INTERNATIONAL JOURNAL OF MEDICAL SCIENCE AND DENTAL HEALTH

Volume03 Issue07, July-2017, pg. 01-04

Published Date: - 04-07-2017

E-ISSN: 2454-4191
P-ISSN: 2455-0779

A SULLEN SPILL FROM SKIN TO EYES: A RARE CASE OF STURGE-WEBER SYNDROME

Dr. Mohit Gandhi

Department of Ophthalmology, Deen Dayal Upadhyay Hospital, Delhi, India

Abstract: Sturge-Weber Syndrome (SWS) is a rare and complex neurocutaneous disorder characterized by the presence of facial port-wine stain (nevus flammeus) and leptomeningeal angiomas. We present a unique and challenging case of Sturge-Weber Syndrome in a 6-year-old male with the hallmark facial port-wine stain and ocular manifestations. The patient presented with recurrent seizures and visual disturbances, prompting a thorough clinical evaluation. Magnetic resonance imaging (MRI) revealed leptomeningeal angiomas, confirming the diagnosis of SWS. This case highlights the importance of early recognition and multidisciplinary management to optimize patient outcomes and quality of life in individuals with Sturge-Weber Syndrome.

Keywords: Sturge-Weber Syndrome, neurocutaneous disorder, port-wine stain, nevus flammeus, leptomeningeal angiomas, seizures, visual disturbances, magnetic resonance imaging, multidisciplinary management, rare case.

INTRODUCTION

Sturge-Weber Syndrome (SWS), also known as encephalotrigeminal angiomatosis, is a rare and complex neurocutaneous disorder characterized by the presence of a facial port-wine stain (nevus flammeus) and leptomeningeal angiomas. This disorder is of great clinical significance as it can lead to various neurological and ophthalmological complications, including recurrent seizures, intellectual disabilities, and visual disturbances. The hallmark facial port-wine stain often presents in the distribution of the trigeminal nerve and is typically seen on one side of the face.

We present a unique and challenging case of Sturge-Weber Syndrome in a 6-year-old male with a distinctive facial port-wine stain and ocular manifestations. The patient's clinical presentation included recurrent seizures and visual disturbances, warranting a comprehensive evaluation to identify the underlying cause and manage the condition effectively. Magnetic resonance imaging (MRI) was crucial in confirming the diagnosis of Sturge-Weber Syndrome by revealing leptomeningeal angiomas.

In this report, we discuss the clinical presentation, diagnostic approach, and multidisciplinary management of this rare case of Sturge-Weber Syndrome. The case emphasizes the importance of early recognition, accurate diagnosis, and prompt multidisciplinary intervention to optimize patient outcomes and enhance the quality of life in individuals with this challenging neurocutaneous disorder.

Published Date: - 04-07-2017

E-ISSN: 2454-4191
P-ISSN: 2455-0779

METHOD

This case report involves a 6-year-old male patient who presented to our pediatric neurology clinic with a history of recurrent seizures and visual disturbances. A detailed medical history, including prenatal and perinatal events, family history, and developmental milestones, was obtained from the patient's caregivers. A thorough physical examination was performed, with particular attention to the presence and characteristics of the facial port-wine stain.

Diagnostic imaging was conducted to evaluate the neurological and ophthalmological aspects of the patient's condition. Magnetic resonance imaging (MRI) of the brain was performed with contrast to visualize the presence of leptomeningeal angiomas, a characteristic feature of Sturge-Weber Syndrome.

In addition to the neuroimaging, the patient underwent ophthalmological evaluation, including visual acuity testing, fundoscopy, and assessment for any ocular abnormalities related to Sturge-Weber Syndrome.

Following the diagnosis of Sturge-Weber Syndrome, a multidisciplinary team, including pediatric neurologists, ophthalmologists, and dermatologists, collaborated to develop an individualized management plan for the patient. The management plan focused on seizure control, visual rehabilitation, and addressing the cosmetic concerns associated with the facial port-wine stain.

Through this case report, we aim to highlight the importance of a comprehensive and collaborative approach to managing Sturge-Weber Syndrome, enabling improved patient outcomes and enhanced quality of life for individuals affected by this rare and complex neurocutaneous disorder.

RESULTS

The case involves a 6-year-old male presenting with a facial port-wine stain and recurrent seizures, prompting a comprehensive evaluation. Clinical examination revealed a characteristic port-wine stain involving the distribution of the trigeminal nerve on one side of the face. Diagnostic imaging with contrast-enhanced MRI confirmed the presence of leptomeningeal angiomas, leading to the diagnosis of Sturge-Weber Syndrome.

DISCUSSION

Sturge-Weber Syndrome is a rare neurocutaneous disorder with a wide spectrum of clinical manifestations. The hallmark facial port-wine stain is often the first clue to the diagnosis. In this case, the presence of the port-wine stain prompted further evaluation, leading to the identification of leptomeningeal angiomas on MRI.

The neurological complications of Sturge-Weber Syndrome can be debilitating, with recurrent seizures being a common and challenging feature. Prompt seizure control is essential to minimize the impact on

Published Date: - 04-07-2017

E-ISSN: 2454-4191
P-ISSN: 2455-0779

the patient's cognitive and developmental outcomes. In this case, the multidisciplinary team collaborated to develop an individualized management plan, including antiepileptic medications tailored to the patient's needs.

Ophthalmological manifestations in Sturge-Weber Syndrome can also be significant and range from refractive errors to more severe complications such as glaucoma and visual field defects. Early ophthalmic assessment and appropriate intervention are vital to address potential visual impairments and prevent irreversible visual loss. The patient in this case underwent thorough ophthalmological evaluation and received visual rehabilitation to optimize visual outcomes.

Multidisciplinary management is essential in Sturge-Weber Syndrome, given its diverse clinical manifestations affecting the skin, eyes, and brain. Collaboration among pediatric neurologists, ophthalmologists, and dermatologists is crucial to develop comprehensive and individualized treatment plans for affected patients.

CONCLUSION

This case report highlights a rare and challenging presentation of Sturge-Weber Syndrome in a 6-year-old male with a characteristic facial port-wine stain and ocular manifestations. The diagnosis was confirmed by MRI, revealing leptomeningeal angiomas, which are diagnostic of the condition.

Early recognition and prompt multidisciplinary intervention are key in managing Sturge-Weber Syndrome to optimize patient outcomes and enhance the quality of life for affected individuals. Seizure control and visual rehabilitation play critical roles in minimizing neurological and ophthalmological complications.

The report underscores the importance of awareness and understanding of Sturge-Weber Syndrome among healthcare professionals to facilitate early diagnosis and timely management. Collaboration between specialists ensures a comprehensive approach, addressing the multi-systemic aspects of this complex neurocutaneous disorder.

As this condition is rare, further research and case reports are essential to expand our knowledge and improve the management of Sturge-Weber Syndrome. Continued efforts to explore therapeutic options and advances in personalized medicine may lead to more effective treatments and better long-term outcomes for individuals with this challenging syndrome.

REFERENCES

1. Comi AM. Update on Sturge–Weber syndrome: diagnosis, treatment, quantitative measures, and controversies. Lymphat Res Biol 2007; 5:257–264.
2. Abdolrahimzadeh S, Scavella V, Felli L, et al. Ophthalmic alterations in the Sturge Weber syndrome, Klippel Trenaunay syndrome and the phakomatosis pigmentovascularis: an independent group of conditions? Biomed Res Int. 2015; Article ID 786519:11 pages.

Volume03 Issue07, July-2017, pg. 01-04

Published Date: - 04-07-2017

E-ISSN: 2454-4191
P-ISSN: 2455-0779

3. Mantelli F, Bruscolini A, La Cava M, Abdolrahimzadeh S, Lambiase A. Ocular manifestations of Sturge-Weber syndrome: pathogenesis, diagnosis, and management. Clin Ophthalmol. 2016 May 13; 10:871-8.
4. Rihani HT, Dalvin LA, Hodge DO, Pulido JS. Incidence of Sturge-Weber syndrome and associated ocular involvement in Olmsted County, Minnesota, United States. Ophthalmic Genet.
5. Sener RN. Sturge-Weber syndrome: a patient with a cervical port-wine nevus. Comput Med Imaging Graph. 1997 Nov-Dec. 21(6):359-60.
6. Enjolras O, Riche MC, Merland JJ. Facial port-wine stains and Sturge-Weber syndrome. Pediatrics. 1985 Jul. 76(1):48-51.
7. Silverstein M, Salvin J. Ocular manifestations of Sturge-Weber syndrome. Curr Opin Ophthalmol.
8. Doucette LP, Rasnitsyn A, Seifi M, Walter MA. The interactions of genes, age, and environment in glaucoma patholgenesis. Surv Ophthal- mol. 2015; 60:310–326.