

Evaluating Retinal and Choroidal Changes in Sturge-Weber Syndrome through Optical Coherence Tomography (OCT) Imaging: Insights and Analysis

Dipanwita Ghosh¹, Dr. Partha Haradhan Chowdhury², Dr. Brinda Shah³

¹Assistant Professor & Head, Department of Optometry,
Swami Vivekananda University, Telinipara, Barasat -
Barrackpore Rd Bara Kanthalia, West Bengal – 700121

²Professor, Department of Optometry, Galgotias University,
Greater Noida, India

³Ph.D. Department of Optometry, Gujarat University

Abstract

Background: Sturge Weber Syndrome is a rare neurocutaneous condition which is characterized by ocular manifestations, neurological abnormalities and facial port wine stains. The aim of the study is to explore the changes in the eye by using Optical Coherence Tomography to examine the alterations in retina and choroid. This study was conducted during March 2023 to August 2024. Data was collected from multi-speciality clinics of Gujarat. All the subjects underwent high resolution spectral domain Optical Coherence Tomography imaging. Subjects were assessed for thickness of macula and choroid and structural abnormalities. Internal comparison was also done between affected and unaffected eyes. To evaluate statistically, tests like t tests, chi square test and correlation tests were incorporated with significance level set at $p < 0.05$. This study found that macular thickness is significantly increased in the affected eyes with value of $(290.3 \pm 15.2 \text{ microns})$ compared to the unaffected eyes $(273.7 \pm 12.5 \text{ microns}, p < 0.01)$. in areas where facial port wine stains are present, choroidal thickness was significantly reduced $(168.6 \pm 20.1 \text{ microns})$ as compared to unaffected areas $(198.4 \pm 18.7 \text{ microns}, p < 0.05)$. Additionally, structural abnormalities like retinal pigment

epithelial changes are seen in 65% and choroidal atrophy in 40% of the affected eyes. Significant positive correlation was found between the severity of choroidal changes and extent of facial port wine stains ($r = 0.62$, $p < 0.01$). This study reveals important differences in macular and choroidal thickness in Sturge Weber Syndrome, along with structural abnormalities. Significant correlation was also found between cutaneous and ocular manifestations. These findings underscore the connection between skin and ocular anomalies which signify the importance of comprehensive ocular examinations. These results also suggest avenues for further research to better understand the condition and to improve its clinical management.

Keywords: Sturge-Weber Syndrome, optical coherence tomography, retinal changes, choroidal changes, ocular manifestations, macular thickness, spectral-domain OCT, neurocutaneous disorder, cohort study, medical, community.

Introduction:

Sturge Weber Syndrome is a rare neuro cutaneous condition which is defined by an identifiable triad of facial port wine stain, neurological anomalies and ocular manifestations. The complex interaction between neurological, cutaneous and ocular elements poses appreciable challenges in knowing the complexity of this syndrome. Among its varied clinical manifestations, its ocular involvement in this syndrome has acquire the increasing attention due to its possible impact on ocular functions. This research focus on unravelling the ocular complexity of SWS by employing OCT for assessment of retinal and choroidal changes.

The ocular manifestations of SWS are known to embrace an array of anomalies, ranging from vascular deformity to structural modifications within the retina and choroid. There is much limited comprehensive research clarifying the limited research on specific retinal and choroidal changes associated with SWS, especially through advanced imaging technique such as OCT. This study addresses the gap by conducting evaluation of 23 subjects diagnosed with SWS, which employs high resolution OCT to examine macular and choroidal characteristics.

OCT with its non-invasive and high-resolution imaging capacity offers a specific possibility to dig into the micro structural modification of retina and choroid in subjects with SWS. The main objective of the study is to examine thickness of macula, choroidal thickness and the existence of structural anomalies in the affected eyes. It provides a significance of the ocular involvement in this syndrome. Comparative assessment between unaffected and affected eyes within the same subjects aim to depict specific changes related with the syndrome.

Basic literature suggest that choroid and retinal changes may serve as important indicator of the progression and severity of SWS related ocular complications. Alterations in the thickness of macula may reflect underlying vascular anomalies while changes in the choroidal thickness may be suggestive of vascular compromise in areas corresponding to facial port wine distribution of stain.

Furthermore, the determination of structural anomalies like RPE alterations and choroidal atrophy, may shed light on pathos physiological mechanisms which underly ocular manifestations in SWS.

The correlativity analysis between the severity of choroidal changes and extent of facial port wine stain introduces a fresh dimension to our understanding of the syndrome which suggest potential links between ocular involvement and cutaneous involvement. Identifying these associations is necessary for early detection, monitoring and strategies for management for ocular complications in subjects with SWS.

Methodology:

In this study, group of 23 subjects who were diagnosed with Sturge Weber Syndrome was precisely selected from specialized neuro cutaneous condition in clinics which ensures a democratic sample for the investigation. Detailed clinical assessments were conducted to conform the diagnosis, emphasizing the presence of facial port wine stains and neurological anomalies. Individuals with ocular comorbidities are excluded. Homogeneity is maintained within the study population. The research engages high

resolution spectral domain OCT, utilizing a standardized protocol. Macular thickness measurements cover central 6*6 mm area and choroidal thickness measurement focused on regions corresponding to facial port wine stain distribution were obtained. To recognize specific changes associated with SWS, internal comparative evaluation was conducted between affected and unaffected eyes within the same individuals. Quantitative analysis of choroidal and macular thickness measurement employed statistical tests with a p level of less than

0.05. Structural anomalies including RPE alterations and atrophy of choroid were identified through precise examination of OCT scans. Furthermore, correlational analysis explores the relationship between extent of severity of choroidal changes and facial port wine stain. Ethical consideration was predominant throughout the study, with approval obtained from ethical committee and commitment to patient privacy and confidentiality. These methodology aims to provide a detailed understanding of the ocular manifestations of SWS, which contributes important insights to the existing body of knowledge.

Results:

The results of our study expose significant insights into the choroidal and retinal changes associated with Sturge Weber Syndrome as assessed through precise OCT and detailed statistical analysis. Macular thickness in affected eyes shows statistically significant increase as compared to unaffected eyes, with a mean thickness of value 290.3 ± 15.2 microns in affected eyes and 273.7 ± 12.5 microns in unaffected eyes with value of ($p < 0.01$). Choroidal thickness shows variability, notably demonstrates a statistically significant reduction in areas corresponding to facial port-wine stain distribution, with a mean thickness value of 168.6 ± 20.1 microns in affected areas compared to 198.4 ± 18.7 microns in unaffected areas ($p < 0.05$). Structural anomalies were prevailing in affected eyes, with retinal pigment epithelial modification observed in 65% of cases and choroidal atrophy present in 40% of cases. Correlation analysis revealed a significant positive correlation between the extent of facial port-wine stain and the severity

of choroidal changes ($r = 0.62$, $p < 0.01$), suggests a possible link between cutaneous and ocular involvement. Relative assessments further highlighting distinct changes connected with SWS, systematically depicting enhanced macular thickness and decreased choroidal thickness in affected eyes compared to their unaffected counterparts.

Table 1: Comparison of Macular Thickness in Affected and Unaffected Eyes

Eye Condition	Mean Macular Thickness (microns)	Standard Deviation
Affected	290.3	15.2
Unaffected	273.7	12.5

Table 1 elaborate the mean macular dimension in affected and unaffected eyes, indicative a statistically significant increase in macular thickness in individuals with Sturge-Weber Syndrome with value of ($p < 0.01$).

Table 2: Comparison of Choroidal Thickness in Affected and Unaffected Areas

Area Condition	Mean Choroidal Thickness (microns)	Standard Deviation
Affected	168.6	20.1
Unaffected	198.4	18.7

Table 2 presents the average choroidal thickness in areas related to facial port-wine stain arrangement, indicating a statistically significant reduction in choroidal thickness in affected areas ($p < 0.05$).

Table 3: Shows Prevalence of Structural Abnormalities in Affected Eyes

Structural Abnormality	Percentage of Cases
Retinal Pigment Epithelial Alterations	65%
Choroidal Atrophy	40%

Table 3 shows the preponderance of structural abnormal

condition in affected eyes, with retinal pigment epithelial modification observed in 65% of cases and choroidal atrophy existing in 40% of cases.

Table 4: shows Correlation Between Facial Port-Wine Stain Extent and Choroidal Changes

Correlation Coefficient	p-value
0.62	<0.01

Table 4 exhibit the correlativity between the extent of facial port-wine stain and the severeness of choroidal changes, inform a significant positive correlation ($r = 0.62$, $p < 0.01$).

Discussion:

Macular Thickness:

The observed increase in thickness of macula in affected eyes as compared to unaffected eyes aligns with previous studies indicating vascular anomalies in SWS. These findings suggest possible alterations in retinal micro vasculature, possibly contributes to the observed increase. The main importance of the increased macular thickness in SWS may have ocular implications for visual function, which shows the need for ongoing observations and intervention scheme to address potential complications associated with macular thickness.

Choroidal Thickness:

The decrease in the thickness of the choroid corresponds to facial port wine stain distribution is a notable discovery. Thinning of choroid may be linked to compromise blood flow, possibly related to underlying vascular abnormalities. The localized nature of choroidal thinning aligns with vascular nature of the disorder which emphasize the need of investigations.

Structural Abnormalities:

The occurrence of structural anomalies like RPE alterations and atrophy of choroid improves our understanding of complex ocular manifestations in SWS. These findings underscore the need of detailed ocular examination in subjects with SWS for detection and management of structural changes at early age.

Correlation Analysis:

Positive correlation was found between the level of facial port wine stain and severity of choroidal changes is novel and interesting part of this study. This correlation suggests possible associations between cutaneous and ocular manifestations which indicates extent of facial involvement. It may serve as a clinical indicator of the severity of ocular changes.

Clinical Implications:

These findings have fundamental clinical implications for the management of Sturge Weber Syndrome. The observed changes in macular and choroidal thickness along with the presence of anomalies of structure highlights the need for specialized optometric care customized to the specific changes by this syndrome. Early detection and monitoring are crucial to monitor and to preserve ocular function and to improve quality of life.

Limitations and Future Directions:

The limitations of this study are relatively small sample size. Further research with larger groups and further longitudinal studies can provide more detailed understanding of the progression of ocular changes. Investigations on the more molecular and genetic factors underly the observed ocular alterations can improve our understanding of the pathophysiology of SWS.

Conclusion:

The multifarious nature of ocular changes in SWS requires ongoing investigations to modify our understanding and improve the management of this complex neuro cutaneous disorder.

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