

## Testing and Diagnosis

Rett syndrome is most often misdiagnosed as autism, cerebral palsy, or non-specific developmental delay. In the past, making the correct diagnosis called not only for a long list of diagnostic tests and procedures to rule out other disorders, but it also took from months to years waiting to confirm the diagnosis as new symptoms appeared over time. Today, we have a simple blood test to confirm the diagnosis. However, since we know that the MECP2 mutation is also seen in other disorders, the presence of the MECP2 mutation in itself is not enough for the diagnosis of Rett syndrome. Diagnosis requires either the presence of the mutation (a molecular diagnosis) or fulfillment of the diagnostic criteria (a clinical diagnosis, based on signs and symptoms that you can observe) or both. Below is a list of labs to share with your ordering physician that can do the MECP2 sequencing + deletion analysis, and the list of diagnostic criteria.

### Revised Diagnostic Criteria for Rett Syndrome (RTT) 2011

#### Requirements

Consider diagnosis when postnatal deceleration of head growth observed.

Required for typical or classic RTT

Required for atypical or variant RTT

A period of regression followed by recovery or stabilization  
All main criteria and all exclusion criteria  
Supportive criteria are not required, although often present in typical RTT

A period of regression followed by recovery or stabilization  
At least 2 of the 4 main criteria  
5 out of 11 supportive criteria

## Criteria

### Main Criteria

Partial or complete loss of acquired purposeful hand skills  
Partial or complete loss of acquired spoken language  
Gait abnormalities: Impaired (dyspraxic) or absence of ability  
Stereotypic hand movements such as hand wringing/squeezing, clapping/tapping, mouthing and washing/rubbing  
automisms

### Exclusion criteria for typical RTT

Brain injury secondary to trauma (peri- or postnatally), neurometabolic disease, or severe infection that causes neurological problems  
Grossly abnormal psychomotor development in first 6 months of life

### Supportive criteria for atypical RTT

Breathing disturbances when awake  
Bruxism when awake  
Impaired sleep pattern  
Abnormal muscle tone  
Peripheral vasomotor disturbances  
Scoliosis/kyphosis

Growth retardation  
Small cold hands and feet  
Inappropriate laughing/screaming spells  
Diminished response to pain  
Intense eye communication—eye pointing

Clinical Diagnosis Revised Criteria Table 2011 155.29 Kb

Revised RTT Diagnostic Criteria Neul ANA 2010 151.37 Kb

## GENETIC TESTING

### Reasons for Genetic Testing

Mutations in another gene on the X-chromosome known as CDKL5 (cyclin-dependent kinase-like 5) can cause an atypical form of Rett Syndrome called the early-onset seizure variant. These individuals have generally tested negative for a MECP2 mutation. Not everyone with a CDKL5 mutation appears as atypical RTT. Other CDKL5 disorders include Infantile Spasms, West Syndrome, Early Onset Seizures, and Autism. CDKL5 mutation testing is not routinely available through most diagnostic labs. If you think your child should have this testing, you should discuss it further with your pediatrician, neurologist, or geneticist. For more information visit <http://www.cdkl5.com/>

### Testing Centers

- DNA Diagnostic Testing Laboratory

Baylor of Medicine

Houston, TX

Tel: 713-798-6555 or 1-800-411-4363 (GENE)

- Dr. Mike Friez

Greenwood Genetic Center

Greenwood, SC

Tel: 864-941-8130 or 1-888-442-4363 (GGC-GENE)

- Iris L. Gonzalez, Ph.D.

Molecular Diagnostics Laboratory

Wilmington, DE

Tel: 302-651-6779/6777

- Center for Human Genetics

Boston University School of Medicine

Boston, MA

Tel: 617-638-7083

- University of Chicago

Genetic Services Laboratories

Chicago, IL

Tel: 888-824-3637 or 1-888-824-3637 (UC-GENES)

- Children's Hospital Boston

DNA Diagnostic Laboratory

Boston, MA

617-355-7582

- GeneDX

207 Gaithersburg, MD 20877

[www.genedx.com](http://www.genedx.com)

301-519-2100

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