ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/25mar1447

The Shrunken and Bright Cerebellum: A Rare Case Series Report in Congenital Disorders of Glycosylation Type 1a and Marinesco-Sjogren Syndrome

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Publication Date: 2025/04/04

Abstract: This case series explores the rare imaging findings of a shrunken and bright cerebellum in patients diagnosed with Congenital Disorders of Glycosylation Type 1a (CDG-Ia) and Marinesco-Sjogren syndrome (MSS). Both disorders are characterized by autosomal recessive inheritance and are associated with cerebellar atrophy, vermian hypoplasia, and cognitive delays. Magnetic Resonance Imaging (MRI) plays a critical role in diagnosing these conditions. This paper presents two case reports, highlighting characteristic imaging findings and genetic investigations, and discusses the underlying pathophysiology of these rare genetic syndromes.

Keywords: *Marinesco-Sjogren Syndrome, Congenital Disorders of Glycosylation, Cerebellar Atrophy, MRI, Genetic Disorders.*

How to Cite: Dr. Harshendra S. G.; Dr. Pradeep Goudar. (2025) The Shrunken and Bright Cerebellum: A Rare Case Series Report in Congenital Disorders of Glycosylation Type 1a and Marinesco-Sjogren Syndrome.
International Journal of Innovative Science and Research Technology,
10(3), 2010-2012. https://doi.org/10.38124/ijisrt/25mar1447.

I. INTRODUCTION

Congenital disorders of glycosylation (CDG) and Marinesco-Sjogren syndrome are both rare, autosomal recessive conditions that primarily affect the cerebellum, among other systems. These syndromes share similar clinical features, including developmental delay, ataxia, and cognitive impairment, but they are distinct in their genetic causes and specific imaging characteristics. This paper aims to enumerate the characteristic MRI findings observed in these disorders and discuss their clinical implications.

II. CASE REPORTS

A. Case 1:

Marinesco-Sjogren Syndrome A 4-year-old female patient presented with developmental delay, walking imbalance, and squinting of the eyes. The patient had no history of seizures or myoclonic jerks, and the birth history was uneventful.

- **MRI Findings:** demonstrated hyperintensity in the cerebellar white matter compared to the supratentorial white matter, as seen on T2-weighted images. T2 hyperintensities were also noted in the dentate nucleus bilaterally. There was a significant reduction in vermian volume, along with folial volume loss. The anterior lobe of the cerebellum showed fissural enlargement, and the inferior vermis was flattened. An enlarged fourth ventricle was also observed.

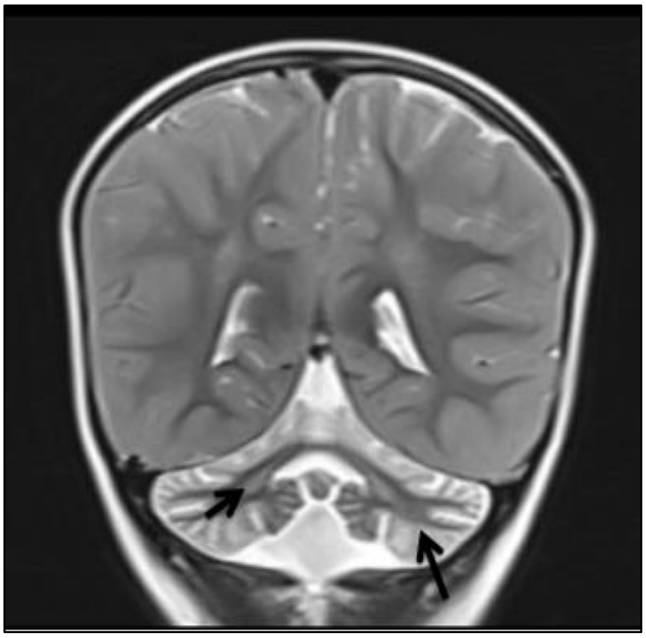

Fig 1: Coronal T2 Image Shows Hyperintensities in the Dentate Nucelus on Both Sides

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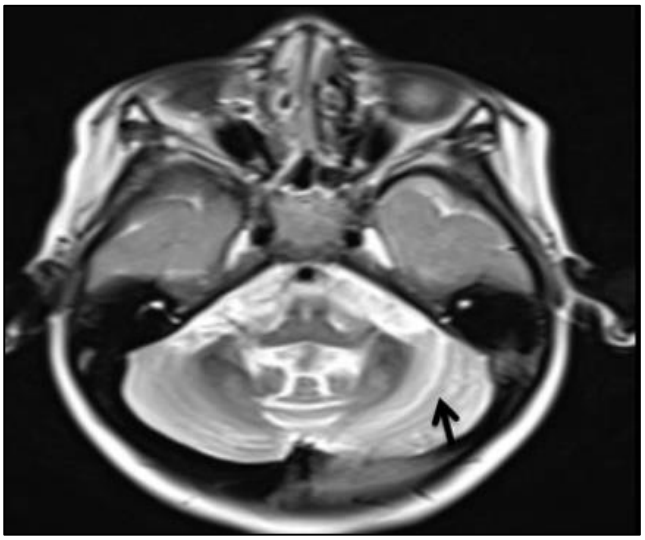

Fig 2: Axial T2 Image Shows Cerebellar with Vermian Hypoplasia

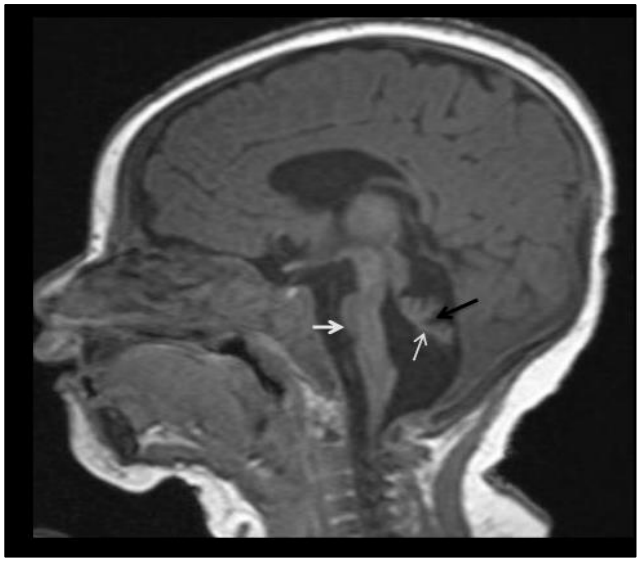

Fig 3: Sagittal Image Shows Shruken Appearance of Vermis

- **Final Diagnosis:** Marinesco-Sjogren syndrome
- **Genetic Workup:** SIL1 gene mutation (autosomal recessive inheritance)

B. Case 2:

- **Congenital Disorders of Glycosylation Type 1a** A 9-month-old girl born to consanguineous parents presented with delayed crying, neonatal seizures, cluster spasms, and developmental delays.
- **MRI Findings:** revealed slight hyperintensities in the cerebellar white matter, as compared to the supratentorial white matter, on T2 and FLAIR sequences. The cerebellum showed atrophy, with widened interfolial spaces. There was a notable reduction in pontine protrusion, and the vermis appeared shrunken with folial volume loss. Fissural enlargement was observed in the anterior lobe, and the inferior vermis was flattened. Additionally, there was an enlargement of the fourth ventricle.

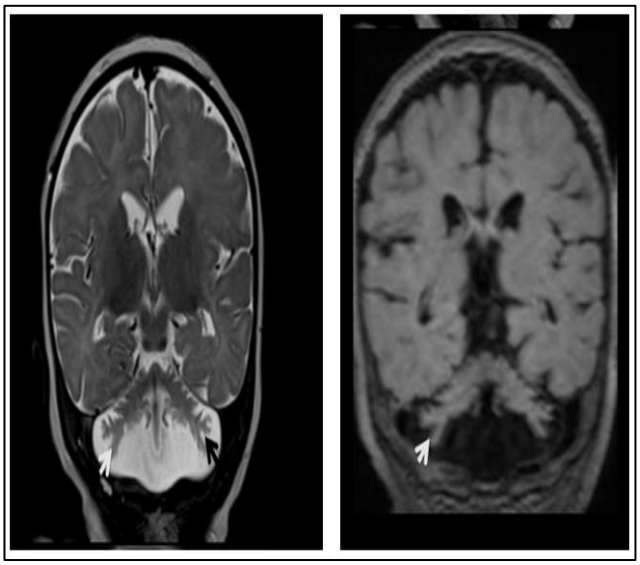

Fig 4: Coronal T2 and Flair Images Show Hyperintesne Cerebellar White Matter with its Atrophy

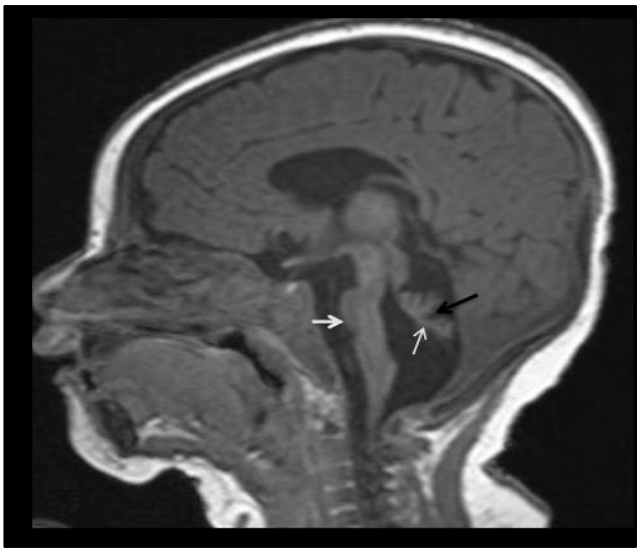

Fig 5: Sagittal Image Shows Reduced Pontine Protuberance with Shrunken Ppearance of Vermis

- **Final Diagnosis:** Congenital Disorders of Glycosylation Type 1a
- **Genetic Workup:** DPM3 gene mutation (Autosomal Recessive Inheritance)

III. DISCUSSION

Both Marinesco-Sjogren syndrome and CDG-Ia present with similar imaging findings that include cerebellar atrophy, vermian hypoplasia, and enlarged fourth ventricle. These findings are critical for differentiating these rare conditions from other neurodegenerative or metabolic disorders.

Marinesco-Sjogren Syndrome (MSS) is an autosomal recessive disorder caused by mutations in the SIL1 gene. Characteristic features of MSS include cerebellar ataxia, congenital cataracts, hypogonadotropic hypogonadism, and developmental delay. Imaging findings typically include T2 and FLAIR hyperintensities in the cerebellar cortex and

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dentate nucleus, widened cerebellar fissures, and pontine hypoplasia. MRI also often reveals vermian hypoplasia and an enlarged fourth ventricle (2 & 3).

Congenital Disorders of Glycosylation Type 1a (CDG-Ia) result from mutations in the PPM2 or DPM3 genes, which impair the glycosylation process. These patients often present with hypotonia, psychomotor retardation, and failure to thrive in infancy. MRI findings in CDG-Ia are similar to those in MSS, including cerebellar atrophy, widened fissures, and pontine hypoplasia. However, CDG-Ia patients are more likely to present with systemic manifestations such as coagulation disorders and liver dysfunction (1).

Both conditions demonstrate a shrunken and bright cerebellum on MRI, a crucial radiological feature for diagnosis. While the clinical presentation and genetic workup help differentiate between these two disorders, imaging plays an essential role in guiding clinicians toward an accurate diagnosis.

IV. CONCLUSION

Marinesco-Sjogren syndrome and Congenital Disorders of Glycosylation Type 1a are rare genetic disorders that manifest with characteristic MRI findings, including cerebellar atrophy, vermian hypoplasia, and enlarged ventricles. These disorders are typically diagnosed through genetic testing, which highlights the importance of a comprehensive evaluation of clinical, genetic, and imaging data. Awareness of these rare conditions is crucial for timely diagnosis and management.

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