



Making Sense of Genetic, Educational, and Psychiatric Diagnoses

Brenda Finucane, MS, LGC
Associate Director,
Autism & Developmental Medicine Institute
Geisinger Health System
Lewisburg, Pennsylvania

GEISINGER
REDEFINING BOUNDARIES

Why Diagnose?

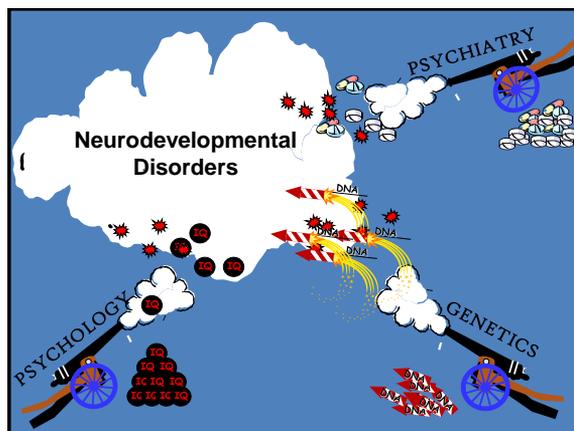
- Reimbursement
- Eligibility for services
- Treatment
- Research

GEISINGER
REDEFINING BOUNDARIES

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

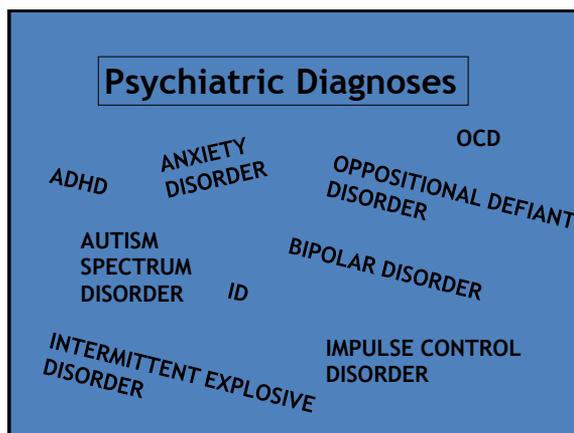
GEISINGER
REDEFINING BOUNDARIES

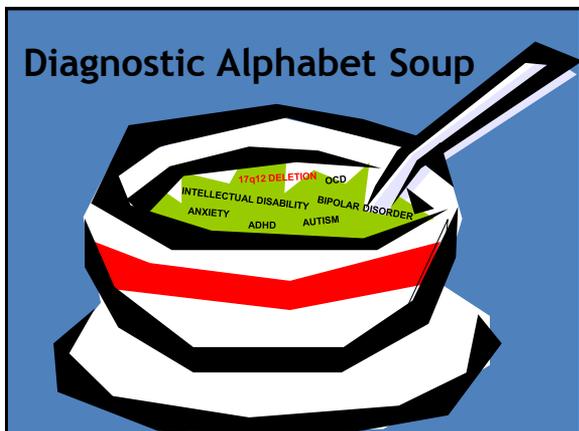


Psychiatric Diagnoses

- 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

GEISINGER
REDEFINING BOUNDARIES





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

GEISINGER
REDEFINING BOUNDARIES

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

GEISINGER
REDEFINING BOUNDARIES

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

GEISINGER
REDEFINING BOUNDARIES

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

GEISINGER
REDEFINING BOUNDARIES

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"

GEISINGER
REDEFINING BOUNDARIES

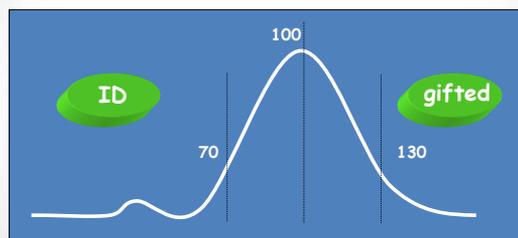
Intellectual Disability (ID)*

- 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

GEISINGER
REDEFINING BOUNDARIES

Distribution of IQ scores



GEISINGER
REDEFINING BOUNDARIES

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

GEISINGER
REDEFINING BOUNDARIES

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

GEISINGER
REDEFINING BOUNDARIES

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

GEISINGER
REDEFINING BOUNDARIES

Acknowledgements

Geisinger Health System

David Ledbetter, PhD	Laina Lusk, BS
Thomas Challman, MD	Steve Martin, BS
Inoka Devapriya, MD	Marissa Mitchell, MS
Megan Epler, BA	Scott Myers, MD
David Evans, PhD	Erin Riggs, MS
Andy Faucett, MS	Missy Stane, MS
Barbara Haas-Givler, MEd	

Bucknell

Heather Bothwell

Yale

Daniel Moreno De Luca, MD

Grant Support

Simons Foundation



GEISINGER
AUTISM & DEVELOPMENTAL MEDICINE

GeisingerADMI.org

...and all the families and individuals with genetic syndromes who partner with researchers to make advances possible

GEISINGER
REDEFINING BOUNDARIES