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EDİTÖRE MEKTUP / LETTER TO THE EDITOR

Bilateral facial and cervical port-wine stain and Sturge-Weber syndrome

İkili yüz ve servikal port-wine lekesi ve Sturge-Weber sendromu

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Dear Editor,

Sturge-Weber syndrome is a rare phakomatosis syndrome. One of the cardinal features of that syndrome is the facial port-wine stain (nevus flammeus). The stain is usually seen within the distribution of the ophthalmic branch of the trigeminal nerve on one side. Extensive and bilateral facial as well as extra-facial (neck) involvement are rare and increase the likelihood of the diagnosis of Sturge-Weber syndrome.

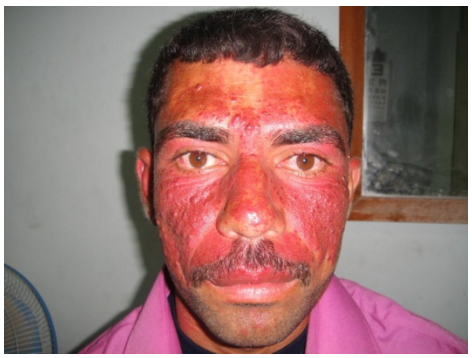

Figure 1. Patient's face. (There are bilateral port-wine stains involving V1 and V2 branches of the trigeminal nerve. In addition, similar "stains" can be noticed on both external ears and the lateral sides of the neck, reflecting extra-facial involvement of C2 and C3 dermatomes. Small scattered agniomas are present on the lower lip and chin. Right bulbar conjunctival/episcleral haemangioma can be seen at the lateral canthus. The patient has Sturge-Weber syndrome).

We report on the case of 30-year-old man who had mental retardation and recurrent seizures; he was diagnosed with cerebral palsy at the age of 7 years. The port-wine stain was bilateral and involved the ophthalmic and mandibular areas of the trigeminal nerves as well as the 2nd and 3rd cervical dermatomes. Examination revealed mental retardation, extensive bilateral facial port-wine stain (Figure 1), and bilateral conjunctival haemangioma.

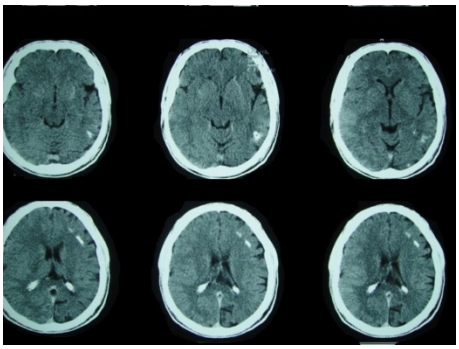

Figure 2. Non-contrast CT brain scan. (Note the left-sided sub-cortical, non-continuous, linear calcification. The overlying sulci seem to be more prominent, in comparison with those of the right cerebral hemisphere. This may well reflect some degree of cortical atrophy. The right cerebral hemisphere grossly appears normal. The patient's family declined doing cranial MRI; this would have provided us with a better image and a clearer delineation of the lesions.)

The intraocular pressure was normal. Funduscopic examination revealed bilateral focal-type of retinal angioma. The patient was uncooperative during perimetry; therefore, we could not assess his visual fields properly. Blood tests were within their normal reference range. Abdominal ultrasonography was normal. Sleep EEG revealed normal background but infrequent bilateral focal and generalized

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epileptiform discharges. Non-contrast brain CT scan was done (figure 2). The family declined doing brain MRI.The patient was diagnosed with Sturge-Weber syndrome (SWS1. Brouwer and colleagues added Sturge-Weber syndrome to the so-called phakomatosis syndromes in the year 1937; this groups is composed of neurofibromatosis, tuberous sclerosis, von Hippel-Lindau syndrome, and Klippel-Trenaunay syndrome[2]. The cardinal features of SWS are leptomeningeal angiomatosis (mainly at the occipital and parietal areas), facial agiomatosis (so-called port-wine stain or nevus flammeus), and ocular changes (e.g., glaucoma and retinal angioma)[3]. Although the syndrome results from a genetic mutation in the GNAQ gene, but it is sporadic and there is no clear-cut inheritance pattern; both genders are equally affected with an incidence of 1:50000 infants[3,4]. Aita, in the year 1966, concluded that during the first embryological trimester, an impaired development of certain cell precursors within the neural crest occurs; this results in the formation of the characteristic and cardinal malformations, which are observed in the central nervous system (CNS), skin, and eyes[5].

The clinical clue in this patient was the port-wine stain (PWS); however, the mere presence of such stains does not always reflect SWS, as they are observed in 1:300 infants[6]; Initially, after birth, these stains may be light pink in colour but they gradually darken. Port-wine stain is a congenital cutaneous capillary malformation which results from overabundance of capillaries around branches of the trigeminal nerve, just under the face. However, Waelchli and co-workers found that facial PWS's distribution appears to follow the embryonic vasculature of the face, rather than the trigeminal nerve itself [7]. Between 8-33% of patients with PWSs have SWS; Maraña Pérez et al analysed 13 patients with SWS and found that leptomeningeal angiomatoses were present in all patients (it was bilateral in 15% only) while facial angiomas were present in 61% of their patients: right (23%), left (38%) and bilateral (7%)[8]. According to Tallman et al, cutaneous extra-facial (cervical) stains were observed in 12% of patients[9]. Bioxeda and colleagues concluded that the presence of bilateral facial PWSs significantly increases the frequency of cutaneous extra-facial PWSs[10]. PWS of the ophthalmic (V_1) branch of trigeminal nerve is one of the cardinal features of SWS and that extension of this stain to the superior eyelid, to other territories of the trigeminal nerve (V_2, V_3), or to the contralateral hemi-face is statistically associated to SWS. According to Tallman et al [10], of those with trigeminal involvement in general, only 8% had CNS and eye involvement; 24% of those with bilateral lesions had eye or CNS involvement compared to only 6% with unilateral lesions. In terms of predicting the syndrome's adverse outcomes (progressive seizures, mental retardation, glaucoma, visual loss etc.), Waelchli and co-workers concluded that the bilateral distribution of PWSs was not an independently significant phenotypic feature and that abnormal cranial MRI was a better predictor of all clinical adverse outcome measures than PWS distribution[7].

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