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DIAGNOSTIC AND STATISTICAL MANUAL OF MENTAL DISORDERS (DSM-5.0)

RELEASED MAY 22 SIGNIFIES CHANGE FOR RETT SYNDROME

The release of the DSM-5.0 marks a decade's-long journey in revising the criteria for the diagnosis and classification of mental disorders. In previous versions of the Diagnostic and Statistical Manual of Mental Disorders, Rett syndrome has been classified as one form of the autism spectrum disorders (ASD). The DSM-5.0 will emphasize that autism is a behavioral diagnosis with multiple causes, both genetic and other unknown. Using this edition, specific syndromes that have autistic features will not be bundled under broad diagnosis of autism, but classified by their specific diagnosis code, which already exists for most genetic syndromes. There have been questions about the new DSM-5.0 edition and the status of Rett syndrome, and Paige Nues, Director of Family Support, easily simplified for us what these changes mean to Rett syndrome:

"It is the definition of autism that is changing, not the definition of Rett syndrome.

Therefore, instead of Rett = ASD, now Rett = Rett, and autism becomes a part of the constellation of diagnoses she may carry.

For example,

Alice has **Rett syndrome**, confirmed by clinical features and MECP2 screening (genetic dx)
AND Alice has **epilepsy**, confirmed by video EEG monitoring and frequency of episodes (neurologic dx)
AND Alice has **dysphagia**, confirmed by swallow study and GI/OT/SLP evaluation (gastroenterologist dx)
AND Alice has autism, as confirmed by autism-specific behavioral evaluations (developmental pediatrician, pediatric neurologist, psychologist, psychiatrist dx)
AND Alice has **Long QT**, as confirmed by EKG monitoring (cardiologist dx)
AND Alice has **scoliosis**, as confirmed by X-ray (orthopedic dx)

From our perspective, the genetic diagnosis should lead as the core identifier for the child. It is the first lens we should apply when assessing treatment, care, services, supports, interventions, community. And from Rett syndrome will stem these other branches of the decision tree which practitioners will then do their differential for the other diagnoses that often accompany Rett syndrome."

How is RTT diagnosed?
The diagnosis of RTT depends on very specific criteria that have been considered by an international group of specialists and revised based on refinement of information. These revised diagnostic criteria have been published broadly and are necessary to maintain diagnostic

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consistency worldwide and to direct public health and public policy. This is an essential element of disease classification. In most cases, classifying RTT as an autism spectrum disorder is not accurate so the decision to reclassify it as a separate diagnosis from autism makes sense.

Why were these changes to the DMS-5.0 made?
The DSM is a clinical classification. The changes to DSM-5.0 were made to permit a behaviorally based diagnosis (autism spectrum disorder - ASD) in the presence of another disorder (such as Rett) IF/WHEN the patient has symptoms that meet criteria for ASD.

The basis of the reclassification of RTT includes the following:
1) Autistic features are usually transient and do not occur in every person with RTT;
2) Physical and neurologic manifestations occur in RTT that are not part of autism;
3) It improves accuracy to use a more specific coded identifier for known genetic disorders including those that may include autism as an additional feature such as fragile X syndrome, tuberous sclerosis, Down syndrome, and RTT.

During the transient period during which individuals with RTT may show autistic features, they could be categorized as RTT and autism (Autism Spectrum Disorder associated with RTT or with a MECP2 mutation).

What should clinicians know?
Specifiers in DSM-5.0 for ASD were added: "Associated with known medical or genetic condition or environmental factor" so that clinicians could indicate that the ASD was related to MECP-2 or other genetic conditions (or associated with epilepsy, or fetal alcohol exposure, etc).

Clinicians should use the specifiers to describe more fully each individual patient's presentation. If they do, the ASD diagnosis will be amplified by information that provides information about the individual's level of functioning, specific strengths and weaknesses, as well as co-occurring conditions.

A final note
While Rett syndrome may not officially be an ASD in the DSM-5.0, the link to autism remains. Many children are diagnosed as autistic before the MECP2 mutation is identified and then the diagnosis is revised to RTT. Rett syndrome also remains a clinical diagnosis, so Rett syndrome can still be diagnosed without an identified MECP2 mutation. Autistic traits do occur, especially during the regression, and sometimes these traits persist. Using the new DSM-5.0, we expect many of the children will carry dual diagnoses of RTT and autism. Not all, but many. We understand that the new DSM-5.0 may cause confusion among many individuals and groups, but this is a natural evolution of the classification process responding to new understanding of the essential elements of each specific entity being classified.