ORIGINAL RESEARCH PAPER

Speech & Hearing

SPEECH AND LANGUAGE MANIFESTATIONS IN STURGE WEBER SYNDROME- A SINGLE CASE REPORT

KEY WORDS: Sturge Weber Syndrome, neurological disorder, speech language assessment

Rahul Kumminimana* Lecturer Institute For Communicative And Cognitive Neurosciences, Kavalappara, Shoranur Palakkad District. Kerala *Corresponding Author

Anuradha S Lecturer Institute For Communicative And Cognitive Neurosciences, Kavalappara, Shoranur Palakkad District. Kerala

Dr. Hamza Mullath Pediatrician Institute For Communicative And Cognitive Neurosciences, Kavalappara, Shoranur Palakkad District. Kerala

ABSTRACT

Sturge Weber Syndrome is a rare congenital neurological disorder that affects the development of certain blood vessels causing abnormalities in the brain, skin and eye. It is characterized by a red or pink mark called port-wine birth mark, a brain abnormality and increased pressure in the eye. A five-year-old girl diagnosed with Sturge Weber syndrome following speech language impairment was investigated. Following a detailed case history, assessment for pre-linguistic skills, Speech and Language skills, Oral Peripheral Mechanism Examination, vegetative functions, semantic skills, pragmatic skills and cognitive skills were assessed informally and formally using Receptive Expressive Emergent Language Scale (REELS) and 3-Dimensional Language Acquisition Test (3-D LAT). Subsequent to examination, the child had poor gross and fine motor skills, diasthemic teeth, red coloration on the tip of tongue with limited range and strength of movement, poor vegetative skills, age inadequate semantics and pragmatics skills. The participant had receptive and expressive language age of 9 to 10months, cognitive age of below 9months, which was age inadequate. The child also underwent detailed assessment for sensory skills. Detailed audiological examination showed child had normal hearing sensitivity bilaterally. On visual skills assessed child was found to have Glaucoma. The child also underwent neurological imaging studies and showed left cerebral atrophy and nonspecific epileptiform dysfunction. Based on the above subject presentation, the study explores communication impairment following Sturge Weber Syndrome and importance of assessment and rehabilitation of speech language skills in restoring their communication skills.

INTRODUCTION

Sturge Weber Syndrome is a rare congenital neurological disorder that affects the development of certain blood vessels causing abnormalities in the brain, skin and eye (Shprintzen, 2000). It is characterized by a red or pink mark called port-wine birth mark, a brain abnormality and increased pressure in the eye. These features may have difference in its severity and also it is not always present in all cases with Sturge Weber Syndrome. This neuro-dermatological disease is named in honor of British physicians Dr. William A Sturge and Dr. Frederick P Weber. Sturge weber syndrome is also known as encephalotrigeminal angiomatosis. The port-wine birth mark is due to the dilation of capillaries near the surface of the skin. The neurological concerns related to this disease is called as leptomeningeal angioma which in turns causes a disruption in the cerebral blood flow and the pressure variations in the eyes causes glaucoma (Adams, 1997). Sturge weber syndrome may be classified as a neurocutaneous syndrome or one of the Phakomatoses (Shields & Shields, 1992). The incidence was reported as 1 in 20000 to 50000 but only 6% of the cases have the classical symptoms of Sturge Weber Syndrome (Di Rocco & Tamburrini, 2006). This syndrome is classified in to 3 variants, type 1 cases will have skin and neurological symptoms and they may or may not have glaucoma. Type 2 exists with skin symptoms and possibly glaucoma and neurological symptoms will be absent. Type 3 consist of neurological involvement without skin abnormalities and glaucoma. Type 3 also known as isolated neurological variant (Roach, 1992). While coming to its genetic basis it is caused by somatic mutation in the GNAQ gene (Comi, 2015). Somatic mutations are not inherited which occur after conception. The GNAQ gene provides instructions for making a protein called guanine-nucleotide binding protein G(q) subunit alpha that regulates signaling pathways to help control the development and function of blood vessels. The affected individuals may have a mosaic pattern of expressions.

On pathophysiology, an atypical development of blood vessels within tissue layer coverings of brain and spinal cord occurs in patients with this disease. This is called as leptomeningeal angioma (Roach & Bodensteiner, 1999) and it leads to decreased blood flow in one or both side of the brain and atrophy and calcification. The reduced blood flow can cause stroke in cases with Sturge weber syndrome and this can cause muscle weakness, visual problems, seizures etc. Generally, these symptoms will begin with 2years of age and the seizures will be focal. The darkening of port-wine birthmark leads to loss of consciousness. While coming to the signs and symptoms, the most apparent feature of Sturge weber syndrome is the port-wine stain or red colored skin on the side of the face. It is due to the dilated blood vessels in the face. Not everyone with port-wine stain has Sturge weber syndrome but all children with Sturge weber syndrome have port-wine stain. They will also have abnormal vessels in brain and in some children, it may lead to seizures, paralysis, weakness on one side of the body, cognitive impairments and developmental delays. 50% of the children were also reported with Glaucoma and it will lead to eye pain and visual impairments (Comi et al., 2005).

Purpose of the study

The neurological impairments of children who were having Sturge weber syndrome will lead to deficiencies speech, language and cognitive skills. Hence to restore communication skills the early identification and intervention of speech and language skills of patients with Sturge weber syndrome is necessary.

Aim & Objectives

The aim of the study is to understand the speech and language characteristics of individuals with Sturge Weber Syndrome

METHOD

A five-year-old girl diagnosed with Sturge Weber syndrome was selected as the subject for the study. The client was reported with the complaint of poor speech output and developmental delay. Open and closed ended questions were compiled to complete case history through a structured interview. Information's regarding birth complications, family

history and developmental history of the child were collected. Followingly, subject's speech and language skills were assessed formally and informally by speech language pathologist. Oral peripheral mechanism examination was assessed to find oral structures and its functions, vegetative functions like blowing, sucking, swallowing, chewing and biting were assessed, clinical observations on pre-linguistic skills were made, reception and expression of language skills and cognitive skills were assessed formally using standardized tests. The formal assessment of language and cognitive skills was done using Receptive Expressive Emergent Language Scale (REELS) and 3-Dimensional Language Acquisition Test (3-D LAT). On detailed audiological evaluation, Brainstem Evoked Response Audiometry and Distortion Product Oto Acoustic Emission were done. The assessment of vision was done by an Ophthalmologist. The psychological assessment was carried out through Vineland Social Maturity Scale (VSMS) and Developmental Screening Test (DST). The subject also underwent Magnetic Resonance Imaging (MRI scan) and Electro Encephalography (EEG).

RESULTS

On the basis of parental interview, following information's have been reported. The appearance of port wine mark is present from the birth, episodes of seizure attacks started from the age of 6 months. No familial incidence of Sturge weber syndrome or any other kind of speech or motor delay reported. On the developmental history the child showed delay in motor and speech language milestones. All motor skills including gross and fine movements are impaired. Oral structures and its functions and vegetative functions are detailed in table 1 and 2 respectively.

Table 1: Oral Peripheral Mechanism Examination

ORGAN	STRUCTURE	FUNCTION	
Lips	Normal	Rounding	Not possible
		Spreading	
		Puckering	
Teeth	Diasthemic	Chewing	Possible
		Biting	Possible
Tongue	Red coloration on the tongue tip	Elevation	Limited
		Depression	Fair
		Protrusion	Good
		Retraction	Good
		Lateral movements	Limited
Hard palate	Normal		
Soft palate	Normal	Symmetrical movements: present	
Uvula	Normal		

Table 2:Vegetative skills

ACTIVITY	Response
Blowing	Not possible
Sucking	Not possible
Swallowing	Possible
Biting/ Chewing	Possible
Intra Oral Breath Pressure	Inadequate

Assessment on semantic skills, and pragmatic skills showed child is having reduced social interaction, poor verbal communication skills, poor receptive and expressive vocabulary. Results of formal assessment is given in table 3, which delineates the child is having age inadequate receptive and expressive speech and language skills with poor cognitive skills. On clinical observation, child shows bruxism, restlessness, mouthing of objects and motor stereotypies.

Table 3: Formal Assessment of Language and Cognitive Skills

REELS	Receptive Language Age	10-11 Months
	Expressive Language Age	9-10 Months
3DLAT	Reception	9-11 Months
	Expression	9-11 Months
	Cognition	<9 Months

On audiological evaluation, Brainstem Evoked Response audiometry results showed bilaterally 5th peak could be elicited at 25dBnHL with good wave morphology and replicability at 27.1 clicks/ second, suggestive of normal hearing sensitivity. DPOAEs were present at all frequencies in both ears, suggestive of normal outer hair cell functioning. Visual skills assessed showed Secondary Glaucoma is present in left eye. The subject had port wine mark on the face, tongue tip, upper limb, neck, hard palate and ears. The mark was more prominent on left side of the body as shown in figure 1.

Figure 1: phenotypic characteristics

On Psychological Investigation Vineland Social Maturity Scale administered the child is having severe level of retardation in social and adaptive functioning. The result of Developmental Screening Test shows severe developmental delay. On imaging studies, Magnetic Resonance Imaging shows atrophy of left cerebral hemisphere and EEG reports Epileptiform abnormalities arising from right centrero-parietal and left parieto-temporal region. Mild to moderate diffuse nonspecific epileptiform dysfunction.

DISCUSSION

The results revealed that the neurological impairments come as a part of Sturge weber syndrome made a developmental delay in the subject's speech and motor milestones. The language test results showed more than 4 years of delay in the child's language age from the chronological age. The psychological assessment revealed that the subject has severe level of retardation in social and adaptive functioning and severe developmental delay. The retardation of speech, language and cognitive skills were the result of cortical atrophy and epileptic discharge arising from right centrero parietal and left parieto-temporal region. This study was limited to a single case report. The study can be extended to include more subjects and discuss the speech language characteristics.

CONCLUSION

The study is concluded with highlighting clinical presentation of Sturge weber syndrome and speech language deficits. Hence, the early identification and intervention of speech and language skills of patients with Sturge weber syndrome is mandatory in the rehabilitation of these patients.

REFERENCES

1. Adams, R. D., Victor, M., Ropper, A. H., & Ropper, A. (1997). Degenerative diseases of the nervous system. *Principles of Neurology*, 1046–1107.
2. Bodensteiner, J. B., & Roach, E. S. (1999). *Sturge-Weber syndrome*. Mt. Freedom: Sturge-Weber Foundation.
3. Comi, A. M., Marchuk, D. A., & Pevsner, J. (2015). Sturge–Weber Syndrome. In R. N. Rosenberg & J. M. Pascual (Eds.), *Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease (Fifth Edition)* (pp. 945–953). Academic Press. https://doi.org/10.1016/B978-0-12-410529-4.00081-4
4. Comi, A. M., Mehta, P., Hatfield, L. A., & Dowling, M. M. (2005). Sturge-Weber syndrome associated with other abnormalities: A medical record and literature review. *Archives of Neurology*, *62*(12), 1924–1927. https://doi.org/10.1001/archneur.62.12.1924
5. Di Rocco, C., & Tamburrini, G. (2006). Sturge-Weber syndrome. *Child's Nervous System: ChNS: Official Journal of the International Society for Pediatric Neurosurgery*, *22*(8), 909–921. https://doi.org/10.1007/s00381-006-0143-2
6. Roach, E. S. (1992). Neurocutaneous syndromes. *Pediatric Clinics of North America*, *39*(4), 591–620. https://doi.org/10.1016/s0031-3955(16)38367-5
7. Shields, J. A., & Shields, C. L. (1994). The Systemic Hamartomatoses (" Phakomatoses"). *PENNSYLVANIA MEDICINE*, *97*, 26-26.
8. Shprintzen, R. J. (2000). *Syndrome Identification for Speech-language Pathologists: An Illustrated Pocket Guide*. Singular Publishing Group.