A rare case of Sturge–Weber syndrome: A case report

Abhinav Ahuja, Alka Bhambri, Syed Moiz Ahmed

Department of Pediatrics, Rohilkhand Medical College, Bareilly, Uttar Pradesh, India

INTRODUCTION

Sturge–Weber syndrome is a rare congenital neurocutaneous disease. The incidence of Sturge–Weber syndrome is 1 in 50,000 live births with somatic single nucleotide variant mutation in the GNAQ gene. It is commonly associated with port-wine stains of the face, glaucoma, convulsions, intellectual disability, headache, and hemiparesis. Sturge–Weber syndrome has features such as facial capillary malformation, ipsilateral leptomeningeal angiomata, and calcifications on CT head. It carries a poor prognosis and needs a multidisciplinary approach for the management.

KEY WORDS: Capillary malformation, neurocutaneous disorder, port-wine stain, Sturge–Weber syndrome

CASE STUDY

A 14-year-old male child came with complaint of weakness on the left side of the body and generalized tonic-clonic type seizures, two such episodes in the last 12 h at an interval of 4 h and headache left side, dull aching type, continues, not associated with any warning signs such as photophobia and phonophobia, but detailed history of the child was suggestive of developmental delay, that is, intellectual disability. The patient also complains of pain in the left eye for the last 1 day. On examination, the child had a port-wine stain on the left side of the face involving upper face and eyelid, as shown in Figure 1. Capillary malformation was also seen in the lower left part of the face. The child also had eleventh and 12th cranial nerve being involved with a deviation of the tongue to the left side. The attendants gave a history of hemangioma on the left side of the face at birth.
Radiograph of the head (NCCT HEAD) had findings of focal calcification alone the left parasagittal parietal cortex and mild atrophic changes in the left cerebral hemisphere compared to the right side, suggestive of focal left cerebral hemisphere gyral calcification with mild diffuse cerebral atrophy, as shown in Figures 2 and 3. The overall clinical and radiological features suggested the diagnosis of Sturge–Weber syndrome.

DISCUSSION

Sturge–Weber syndrome is a sporadic vascular disease having an incidence of 1 in 50,000 live birth with somatic single nucleotide variant mutation in the GNAQ gene.[2] It is an embryonal developmental anomaly due to mesodermal and ectodermal development errors in early stages of the face and brain development, and low flow angiomatosis of the leptomeninges leads to the chronic hypoxic state resulting cortical atrophy and calcifications. The patient mostly presents with seizures, developmental delay, headache, glaucoma, ambylopia, heterochromia iridis, and hemiparesis. Common features include port-wine stain, leptomeningeal angioma, facial capillary malformation, and calcifications on head CT. ROACH scale categorized Sturge–Weber syndrome into three types:[3,4]

a. Type 1: Facial and leptomeningeal angioma can have glaucoma
b. Type 2: Facial angioma (no central nervous system manifestation) can have glaucoma
c. Type 3: Isolated leptomeningeal angioma no glaucoma.

The Sturge–Weber syndrome is diagnosed on the basis of clinical and radiological findings. A port-wine stain is not the characteristic sign present at birth in babies of SWS. However, the child may develop a port-wine stain shortly after birth. Treatment for Sturge–Weber syndrome is decided on the basis of symptoms present in child. The management include involvement of various specialities. convulsions can be easily controlled by anticonvulsions, glaucoma can be managed with topical eye drops such as latanoprost or if not relieved surgeries for it can be opted. Physiotherapy for strengthening weak muscles and speech therapy for overcoming intellectual disability. Dermatologist consultation can be obtained for the management of port-wine stain using Nd:Yag laser. if the seizures are not controlled by medications then hemispherectomy can be an option. The patient may suffer from various complications such as stroke, seizures, epilepsy, cognitive delay, neurobehavioral abnormalities, and intellectual impairment.[5,6]

CONCLUSION

Sturge–Weber syndrome is a rare sporadic vascular disease and needs early identification and multidisciplinary intervention. The patient needs to be started on antiepileptics, analgesics, and physiotherapy and other supportive managements with frequent consultation with neurosurgeon, dermatologist, and ophthalmologist. The patient experience cerebral atrophy and convulsions in 1st year of life with the overall prognosis of the syndrome being poor.

REFERENCES

