

# Electronic Medical Record (EMR) Summary

Patient ID: PID69083906

Name: Sara

Age: 2, Sex: Female

Visit ID: VISIT61831716

Date: 2025-05-17 14:36

## Clinical Reasoning Summary

### **\*\*Definition & Key Concerns\*\***

The clinical features described suggest a neurodevelopmental disorder known as Rett syndrome. This is a rare genetic disorder that primarily affects girls and leads to severe cognitive and physical impairments. Key concerns include developmental regression, loss of purposeful hand skills replaced with repetitive movements (such as hand-wringing), slowed growth, and autistic-like behaviors.

### **\*\*Differential Diagnosis\*\***

1. Rett Syndrome: The combination of developmental regression, loss of hand skills, hand-wringing movements, and autistic-like behaviors is highly characteristic of Rett syndrome.
2. Childhood Disintegrative Disorder: This condition also involves regression of skills, but it typically occurs later (around age 3-4 years) and does not usually involve the characteristic hand movements seen in Rett syndrome.
3. Autism Spectrum Disorder: While some symptoms overlap, the loss of purposeful hand movements and the specific hand-wringing behavior are more characteristic of Rett syndrome.

### **\*\*Can't-Miss Diagnosis\*\***

Rett Syndrome is the can't-miss diagnosis in this case due to the characteristic clinical features described.

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## **\*\*Suggested Investigations\*\***

1. Genetic testing: A mutation in the MECP2 gene is found in the majority of girls with Rett syndrome.
2. Neuroimaging: To rule out other causes of developmental regression.
3. EEG: Some girls with Rett syndrome may have seizures, so an EEG may be useful.

## **\*\*Management Plan\*\***

There is currently no cure for Rett syndrome, so management is supportive and includes:

1. Physical therapy: To maintain mobility and prevent contractures.
2. Speech therapy: To maximize communication skills.
3. Occupational therapy: To promote independence in daily activities.
4. Nutrition management: To ensure adequate growth and development.
5. Regular follow-up with a neurologist: To manage any associated neurological issues such as seizures.

## **\*\*Reference Insight\*\***

According to UpToDate (2023), the diagnosis of Rett syndrome is based on clinical criteria, including the loss of purposeful hand skills between ages 5 and 30 months, stereotypic hand movements (such as hand-wringing), and deceleration of head growth. Genetic testing can confirm the diagnosis in the majority of cases.

## **Rare Disease Alerts**

None triggered

## **Prescription**

None provided