

Diagnostic Issues in Rett Syndrome: DSM-V, ICD-11, Rett Terminology

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Classifications of Diseases

- **International Classification of Diseases (ICD) system (WHO):** International classification of all disorders, used for diagnosis and billing. Currently using ICD-9. ICD 11 is next major revision.
- **Diagnostic and Statistical Manual of Mental Disorders (DSM) system (APA):** U.S.-based classification of (neuro)psychiatric disorders, which is used internationally. The next revision, DSM-V, is being developed.
- DSM-V will constitute the psychiatric disorders section of ICD-11.

Classification of Rett Syndrome

**Do we need to improve current classifications:
ICD-9 & DSM-IV?**

**RettSearch, the International Consortium of
Rett Syndrome Clinical Researchers, surveyed
its members: Yes, we need to improve ICD-9
and DSM-IV classifications of Rett.**



Classification of Rett Syndrome

- **ICD-9 and Rett:**

No specific code, under:

330.8 Other specified cerebral degenerations
 in childhood

Change: Rett syndrome is NOT a degenerative disorder.

Classification of Rett Syndrome

- **DSM-IV and Rett:**

A specific code under the Pervasive Developmental Disorder category (along Autism, Childhood Disintegrative Disorder, and Asperger)

299.80 Rett's Disorder

Change: Rett syndrome is NOT an autistic disorder.

Autistic features are transient

There are other (than autism) neurologic manifestations.

Classification of Rett Syndrome

- **Proposed for DSM-V (release: May 2012):**

No specific code for Rett

Diagnosis of Autism will include associated genetic disorder (Rett/*MECP2* mutation, Fragile X, etc.).

So, a girl with Rett could meet criteria for “Autism associated with *MECP2* mutation” during the regression period.

Classification of Rett Syndrome

- **DSM-V & Rett:**

1. Autism is a behavioral diagnosis, with multiple genetic (and other) causes.
2. New DSM-V guidelines for Autism may decrease the number of Rett girls meeting criteria.

Classification of Rett Syndrome

- **Rett and the “new genes”:**
 1. We are re-examining criteria for typical and atypical Rett. Emphasis: diagnosis is clinical.
 2. We are considering how new genes implicated in Rett (e.g., *CDKL5*) will be incorporated into classification.

Rett Syndrome in DSM-IV

PERVASIVE DEVELOPMENTAL DISORDERS

Pervasive Developmental Disorders are characterized by severe and pervasive impairment in several areas of development: reciprocal social interaction skills, communication skills, or the presence of stereotyped behavior, interests, and activities. The qualitative impairments that define these conditions are distinctly deviant relative to the individual's developmental level or mental age. This section contains Autistic Disorder, Rett's Disorder, Childhood Disintegrative Disorder, Asperger's Disorder, and Pervasive Developmental Disorder Not Otherwise Specified. These disorders are usually evident in the first years of life and are often associated with some degree of Mental Retardation, which, if present, should be coded on Axis II. The Pervasive Developmental Disorders are sometimes observed with a diverse group of other general medical conditions (e.g., chromosomal abnormalities, congenital infections, structural abnormalities of the central nervous system). If such conditions are present, they should be noted on Axis III. Although terms like "psychosis" and "childhood schizophrenia" were once used to refer to individuals with these conditions, there is considerable evidence to suggest that the Pervasive Developmental Disorders are distinct from Schizophrenia (however, an individual with Pervasive Developmental Disorder may occasionally later develop Schizophrenia).

299.00 Autistic Disorder

Diagnostic criteria for 299.00 Autistic Disorder

- A. A total of six (or more) items from (1), (2), and (3), with at least two from (1), and one each from (2) and (3):
1. qualitative impairment in social interaction, as manifested by at least two of the following:
 - a. marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
 - b. failure to develop peer relationships appropriate to developmental level
 - c. a lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., by a lack of showing, bringing, or pointing out objects of interest)
 - d. lack of social or emotional reciprocity

Rett Syndrome in DSM-IV

299.80 Rett's Disorder

Diagnostic criteria for 299.80 Rett's Disorder

- A. All of the following:
 - 1. apparently normal prenatal and perinatal development
 - 2. apparently normal psychomotor development through the first 5 months after birth
 - 3. normal head circumference at birth
- B. Onset of all of the following after the period of normal development:
 - 1. deceleration of head growth between ages 5 and 48 months
 - 2. loss of previously acquired purposeful hand skills between ages 5 and 30 months with the subsequent development of stereotyped hand movements (e.g., hand-wringing or hand washing)
 - 3. loss of social engagement early in the course (although often social interaction develops later)
 - 4. appearance of poorly coordinated gait or trunk movements
 - 5. severely impaired expressive and receptive language development with severe psychomotor retardation

Autism in DSM-V (in progress)

	Social Communication	Fixated Interests and Repetitive Behaviors
Most severe ASD	Minimal or no social communication	Nearly constant, complete preoccupation, strongly resists interference w/ ritual
Moderately severe ASD	Some social communication but interactions noticeably disturbed	Frequent and interfering rituals, repetitive behaviors and fixated interests
Less severe ASD	Clear impairments in social communication. Meets all diagnostic criteria including symptom severity greater than threshold	Occasional rituals, repetitive behaviors and fixated interests; some interference
Subclinical AS Symptoms	Has some symptoms from one or both domains but no significant interference or impairment.	Odd mannerisms, some excessive preoccupations but distractible, may have ritualized behaviors but they don't interfere with daily activities
Normal Variation	Socially isolated or "awkward"	Some ritualized behaviors and preoccupations but these are normal for developmental stage and cause no interference