

Making Sense of Genetic, Educational, and Psychiatric Diagnoses

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Why Diagnose?

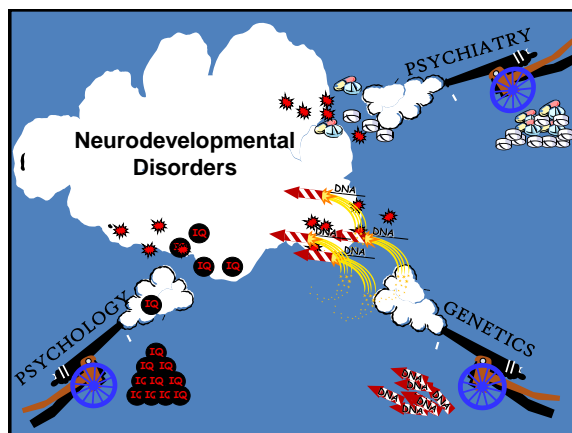
- Reimbursement
- Eligibility for services
- Treatment
- Research

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What is Etiology?

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

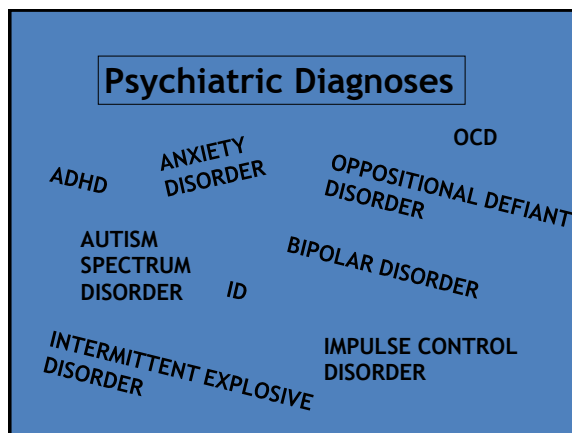
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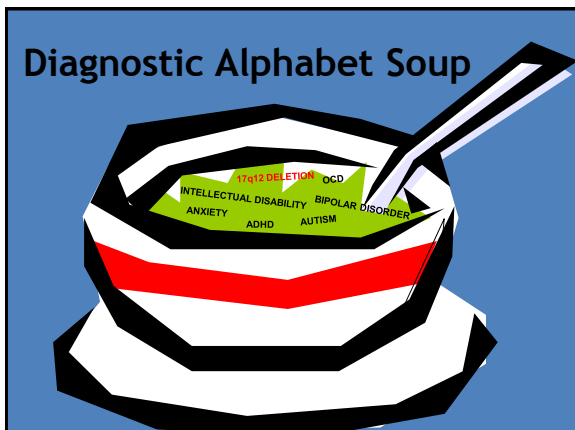


Psychiatric Diagnoses

- based upon observed, recognizable patterns of human behavior
- diagnosed using criteria found in the DSM (Diagnostic & Statistical Manual)
- symptom diagnoses that do not emphasize etiology
- not diagnosed using laboratory tests

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Autism Spectrum Disorders

- Autism Spectrum Disorders (ASD) – as of May 2013
- Formerly Pervasive Developmental Disorders (PDD)
- Previous subtypes under PDD now gone:
 - Autistic Disorder (a.k.a. autism)
 - Asperger's Disorder
 - Childhood Disintegrative Disorder
 - Rett's Disorder
 - PDD-NOS



The ASD Umbrella

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Autism Spectrum Disorder

- A. Persistent deficits in social communication and social interaction across multiple contexts, as manifested by deficits in:
- social or emotional reciprocity
 - use of nonverbal communicative behaviors used for social interaction
 - developing, maintaining, and understanding relationships

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Autism Spectrum Disorder

- B. Restricted, repetitive patterns of behavior, interests, or activities, as manifested by at least 2 of the following:
- Stereotyped or repetitive motor movements, use of objects, or speech
 - Insistence on sameness, routine, ritualized patterns of verbal or nonverbal behavior
 - Highly restricted, fixated interests that are abnormal in intensity or focus
 - Hyper- or hyposensitivity to sensory input or unusual interest in sensory aspects of the environment

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Autism Spectrum Disorder

- C. Symptoms must be present in the early developmental period
- D. Symptoms must cause clinically significant impairment in social, occupational, or other important areas of current functioning
- E. Not better explained by intellectual disability or global developmental delay

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Autism Spectrum Disorder

- Severity levels for
- Social Communication
 - Restricted, repetitive behaviors

- Level 3: "Requiring very substantial support"
 Level 2: "Requiring substantial support"
 Level 1: "Requiring support"

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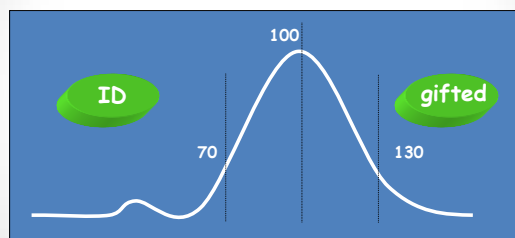
Intellectual Disability (ID)*

- Deficits in intellectual functioning (typically IQ of 70 or below)
- Deficits or impairments in adaptive functioning
 - Communication
 - Social skills
 - Personal independence at home or in community settings
 - School or work functioning
- Onset during the developmental period (up to ~age 18)

* Formerly known as mental retardation

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Distribution of IQ scores



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Conclusions

- Diagnostic confusion abounds!
- Psychiatric / behavioral / cognitive symptoms: Found in association with many genetic disorders, including 17q12 deletions and duplications
- Causes vs. Symptoms: Important for parents and professionals to understand distinction

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Conclusions

- Educational and behavioral diagnoses, not genetic diagnoses, determine eligibility and services within the school setting
- Individuals with 17q12 deletions and duplications often meet criteria for one or more behavioral / educational diagnoses
- Use these diagnoses for everything they're worth, realizing that 17q12 del /dup is the underlying cause of the learning / behavioral symptoms

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Conclusions

- Schools and teachers unlikely to be familiar with the 17q12 deletions and duplications
- This does NOT necessarily mean they are unable to provide excellent services
- An open mind, willingness to learn about 17q12, and a creative approach to meeting a child's needs are just as important as experience with the syndrome

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