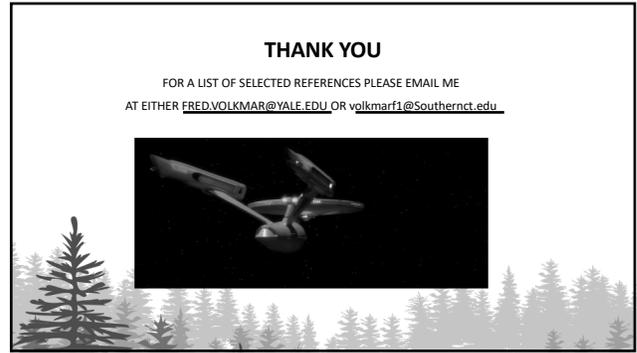
- Clearly much progress since 1940's
  - Recognition of autism/ASD
  - Importance of treatments
  - Growing body of basic and applied research
- Some continuing and some new challenges
  - Boundaries – broad vs. narrow
  - Asperger's and autism – same or different
  - Age and developmental factors
  - Gender – probably a major underappreciated variable
  - Cultural issues
- Need for better biomarkers,
- Working with rather than against complexity!
- Need to balance research AND SERVICE needs
- + **some basic good sense**
- Many important needs for DSM-5.1. !!!!!



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