



An Interesting MRI Brain Finding in a Young Male Patient Presented with Slowly Progressive Spastic Quadriparesis

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Received Date: June 10, 2024; **Published Date:** June 26, 2024

Abstract

A 29-year-old male presented with progressive spastic quadriparesis over two years, predominantly affecting the lower limbs, with associated stiffness, gait disturbances, and urinary urgency. Neurological examination revealed symmetric spastic quadriparesis, hyperreflexia, and a positive Babinski sign, but no sensory or cognitive deficits. MRI brain imaging displayed the distinctive “wine glass” sign, indicating symmetric hyperintensities in the corticospinal tracts, suggestive of bilateral corticospinal tract involvement. This radiological feature is associated with hereditary spastic paraplegia (HSP), particularly autosomal recessive spastic paraplegia type 7 (SPG7). The patient was managed with physical therapy and antispasticity medications. The “wine glass” sign is a significant radiological marker in diagnosing neurodegenerative conditions like HSP, primary lateral sclerosis (PLS), and leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation (LBSL). This case underscores the importance of this MRI finding in guiding the diagnosis and management of spastic quadriparesis in young patients.

Keywords: Spastic Quadriparesis; Wine Glass Sign; Posterior Limb; Spastic Quadriparesis

Abbreviations: HSP: Hereditary Spastic Paraplegia; SPG7: Spastic Paraplegia Type 7; PLS: primary lateral sclerosis; LBSL: leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation.

Presentation

A 29-year-old male presented with slowly progressive symmetric weakness predominantly involving the lower limbs and stiffness in all four limbs over the past two years. The patient reported difficulty in walking, frequent falls, and stiffness of all 4 limbs. He also had difficulty in micturition in the form of urgency, hesitancy and occasional urge incontinence. There were no sensory deficits, and cognitive functions were intact. The family history was not significant.

On examination, the patient exhibited symmetric spastic quadriparesis with hyperreflexia including bilateral patellar and ankle clonus. Babinski sign was positive bilaterally. He had spastic dysarthria and UMN type bladder involvement. No signs of cognitive, sensory or cranial nerve involvement. Fundoscopy was normal. His cerebellar function was normal and there was no extrapyramidal or autonomic sign. His gait was found to be spastic ataxic type.

Laboratory and Imaging Studies

Routine blood tests, including metabolic panel and vitamin levels, were normal. His MRI spinal cord screen didn't document any cord compression. Genetic testing was pending at the time of initial evaluation.

MRI Findings

An MRI of the brain, T2-weighted axial images and FLAIR images revealed symmetric hyperintensity in the bilateral midbrain, pons, and thalamus, posterior limb of internal capsule and splenium of the corpus callosum suggesting selective involvement of the white matter tracts (Figures 1 & 2). Rest of the other brain imaging was normal. This kind

of distinctive MRI brain finding is known as “wine glass” sign. This finding was suggestive of bilateral corticospinal tract involvement. The “wine glass sign” is characterized by symmetric hyperintensities on T2-weighted MRI images of the areas corresponding to the corticospinal tracts. This shape resembles a wine glass, with the “stem” representing the corticospinal tracts.

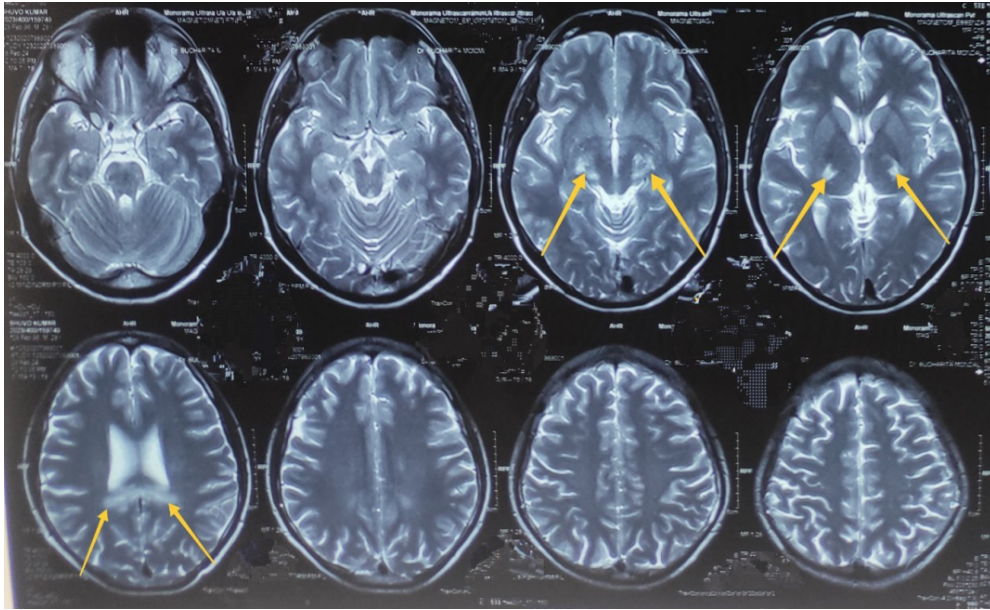


Figure 1: MRI of the brain, T2-weighted axial images showing symmetric hyperintensities in the white matter tracts of brain (yellow arrow).

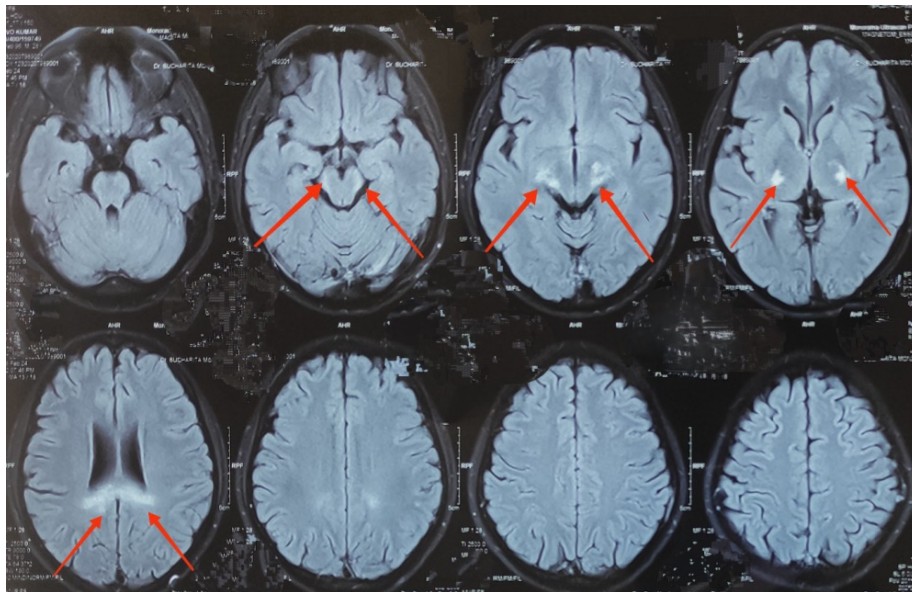


Figure 2: MRI of the brain, FLAIR images showing symmetric hyperintensities in the white matter tracts of brain (red arrow).

Diagnosis

Based on the clinical presentation and MRI findings, a provisional diagnosis of hereditary spastic paraplegia (HSP) possibly autosomal recessive spastic paraplegia type 7 related to the SPG7 gene mutation was made, pending confirmation from genetic testing.

Management and Follow-up

The patient was started on physical therapy and antispasticity medications, including baclofen. Genetic counseling was provided. The confirmation of genetic testing couldn't be done as the patient was lost to our follow up.

Review of Literature

The "wine glass sign" on MRI brain imaging is an important diagnostic feature in identifying several neurological disorders, particularly those involving the corticospinal tracts. This review expands on the significance of this sign in conditions such as hereditary spastic paraplegia (HSP), primary lateral sclerosis (PLS), and leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation (LBSL).

Hereditary Spastic Paraparesis (HSP)

HSP encompasses a group of genetic disorders characterized by progressive spasticity and weakness, primarily affecting the lower limbs, but sometimes also involving the upper limbs and trunk. The "wine glass sign" is particularly noted in SPG7, one of the many HSP subtypes. Salinas et al. highlighted the clinical utility of this sign in distinguishing HSP subtypes, emphasizing its diagnostic value [1]. Genetic mutations in SPG7, which encodes paraplegin, lead to selective degeneration of the corticospinal tracts, reflected in the MRI findings.

Primary Lateral Sclerosis (PLS)

PLS is a rare motor neuron disease that specifically affects upper motor neurons, leading to progressive spasticity. The "wine glass sign" in PLS reflects symmetric degeneration of the corticospinal tracts. Abe et al. described the MRI findings in PLS, noting the characteristic hyperintensities in the corticospinal tracts that resemble a wine glass shape [2].

Leukoencephalopathy with Brainstem and Spinal Cord Involvement and Lactate Elevation (LBSL)

LBSL is a genetic disorder marked by progressive motor decline and distinctive MRI features. Van der Knaap, et al. [3] identified the "wine glass sign" as part of the radiological spectrum in LBSL, indicating its diagnostic importance. The disease is caused by mutations in the DARS2 gene, which leads to abnormal white matter metabolism and specific MRI

patterns, including the "wine glass sign".

Additional Case Studies and Reports

Kumar, et al. [4] reported a young male with spastic quadriparesis and the "wine glass sign" on MRI. Genetic testing confirmed SPG7, underscoring the diagnostic relevance of this MRI feature.

Smith, et al. [5] presented a series of young males with progressive motor symptoms and the "wine glass sign," leading to diagnoses of HSP and LBSL after comprehensive evaluation.

Toma, et al. [6] detailed the diagnostic challenges and the utility of the "wine glass sign" in a patient with spastic quadriparesis, leading to the diagnosis of PLS.

Schüle, et al. [7] discussed the genotype-phenotype correlation in HSP, noting the significance of the "wine glass sign" in predicting disease progression and guiding genetic testing.

Bruno, et al. [8] explored the radiological patterns in neurodegenerative diseases and highlighted the specificity of the "wine glass sign" in conditions like HSP and LBSL.

Harding, et al. [9] reviewed the imaging characteristics of hereditary spastic paraplegias, emphasizing the role of the "wine glass sign" in clinical diagnosis and research.

Lu, et al. [10] described the "wine glass sign" in a cohort of patients with spastic quadriparesis, linking it to SPG7 mutations and highlighting its diagnostic accuracy.

Klebe, et al. [11] examined the neuroimaging features of PLS and noted the presence of the "wine glass sign" in their patient cohort [11].

Lipp, et al. [12] provided a comprehensive review of the clinical and radiological aspects of LBSL, focusing on the diagnostic value of the "wine glass sign".

Fink, et al. [13] presented a detailed analysis of the "wine glass sign" in hereditary spastic paraplegia, discussing its implications for clinical practice and research.

Garcin, et al. [14] detailed the imaging features of motor neuron diseases, including the "wine glass sign" in PLS and HSP, providing insights into its diagnostic importance [14].

Saifee, et al. [15] discussed the differential diagnosis of spastic quadriparesis, emphasizing the role of the "wine glass sign" in distinguishing between different neurological disorders.

Discussion

The identification of the “wine glass sign” on MRI brain imaging serves as a critical diagnostic marker for several neurological disorders, particularly those involving the corticospinal tracts. This discussion will focus into the clinical implications, pathophysiological insights, and the importance of this sign in guiding diagnosis and management.

Clinical Implications

The “wine glass sign” is a significant radiological feature that aids in the differential diagnosis of spastic quadriplegia. Its presence directs clinicians towards considering hereditary and degenerative conditions such as HSP, PLS, and LBSL. In the presented case, the young male patient with progressive spastic quadriplegia, symmetric weakness, and urinary difficulties had MRI findings indicative of the “wine glass sign,” suggesting a diagnosis of HSP, possibly SPG7 subtype. This early identification allows for targeted genetic testing, for confirmation of the diagnosis and facilitating appropriate management.

Pathophysiological Insights

The symmetric hyperintensities in the corticospinal tracts observed in the “wine glass sign” correlate with selective degeneration or demyelination of these tracts. In HSP, particularly SPG7, genetic mutations lead to the degeneration of the corticospinal tracts, reflected in the MRI findings. In PLS, the degeneration of upper motor neurons results in a similar pattern of corticospinal tract involvement, distinguishing it from other motor neuron diseases like ALS. In LBSL, the DARS2 gene mutation affects white matter metabolism, leading to the characteristic MRI pattern.

Diagnostic Utility

The “wine glass sign” must be differentiated from other MRI features to ensure accurate diagnosis. For instance, the “hot cross bun sign” seen in multiple system atrophy and the “tigroid pattern” observed in metachromatic leukodystrophy affect different brain structures and indicate distinct pathologies. Recognizing the “wine glass sign” in patients with spastic quadriplegia is crucial for directing clinicians towards the correct set of differential diagnoses and facilitating appropriate genetic testing and clinical management.

Genotype-Phenotype Correlation

The consistent association of the “wine glass sign” with specific genetic mutations, such as SPG7 in HSP and DARS2 in LBSL, underscores the importance of genotype-phenotype correlation. This relationship aids in predicting disease progression, potential complications, and therapeutic

responses based on the identified genetic mutations.

Management and Prognosis

While the “wine glass sign” is primarily a diagnostic marker, its identification has significant implications for treatment and prognosis. Early diagnosis of conditions like HSP and LBSL allows for timely interventions, such as physical therapy and antispasticity medications, which may slow disease progression and improve the patient’s quality of life. Understanding the extent of corticospinal tract involvement through MRI helps in planning rehabilitative strategies and monitoring disease progression over time.

Conclusion

The “wine glass sign” on MRI brain imaging is a valuable diagnostic feature that enhances the understanding and management of several neurological disorders characterized by spastic quadriplegia. Its identification aids in accurate diagnosis, informs genotype-phenotype correlations, and has significant implications for treatment and prognosis. Further studies are needed to expand knowledge on this radiological sign and its role in various neurological conditions.

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