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Case Report

Beyond the Port-Wine Stain: Unveiling Hypothyroidism in a Child with Type II Sturge-Weber Syndrome: A Case Report from Iraq

Wasnaa Hadi Abdullah*, Abdulameer Jasim Jawad al-Gburi
Department of Pediatrics, College of Medicine, Mustansiriyah University, Baghdad, Iraq
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Abstract

Facial port-wine stain, leptomeningeal capillary abnormalities, and ocular vascular anomalies are prominent features of the rare neurocutaneous disorder known as Sturge-Weber Syndrome (SWS). While the syndrome is primarily characterized by its dermatologic, ophthalmologic, and neurological features, multi-systemic involvement, including endocrinologic dysfunction, has also been reported. In this case report, we describe a 12-year, and 9-month-old boy diagnosed with SWS who was also found to have central hypothyroidism.

Keywords: Central hypothyroidism, Endocrine, Port-wine stain, Sturge-Weber Syndrome.

ما وراء وصمة بورت واين: الكشف عن قصور الغدة الدرقية لدى طفل مصاب بمتلازمة سمك الحفش من النوع الثاني: تقرير حالة من العراق

الخلاصة

تعد بقع نبيذ الوجه، والتشوهات الشعرية البريمية السحائية، وتشوهات الأوعية الدموية العينية من السمات البارزة للاضطراب العصبي الجلدي النادر المعروف باسم متلازمة ستورج ويبر (SWS). في حين أن المتلازمة تتميز في المقام الأول بخصائصها الجلدية والعيون والعصبية، فقد تم الإبلاغ أيضا عن تورط متعدد الأجهزة، بما في ذلك الخلل الوظيفي في الغدد الصماء. في تقرير الحالة هذا، نصف صبيا يبلغ من العمر 12 عاما و 9 أشهر تم تشخيص إصابته ب SWS والذي وجد أيضا أنه مصاب بقصور الغدة الدرقية المركزي.

* ***Corresponding author***: Wasnaa H. Abdullah, Department of Pediatrics, College of Medicine, Mustansiriyah University, Baghdad, Iraq; Email: wasnaa.hadi@uomustansiriyah.edu.iq
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INTRODUCTION

One in 50,000 live infants is believed to have the rare neurocutaneous illness known as Sturge-Weber Syndrome (SWS). In 1860, Schirmer was the first to identify the syndrome; in 1879, Sturge officially recognized it [1]. Postzygotic somatic mutations in the GNAQ gene, which codes for the G protein α subunit (Gαq), are the main cause of SWS. Another factor in the disorder's etiology, according to recent research, is random gene mutations (GNA11 and GNB2) [2]. SWS typically manifested as port-wine staining of the face and often impacts the distribution of the trigeminal nerve. Additionally, there might be leptomeningeal capillary anomalies and ocular vascular abnormalities. Among the severe neurological and ophthalmologic complications that might develop from these diseases are glaucoma and epilepsy [3]. The degree of systemic effects determines whether SWS is complete or partial. In contrast to partial SWS, which impacts just one of the aforementioned areas, complete SWS incorporates both facial and leptomeningeal angiomas. The three clinical types of the condition are commonly classified using the Roach scale [4]; In type I, glaucoma is present in addition to face and leptomeningeal angiomas. Isolated facial angiomas, whether or not glaucoma is present, constitute Type II. Isolated leptomeningeal angiomas, typically without involving the eyes, are Type III. Nearly 40% of cases include abnormalities in the mouth, such as hemiatrophy of the floor of the mouth, palatal mucosa, buccal mucosa, or angiomatosis of the lips, which causes macrocheilia [5]. We report here a patient with central hypothyroidism and Sturge-Weber Syndrome. To the best of our knowledge, this is the first reported instance of such a connection in Iraq, drawing attention to an unusual endocrine symptom in a patient suffering from a mostly neurocutaneous disease.

Case Presentation

The pediatric endocrinology outpatient clinic received a referral of a 12-year-and-9-month-old boy who complained of chronic exhaustion and trouble focusing on schoolwork. There was no correlation between the symptoms and any changes in the patient's visual or auditory perception, gait, limb strength, headaches, seizures, or bowel habits. The patient's mother reported that her child had a bluish-red discoloration on their face that had been there from birth but had become darker over the years. Gingival hypertrophy and bleeding

episodes when brushing teeth were other symptoms she saw. The patient had mouth expansion (mandibular hypertrophy) and crowding of the teeth, for which odontoplasty was done when he was five years old. He had no prior history of drug use or epilepsy. The mother had hypothyroidism, and an aunt on her mother's side had hyperthyroidism, two thyroid illnesses that were prominent in family history. Nobody in the family has any skin lesions like that. Several branches of the right trigeminal nerve were involved in a port-wine stain (PWS) that was discovered during the physical examination. It had spread from the middle of the right forehead to the upper neck, lower lip, mandible, cheek, philtrum, and lateral aspect of the right eye. We saw clinical evidence of mandibular hypertrophy (Figure 1 A and B).

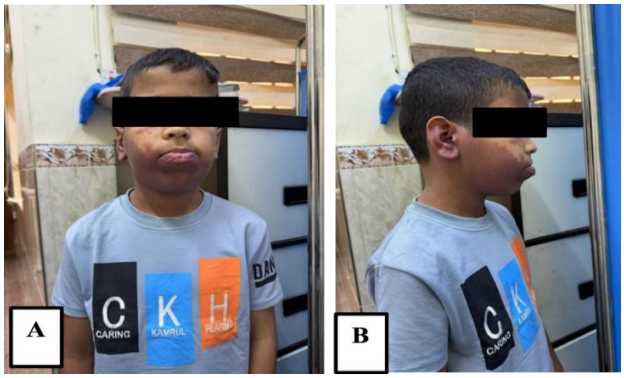

Figure 1: **A**) The child's overall appearance. **B**) Where lesions stained with port wine appear on the skin.

Iris color alterations and conjunctival telangiectasia were not detected during the ophthalmologic examination. A thorough ophthalmic evaluation was ordered for the patient because of the increased risk of glaucoma in Sturge-Weber syndrome; however, the results came back to normal. Gingival overgrowth of the lower teeth was seen during the intraoral exam as reddish pink. No detectable goiter was found during the thyroid exam. The results of the neurological evaluation were normal, with no obvious abnormalities. The patient's vital signs were within normal range, his height was 143 cm, and his body weight was 36 kg. As far as Tanner sexual maturity rating (SMR) was concerned, it was 1.

Investigations

A low free thyroxine (fT4) level of 0.4 ng/dL (reference range: 0.7-1.6 ng/dL) and an unexpectedly normal thyroid-stimulating hormone (TSH) level of 1.1 IU/mL (reference range: 0.47-4.9 IU/mL) were found during thyroid function testing, which indicates the possibility of central (secondary) hypothyroidism. Additional examination of pituitary function was conducted in light of these findings. It was ruled out that there is no adrenal insufficiency because both the morning serum cortisol (501 nmol/L) and serum adrenocorticotropic hormone (ACTH) were within the normal range (166-507 nmol/L and 51 pg/mL, respectively). It was recommended to closely observe the growth velocity due to the presence of central hypothyroidism. In order to rule out concomitant growth hormone (GH) insufficiency, a stimulation test should be conducted if growth slowing is detected. Neuroimaging was conducted to assess for intracranial involvement in light of laboratory data that indicated central hypothyroidism in the context of Sturge-Weber syndrome. There were no abnormalities detected on gadolinium contrast-enhanced MRI and brain CT scans, including the absence of leptomeningeal enhancement and calcifications. Type II Sturge-Weber syndrome, defined by facial capillary abnormalities absent from the central nervous system, is supported by these findings.

Treatment

Levothyroxine, at a dosage of 2-4 µg/kg/day, or 75 µg per day, was initiated as a thyroid hormone replacement therapy for the patient. It was advised to closely observe the thyroid's function and growth indicators.

DISCUSSION

Facial port-wine stains, ocular abnormalities, and leptomeningeal angiomas are the hallmarks of the uncommon neurocutaneous condition Sturge-Weber Syndrome [1]. In SWS, it is not uncommon for multiple systemic symptoms to co-occur, including symptoms related to dermatology, neuropsychiatry, ophthalmology, and endocrinology [6]. A genetic capillary malformation called nevus flammeus, or port-wine stain (PWS), affects about 1 in 300 live infants. The predicted chance of developing SWS among children born with a PWS ranges from seven percent to twenty-eight percent. Typically, SWS patients only experience unilateral facial PWS. However, it is worth noting that around 15% of instances have documented bilateral involvement [6]. A characteristic aspect of Sturge-Weber Syndrome is the involvement of the ocular division of the trigeminal nerve, which is followed dermatatomally by the ophthalmic (V1) and maxillary (V2) branches. Port wine stains can darken, grow more nodular, and even become hypertrophic with time [7]. The majority of patients with SWS who develop epilepsy begin experiencing seizures within the first year of life, and this number rises to 75% to 90% when there is intracranial involvement. Cognitive impairment and diminished neurodevelopmental quality of life are hallmarks of early-onset epilepsy. Because uncontrolled seizures raise the risk of stroke-like episodes and progressive brain injury, effective seizure prevention is critical in the management of SWS [8]. Growth abnormalities, typically caused by underlying endocrine problems, are another possible symptom in patients with SWS, alongside the more common neurological and cutaneous signs. Significantly, there is evidence that growth hormone insufficiency is up to 18 times more common in SWS patients than in the normal population. This may explain why affected children show signs of reduced linear growth [9]. Central hypothyroidism was present in 2.4% of SWS patients, a significantly higher rate than the general population [10]. Hypothyroidism is a major endocrine abnormality that is seen in many syndromic disorders, such as Moyamoya syndrome and Prader-Willi syndrome. It plays a big role in the overall clinical picture of these ailments and needs to be managed carefully [11–13]; given the possible disturbance of the

hypothalamic-pituitary axis, it is important to maintain a high index of suspicion for hypothyroidism in patients with SWS, as shown in our indexed case. The fact that endocrine dysfunction can happen even when leptomeningeal involvement is not present is what makes this case noteworthy. Regardless of the results of neuroimaging, it is crucial to check for endocrine disorders such as central hypothyroidism and growth hormone insufficiency routinely. The importance of endocrine examination in patients with hypothyroidism was highlighted in a case report by Saroj et al. involving an 11-year-old girl with prominent oral and facial symptoms of Sturge-Weber Syndrome [14]. SWS has also been found to be associated with aortic diseases [15,16]. Although neuroimaging usually shows normal hypothalamic and pituitary structures, patients with SWS have been found to have central hypothyroidism and growth hormone insufficiency, according to a review by Ramirez EL. Uncertainty surrounds the pathophysiology of this association, which may be due to a functional rather than anatomical disturbance of the hypothalamic-pituitary axis [6].

Conflict of interests

No conflict of interest was declared by the authors.

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Data sharing statement

Supplementary data can be shared with the corresponding author upon reasonable request.

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