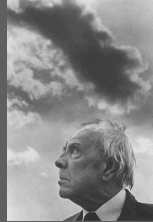


### On Exactitude in Science . . .

In that Empire, the Art of Cartography attained such Perfection that the map of a single Province occupied the entirety of a City, and the map of the Empire, the entirety of a Province. In time, those Unconscionable Maps no longer satisfied, and the Cartographers Guilds struck a Map of the Empire whose size was that of the Empire, and which coincided point for point with it. The following Generations, who were not so fond of the Study of Cartography as their Forebears had been, saw that that vast Map was Useless, and not without some Pittilessness was it, that they delivered it up to the Inclemencies of Sun and Winters. In the Deserts of the West, still today, there are Tattered Ruins of that Map, inhabited by Animals and Beggars; in all the Land there is no other Relic of the Disciplines of Geography.

*Suarez Miranda, Viajes de varones prudentes, Libro IV, Cap. XLV, Lerida, 1658*  
From Jorge Luis Borges, *Collected Fictions*, Translated by Andrew Hurley Copyright Penguin 1999.



## What's In a Name ?

### Dilemmas and Alternatives in the Diagnostic Classification of Neurodevelopmental Disorders

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## Goals

- ⇒ To review the evolution of our current diagnostic system and the empirical evidence supporting it
- ⇒ To examine the limits of our current diagnostic approach for understanding specific cases, especially persons with neurodevelopmental disabilities
- ⇒ To discuss some general theories diagnostic classification and some metaphors for thinking about diagnosis
- ⇒ To explore the possibility of other diagnostic approaches that may have greater clinical and research utility

## What I'm NOT intending to do:

- ⇒ Suggest that our current diagnostic system is without utility
- ⇒ Suggest that our current diagnostic system does not reflect some underlying reality (e.g. that neurodevelopmental or other psychiatric disorders are a socially constructed myth)
- ⇒ Suggest that diagnosis per se serves no purpose
- ⇒ Suggest that all neurodevelopmental or psychiatric disorders are somehow equivalent - in severity, in their impact on families and in their effect on persons suffering with them.

## Historical Background

- ⇒ A systems of classifying disease is called a "nosology"
- ⇒ Historically there have been many ways of doing this
  - The Greeks had 4 "humours" that could be imbalanced to create disease
- ⇒ Now we have two major systems in western medicine:
  - ICD-9 which covers physical and mental disease
  - DSM-IV (TR) which covers only mental disease
- ⇒ However, there are many different ways in which one can create a nosology

## Etiological approach to classification

- ⇒ In formally diagnosing stroke, there is little emphasis on symptoms per se.
  - In fact, cases can look wildly different in terms of their symptom presentation.
  - One person loses speech, another mobility, another memory, etc.
- ⇒ What they share is a common underlying cause ("etiology") that is used to make the diagnosis.
- ⇒ A similar approach used to diagnose epilepsy, strep throat, cancer, etc.
  - Symptoms can vary but the diagnosis remains.

## Symptomological approach to classification

- ⇒ Overt symptoms used as criteria for defining disorders
  - Really "syndromes" which are clusters of symptoms
- ⇒ Asthma defined by the difficulties breathing, the particular pattern of onset, etc.
  - Can be difficult to diagnose when a patient's symptoms do not meet the diagnostic prototype exactly
- ⇒ Nearly all developmental and mental health disorders are defined this way

## From Symptoms to Cause

- ⇒ In many cases physical disorders move in diagnosis from symptomological descriptions to etiological ones
  - AIDS initially a set of symptoms and "rule outs"
  - Once HIV found as cause, it's presence became defining criteria
  - Sometimes this understanding of cause restructures the diagnosis itself
    - reveals ignored cases/symptoms (e.g. patients who are "HIV+") and removes previously diagnosed cases
- ⇒ For some neurodevelopmental disorders we may be coming to the same place
  - e.g. the genetic marker for Rett's Syndrome

## The evolution of the DSM-IV

- ⇒ The Diagnostic and Statistical Manual of the American Psychiatric Association – 4<sup>th</sup> Edition
- ⇒ Defines all mental disorders in terms of a list of symptom sets.
  - Largely (though not entirely) a classical categorical system
- ⇒ An evolution from earlier editions which retains many of their historical nosological distinctions.
- ⇒ Diagnostic categories in it are not statistically derived.
  - In fact, many researchers have expressed concern about the inability to replicate the diagnostic distinctions statistically

## DSM - IV is symptom-based

- ⇒ In DSM-III a symptom-based nosology was adopted because previous versions made unsubstantiated claims about the etiology (causes) of disorders
  - This helped to improve reliability and made the DSM acceptable to those with varying theoretical viewpoints.
  - It also carries certain risks – that these symptom based distinctions will be largely meaningless, that they will not "slice the pie" in a relevant or useful way

## The problem of co-morbidity

- ⇒ Many disorders seem inextricably interconnected:
  - Epidemiological studies show that a majority of patients are diagnosable with two or more disorders.
  - Across the lifespan most individuals move in and out of these diagnostic categories over the lifespan.
    - Especially where they relate to severity (dysthymia/depression)
  - Many disorders are over-represented in the twins, siblings parents and children of diagnosed individuals suggesting a common etiology across disorders
  - There is increasing direct evidence for common genetic underpinnings
  - Many symptoms themselves occur on some sort of spectrum

## The Problem of Heterogeneity

- ⇒ Disorders initially thought to be unitary appear to need subdivision
  - Autism into Autism, AS, Rett's, CDD, etc.
  - Bipolar into I and II
  - Anxiety disorders into phobias, GAD, panic, OCD, etc.
- ⇒ Two persons with the same diagnosis can look VERY different.
  - This is a clear problem in the ASD's where the label often seems too broad to meaningfully direct treatment
  - However this clear affects other dx categories like the personality disorders

## The Problem of Subclinical Syndromes

- ⇒ While we clinically diagnose many disorders with high frequency, rigorous epidemiological studies suggest that many common disorders occur at very low levels in the general population
  - Bipolar
  - OCD
- ⇒ Clinicians recognize that even below strict cutoffs the symptoms cause distress, are predictive of later more severe dysfunction, and are treatable.

## The problem of exclusion rules

- ⇒ DSM-IV somewhat arbitrarily imposes restrictions to address comorbidity
  - Can't diagnose ADHD and autism
  - Can diagnose OCD and autism?!
  - Can diagnose Tourette's and autism?!
- ⇒ This has odd research effects where, for example no one discusses comorbidity of ADHD and Autism though most practitioners see clear overlap

## Autism Spectrum Disorders

- ⇒ Autistic Disorder
- ⇒ PDD-NOS
- ⇒ Asperger's Syndrome
- ⇒ Childhood Disintegrative Disorder
- ⇒ Rett's Syndrome?

## Diagnoses showing symptom overlap with ASD's

- ⇒ ADHD
- ⇒ Language disorders
- ⇒ Mental retardation
- ⇒ Bipolar Disorder (especially childhood onset)
- ⇒ OCD
- ⇒ Tourette's syndrome
- ⇒ Social phobia
- ⇒ Schizoid personality disorder
- ⇒ Epilepsy (in various manifestations)
- ⇒ Etc.

## In children with autism...

### As many as:

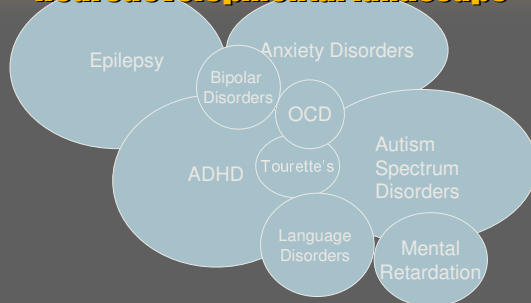
- ⇒ 60% with poor attention/concentration
- ⇒ 40% hyperactive
- ⇒ 88% with morbid or unusual preoccupations
- ⇒ 37% with obsessive thinking
- ⇒ 86% with rituals
- ⇒ 89% with stereotyped language
- ⇒ 74% with sig fears/anxiety
- ⇒ 44% with depressed mood, irritability and agitation
- ⇒ 11% with sleep problems
- ⇒ 43% with self injury
- ⇒ 10% with tics

From Tsai, L. (2000) Focus on autism and other developmental disabilities

## Is a "spectrum" too simple?

- ⇒ Autism "spectrum" suggests a two dimensional range from mild to severe.
- ⇒ Yet even within the known ASD's we have trouble determining what we mean by severity
  - Is language the measure of severity? IQ? Functional skills?
  - By strict standards many people currently diagnosed AS should be diagnosed autistic anyhow.
  - Not clear that language onset is critical to outcome for these persons

## A "map" of the neurodevelopmental landscape



## The neurodevelopmental landscape

- ⇒ It is critical to remember that these diagnoses are just categories defined by professional consensus and historical convention
  - Neither arbitrary NOR empirically derived.
  - They serve as a "map", to the real "landscape" of symptom presentation.
  - In the actual landscape of people's symptoms, these boundaries between disorders (like the state lines on a map) do not exist.
  - Rather, symptom clusters flow naturally into each other. Not just a spectrum between Autism and Asperger's, but between Asperger's and Bipolar, Tourette's and OCD, ADHD and AS

## The Current Map

- ⇒ Our current diagnostic "map" forces us into dichotomous choices about whether disorders exist or not
  - Critical data about severity, and prototypically of individual cases is lost
  - We are not allowed fuzzy boundaries to these classifications
- ⇒ When persons have symptoms of two disorders we talk about co-morbidity
  - Failing to recognize that, for example, Bipolar and Autism or Aspergers and GAD be fundamentally linked disorders.

## So what?!

- ⇒ This is all a bit abstract, so why does it matter?
  - You can't diagnose autism by its cause anyhow.
  - We need diagnosis for a range of good reasons (reimbursement, patient tracking, research selection, educational placement, treatment selection, etc.)
- ⇒ Because, when we confuse the "map" with the "landscape" it represents, it results in critical real-world consequences for clinical and research work.

## The Feverish Spectrum

- ⇒ Imagine someone taking a similar categorical approach, decided to examine the "fever spectrum"
- ⇒ They divide children into high, medium and low fever groups and are able to measure this with exacting reliability.
- ⇒ They even show the validity of these classifications by noting the symptom similarities the children share
  - High fever kids are lethargic, vomit, hallucinate, fall unconscious, etc....
- ⇒ Now a series of studies are undertaken to determine the cause of high fever... its outcome... its precursors... its treatment...

## A parallel for ASDs

- ⇒ When we look for causes of autism, have we divided up the cases correctly to find that cause?
- ⇒ The DSM divides ASDs primarily by onset of expressive language, but is this the relevant factor?
- ⇒ Maybe there are 10 or 100 (or 3) types of autism differing in IQ level, or activity level, or age of onset, or level of social engagement, or sensory style, or ....
- ⇒ If we don't "slice the pie" correctly, the causes for each subgroup will be swamped by the data from the others
- ⇒ Boiling our clinical information down to a single yes/no classification decision eliminates essential data

## Clinical implications

- ⇒ Clinicians are good at agreeing on “prototypical” cases but often disagree when forced to make yes/no decisions near the “edges”
  - Our current system has no method for noting how “central” a case is to the diagnostic label
  - Different labels can lead to dramatic differences in provided services, even when there is basic agreement about what services are needed for an individual case
- ⇒ On the other hand, when diagnoses are taken as gospel, similar diagnoses can lead us away from thinking about individual differences
  - Despite legal safeguards, children are often given programs for “ADHD” or “autism”, rather than those tailored to their unique needs.
  - Again, this is especially problematic for those on the diagnostic boundaries.

## More Implications

- ⇒ As you examine research and clinical practice with the realization that these categories have limited empirical backing it occurs that the entire field is involved in a bootstrapping effort.
  - The validity of tests is compared to other tests which are compared to the ratings of clinicians on diagnostic criteria which may have little validity themselves.
  - Clinicians debate diagnoses when there is little evidence that the features most relevant to outcome, treatment, etc. are even included in the diagnostic criteria.
  - Children are sent for expensive evaluations to make a differential diagnosis between PDD and Autism as if that distinction was fundamentally clear
  - This is the map being mistaken for the landscape it represents

## So what else could we do?

- ⇒ A spectrum or “dimensional” based approach:
  - Diagnosis where we rate children on a range of factors – activity level, language, cognitive function, social communication, etc...
  - Fits the data better, provides a richer description, provides more statistical power to research
  - Gets rid of the awkward “NOS” categories that crop up all over the DSM
  - Addresses “fuzzy” boundaries and clinical overlap
  - Allows diagnosis of “sub-clinical” disorder

## Problems with a dimensional approach

- ⇒ Difficult to use in practice, “Timmy’s a 9-activity, 3-social, 6-language”
- ⇒ Still faced with what dimensions to use
- ⇒ Many treatment and policy decisions are inherently categorical
  - e.g. access to special ed, to regional center services, to insurance reimbursement, etc

## Another “taxonic” approach...

- ⇒ A prototype approach
  - In fact, DSM-IV has moved more toward this with fewer symptoms that are “defining” and more of the “cafeteria” approach (2 of the following 4 symptoms...)
- ⇒ However, research has suggested that in practice clinicians don’t really follow the rules
  - There has been little increase in diagnostic reliability in day to day clinical practice (though it has improved for research)
  - Clinicians get the “gist” of the symptom set and then assign diagnosis by how well they feel patients fit them, often going back to the DSM after the fact.

## Prototype diagnosis

- ⇒ Westen, et. Al. (2003) suggest an approach based on “prototypes” that acknowledges clinicians tendency to diagnose by “feel” and capitalizes on this tendency.
  - Use statistics (factor analysis and cluster analysis) applied to symptom lists to empirically derive syndromes that seem to “hang together”.
  - Take those syndromes and associated symptoms and develop a list of 10-20 descriptive statements about a prototypical patient with the disorder
  - Rather than asking clinicians to rate each statement as true or false, have them rate their global impression of the child’s similarity to the prototype on a 5 point scale.

## A Possible Autism Prototype

- ⊖ Delayed onset of language not accompanied by other attempts to communicate
- ⊖ Impairment in nonverbal communication (eye contact, gestures, etc.)
- ⊖ Lack of social reciprocity
- ⊖ Limited or abnormal peer relationships
- ⊖ Abnormal sensory processing (hyper or hypo sensitivity)
- ⊖ Self injurious behaviors
- ⊖ Repetitive motor mannerisms
- ⊖ Insistence on routine
- ⊖ Impairment or delay in development of joint attention
- ⊖ Lack of sharing for enjoyment
- ⊖ Impairment in social referencing
- ⊖ Inability to take perspective of others or have insight into own behavior
- ⊖ Deficits in episodic memory
- ⊖ Odd or unusual social affect or markedly flat affect

## Westen et. al. Rating Scale

- ⊖ 5 - very strong match (patient has a prototypical case of the disorder)
- ⊖ 4 - strong match (patient has the diagnosis; categorical diagnosis applies)
- ⊖ 3 - moderate match (patient has significant features of this disorder)
- ⊖ 2 - slight match (patient has minor features of this disorder)
- ⊖ 1 - no match

## Advantages of this type of prototype diagnosis

- ⊖ Many of the statements could be the same criteria used in the current DSM-IV (just without the "cafeteria" restrictions)
- ⊖ Clinicians would not be forced into dichotomous choices about symptoms' presence or absence
  - How much impairment is "marked impairment"?
- ⊖ Allows acknowledgement of sub-clinical syndromes while still providing categorical diagnoses
- ⊖ Syndrome overlap would be more recognized leading to better treatment and research.
- ⊖ The diagnostic process would more accurately match the natural cognitive process of good diagnosticians.

## Disadvantages

- ⊖ It adds an element of subjectivity
- ⊖ It still does not solve the problem of symptomological diagnoses
  - Best would be a combination of this and etiological diagnoses when that information is present
- ⊖ Does not work as well as a profile approach in dealing with "outliers"
  - Two children rated "3" on Autism still may look quite different from each other (though equally similar to the prototype)
- ⊖ It would likely reduce the total number of diagnoses, changing people's current diagnoses and this is likely to be resisted.

## Final Thoughts

- ⊖ There is much concern among researchers about the direction DSM-V will take
- ⊖ There is increased pressure for a firm scientific basis for diagnostic categorization given the increasing impact of mental health in western society
  - This is completely unlike the scrutiny previous versions received
- ⊖ However there is also increased political pressure (for the same reasons), which may work against a rational/scientific resolution of these problems

"Wherever we try to mark out the frontier between mental health and disease, we find a neutral territory, in which the imperceptible change from the realm of normal life to that of obvious derangement takes place"

- Emil Kraepelin, 1917