


Diagnostic Alphabet Soup: Understanding How Fragile X Fits in With Educational and Behavioral Diagnoses

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Why Diagnose?

- Reimbursement
- Eligibility for services
- Treatment
- Research

What is Etiology?

- Etiology: Underlying cause
- Developmental disorders are symptoms for which there are many different etiologies
- Genetic and / or medical factors play a role in the etiology of most developmental disorders

Why is Etiology Important?

- Genetic counseling for families
- Alleviates guilt, misconceptions
- Anticipation of medical needs
- Insight into behavior, learning styles
- Support groups

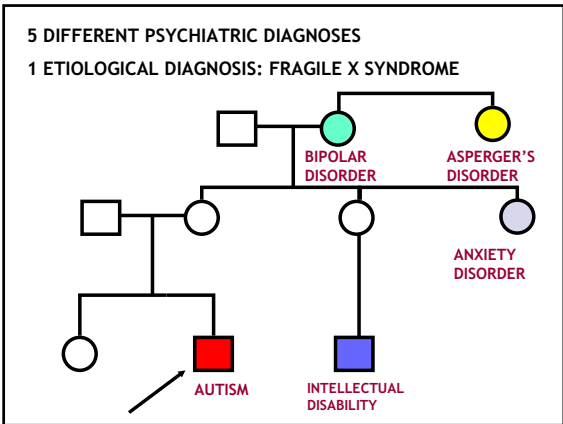
Psychiatric Diagnoses

ADD ANXIETY DISORDER OPPOSITIONAL DEFIANT DISORDER OCD

ADHD BIPOLAR DISORDER

AUTISTIC DISORDER MR (ID) IMPULSE CONTROL DISORDER

INTERMITTENT EXPLOSIVE DISORDER



Diagnostic Alphabet Soup



Etiological Diagnoses

In the school setting:

- Etiological diagnoses often considered irrelevant
- Educational / psychiatric diagnoses determine services and treatment approaches

Psychiatric Diagnoses

- based upon observed, recognizable patterns of human behavior
- diagnosed using criteria found in the DSM (Diagnostic & Statistical Manual)
- symptom diagnoses which do not emphasize etiology
- never diagnosed using laboratory tests

PSYCHIATRIC

CHILDHOOD DISORDERS

Attention Deficit /
Disruptive Behavior Disorders

Learning Disorders

Mental Retardation

Pervasive Developmental
Disorders

Tic Disorders

Etc., etc., etc.

Attention Deficit / Disruptive Behavior Disorders

- HD, ADD
- ADHD, ADHD-NOS,
- Conduct Disorder
- ODD (Oppositional Defiant Disorder)
- Disruptive Behavior - NOS

HA / ADD / ADHD

- Characterized by a majority of the following symptoms being present in either category (inattention or hyperactivity).
- Symptoms are inconsistent with the child's developmental level.

Symptoms of Inattention

- Fails to give close attention to details / makes careless mistakes
- Difficulty sustaining attention on tasks
- Does not seem to listen when spoken to directly
- Does not follow through on instructions / fails to finish schoolwork, chores, etc.

Symptoms of Inattention

- Avoids, dislikes tasks requiring sustained mental effort
- Loses things necessary for tasks, activities
- Easily distracted by extraneous stimuli
- Forgetful in daily activities
- Fidgets with hands / feet, squirms in seat

Symptoms of Hyperactivity

- Leaves seat in class / other situations when required to remain seated
- Runs about or climbs excessively in inappropriate situations
- “On the go”, acts as if “driven by a motor”
- Talks excessively

Symptoms of Impulsivity

- Blurts out answers before questions have been completed
- Has difficulty awaiting turn
- Interrupts or intrudes on others

HA / ADD / ADHD

- Symptoms have been present \geq 6 months
- Some symptoms present by age 7 years
- Symptoms must exist in at least 2 separate settings
- Symptoms create significant impairment in social, academic or occupational functioning or relationships

THE PDD UMBRELLA

- Pervasive Developmental Disorders (PDD): Formal DSM-IV Designation
- Autism Spectrum Disorders (ASD)
- In practice: ASD = PDD
- Subtypes under ASD, PDD umbrella:
 - Autistic Disorder (a.k.a. autism)
 - Asperger's Disorder
 - Childhood Disintegrative Disorder
 - Rett's Disorder
 - PDD-NOS



Autistic Disorder (Autism)



(I) Need 6 or more items from section A, B, and C with at least 2 from A and 1 each from B and C.

Autistic Disorder (Autism)



A) Qualitative impairment in social interaction as manifested by at least 2 of the following:

- Impairment in use of nonverbal behaviors
- Failure to develop peer relationships
- Lack of spontaneous seeking to share enjoyment, interests, etc. with others
- Lack of social or emotional reciprocity

Autistic Disorder (Autism)



B) Qualitative impairment in communication as manifested by at least 1 of the following:

- Delay in, or total lack of, spoken language
- Impairment in ability to initiate or sustain a conversation with others
- Stereotyped, repetitive, or idiosyncratic language
- Lack of make-believe or imitative play

Autistic Disorder (Autism)



C) Restricted, repetitive and stereotyped patterns of behavior, interests, and activities, as manifested by at least 2 of the following:

- Preoccupation with one or more stereotyped / restricted patterns of interest
- Adherence to routine, rituals
- Stereotyped / repetitive motor mannerisms
- Preoccupation with parts of objects

Autistic Disorder (Autism)



(II) Delays or abnormal functioning in at least 1 of the following areas, with onset prior to age 3 years:

- A. social interaction
- B. language as used in social communication
- C. symbolic or imaginative play

(III) Not better accounted for by Rett's or Childhood Disintegrative Disorder

Proposed Changes to the DSM



- DSM-V to be published in May, 2013
- PDD category to be replaced by ASD
- Subtypes of PDD eliminated (Asperger's, PDD-NOS, Rett syndrome, etc.) All will now fall under ASD category.
- MR to become ID; no specific IQ cutoff; instead, based on % of deviation from age-matched, culturally appropriate peers
- Intent is to clarify, streamline behavioral diagnostic categories

Intellectual Disability



- Significantly subaverage intellectual functioning (IQ of 70 or below)
- Deficits in adaptive functioning
- Onset prior to age 18

Dual Diagnosis



The co-occurrence of intellectual disability and psychiatric diagnoses in the same person

Conclusions



- Diagnostic confusion abounds!
- Psychiatric / behavioral symptoms:
Found in association with many genetic disorders, including fragile X syndrome
- Causes vs. symptoms: Important for parents and professionals to understand distinction

Conclusions



- Educational and behavioral diagnoses, not genetic diagnoses, determine eligibility and services within the school setting
- Individuals with fragile X syndrome often meet criteria for one or more behavioral / educational diagnoses

Conclusions



- Use these diagnoses for everything they're worth, realizing that fragile X syndrome is the underlying cause of the behavioral and intellectual symptoms

Conclusions



- School districts and teachers are unlikely to be familiar with fragile X
- This does NOT necessarily mean they are unable to provide excellent services
- An open mind, willingness to learn about fragile X syndrome, and a creative approach to meeting a child's needs are just as important as experience with the syndrome