

# Electronic Medical Record (EMR) Summary

Patient ID: PID69083906

Name: Sara

Age: 2, Sex: Female

Visit ID: VISIT31846752

Date: 2025-05-17 16:37

## Clinical Reasoning Summary

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

The clinical features presented by the patient suggest a neurodevelopmental disorder. The specific syndrome that these symptoms suggest is Rett Syndrome, a rare genetic disorder that primarily affects girls and leads to severe cognitive and physical impairments.

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

1. Rett Syndrome: This is the most likely diagnosis given the loss of purposeful hand skills, development of repetitive hand movements (hand-wringing), loss of speech, and deceleration of head growth after a period of normal development.
2. Childhood Disintegrative Disorder: This condition also involves a period of normal development followed by significant loss of skills. However, it typically occurs later (around 2-4 years of age) and does not typically involve the characteristic hand movements seen in Rett Syndrome.
3. Autism Spectrum Disorder: While some autistic-like behaviors are present, the loss of purposeful hand movements and deceleration of head growth are not typical features of autism.

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

Rett Syndrome is the critical high-risk condition that must be ruled out given the patient's symptoms.

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

1. Genetic Testing: A definitive diagnosis of Rett Syndrome can be made through identification of a mutation in the MECP2 gene, which is found in the majority of cases.
2. EEG, MRI, and other neuroimaging may be helpful to rule out other causes of developmental regression.

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

There is no cure for Rett Syndrome, so management is supportive and aimed at maximizing the patient's abilities and quality of life. This may include:

1. Physical, occupational, and speech therapy to promote skills and abilities.
2. Management of associated symptoms and complications (e.g., antiepileptic drugs for seizures, laxatives for constipation).
3. Regular follow-up with a multidisciplinary team including neurology, genetics, and developmental pediatrics.

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

According to UpToDate (2023), the diagnosis of Rett Syndrome is primarily clinical, based on the characteristic pattern of normal development followed by regression and the development of distinctive hand movements. However, genetic testing can confirm the diagnosis in the majority of cases. The management of Rett Syndrome is supportive and involves a multidisciplinary approach.

## **Rare Disease Alerts**

Rett Syndrome (matched 3 symptoms)

## **Prescription**

None provided