



Empowering Children. Supporting Families. Building Brighter Futures.

Resource 066 | Genetic and chromosomal conditions

Rett syndrome

What it means

Rett syndrome is a genetic neurodevelopmental condition often recognized when a child loses previously acquired hand skills and spoken language. Movement, communication, breathing patterns, and other body functions may be affected. It occurs mainly in girls, but boys can also be affected. Abilities require careful individual assessment. [1]

Causes and risk factors

Many cases involve a disease-causing change in the MECP2 gene. The change is often new in the child rather than inherited. MECP2 is important for brain function. Not every MECP2-related condition is classic Rett syndrome, and the genetic finding must be interpreted with clinical features. [1,2]

General family education. A concern deserves assessment, but this guide does not diagnose a child or prescribe treatment.



Signs and everyday impact

After a period of development, a child may lose purposeful hand use or speech and develop repetitive hand movements. Walking difficulties, altered breathing while awake, sleep problems, feeding concerns, seizures, and scoliosis may occur. Limited speech or movement should not be treated as a reliable measure of understanding. [1,2]

Assessment and diagnosis

A specialist evaluates the developmental course and clinical diagnostic criteria, with genetic testing to support clarification. Assessment also considers other causes of regression. Ongoing review includes nutrition, mobility, breathing, heart rhythm, bone health, seizures, and communication access. Care plans should reflect the child's actual abilities and needs. [1,2]

Treatment and therapy

Care is multidisciplinary, including seizure management, nutrition, physical and occupational therapy, orthopedic care, and AAC adapted to motor access. Eye-gaze systems may help selected children communicate. Where approved and appropriate, a specialist may discuss trofinetide, including benefits and adverse effects. It is not a cure and does not replace comprehensive supportive care. [1,2,3]

Support at home

Offer choices through accessible communication and allow ample response time. Follow individualized positioning, mobility, feeding, and seizure plans. Keep enjoyable social activities part of everyday life. Report discomfort, changed breathing, constipation, and sleep problems, because limited speech may make physical symptoms harder to express.

When to seek help

Seek prompt assessment for skill loss, new seizures, feeding deterioration, or major changes in breathing. Emergency services are needed for severe breathing difficulty, collapse, or prolonged seizures according to the action plan.

Questions for your care team

- Which communication system matches my child's access needs?
- What heart, bone, feeding, and breathing monitoring is due?
- Are any condition-specific treatments appropriate and available?



Podcast preparation

Use this outline to develop a later podcast script.

Suggested opening

Welcome to Mariel Early Intervention. Today we discuss rett syndrome. Rett syndrome is a genetic neurodevelopmental condition often recognized when a child loses previously acquired hand skills and spoken language. This is general education; each child needs individualized care.

Opening: Define the topic and its developmental context.

Understanding: Explain causes and signs without inviting self-diagnosis.

Assessment: Describe evaluation and distinguish screening from diagnosis.

Support: Discuss treatment, therapy, and practical family supports.

Closing: Explain urgent signs and one clear next step.

Misconception to address: Myth: little speech means little understanding. Communication access and careful assessment are essential.

Before recording: update clinical references and local service details, arrange clinical review, and use fully consented or fictional examples.

Terms in plain language

- MECP2: a gene involved in regulating brain-cell function.
- Stereotyped movements: repeated movements with a similar pattern.
- Eye-gaze AAC: a communication system controlled through eye movement.

Sources and further reading

Sources accessed 3 October 2026. Numbered references support the clinical overview. Treatment availability and referral routes vary by country.

[1] Rett syndrome: MedlinePlus Medical Encyclopedia

<https://www.medlineplus.gov/ency/article/001536.htm>

[2] Rett Syndrome

<https://www.ninds.nih.gov/health-information/disorders/rett-syndrome>

[3] Epilepsy and Seizures

<https://www.ninds.nih.gov/node/647>