

## Imaging of Phacomatosis: A Retrospective Study of 24 Patients

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### Abstract

### Original Research Article

Our study delves into the radiological characteristics of phacomatoses, leveraging advanced imaging techniques like MRI and CT to differentiate and diagnose various neurocutaneous syndromes, including Sturge-Weber syndrome, neurofibromatosis types 1 and 2, tuberous sclerosis complex, and Von Hippel-Lindau disease. Conducted as a retrospective analysis at the CHU Mohammed VI's Mother and Child Hospital, our research spans over four years and eight months, examining imaging findings from 24 patients. We highlight specific radiological signatures that aid in the identification and management of these complex conditions. Our findings not only corroborate established radiological markers but also introduce new insights into the diverse manifestations of phacomatoses. This study emphasizes the critical role of imaging in enhancing diagnostic accuracy, informing therapeutic decisions, and monitoring disease progression, thereby contributing to improved patient outcomes in managing these genetic disorders.

**Keywords:** phacomatoses, imaging techniques, diagnostic.

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## INTRODUCTION

Phacomatoses, also known as neurocutaneous syndromes, encompass a group of genetic disorders that primarily affect the nervous system and the skin. Among these, neurofibromatoses stand out as the most common conditions. Imaging plays a pivotal role in the diagnosis, monitoring, and management of these diseases, aiding in determining the appropriate type of examination and the necessary frequency for each condition. The aim of our study is to identify the radiological elements that facilitate the diagnosis of phacomatosis and to clarify the distinguishing features among the various types of phacomatosis.

The comprehensive evaluation and management of phacomatoses require a nuanced understanding of their radiological characteristics. Advanced imaging techniques, including magnetic resonance imaging (MRI), computed tomography (CT), and positron emission tomography (PET), have revolutionized the approach to these disorders, enabling detailed visualization of both neurological and dermatological manifestations. This article delves into the critical role of imaging in the context of phacomatoses, highlighting the specific radiological signatures associated with different types of these

syndromes. By examining the latest advancements and research in medical imaging, we aim to provide a thorough overview that not only aids in the accurate diagnosis of these complex conditions but also sheds light on the potential for imaging to guide therapeutic decision-making and monitor disease progression. Through this exploration, our study seeks to contribute to the broader understanding of phacomatoses, facilitating improved patient outcomes through precise diagnostic and management strategies.

## METHODOLOGY

This research was designed as a retrospective study, aimed at evaluating the radiological characteristics of phacomatoses by analyzing patient records at the CHU Mohammed VI's Mother and Child Hospital's Radiology Department. The study period extended over four years and eight months, providing a substantial timeframe to collect and analyze data relevant to the diagnostic imaging of phacomatoses.

The cohort comprised 24 patients diagnosed with various forms of phacomatoses, identified through a rigorous selection process. Eligibility criteria included patients of any age and sex who underwent CT and/or MRI brain scans as part of their diagnostic workup or follow-up care for phacomatoses during the study period.

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Patients were excluded if they had incomplete medical records or if their imaging studies were not available for review.

The collected data focused on imaging findings obtained through CT and MRI, with attention to the presence, type, and location of neurological and dermatological manifestations characteristic of phacomatoses. The analysis aimed to identify patterns and specific radiological features that could aid in the differentiation and diagnosis of phacomatoses types.

Data collection was comprehensive, encompassing patient demographics (age, sex), clinical history, type of phacomatosis diagnosed, details of the imaging modality used (CT, MRI, or both), and the radiological findings. The collected information was anonymized to ensure patient confidentiality and compliance with ethical standards.

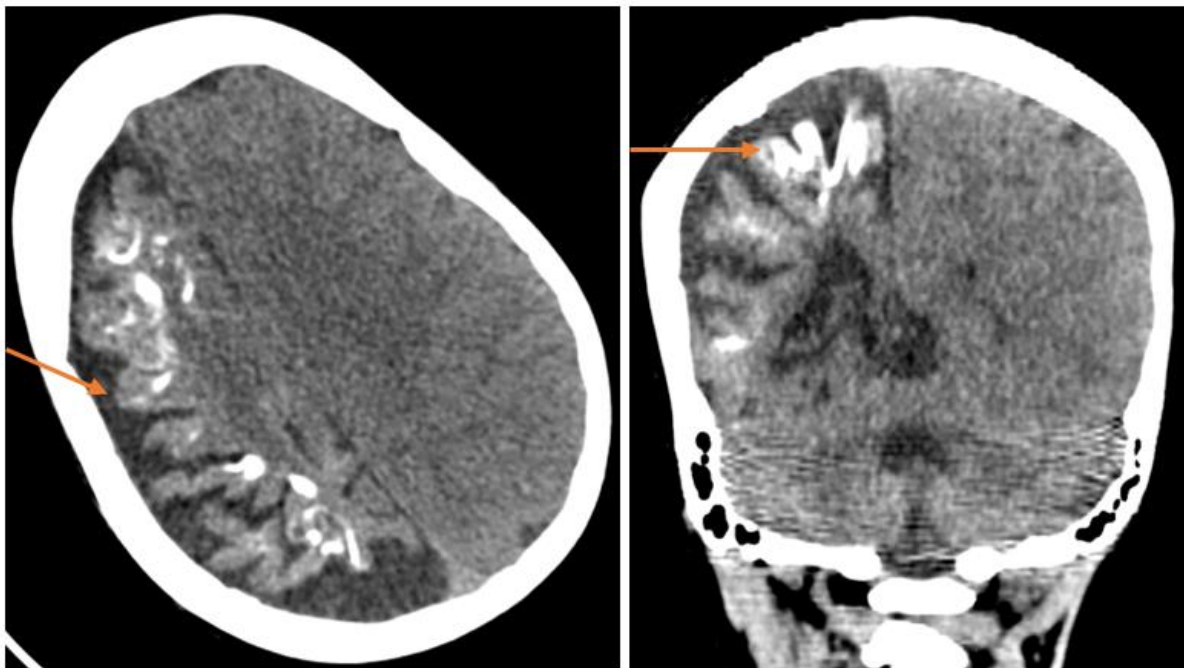
Descriptive statistics were employed to summarize the data, including frequencies and percentages for categorical variables and means or medians for continuous variables. The objective was to elucidate the prevalence of specific radiological findings and their association with different types of phacomatoses.

By leveraging a detailed retrospective analysis, this study aims to enrich the understanding of phacomatoses' radiological spectrum, facilitating early diagnosis and tailored management approaches for affected patients.

## RESULTS

The average age of patients in our study was 17 years, with a total of 4 undergoing CT brain scans and 11 undergoing MRI brain scans. The imaging outcomes revealed specific findings related to different types of phacomatoses, as detailed below.

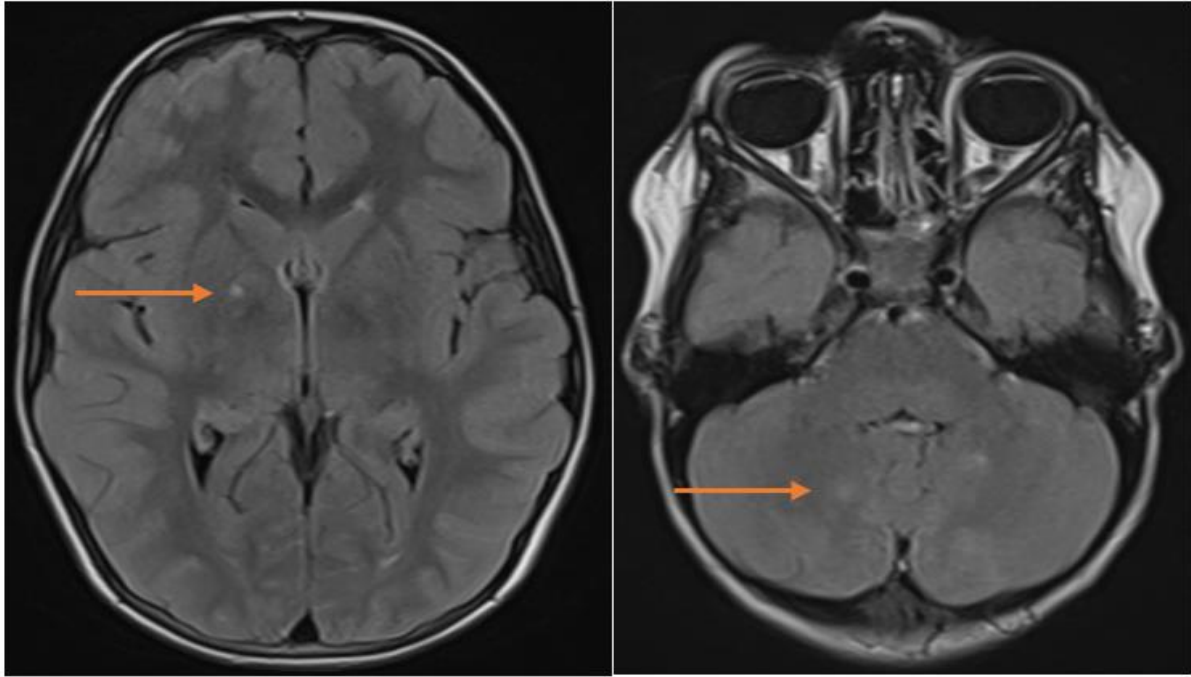
Our study included 3 patients diagnosed with Sturge-Weber syndrome, characterized by a constellation of radiological findings. All patients demonstrated cortical and subcortical parenchymal atrophy, which is indicative of the progressive nature of the disease. Leptomeningeal contrast enhancement was observed consistently, suggesting abnormal leptomeningeal capillary-venous malformations associated with SWS. Cortical calcifications, a hallmark of SWS, were evident and correlate with the disease's epileptogenic potential. One case stood out due to hypertrophy of the choroid plexus and associated cranial vault thickening, which may suggest an underlying abnormal cerebrospinal fluid dynamics or a response to chronic seizure activity.



**Brain CT scan in axial and coronal sections showing right hemispheric parenchymal atrophy, both cortical and subcortical, with gyriform cortical calcifications Related to Sturge-Weber syndrome**

In the cohort of 5 patients with NF1, the presence of unidentified bright objects (UBOs) on MRI was a common finding. These lesions, while not typically

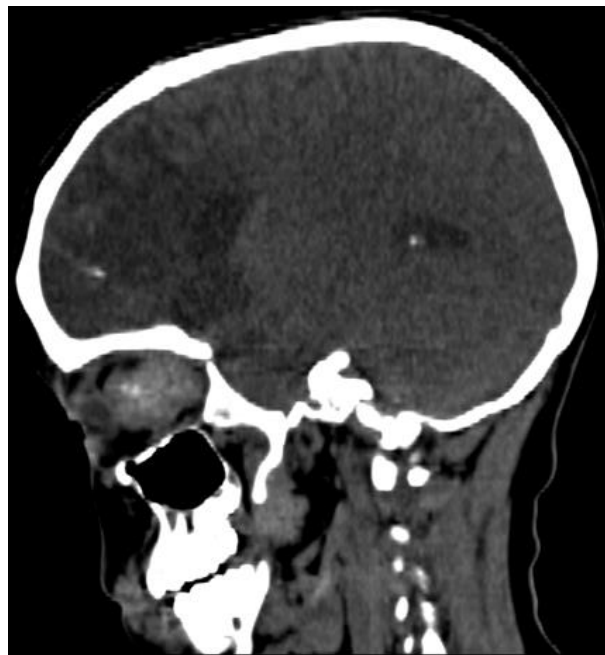
associated with clinical symptoms, indicate the disease's widespread impact on the nervous system.



**Brain MRI in FLAIR sequence showing nodular signal abnormalities visible in the right pallidum and cerebellar peduncles as FLAIR hyperintensities, related to unidentified bright objects (UBOs) in the context of Neurofibromatosis type 1**

The case with an optic nerve glioma underscores the risk of vision-threatening tumors in NF1,

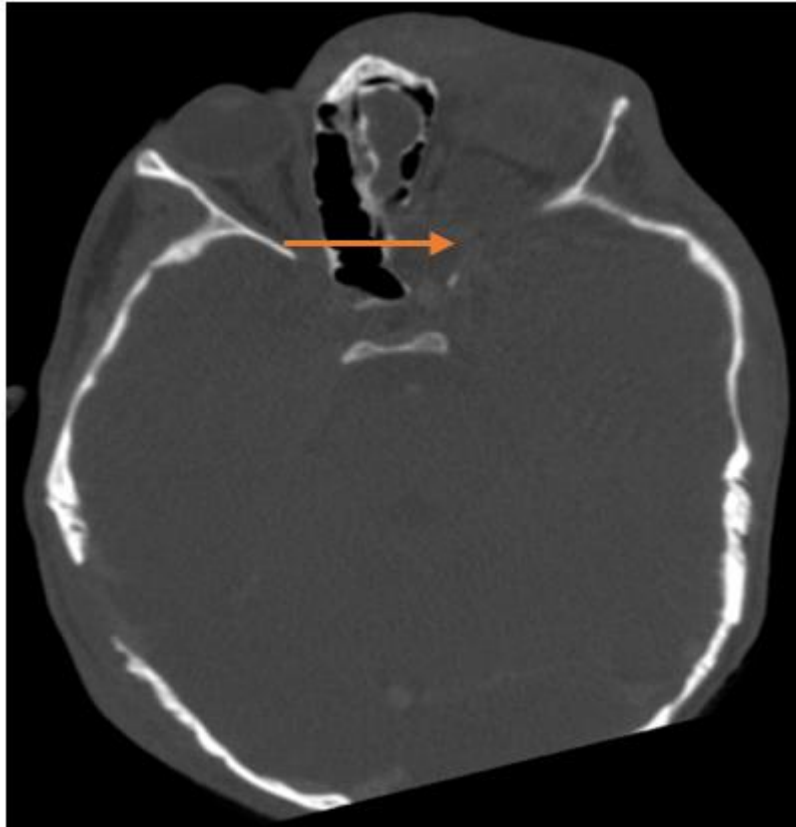
necessitating vigilant ophthalmologic and neurological monitoring.



**Brain CT scan in sagittal section showing intra-orbital lesion process related to an optic nerve glioma associated with Neurofibromatosis type 1**

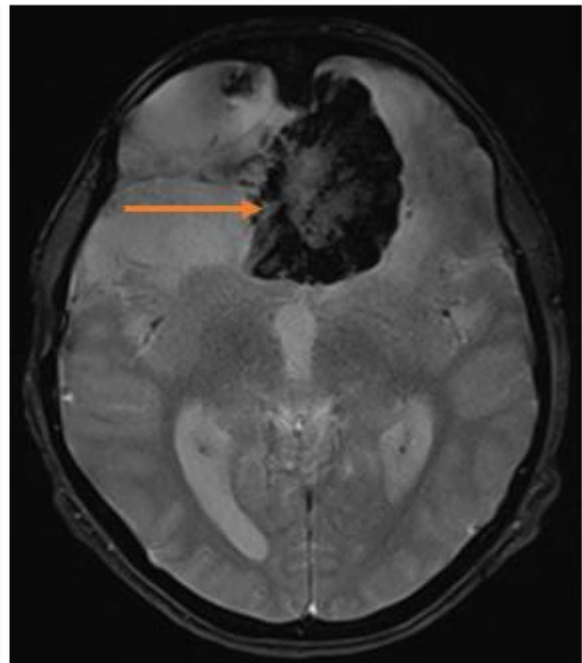
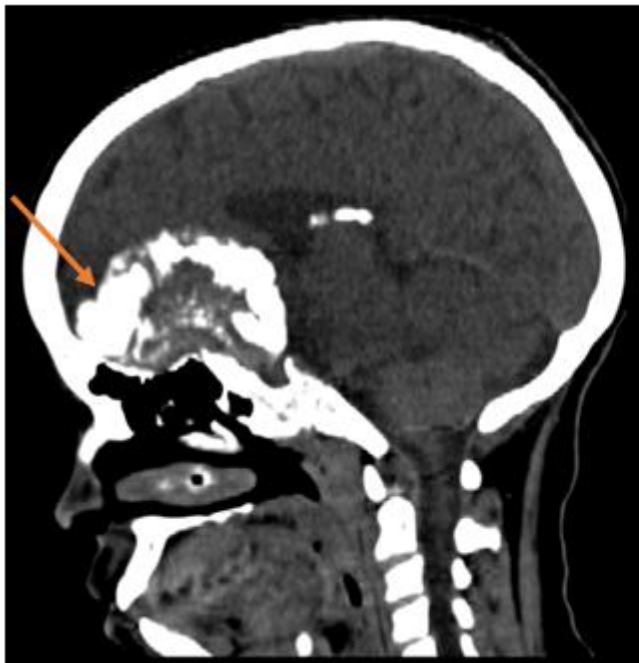
Spheno-orbital dysplasia and vascular dysplasia were identified in one case each, highlighting the diverse

manifestations of NF1 and the importance of a multidisciplinary approach to care.



**Brain CT scan in axial section, bone window, showing sphenoorbital dysplasia as part of Neurofibromatosis type 1**

The 2 patients with NF2 in our study exhibited meningiomatosis, reflecting the propensity for multiple intracranial tumors in this condition.



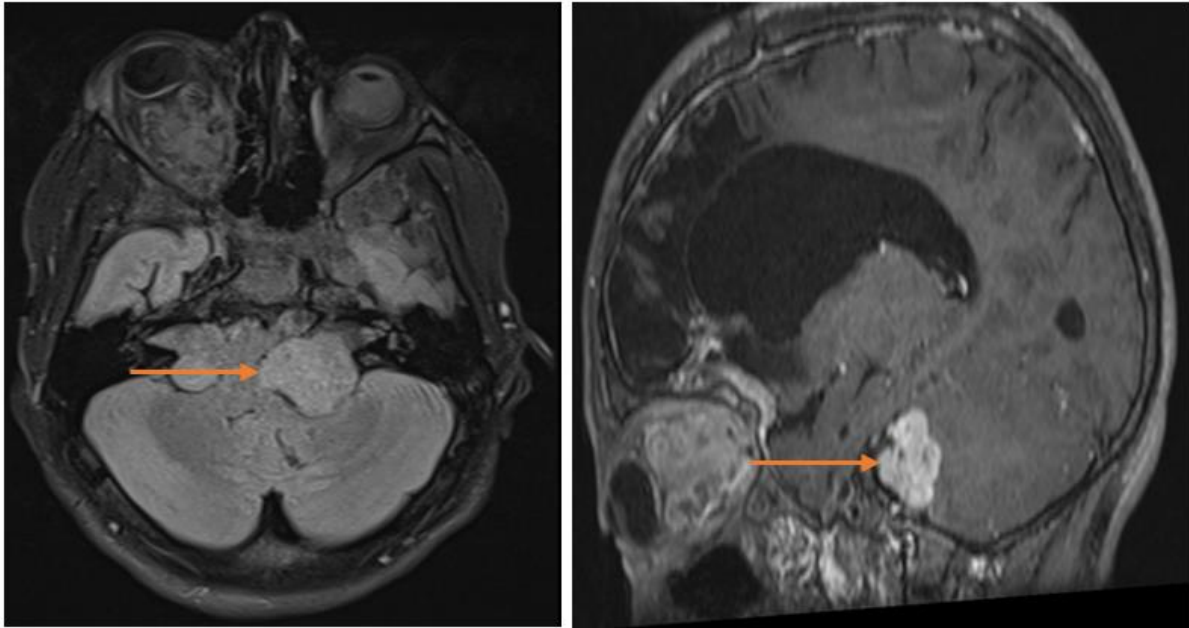
**Brain CT and MRI in T2\* sequence showing a largely calcified extra-axial lesion related to meningiomatosis as part of Neurofibromatosis type 2**

The presence of a schwannoma of the auditory nerve in one patient aligns with the characteristic

vestibular schwannomas that can lead to hearing loss and balance problems in NF2. These findings emphasize the



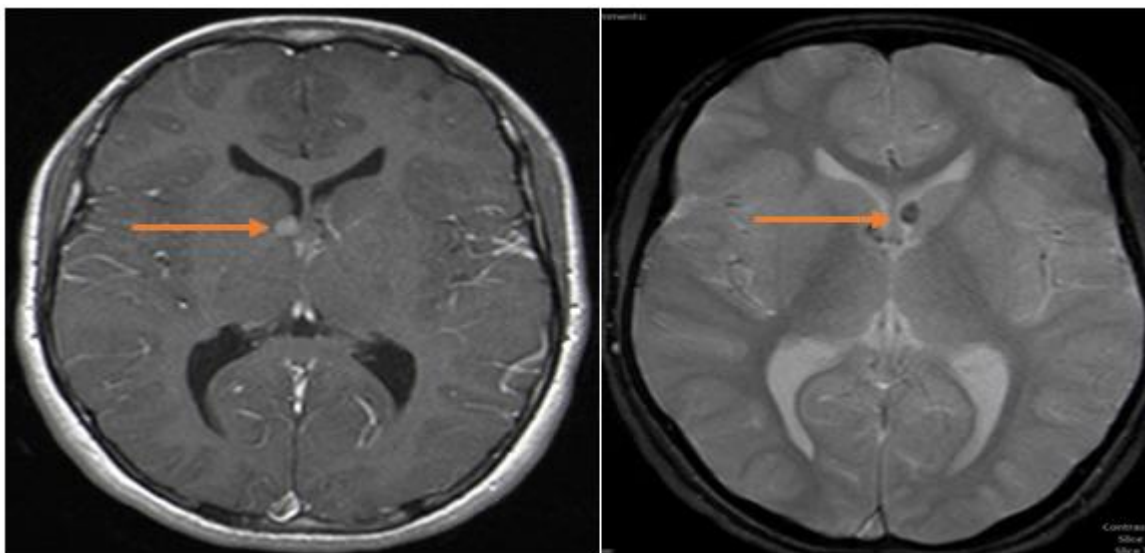
critical role of MRI in the early detection and management of NF2-associated tumors to preserve neurological function and quality of life.



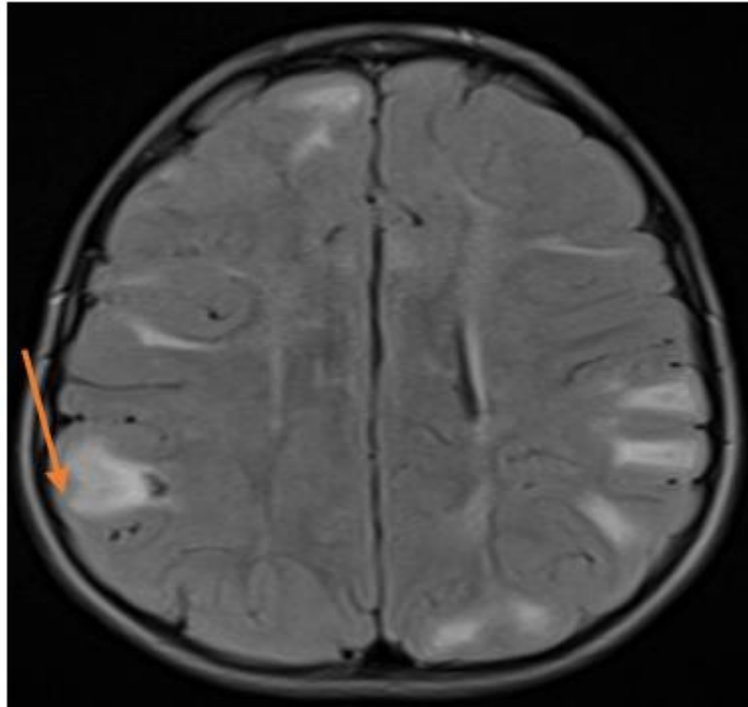
**Brain MRI in FLAIR and T1 sequences after gadolinium injection showing two processes in the cerebellopontine angles causing an enlargement of the internal auditory canals, related to vestibular schwannomas associated with Neurofibromatosis type 2**

Our cohort revealed a variety of central nervous system manifestations in patients with Tuberous Sclerosis Complex. White matter signal abnormalities detected in 4 cases represent the widespread impact of TSC on brain architecture and function. Subependymal nodules and cortical tubers, found in 3 and 4 cases

respectively, are key diagnostic criteria for TSC and have implications for seizure development and neurocognitive outcomes. The detection of giant cell astrocytomas in one case highlights the potential for tumor growth in TSC, necessitating regular surveillance and potentially surgical intervention.



**Brain MRI in T2\* and contrast-enhanced T1 sequences revealing subependymal nodules, some of which are calcified, consistent with Tuberous Sclerosis Complex**



**Brain MRI in FLAIR sequence revealing cortical tubers some of which are sites of signal void areas associated with calcifications, in the context of Tuberous Sclerosis Complex**

Interestingly, our VHL cohort did not present with cerebral manifestations, which is somewhat atypical given the disease's association with hemangioblastomas in the brain and spinal cord. This finding may reflect the variability in VHL expression or possibly the early stage of disease in our patients. Continuous monitoring through imaging is essential for early detection of CNS lesions, which can significantly impact management and prognosis.

## DISCUSSION

Our study presents a detailed examination of the radiological manifestations of various phacomatoses, including Sturge-Weber syndrome (SWS), neurofibromatosis types 1 and 2 (NF1 and NF2), tuberous sclerosis complex (TSC), and Von Hippel-Lindau disease (VHL). By meticulously analyzing imaging data from a cohort of 24 patients, we have identified several distinctive radiological features that not only aid in the differential diagnosis of these complex conditions but also provide insight into their underlying pathophysiology.

Comparing our findings with established literature reveals both corroborative evidence and new perspectives on the imaging characteristics of phacomatoses. For example, our observations of cortical and subcortical atrophy, leptomeningeal contrast enhancement, and cortical calcifications in SWS patients align with previous descriptions of the condition's radiological profile. However, our report of choroid plexus hypertrophy and associated cranial vault thickening in a case of SWS suggests an underexplored aspect of the disease's impact on cerebrospinal fluid

dynamics, inviting further investigation into its clinical significance.

In the realm of NF1, the common appearance of unidentified bright objects (UBOs) on MRI in our study corroborates the literature [1], underscoring their utility as a diagnostic marker. Yet, our identification of sphenoorbital dysplasia and vascular dysplasia in individual cases highlights the condition's broad radiological spectrum and underscores the necessity for comprehensive imaging evaluations. These findings echo the diverse manifestations of NF1, as noted in the provided study, and emphasize the importance of vigilant monitoring for complications such as optic nerve gliomas.

Our analysis of NF2 patients reveals a consistent presentation of meningiomatosis and auditory nerve schwannomas, supporting the established diagnostic criteria for this condition. The critical role of MRI in detecting NF2-associated tumors, as demonstrated in our study, reinforces the value of this modality in the early intervention and management strategies to preserve patient quality of life, aligning with previous research findings.

The TSC-related imaging outcomes in our cohort, including white matter signal abnormalities, subependymal nodules, cortical tubers, and giant cell astrocytomas, are in agreement with documented diagnostic criteria. These results highlight the condition's extensive neurological impact and the importance of regular surveillance to manage seizure development and

neurocognitive outcomes, a theme that resonates with the broader literature on TSC.

Interestingly, the absence of cerebral manifestations in our VHL cohort offers a point of divergence from expected findings, suggesting variability in disease expression and the potential influence of disease stage on imaging outcomes. This observation prompts a reevaluation of monitoring protocols and underscores the heterogeneity of VHL, as discussed in the literature [2].

The detailed imaging findings in our study elucidate the complex radiological spectrum of phacomatoses. From the distinctive leptomeningeal enhancements in Sturge-Weber syndrome to the varied central nervous system tumors in neurofibromatosis and tuberous sclerosis complex, these results underscore the indispensable role of advanced imaging in the accurate diagnosis, management, and understanding of these conditions. Furthermore, the absence of cerebral manifestations in our Von Hippel-Lindau cohort prompts a discussion on the heterogeneity of disease expression and the importance of personalized monitoring strategies. Our findings contribute to the growing body of knowledge on phacomatoses, highlighting the critical intersection of radiology with genetic and neurocutaneous disorders.

In conclusion, our study not only reaffirms several established radiological features of phacomatoses but also introduces novel insights into their diverse manifestations. By leveraging advanced imaging techniques, we have elucidated critical aspects of these conditions that enhance diagnostic accuracy and inform therapeutic strategies. Our findings contribute to the growing body of knowledge on phacomatoses, emphasizing the indispensable role of radiology in the management of these complex genetic disorders.

We have traversed a significant landscape of neurocutaneous syndromes, meticulously exploring the diverse and complex radiological signatures associated with Sturge-Weber syndrome (SWS) [3], neurofibromatosis types 1 and 2 (NF1 and NF2), tuberous sclerosis complex (TSC), and Von Hippel-Lindau disease (VHL). Our investigation, rooted in a detailed retrospective analysis of imaging findings, has illuminated the pivotal role of advanced imaging techniques in the diagnosis, differentiation, and management of these genetic disorders.

Through the lens of our study, the critical importance of magnetic resonance imaging (MRI), computed tomography (CT), and, where applicable, positron emission tomography (PET) in elucidating the pathological underpinnings and clinical manifestations of phacomatoses has been underscored. Our findings

reaffirm the diagnostic hallmarks of these conditions [4], such as the leptomeningeal enhancements in SWS, the characteristic unidentified bright objects (UBOs) in NF1, the meningiomatosis and schwannomas in NF2, and the neurocognitive impact of tubers in TSC. Additionally, our research contributes new insights into the variability of disease expression, as highlighted by the unexpected absence of cerebral manifestations in our VHL cohort, suggesting a nuanced understanding of this condition.

The convergence of our study's outcomes with existing literature not only validates the established radiological criteria for phacomatoses but also propels the field forward by shedding light on less explored aspects of these diseases. For instance, the identification of specific patterns of brain involvement and novel imaging findings invites further research and underscores the necessity for ongoing vigilance in the clinical monitoring of affected patients.

Moreover, our study accentuates the indispensable role of radiology not just in the diagnostic phase but also in guiding therapeutic decisions and monitoring disease progression. The advancements in imaging technology and technique have equipped clinicians with the tools necessary to offer more personalized and effective care, ultimately aiming to improve the quality of life for patients with phacomatoses.

## CONCLUSION

In summary, this exploration of the radiological spectrum of phacomatoses enriches our understanding of these complex syndromes and reinforces the integral role of imaging in their comprehensive management. As we continue to unravel the intricacies of these genetic disorders, it is imperative that we leverage the full potential of radiological science to enhance diagnostic accuracy, inform treatment strategies, and foster improved patient outcomes. Our study, by contributing to the body of knowledge on phacomatoses, underscores the ongoing need for research and innovation in the field of neurocutaneous syndromes, paving the way for future advancements in care and treatment.

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