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Describing the Phenotypic Spectrum of Sturge Weber Syndrome in a Cohort of Children Attending the Neurology Clinic at a Tertiary Care Center

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Abstract

Background: Sturge Weber syndrome (SWS) is a neurocutaneous disorder that is associated with facial capillary malformation (Port wine stain [PWS]), glaucoma, and leptomeningeal angioma in its complete form. Its incidence is estimated to be 1 per 20 000-50 000 live births.

Aim of Study: To assess and evaluate the spectrum and variability among patients with Sturge Weber syndrome in terms of clinical manifestations and to correlate these clinical manifestations with radiological findings including gyriform calcifications in CT brain and leptomeningeal enhancement in MRI brain with contrast.

Patients and Methods: This cross-sectional survey study will be conducted on 20 cases of Sturge-Weber syndrome, a rare disease, selected based on specific inclusion criteria. The study population will be drawn from patients attending the Pediatric Neurology Outpatient Clinic at the Faculty of Medicine, Cairo University, and the Pediatric Outpatient Clinic at Souad Kafafi University Hospital, MUST University, both located in Cairo, Egypt.

Results: The study on Sturge-Weber syndrome (SWS) identified significant clinical and radiological findings. Skin lesions were mostly left-sided (55%), with 70% of patients also having glaucoma, strongly linked to lesion distribution (r=0.438, p=0.037). Developmental delays were common, with 50% showing motor delays and 65% mental delays. Cognitive challenges were evident, with a mean IQ of 73.5 and 40% of patients having an extremely low IQ. Leptomeningeal angiomas were present in 85% of cases, while gyriform calcifications were found in 65%, correlating with motor deficits in 70% of patients. Seizures, particularly intractable epilepsy, were prominent in 55% of patients, necessitating complex, individualized treatment plans. EEG abnormalities were noted in 33% of patients, correlating with seizures and mental delay, though EEG was less effective in predicting motor and cognitive outcomes. CT and MRI showed good to moderate predictive ability for seizures and motor deficits but were less reliable for mental delay and did not correlate with seizure intractability. These findings highlight the importance of comprehensive assessments and personalized management in improving outcomes for SWS patients.

Conclusion: Our study provides valuable insights into the clinical and radiological features of Sturge-Weber syndrome and their associations with clinical outcomes. We found significant correlations between calcifications in CT and MRI findings with seizures and mental delay, highlighting the importance of these imaging modalities in predicting these outcomes. However, EEG findings showed limited predictive value for clinical outcomes. The presence of glaucoma was also found to be correlated with specific patterns of skin lesions.

Our findings underscore the importance of comprehensive clinical and radiological assessments in the management of Sturge-Weber syndrome, and further research is warranted to validate these findings and improve our understanding of this complex condition.

Key Words: *Phenotypic Spectrum – Sturge Weber Syndrome.*

Introduction

STURGE Weber syndrome (SWS) is a neurocutaneous disorder that is associated with facial capillary malformation [Port wine stain (PWS)], glaucoma, and leptomeningeal angioma in its complete form. Its incidence is estimated to be 1 per 20 000-50 000 live births **[1].**

Sturge Weber syndrome can be further classified according to the Roach scale to 3 types: Type I with facial PWS and leptomeningeal angiomatosis, with

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or without associated glaucoma, corresponding to classical SWS; type ii, which is more common, with facial PWS and no leptomeningeal involvement, with or without presence of glaucoma; and type iii, which is the least frequent form, with presence only of leptomeningeal angiomatosis **[2].**

The neurological complications of SWS include epilepsy, stroke-like episodes, migraine, learning, and behavioral difficulties affecting every aspect of the individual's life. Efforts have been made to predict who is at greatest risk for these symptoms. The strongest predictors of neurological complications include both distribution (involvement of the forehead and upper eyelid) and size of the port-wine birthmark and the magnetic resonance imaging (MRI) findings (unilateral versus bilateral brain involvement) **[3].**

Facial nevus (port-wine stain) is another common finding and is typically seen along the ophthalmic or maxillary segment of the trigeminal nerve (forehead, cheeks). It is typically unilateral, present at birth, and does not change with the age of the patient. It slowly grows and is followed by involution. A child with a facial port-wine stain has a 10% to 35% risk of brain involvement. If there is involvement of both upper and lower eyelids, then the risk of glaucoma increases up to 50% **[4].**

Glaucoma remains the most common ocular complication of SWS, which occurs in 30–70% of the patients. In SWS it has a bimodal presentation: early onset glaucoma in 60% of the cases and late onset glaucoma in 40% of patients. In 60% of the early forms, glaucoma is caused by abnormalities in the angle of the anterior chamber, whereas the cause of glaucoma in 40% of young people and young adults is due to elevated episcleral venous pressure **[5].**

Computed tomogram is the best modality to detect calcifications and show the other changes such as cortical atrophy and leptomeningeal enhancement on the post-contrast studies. MRI brain with contrast is the recommended imaging modality of choice. The most common locations of angiomatosis are occipital and posterior parietal/temporal lobes. Depending on the location of the leptomeningeal angiomas, and the secondary effects of the angioma, the neurologic manifestations vary. A part of evaluating a patient with SWS also includes IQ testing to detect the presence of intellectual disability **[4].**

Aim of the work:

To assess and evaluate the spectrum and variability among patients with Sturge Weber syndrome in terms of clinical manifestations and to correlate these clinical manifestations with radiological findings including gyriform calcifications in CT brain and leptomeningeal enhancement in MRI brain with contrast.

Patients and Methods

Study design: Cross sectional survey study.

Population and location of the study: Sturge Weber syndrome is a rare disease, 20 cases fulfilling the inclusion criteria following in the Pediatric Neurology Outpatient Clinic, Faculty of Medicine, Cairo University, Cairo, Egypt and Pediatric outpatient clinic, Souad Kafafi University Hospital, MUST university, Cairo, Egypt from April 2023 – April 2024 will be enrolled in the study.

Inclusion criteria:

Patients with facial nevus (port wine stain), patients with leptomeningeal angiomas on MRI brain, patients with typical SWS calcifications on CT brain, both sexes are included and age 0-18 years old.

Exclusion criteria:

Patients with other neurocutaneous syndromes, patients with other causes of intellectual disability and patients with calcifications due to other causes eg. TORCH infections, tuberous sclerosis, neurofibromatosis, endocrine or metabolic problems.

Patients will be subjected to the following:

Medical history assessment: Data will be collected by reviewing medical records as well as by direct patient/parent interviewing and thorough history taking will be obtained including: Age, gender, consanguinity, history of seizures, vision affection and intellectual disability.

Intelligence quotient: An IQ test will be performed on all patients to determine, if present, the degree of mental deficit. Using WISC-V which classifies the scores as follows: Extremely high: 130 and above, very high: 120-129, high average: 110-119, average: 90-109, low average: 80-89, very low: 70-79 and extremely low: 69 and below.

Ophthalmological examination: As glaucoma incidence in patients with SWS is high, an eye examination will be done to exclude or diagnose its presence.

Imaging: CT brain to detect the presence of tramline (serpentine) calcifications consistent with the diagnosis of SWS and MRI brain with IV contrast will also be performed to detect the presence of leptomeningeal angiomas. Results will be retrieved from the patients' medical records if available.

Research involve: Human participants.

Type of consent of study participants: Written consent.

Potential risks: Allergic reaction upon use of IV contrast

Confidentiality of data: The Confidentiality of the Research Participants will be preserved in our research by: Data will be collected from the patients after signing written consents by any of the parents, any data that makes the patient identifiable will be omitted, patients will be numerically coded during the data collection phase, these codes will be used in all subsequent research phases and files will be locked and secured.

Study outcomes:

Primary outcomes: To better define and describe the spectrum of manifestations and various ways of presentation of Sturge Weber syndrome.

Secondary outcomes: To find correlation between clinical manifestations including seizures and motor deficits with radiological and EEG findings.

Sample size: Using EPI INFO sample size calculator for observation studies; with 0.05 alpha error and power of the study 0.80, confidence interval of 95%. According to literature clinical manifestations of patients with Sturge-Weber syndrome all patients had seizures; they were well controlled in (73.3%) and 30% of them had glaucoma (Sujit Jagtap et al., 2012) Sample size calculated to Study Phenotypic Spectrum of Sturge Weber syndrome is 20 patients.

Sampling technique: A convenient sample of patients coming to the Pediatric Neurology Clinic, Faculty of Medicine, Cairo University and the Pediatric outpatient clinic, Souad Kafafi University Hospital, MUST University, with the inclusion and exclusion criteria will be assigned into the study till reaching total sample size calculated.

Statistical analysis: Recorded data were analyzed using the statistical package for social sciences, version 23.0 (SPSS Inc., Chicago, Illinois, USA). The quantitative data were presented as mean ± standard deviation and ranges when their distribution was parametric (normal) while non-normally distributed variables (non-parametric data) were presented as median with inter-quartile range (IQR). Also qualitative variables were presented as number and percentages. Data were explored for normality using Kolmogorov-Smirnov and Shapiro-Wilk Test.

Results

This study aims to assess and evaluate the spectrum and variability among patients with Sturge Weber syndrome in terms of clinical manifestations and to correlate these clinical manifestations with radiological findings including gyriform calcifications in CT brain and leptomeningeal enhancement in MRI brain with contrast.

This study will be conducted on 20 patients.

Table (1): Demographic data among the studied patients.

	Studied patients (n=20)	
	N	%
Age:		
Mean ± SD	6.1±3.35	
Range	2-12	
Gender:		
Male	10	50
Female	10	50
Consanguinity:		
Yes	3	15
No	17	85

Table (1) provides demographic characteristics of the studied patients. With a sample size of 20 patients, the data presents a balanced gender distribution, with an equal representation of males and females. The mean age of 6.1 years, with a standard deviation of 3.35 years, suggests a relatively young cohort, spanning from 2 to 12 years old. Regarding consanguinity, 15% of patients reported familial relatedness, while the majority (85%) did not.

Table (2): Distribution of skin lesion and glaucoma for the studied patients.

	Studied patients (n=20)	
	N	%
Skin Lesion:		
Lt facial	11	55
Rt facial	7	35
Bilateral	2	10
Glaucoma:		
No glaucoma	6	30
Lt eye	4	20
Rt eye	5	25
Bilateral	5	25

Table (2) reveals that a significant proportion of patients (55%) exhibited left facial skin lesions, while 35% presented with lesions on the right side, and 10% showed bilateral involvement. Regarding glaucoma, 30% of patients had no eye affection. However, a notable portion exhibited glaucoma in various degrees of severity: 20% in the left eye, 25% in the right eye, and 25% in both eyes while 30% had no evidence of the disease upon examination.

Table (3): Correlation between skin lesion and glaucoma.

	Glaucoma
Skin lesion:	
r	0.438
p-value	0.037

r: Pearson correlation coefficient. *p*-value <0.05 is significant.
p-value >0.05 is insignificant. *p*-value <0.01 is highly significant.

Table (3) presents the correlation between skin lesions and glaucoma among the studied patients, aiming to elucidate potential associations between dermatological and ocular manifestations. A significant positive correlation was observed between skin lesions and glaucoma (r=0.438, p=0.037), indicating that the presence of skin lesions may be associated with an increased likelihood of developing glaucoma.

Table (4): Motor and mental development of the studied patients.

	Studied patients (n=20)	
	N	%
Motor Development:		
Normal	10	50
Delayed	10	50
Mental Development:		
Normal	7	35
Delayed	13	65

Table (4) presents data on the development of the studied patients, providing insights into both motor and mental developmental milestones within the cohort. Half of the patients demonstrated normal motor development, while the remaining 50% experienced delays in reaching motor milestones. Similarly, mental development exhibited a similar trend, with 35% of patients achieving normal milestones, and a higher proportion, 65%, experiencing delayed mental development.

Table (5): Intelligence Quotient (IQ) test for the studied patients.

	Studied patients (n=10)	
	N	%
IQ test:		
Mean ± SD	73.5±14.9	
Range	55-96	
IQ test Classification:		
Average (90-109)	3	30
Low Average (80-89)	1	10
Very low (70-79)	2	20
Extremely low (<70)	4	40

Table (5) outlines the results of Intelligence Quotient (IQ) tests conducted on a subset of the studied patients. The mean IQ score was 73.5, with a standard deviation of 14.9, indicating a wide range of cognitive functioning within the cohort. The IQ scores ranged from 55 to 96, showcasing variability in intellectual capabilities among the patients. In terms of classification, the majority of patients fell into the lower range of IQ scores: 30% scored within the “Average” range (90-109), a larger portion (40%) scored in the “Extremely low” range (<70). Additionally, 20% scored in the “Very low” range (70-79), while 10% fell into the “Low Average” range (80-89).

Table (6): Radiological findings and motor deficits for the studied patients.

	Studied patients (n=20)	
	N	%
LMA in MRI:		
No LMA	3	15
Lt LMA	8	40
Rt LMA	7	35
Bilateral	2	
Calcifications in CT:		10
No Calcification	7	35
Left cortical calcifications	6	30
Right cortical calcifications	6	30
Bilateral cortical calcifications	1	
Deficit:		5
No weakness	6	30
Rt side weakness	8	40
Lt side weakness	5	25
Quadriplegia	1	5

Table (6) presents radiological findings among the studied patients. Magnetic Resonance Imaging (MRI) revealed the presence of Leptomeningeal angiomas (LMA) in 85% of patients, with varied distribution: 40% exhibited LMA on the left side, 35% on the right side, and 10% bilaterally. Computed Tomography (CT) scans unveiled gyriform calcifications in 65% of patients, with equal proportions showing calcifications on the left and right sides, and a small percentage demonstrating bilateral calcifications. Furthermore, deficits were observed in 70% of patients, with 40% experiencing right-sided weakness, 25% left-sided weakness, and 5% quadriplegia.

Table (7) illustrates seizure characteristics among 20 studied patients. 30% experienced right focal seizures, 15% had left focal seizures, and 15% had generalized tonic-clonic seizures. Mixed seizure types, such as right or left focal seizures with secondary generalization, were observed in 20% of patients, while 20% did not experience seizures. Intractable seizures were reported in 55% of patients, highlighting the challenge in achieving seizure control. The median number of antiepileptic drugs (AEDs) used was 2, ranging from 1 to 3. Recovery periods post-seizure varied, with 37.5% of patients experiencing a seizure within days to weeks prior to data collection, 25% of patients had a recovery period of months, while 37.5% experienced their last seizure years prior to data collection.

Table (7): Seizures characteristics for the studied patients.

	Studied patients (n=16)	
	N	%
Type:		
No seizures	4	20
Lt focal	3	15
Rt focal	6	30
GTC	3	15
Rt focal with 2ry generalization	3	15
Lt focal with 2ry generalization	1	5
Intractable:		
Yes	9	55
No	7	45
No. of AED:		
Median (IQR)	2 (1-3)	
Range	1-3	
No. of AED:		
1	6	37
2	3	19
3	7	44
Recovery period mean value:		
Mean ± SD	24.0±37.3	
Range	0.13-120	
Recovery period:		
Days-weeks	6	37.5
Months	4	25
Years	6	37.5

Table (8): EEG findings for the studied patients.

	Studied patients (n=16)	
	N	%
EEG:		
Normal	10	67
Atypical hypsarrhythmia	1	5
pattern + rt temporal focus	5	28
Asymmetric background		

Table (8) presents electroencephalogram (EEG) findings among the studied patients, offering insights into their neurological status. The majority of patients (67%) exhibited normal EEG results, suggesting typical brain activity. However, abnormalities were observed in a subset of patients, including asymmetric background (28%) and atypical hypsarrhythmia pattern with a right temporal focus (5%).

Table (9) illustrates the correlation between mental delay, motor deficits, and seizures, alongside the EEG findings, calcifications in CT scans and MRI results among the studied patients. Significant positive correlations were observed between calcifications in CT scans and seizures (r=0.495, p=0.016), as well as mental delay (r=0.725, p<0.001), indicating a strong relationship between these factors. Additionally, MRI results showed significant correlations with seizures (r=0.462, p=0.026) and mental delay (r=0.540, p=0.008), suggesting a notable association between MRI abnormalities and these clinical parameters. However, no significant correlation was found between EEG findings and any of the studied variables.

Table (9): Correlation between Radiological findings and EEG findings with Motor deficit, Mental delay and Seizures.

	Seizures	Mental Delay	Motor Deficit
Calcifications in CT:			
r	0.495	0.725	0.437
p-value	0.016	>0.001	0.054
EEG Findings:			
r	0.357	0.019	0.323
p-value	0.146	0.940	0.223
LMA in MRI:			
r	0.462	0.540	0.304
p-value	0.026	0.008	0.192

r: Pearson correlation coefficient.
p-value >0.05 is insignificant.
p-value <0.05 is significant.
p-value <0.01 is highly significant.

Table (10) and Fig. (1) illustrate the correlation between EEG findings and motor deficits, seizures, and mental delay. The AUC values indicate that EEG has the highest predictive power for seizures (0.656), followed by mental delay (0.607), and motor deficits (0.576). Sensitivity is highest for seizures and mental delay, both at 100%, but specificity is highest for mental delay at 78.6%. The ROC curve visually confirms these findings, with the seizure curve (green) demonstrating the best performance, followed by mental delay (blue), and motor deficits (red). These results highlight EEG's varying effectiveness in predicting different neurological conditions.

Table (10): Correlation between EEG Findings and motor deficits, seizure, and mental delay.

EEG	AUC	Specificity	Sensitivity
Motor Deficits	0.576	56.3%	71.4%
Seizure	0.656	68.8%	100 %
Mental Delay	0.607	78.6%	100%

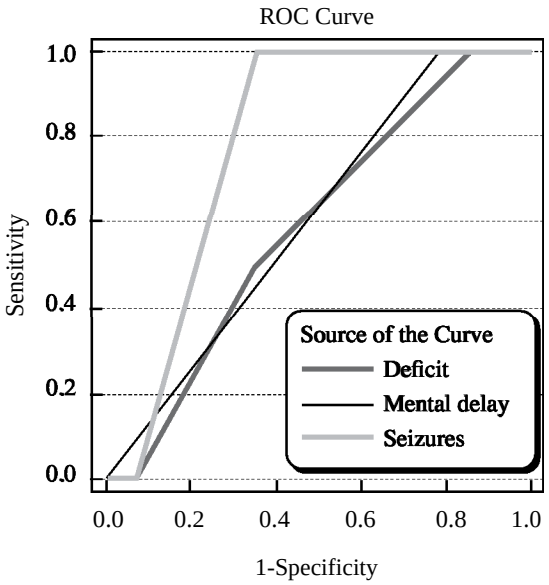

Fig. (1): Roc curve showing the correlation between EEG Findings and Motor Deficits, Seizure, and Mental Delay.

Table (11): Correlation between Calcifications in CT and Motor Deficits, Seizure, and Mental Delay.

Calcifications in CT	AUC	Specificity	Sensitivity
Motor Deficits	0.839	64.3%	83.3%
Seizure	0.810	71.4%	100%
Mental Delay	0.631	57.1%	83.3%

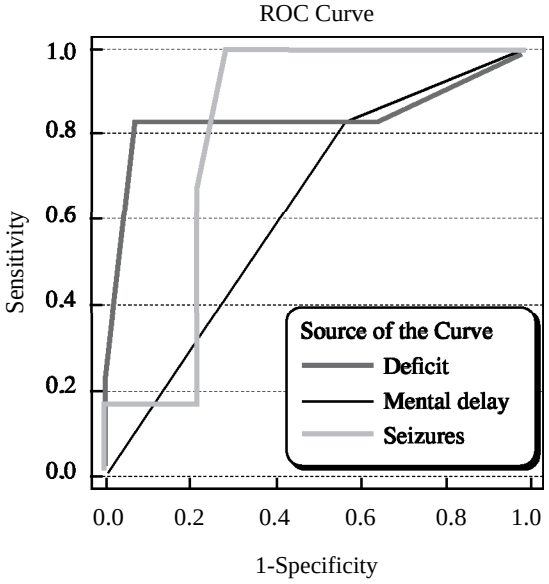

Fig. (2): Roc curve showing the correlation between Calcifications in CT and Motor Deficits, Seizure, and Mental Delay.

Table (11) and Fig. (2) illustrate the correlation between CT scan findings of calcifications and motor deficits, seizures, and mental delay. Calcifications in CT scans show the highest predictive power for seizures (AUC 0.810), followed by motor deficits (AUC 0.839), and mental delay (AUC 0.631). Seizure prediction exhibits the highest sensitivity (100%) and good specificity (71.4%). Motor deficits have high sensitivity (83.3%) and moderate specificity (64.3%). Mental delay shows moderate sensitivity (83.3%) and lower specificity (57.1%). The ROC curve visually supports these findings, highlighting CT's efficacy, especially for predicting seizures and motor deficits compared to mental delay.

Table (12): Correlation between MRI and Motor Deficits, Seizure, and Mental Delay.

MRI	AUC	Specificity	Sensitivity
Motor Deficits	0.665	53.8	85.7
Seizure	0.780	69.2	100
Mental Delay	0.659	53.8	85.7

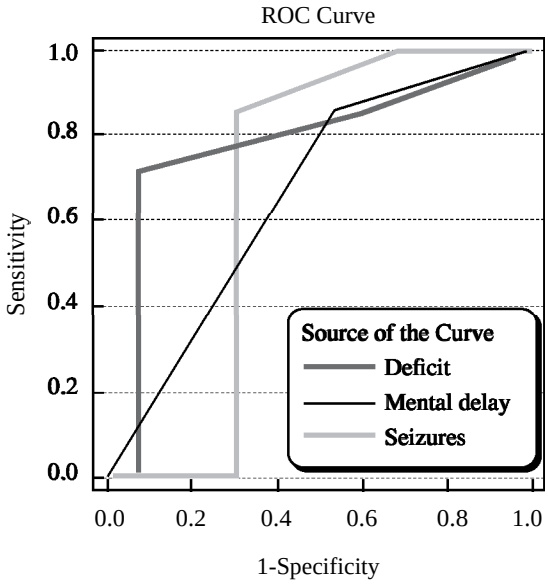

Fig. (3): Roc curve showing the correlation between MRI and Motor Deficits, Seizure, and Mental Delay.

Table (12) and Fig. (3) present the correlation between MRI findings and motor deficits, seizures, and mental delay. The AUC values indicate that MRI has the highest predictive power for seizures (0.780), followed by motor deficits (0.665), and mental delay (0.659). Seizures show the highest sensitivity (100%) and reasonable specificity (69.2%), indicating MRI's effectiveness in seizure detection. Motor deficits and mental delay exhibit moderate sensitivity (85.7%) but lower specificity (53.8%). The ROC curve confirms these results, with the seizure curve (green) outperforming others. Overall, MRI is particularly useful for predicting seizures compared to motor deficits and mental delay.

Table (13): Correlation between seizures intractability and radiological findings.

	Seizures intractability
Calcifications in CT:	
r	0.272
p-value	0.275
EEG:	
r	0.180
p-value	0.475

r: Pearson correlation coefficient.
p-value >0.05 is insignificant.
p-value <0.05 is significant.
p-value <0.01 is highly significant.

Table (13) presents the correlation between seizures intractability and radiological findings. The correlation coefficient between seizures intractability and calcifications in CT scans was found to be 0.272, with a corresponding *p*-value of 0.275, indicating no statistically significant correlation between these variables. Similarly, the correlation coefficient between seizures intractability and EEG findings was 0.180, with a *p*-value of 0.475, suggesting no significant association between seizures intractability and EEG abnormalities.

Discussion

The SWS is characterized by hamartomas of the eye, skin, and brain that manifest at different times during life. Some patients develop all features, while others show partial expression of the syndrome [6].

The presumed pathogenesis of SWS is early embryological malformation of vascular development, affecting skin, brain, and ocular structures [7].

The current study aimed to assess and evaluate the spectrum and variability among patients with Sturge Weber syndrome in terms of clinical manifestations and to correlate these clinical manifestations with radiological findings including gyriform calcifications in CT brain and leptomeningeal enhancement in MRI brain with contrast.

The current study founded an equal distribution of males and females between studied patients which is consistent with what was described in the literature [8].

The mean age of 6.1 years aligned with Alkonyi et al. [9] Fifteen children with SWS and unilateral hemispheric involvement (age range: 3-12.4 years) were studied. Consanguinity was reported in 15% of cases.

Also in agreement with HAO et al. [10] founded that 27 children who presented with facial port-wine stains, a type of vascular birthmark. Among them, there were 17 males and 10 females, with ages ranging from 2 days to 10 years and 7 months, averaging at 2.54 years.

The current study showed the distribution of skin lesions was predominantly on the left facial side (55%), followed by the right facial side (35%), with a smaller proportion exhibiting bilateral lesions (10%). Glaucoma was present in 70% of patients, with varying degrees of involvement in each eye. These findings highlight the characteristic distribution of skin lesions in Sturge-Weber syndrome, typically following the distribution of the trigeminal nerve, and underscore the high prevalence of glaucoma in affected individuals. The presence of glaucoma warrants close ophthalmological monitoring and highlights the importance of early detection and intervention to prevent vision loss in these patients.

In consistent with the current study results Hassanpour et al. [5] revealed that Glaucoma remains the most common ocular complication of SWS, which occurs in 30–70% of the patients.

Glaucoma in SWS has a bimodal presentation: early onset glaucoma in 60% of the cases and late onset glaucoma in 40% of patients. Also HAO et al. [10] highlighted associated conditions observed in the patients. Seventeen children of 27 had ocular choroidal vascular malformations, leading to various eye conditions such as congenital glaucoma (present in 14 cases) and high intraocular pressure (present in 5 cases). One patient exhibited optic nerve atrophy accompanied by transient blindness.

In the study done by HAO et al. [10] showed that the port-wine stains displayed varying colors, from light red to purple-red, and were distributed across the facial midline. Notably, 21 children had predominantly unilateral stains, while 6 presented with bilateral symmetrical stains.

The current study investigated the relationship between glaucoma and skin lesions in Sturge-Weber syndrome, we found a significant correlation between the presence of glaucoma and the distribution of skin lesions (r=0.438, p=0.037). This suggests that there may be a relationship between the location or extent of skin lesions and the development of glaucoma in patients with Sturge-Weber syndrome. The findings highlight the importance of close ophthalmological monitoring in patients with specific patterns of skin lesions, as they may be at higher risk for developing glaucoma.

In the same line Ha et al. [11] assessed the relationship between the cutaneous manifestations and other features of this disorder in 35 patients the study observed that all patients with the facial nevus flammeus had involvement of the upper eye lid or forehead. Bilateral cutaneous involvement was common and when present, was often associated with extensive lesions of the trunk and extremities.

The current study observed that 50% of patients had normal motor development, while the other 50% exhibited delayed motor development. Similarly, 35% of patients had normal mental development, while 65% showed delayed mental development. These findings underscore the variability in developmental outcomes among individuals with Sturge-Weber syndrome, highlighting the need for individualized management approaches. The high proportion of patients with delayed motor and mental development emphasizes the importance of early intervention and comprehensive care to optimize outcomes for these patients.

In the same line Luat et al. *[3]* showed that seizure onset prior to age 1 year has a profound effect on severity of cognitive and motor dysfunction in children with SWS; however, the effect of seizures on the type of cognitive deficit is influenced by laterality of brain involvement. Furthermore, seizure onset at or before 7 months of age, along with early frontal lobe involvement, is also a predictor of motor deficit.

These findings may have important clinical implications for a more accurate prognostication of the severity and pattern of neurocognitive outcome in young children with unilateral SWS.

The current study founded a mean IQ score of 73.5, indicating a range of cognitive abilities among the patients. The classification of IQ scores revealed that 10% of the patients fell within the low average range (80-89), while 30% were classified as having an average IQ (90-109). However, a substantial proportion of patients had below-average IQ scores, with 20% classified as very low (70-79) and another 40% classified as extremely low (<70). These findings highlight the cognitive challenges faced by individuals with Sturge-Weber syndrome and underscore the importance of comprehensive neuropsychological assessments and tailored interventions to support their cognitive development and quality of life.

In accordance Powell et al. *[12]* showed that nearly half (49.6%) experienced intellectual disability (ID), with 20.3% classified as severe. Notably, 28.6% showed no intellectual impairment, while varying degrees of impairment were observed in the rest, ranging from borderline to profound. This underscores the diverse cognitive challenges faced by individuals with Sturge-Weber syndrome, necessitating tailored support and intervention strategies.

The current study founded the presence of leptomeningeal angiomas (LMA) was observed in 85% of cases, with 40% showing involvement on the left side, 35% on the right side, and 10% bilateral. Gyriform calcifications were present in 65% of patients, with 30% showing calcifications on the left side, 30% on the right side, and 5% bilateral. Motor deficits were noted in 70% of patients, with 40% exhibiting right side weakness, 25% left side weakness, and 5% quadriplegia. These findings emphasize the variable and often asymmetric presentation of Sturge-Weber syndrome, emphasizing the importance of comprehensive imaging and clinical assessments to tailor management strategies for each patient. The high prevalence of motor deficits highlights the significant impact of this condition on motor function particularly in those with abnormal MRI brain with contrast and CT brain results, therefore the need for targeted interventions to improve quality of life for affected individuals is needed.

In consistent Powell et al. *[12]* showed that most patients had a PWS (85.7%). Three-quarters had unilateral angioma (75.7%), with approximately equal numbers having unilateral right or left angioma. Common neurological features included: Epilepsy (90%), hemiplegia (57.8%), headaches (57.1%) and visual field deficits (36.4%). Over half had experienced an episode of status epilepticus (SE) (57.1%). Neuropsychological impairments were common including intellectual disability (47.1%) and language disorder (40.7%).

The current study founded seizures were a prominent feature, with various types observed. Focal seizures were the most common, with 30% occurring on the right side and 15% on the left side, while 15% presented with generalized tonic-clonic seizures (GTC). Mixed seizure types were also observed, such as focal seizures on either side with secondary generalization in 20%. The development of GTCs in patients with focal convulsions is one of reasons that explains the difficulty of choosing the correct AED and the inability of monotherapy to control seizures. Intractable seizures were reported in 55% of patients, highlighting the challenge of managing epilepsy in this population, in some patients with intractable seizures the use of AEDs alone was not enough and further management including diet alterations (Ketogenic diet) and surgical operations have been offered. The median number of anti-epileptic drugs (AEDs) used was 2, with some patients requiring up to 3 AEDs particularly in those with intractable seizures. The recovery period from seizures was variable, ranging from days to years, with a mean of 24.0 days. These findings show the complexity of managing seizures in Sturge-Weber syndrome and the need for individualized treatment approaches to achieve seizure control and improve patient outcomes.

In the same line Powell et al. *[12]* revealed in retrospective review at Great Ormond Street Hospital focused on Sturge-Weber syndrome (SWS) patients. Seizure prevalence was notably high at 90%, emphasizing the importance of specialized care and multidisciplinary approaches in managing SWS.

The current study founded EEG findings revealed a range of abnormalities, with 67% of patients showing normal results. However, abnormal-

ities were observed in 33% of patients, including asymmetric background attenuation and atypical hypsarrhythmia pattern. These findings suggest that while a majority of patients may have normal EEGs, a significant proportion exhibit abnormal patterns, indicating underlying cortical dysfunction. These results highlight the importance of EEG monitoring in patients with Sturge-Weber syndrome to assess for seizure activity and cortical abnormalities, guiding treatment decisions and improving outcomes for these patients.

In alignment Pérez et al. *[4]* showed that the most common EEG abnormalities in SWS include spikes, spike-waves, or polyspike-waves, which were observed in 46% of patients. Low-voltage areas were also a common finding, observed in 31% of patients. These EEG findings are thought to be related to the leptomeningeal angiomatosis and the associated neurological symptoms, including seizures, which are present in 100% of SWS patients

The current study investigated the correlations between clinical manifestations (seizures, mental delay, motor deficit) and radiological findings (calcifications in CT, EEG findings, MRI findings). We found significant correlations between calcifications in CT and seizures (r=0.495, p=0.016), as well as mental delay (r=0.725, p<0.001), indicating that the presence of calcifications may be associated with a higher likelihood of experiencing seizures and mental delay in these patients. However, no significant correlation was found between calcifications in CT and motor deficit (r=0.437, p=0.054). EEG findings did not show significant correlations with any of the clinical manifestations, although there was a trend towards significance with seizures (r=0.357, p=0.146). Similarly, MRI findings showed a significant correlation with seizures (r=0.462, p=0.026) and mental delay (r=0.540, p=0.008), but not with motor deficit (r=0.304, p=0.192). These findings suggest that while CT and MRI findings may be useful in predicting seizures and mental delay in patients with Sturge-Weber syndrome, EEG findings may not be as predictive.

In agreement Kossoff et al. *[13]* demonstrated that EEG in patients with SWS does appear to evolve over time, becoming more abnormal with more frequent epileptiform activity, as suspected in smaller studies decades ago. This progressive change, however, did not correlate with the child's neurologic function or seizure frequency.

The current study evaluated the utility of EEG in predicting clinical outcomes in Sturge-Weber syndrome, we found that EEG had moderate discriminatory ability for predicting seizures (AUC=0.656), with a specificity of 68.8% and sensitivity of 100%. However, EEG showed poor discriminatory ability for predicting motor deficits (AUC=0.576) and mental delay (AUC=0.607), with specificity values of 56.3% and 78.6%, and sensitivity values of 71.4% and 100%, respectively. These results suggest that while EEG may be a valuable tool for predicting seizures in patients with Sturge-Weber syndrome, it may be less reliable for predicting motor deficits and mental delay.

In accordance Bar et al. *[14]* showed that Spikes on EEG might be a useful marker to identify patients with SWS at risk of developing epilepsy. Their predictive value should be assessed in larger prospective studies.

The current study investigated the predictive value of calcifications in CT for clinical outcomes in Sturge-Weber syndrome, we found that CT had good discriminatory ability for predicting motor deficits (AUC=0.839) and seizures (AUC=0.810), with specificity values of 64.3% and 71.4%, and sensitivity values of 83.3% for both outcomes. However, CT showed poorer discriminatory ability for predicting mental delay (AUC=0.631), with a specificity of 57.1% and sensitivity of 83.3%.

These findings suggest that calcifications in CT may be a valuable tool for predicting motor deficits and seizures in patients with Sturge-Weber syndrome, but may be less reliable for predicting mental delay.

In the same line Pilli et al. *[15]* demonstrated that brain calcifications are common and progress faster in children with SWS with early epilepsy onset, and are associated with a variable degree of hypometabolism, which is typically more extensive than the calcified area. Higher calcified brain volumes may indicate a risk for poorer neurocognitive outcome, so calcifications in CT may be a valuable tool for predicting motor deficits and seizures.

The current study evaluated the predictive value of MRI with contrast findings for clinical outcomes in Sturge-Weber syndrome, we found that MRI had moderate discriminatory ability for predicting seizures (AUC=0.780) and motor deficits (AUC=0.665), with specificity values of 69.2% and 53.8%, and sensitivity values of 100% and 85.7%, respectively. However, MRI showed poorer discriminatory ability for predicting mental delay (AUC=0.659), with a specificity of 53.8% and sensitivity of 85.7%. These results suggest that while MRI may be useful for predicting seizures and motor deficits in patients with Sturge-Weber syndrome, it may be less reliable for predicting mental delay.

Bosnyák et al. *[16]* suggested that while there may be a weak association between the extent of brain involvement on MRI and cognitive outcomes in terms of GIQ scores, this relationship does not extend to the frequency of seizures experienced by the individuals with Sturge-Weber syndrome.

Zallmann et al. *[17]* showed that there is no evidence to verify that early MRI (before 6 months)

results in better neurodevelopmental outcomes for infants with SWS.

The current study examined the relationship between seizure intractability and radiological and EEG findings in Sturge-Weber syndrome, we found no significant correlations between seizure intractability and calcifications in CT (r=0.272, p=0.275) or EEG findings (r=0.180, p=0.475). These results suggest that the presence of calcifications in CT or specific EEG patterns may not be reliable predictors of seizure intractability in patients with Sturge-Weber syndrome. The lack of significant correlations highlights the complexity of seizure control in this population.

Conclusion:

Our study provides valuable insights into the clinical and radiological features of Sturge-Weber syndrome and their associations with clinical outcomes. We found significant correlations between calcifications in CT and MRI findings with seizures and mental delay, highlighting the importance of these imaging modalities in predicting these outcomes. However, EEG findings showed limited predictive value for clinical outcomes. The presence of glaucoma was also found to be correlated with specific patterns of skin lesions. Our findings underscore the importance of comprehensive clinical and radiological assessments in the management of Sturge-Weber syndrome, and further research is warranted to validate these findings and improve our understanding of this complex condition.

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وصف النمط الظاهرى لمتلازمة ستيرج ويبر فى مجموعة من الأطفال الذين يحضرون إلى عيادة طب الاعصاب فى مركز رعاية ثالثية

الخلفية: يُعَدّ متلازمة ستيرج ويبر اضطرابًا عصبيًا جلديًا يرتبط بتشوهات الشعيرات الدموية فى الوجه (وحمة النبيذ البورتو)، الجلوكوما، والأورام الوعائية السحائية فى صورتها الكاملة. يُقدّر معدل حدوثها بحالة واحدة لكل ٢٠,٠٠٠–٥٠,٠٠٠ ولادة حية.

الهدف من الدراسة: تقييم ودراسة تنوع الطيف السريري بين مرضى متلازمة ستيرج ويبر وربط هذه المظاهر السريرية بالنتائج الإشعاعية بما فى ذلك التكلسات القشرية فى التصوير المقطعى للدماغ وتعزيز السحايا فى التصوير بالرنين المغناطيسى مع التباين.

المرضى وطرق البحث: ستُجرى هذه الدراسة المقطعية على ٢٠ حالة من حالات متلازمة ستيرج ويبر، وهى مرض نادر، تم اختيارها بناءً على معايير إدراج محددة. سيتم اختيار مجموعة الدراسة من المرضى الذين يترددون على عيادة الأعصاب للأطفال بكلية الطب، جامعة القاهرة، وعيادة الأطفال بمستشفى سعاد كفافى الجامعى، جامعة MUST، وكلاهما يقع فى القاهرة، مصر.

النتائج: كشفت الدراسة عن نتائج سريرية وإشعاعية هامة. كانت الآفات الجلدية فى الغالب على الجانب الأيسر من الوجه (٥٥٪)، مع وجود الجلوكوما فى ٧٠٪ من المرضى، وكانت مرتبطة بشكل كبير بتوزيع الآفات الجلدية (r=٠,٤٣٨، p=٠,٠٣٧). كانت التأخيرات فى النمو شائعة، حيث أظهر ٥٠٪ من المرضى تأخرًا فى النمو الحركى و٦٥٪ تأخرًا في النمو العقلى. كانت التحديات الإدراكية واضحة، حيث كان متوسط معدل الذكاء ٧٣,٥ مع تصنيف ٤٠٪ من المرضى بأنهم يعانون من معدل ذكاء منخفض جدًا. كانت الأورام الوعائية السحائية موجودة فى ٨٥٪ من الحالات، في حين تم العثور على التكلسات القشرية فى ٦٥٪، وغالبًا ما ترتبط بضعف الحركة الموجود فى ٧٠٪ من المرضى. كانت النوبات، خاصة الصرع المستعصى، بارزة فى ٥٥٪ من المرضى، مما يتطلب خطط علاجية معقدة وفردية. لوحظت تشوهات فى تخطيط كهربية الدماغ (EEG) فى ٣٣٪ من المرضى، وارتبطت بالنوبات والتأخر العقلى، على الرغم من أن تخطيط كهربية الدماغ كان أقل فعالية فى التنبؤ بالنتائج الحركية والإدراكية. أظهر التصوير المقطعى والتصوير بالرنين المغناطيسى قدرة تنبؤية جيدة إلى متوسطة للنوبات وعيوب الحركة، ولكنهما كانا أقل فعالية فى التنبؤ بالتأخر العقلى ولم يرتبطا باستعصاء النوبات. تبرز هذه النتائج أهمية التقييمات الشاملة والشخصية فى تحسين النتائج للمرضى الذين يعانون من متلازمة ستيرج ويبر.

الاستنتاج: توفر دراستنا رؤى قيمة حول السمات السريرية والإشعاعية لمتلازمة ستيرج ويبر وعلاقتها بالنتائج السريرية. وجدنا ارتباطات مهمة بين التكلسات في التصوير المقطعى والتصوير بالرنين المغناطيسى مع النوبات والتأخر العقلى، مما يبرز أهمية هذه الوسائل التصويرية فى التنبؤ بهذه النتائج. ومع ذلك، أظهرت نتائج تخطيط كهربية الدماغ قيمة تنبؤية محدودة للنتائج السريرية. كما وُجد ارتباط بين وجود الجلوكوما وأنماط محددة من الآفات الجلدية. تؤكد نتائجنا على أهمية التقييمات السريرية والإشعاعية الشاملة فى إدارة متلازمة ستيرج ويبر، ويجب إجراء المزيد من الأبحاث لتأكيد هذه النتائج وتحسين فهمنا لهذه الحالة المعقدة.