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Combination of Sturge-weber Syndrome, Klippel-trenaunay Syndrome and Widespread Dermal Melanocytosis in a Female Infant: A Rare Association

Leila Tajziehchi*

Department of Pediatrics, Bahrami Hospital, Iran

ABSTRACT

Phacomatosis Pigmentovascularis (PPV) is a rare sporadic genodermatosis characterized by the combination of an extensive pigmentary nevus with a widespread vascular abnormality. PPV had originally been classified into four types and a fifth type was added later. Each type of PPV has two subtypes with subtype 'a' for cutaneous involvement only and subtype 'b' for cutaneous and systemic involvement. Here in we describe a case of PPV (type 2b) which presented with sturge-weber syndrome, widespread dermal melanocytosis, klippel-trenaunay syndrome. To the best of our knowledge, very few cases with these associations have been reported to date. Our case had widespread dermal melanocytosis (nearly most of the body), which made it unique.

Keywords: Phacomatosis pigmentovascularis; Klippel-trenaunay syndrome; Dermal melanocytosis

INTRODUCTION

Phakomatosis Pigmentovascularis (PPV) is a rare condition first described by Ota et al., in 1947 [1]. Since then around 250 cases been reported, being mostly sporadic [2]. It is a rare genodermatosis characterized by the combination of an extensive pigmentary nevus with a widespread vascular abnormality [3]. In 1985 it has been classified by Hasefawa and Yasuhara into 4 types according to the different characteristics of the vascular and pigmentary malformations: Type I, nevus flammeus (port-wine stain) with nevus pigmentosus et verrucosus; Type II, nevus flammeus with aberrant dermal melanocytosis; Type III, nevus flammeus with nevus spilus; Type IV, nevus flammeus with both aberrant dermal melanocytosiss and nevus spilus [3-5]. In 2003, Torrelo et al., described PPV type V with Cutis Marmorata Telangiectatica Congenita (CMTC) associated with aberrant mongolian spot [6]. Each type of PPV has two subtypes with subtype 'a' for cutaneous involvement only and subtype 'b' for cutaneous and systemic involvement. In 2005, Happle proposed a new simplified classification into four groups, utilizing descriptive terms: Phacomatosis Cesioflamea (association of the nevus flameus and aberrant Mongolian spots or blue nevus); Phacomatosis Spilorosea (coexistence of Nevus Spilus and telangiectatic nevus), Phacomatosis Cesiomarmorata (association of aberrant Mongolian spots or blue nevus with Cutis Marmorata Telangiectatica Congenita) and Phacomatosis Pigmentovascularis of non-classifiable type. Differentiation between cases with or without extracutaneous features was eliminated [7]. Approximately 50% of PPV patients have associated one or more comorbidities, including Sturge-Weber Syndrome (SWS), Klippel-Trenaunay syndrome, congenital glaucoma, mental retardation, hydrocephalus, congenital triangular alopecia and skeletal abnormalities [5]. Here, we report an infantile case of, Phakomatosis pigmentovascularis with Sturge-Weber Syndrome (SWS), Klippel-Trenaunay syndrome and widespread dermal melanocytosis.

CASE PRESENTATION

A 11-month-old female infant presented to our department with focal seizure. She was born from related parents (first degree parental consanguinity) at term with cesarian section because of fetal distress. At physical examination, she had delayed developmental milestones. We noticed widespread Mongolian spots involving the abdomen, chest, back, buttocks, extensor

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Corresponding author Leila Tajziehchi, Department of Pediatrics, Bahrami Hospital, Iran, E-mail: dr_l_tajziechi@yahoo.com

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surfaces of the upper and lower extremities. She had port-wine stain on the right side of face and hypertrophy of right upper limb (Figure 1). The patient had a history of right-eye glaucoma operation at 3 months old. Neuroradiological findings in these patients showed leptomeningeal enhancement. For seizure treatment phenytoin (50 mg/day) and phenobarbital (25 mg/day) prescribed.

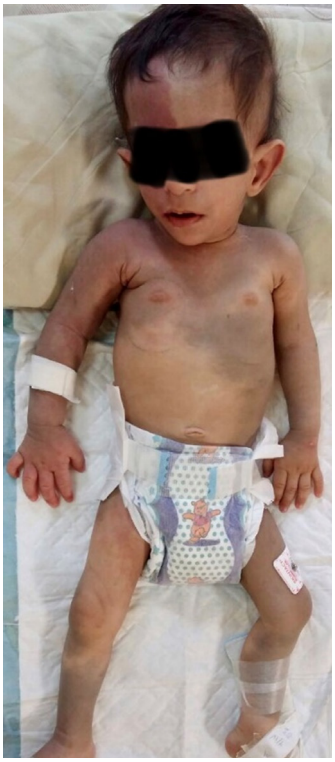

Figure 1: Port-wine stain on the right side of face and hypertrophy of right upper limb

RESULTS AND DISCUSSION

Sturge-Weber syndrome is a neurocutaneus syndrome characterized by port-wine nevus covering face and cranium (usually the area supplied by first branch of trigeminal nerve) along with atrophy and calcification of cerebral hemisphere, homolateral to the skin lesion. It is a rare sporadic syndrome occurs with a frequency of approximately 1 in 50,000 births. Sturge-Weber syndrome is associated with glaucoma, seizure, paresis, and neurodevelopmental delay. Most of the patients with epilepsy have their first seizure before one year old [8]. Klippel-Trenaunay Syndrome (KTS) is characterized by 3 clinical features, namely cutaneous capillary malformations, venous malformations, and soft tissue and/or bony hypertrophy of the extremity. At least two of the three above major clinical features are necessary for diagnosis of KTS. KTS mostly occurs sporadically, with only rare cases of familial background [9].

Dermal melanocytosis is characterized by the presence of ectopic melanocytes in the dermis. The most common forms include the Mongolian spot, blue nevus, nevus of Ota, and nevus of Ito [10]. Historically, Mongolian spot have been regarded as benign. Recent data, show that Mongolian spot may be associated with other conditions like various inborn errors of metabolism, Sturge-Weber syndrome, Klippel-Trenaunay syndrome, cutis marmorata telangiectatica congenita and segmental cafe-au-lait [11].

In the realm of pediatric medicine, encountering a confluence of rare syndromes in a single patient is an exceptional rarity. Such is the case with a female infant presenting a distinctive amalgamation of Sturge-Weber Syndrome (SWS), Klippel-Trenaunay Syndrome (KTS), and widespread dermal melanocytosis. This unique association poses a complex clinical challenge, requiring a multidisciplinary approach for comprehensive management. Sturge-Weber Syndrome, characterized by facial port-wine stains and neurological abnormalities, has crossed paths with Klippel-Trenaunay Syndrome, a disorder marked by vascular malformations and soft tissue overgrowth. These two conditions, individually uncommon, share a common denominator of vascular anomalies but rarely manifest concurrently in a single patient. The addition of widespread dermal melanocytosis further complicates the clinical picture, as this condition involves the abnormal proliferation of pigment-producing cells in the skin.

The challenges presented by this triad of conditions extend beyond their individual complexities. Neurological implications from SWS, vascular anomalies from KTS, and pigmentation irregularities from dermal melanocytosis collectively demand a holistic and coordinated medical approach. Early intervention and close monitoring become imperative to mitigate potential complications, such as seizures, developmental delays, or circulatory issues. Treatment strategies may include a combination of medical, surgical, and supportive interventions. Laser therapy may address the cosmetic aspects of port-wine stains, while anticonvulsant medications could help manage seizures associated with SWS. Addressing the vascular component of KTS may require surgical intervention, and vigilant dermatologic care is essential to monitor and manage dermal melanocytosis. The rarity of this association underscores the importance of heightened awareness among healthcare professionals, prompting timely diagnosis and tailored therapeutic strategies. Collaborative efforts between neurologists, dermatologists, and vascular specialists are crucial to navigating the intricate web of symptoms these syndromes present. In unravelling this rare tapestry, medical science advances not only in understanding the nuances of these conditions but also in refining the collective approach to complex pediatric cases.

CONCLUSION

Our case had features that made it unique. She presented with widespread dermal melanocytosis, which covered trunk, upper and lower extremities. She had right side hemihypertrophy, portwine stain on the face, focal motor neuron seizure, history of glaucoma. Although rare cases of dermal melanocytosis, Sturge-Weber syndrome and Klippel-Trenaunay syndrome combination have been reported to date, but our case is the first one with such widespread dermal melanocytosis.

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CONFLICT OF INTEREST

The author's declared that they have no conflict of interest.

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