

Electronic Medical Record (EMR) Summary

Patient ID: PID69083906

Name: Sara

Age: 2, Sex: Female

Visit ID: VISIT31846752

Date: 2025-05-17 16:39

Clinical Reasoning Summary

****Definition & Key Concerns****

The clinical picture presented suggests a neurodevelopmental disorder, specifically Rett syndrome. This is a rare genetic disorder that predominantly affects females and leads to severe cognitive and physical impairments. Key features include normal early development followed by loss of purposeful hand skills and speech, stereotypic hand movements (such as hand-wringing), and deceleration of head growth.

****Differential Diagnosis****

1. Rett syndrome: This is the most likely diagnosis given the characteristic loss of purposeful hand movements, development of stereotypic hand movements, and deceleration of head growth.
2. Autism Spectrum Disorder: Some of the symptoms such as loss of social skills and language could suggest autism, but the specific hand movements and deceleration of head growth are more characteristic of Rett syndrome.
3. Childhood disintegrative disorder: This is a rare condition characterized by late onset (usually between 2 and 4 years of age) of developmental delays in language, social function, and motor skills. However, the hand-wringing is not a typical feature of this disorder.

****Can't-Miss Diagnosis****

Rett syndrome is the critical high-risk condition that must be ruled out given the characteristic symptoms and the potential for severe cognitive and physical impairments.

****Suggested Investigations****

1. Genetic testing: A definitive diagnosis of Rett syndrome can be made through identification of a mutation in the MECP2 gene.

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2. EEG: This may be performed to evaluate for seizures, which are common in Rett syndrome.
3. MRI: This can be used to rule out other causes of neurodevelopmental regression.

****Management Plan****

There is currently no cure for Rett syndrome. Management is supportive and includes:

1. Physical therapy to help with motor skills.
2. Speech therapy to improve any remaining language skills.
3. Occupational therapy to help with self-care tasks.
4. Nutritional support, as feeding difficulties are common.
5. Regular monitoring for scoliosis and cardiac issues, which are common in Rett syndrome.

****Reference Insight****

According to UpToDate, the diagnosis of Rett syndrome is primarily clinical, based on the characteristic history and physical findings. However, genetic testing can confirm the diagnosis in the majority of cases. The management of Rett syndrome is multidisciplinary and focuses on improving the quality of life and maximizing the child's abilities.

Reference: UpToDate. Rett syndrome: Clinical features and diagnosis. Updated 2023.

Rare Disease Alerts

Rett Syndrome (matched 2 symptoms)

Prescription

None provided