

Diagnostic Criteria for Rett Syndrome

Rett Syndrome is a clinical diagnosis. Please check all of the following diagnostic criteria that apply for your client.

Typical Rett Syndrome Diagnosis:

Essential criteria:

Must have had a period of normal development until between 6 to 18 months followed by a loss of skills, then recovery or stabilization of skills

Partial or complete loss of purposeful hand skills

Partial or complete loss of spoken language

Dyspraxic gait (often an unsteady, wide-based and stiff-legged gait)

Repetitive hand movements (such as hand wringing or squeezing, clapping or tapping, or hand mouthing movements)

Exclusion criteria:

A child with any of the following criteria does not have Rett syndrome:

Neurometabolic disease or other inherited degenerative disorder

Neurological disorder resulting from severe infection or head trauma

Evidence of brain damage acquired after birth

Grossly abnormal development in the first six months of life

Atypical Rett Syndrome Diagnosis:

Essential criteria:

Must have had a period of normal development until between 6 to 18 months followed by a loss of skills, then recovery or stabilization of skills.

Require 2 of the 4 Essential Criteria AND 5 of the 11 Supportive Criteria

Partial or complete loss of purposeful hand skills

Partial or complete loss of spoken language

Dyspraxic gait (often an unsteady, wide-based and stiff-legged gait)

Repetitive hand movements (such as hand wringing or squeezing, clapping or tapping, or hand mouthing movements)

Supportive criteria:

These include symptoms that are not necessary for a diagnosis of Typical Rett syndrome but are often present in people with the condition. A child who has supportive criteria, but NO essential criteria does not have Rett syndrome.

Breathing irregularities while awake, such as apnea, hyperventilation and air swallowing

Teeth-grinding

Abnormal sleep patterns

Abnormal muscle tone (hypotonia, rigidity or spasticity)

Poor circulation in the hands and feet, with cold and bluish to red hands and feet

Scoliosis or kyphosis (curvature of the spine)

Delayed growth

Small hands and feet

Inappropriate laughing or screaming spells

Reduced response to pain

Intense eye communication or “eye pointing”

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Neul, J., Kaufmann, W., Glaze, D., Christodoulou, J., et al. (2010). Rett Syndrome: Revised Diagnostic Criteria and nomenclature. Ann Neurol. Available at PMC 2011 December 1.