Case 16

An 18-year-old woman, from Bangkok.

Chief complaint: Erythematous patches on her cheeks and left forearm since birth.

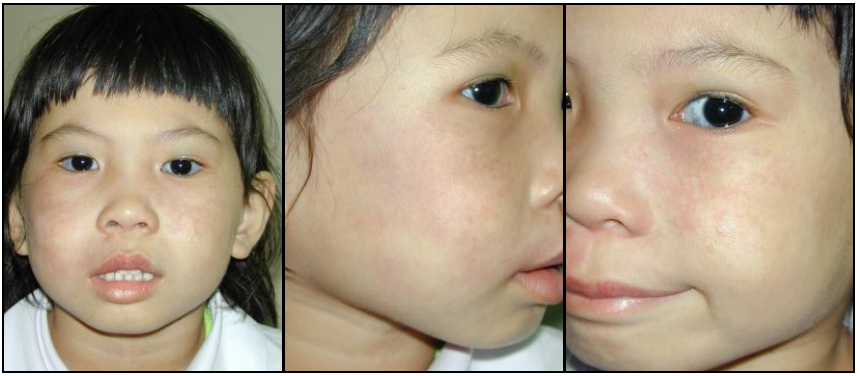

Fig 1. Pinkish-erythematous patch at right cheek, multiple brownish macules at both cheeks and bilateral bluish sclera.

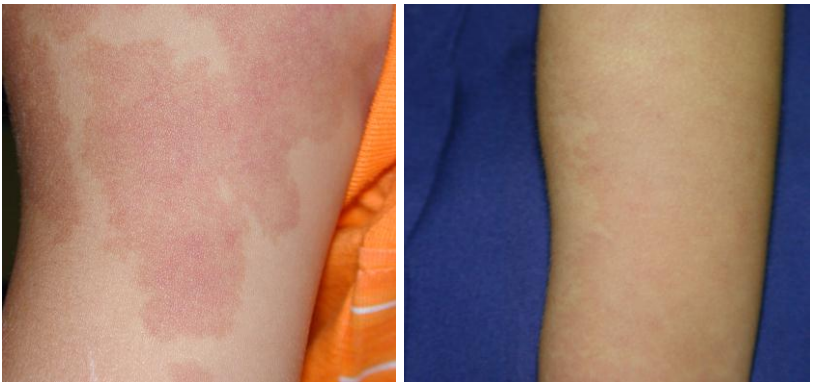

Fig 2. Well-defined irregular border erythematous patches at left forearm.

Present illness: The patient had erythematous patches on her cheeks with bilateral bluish sclera, and also had several well-defined irregular border erythematous patches at left forearm.

Past history
In May 2013, she was diagnosed with breast masses and underwent excisional biopsy, pathological examination showed phyllodes tumor.

Physical examination
HEENT: no pale conjunctiva, anicteric sclera, hyperpigmented patches at sclera both eyes .
no alopecia .
Breast: no masses, no nipple retraction or discharge
Heart : normal S1S2, no murmur
Lung : normal breath sound , no adventitious sound
Abdomen : soft, not tender, no hepatosplenomegaly.
Extremities : symmetrical extremities.
Neurologiacal : good consciousness, no muscle weakness.

Skin examination
- ill-defined ,faint pinkish-erythematous patch at right cheek
- multiple faint brownish macules at both cheeks with bilateral scleral involvement.
- Well-defined , irregular border, erythematous patches at left forearm.

Diagnosis: Phakomatosis pigmentovascularis type IIa
Treatment: QS Nd:YAG (1064) LASER

Presenter: Natnicha Girdwichai, M.D.
Consultant: Somsak Tanrattankorn, M.D.

Discussion

Phakomatosis pigmentovascularis (PPV) is a group of congenital skin disorders combining extensive nevi of the capillary type and various forms of (epi)dermal melanocytosis,with or without additional cutaneous features .[1] PPV was first published in 1910, but it was described by Ota in 1947.[2]

PPV is very rare ,sporadic syndrome. The true prevalence is unknown with a slight female predominance (ratio of female : male was approximately 1.34:1) .[3]

PPV is mainly diagnosed by clinical of skin manifestations ,that were classified into 5 groups according to associated pigmentary anomalies by Enjorlas and Mulliken in 2000.[4]And are classified into 4 groups by Happle in 2005.[5]

In this classification Phakomatosis spilorosea lacks Mongolian spots and Phakomatosis cesiomarmorata lacks naevus flammeus.[1]

Table 1. Classification of PPV[5-7]

Original classification	**Happle's shortened classification**
• **Type I**: Nevus flammeus and pigmented linear epidermal nevi • **Type II**: Nevus flammeus and Mongolian spots and/or nevus anemicus • **Type III**: Nevus flammeus and nevus spilus and/or nevus anemicus • **Type IV**: Nevus flammeus, Mongolian spots, and nevus spilus and/or nevus anemicus • **Type V**: Cutis marmorata telangiectatica congenital and Mongolian spots • **Unclassifiable** • **Type A or B** depending on whether or not there is systemic involvement	• **Phacomatosis cesioflammea**: Nevus cesius (blue spot) and nevus flammeus • **Phacomatosis spilorosea**: Nevus spilus and telangiectatic nevus of a pale-pink type (nevus roseus) • **Phacomatosis cesiomarmorata**: Nevus cesius (blue spot) and cutis marmorata telangiectatica congenital • **Phacomatosis pigmentovascularis, unclassifiable type** **Correspondence between 2 systems** • Cesioflammea IIA, IIB • Spilorosea IIIA, IIIB • Cesiomarmorata VA, VB • Unclassifiable IA, IVB • Type I from the first classification is eliminated

The most common type of PPV is IIb , followed by IIa .The others are much less frequent.[8]

Patients with PPVs may presented with other associated skin and systemic manifestations in PPV (table2).

Table 2. Phakomatosis cesioflammea: published association[3, 5, 9-21]

Cutaneous lesions	Nevus anemicus Café-au-lait spots Generalized vitiligo
Vascular abnormalities	Sturge-Weber Klippel-Trénaunay
Neurologic abnormalities	Seizures Cortical atrophy Arnold-Chiarri type I Bilateral deafness Idiopathic facial paralysis Hydrocephalia Diabetes insipidus Plexiform neurofibroma Delay in psychomotor development Encephalogram alterations
Ocular alterations	Melanosis oculi Iris mammilations Iris hamartomas Glaucoma Prominent vessels in sclera Chronic edema in the cornea Pigmentary alterations in retina Pigmentary cataract
Miscellaneous	Discrepancy in the length of extremities Scoliosis Spinal dysraphism Hemihypertrophy Syndactilia Macrocephalia Renal agenesia Renal angiomatosis Hepatosplenomegaly Pyogenic granuloma Cavernous hemangioma Umbilical hernia Hypoplasia of leg veins IgA deficit Hyper-IgE syndrome Eczemas Premature eruption of the teeth

The most common dermal melanosis is Nevus of Ota and aberrant Mongolian spots.[8] However, the real extent of the associated clinical spectrum is not well defined.[22]

The etiology is unknown. Sporadicity and mosaic distribution of skin lesions suggest twin spotting (didymodisis). Twin spots are two different cutaneous areas that must be adjacent to one another,formed by mutant tissues that also differ from normal tissue surrounding them.[23]

Histopathology is seldom necessary to make diagnosis.[1]

Laser is treatment for patient with aesthetic nuisance. Ono and Tateshita reported one case treated with a Q-switched ruby laser and a dye laser.[24] A combined laser approach (a Q-switched Ruby laser, a QAL, and a flashlamp pumped pulsed-dye laser) is also effective in PPV type IIa patient .[25] Port-wine stain (PWS) can be treated with long-pulsed dye laser (LPDL) .[26] And Mongolian spots can be treated with Q-switched ruby and Alexandrite lasers.[27] For Cutis marmorata telangiectatica congenital (CMTC) .There are reports that frequency –doubled Nd:YAG/neodymium-doped yttrium aluminum garnet failed to improve CMTC because of dilated veins extensive large, deep capillaries.[28] According to the same reason, LPDL therapy was not effective for CMTC.[4]

The prognosis of PPV depends on type and severity of associated abnormalities.[1]

In this patient, she was diagnosed with phakomatosis pigmentovascularis nevus type IIa (nevus of Ota bilaterally, PWS at right cheek, nevus flammeus at left arm). Annual eye examination revealed normal visual acuity. She was treated with Nd:YAG laser 1064 nm.

References

1. Ruiz-Maldonado R, Duràn-McKinster C, Orozco-Covarrubias L , Saez-De-Ocariz M. Phakomatosis Pigmentovascularis. In: M. Ruggieri, I. Pascual-Castroviejo and C. Rocco editors. Neurocutaneous Disorders Phakomatoses and

Hamartoneoplastic Syndromes: Springer Vienna; 2008. p. 449-54.
2. Ota N. Phakomatosis pigmentovascularis. Jpn J Dermatol 1947;57:1-3.
3. Vidaurri-de la Cruz H, Tamayo-Sanchez L, Duran-McKinster C, Orozco-Covarrubias Mde L , Ruiz-Maldonado R. Phakomatosis pigmentovascularis II A and II B: clinical findings in 24 patients. J dermatol 2003;30:381-8.
4. Adachi K, Togashi S, Sasaki K , Sekido M. Laser therapy treatment of phacomatosis pigmentovascularis type II: two case reports. J Med Case Rep 2013;7:1-5.
5. Happle R. PHacomatosis pigmentovascularis revisited and reclassified. Arch dermatol 2005;141:385-8.
6. Hasegawa Y , Yasuhara M. Phakomatosis pigmentovascularis type IVa. Arch dermatol 1985;121:651-5.
7. Enjolras O MJVmIHJ, Oranje A, Prose N eds. Textbook of Pediatr Dermatol . Oxford: Blackwell Science, 2000: 975–96.
8. Fernandez-Guarino M, Boixeda P, de Las Heras E, Aboin S, Garcia-Millan C , Olasolo PJ. Phakomatosis pigmentovascularis: Clinical findings in 15 patients and review of the literature. J Am Acad Dermatol 2008;58:88-93.
9. Colin TS , Kumarasinghe Sujith PW. Phakomatosis Pigmentovascularis Type IIB: a case report. J dermatol 2004;31:415-8.
10. Cordisco MR CA, Castro C, Bottegal F, Bocian M, Persico S. Phakomatosis pigmentovascularis: report of 25 cases. Pediatr dermatol 2001.
11. Hagiwara K, Uezato H , Nonaka S. Phacomatosis pigmentovascularis type IIb associated with Sturge-Weber syndrome and pyogenic granuloma. J dermatol 1998;25:721-9.
12. Ruiz-Maldonado R, Tamayo L, Laterza AM, Brawn G , Lopez A. Phacomatosis pigmentovascularis: a new syndrome? Report of four cases. Pediatr dermatol 1987;4:189-96.
13. Al Robaee A, Banka N , Alfadley A. Phakomatosis pigmentovascularis type IIb associated with Sturge-Weber syndrome. Pediatr dermatol 2004;21:642-5.
14. Kim HJ, Park KB, Yang JM, Park SH , Lee ES. Congenital triangular alopecia in phakomatosis pigmentovascularis: report of 3 cases: Acta Derm Venereol. 2000 May;80(3):215-6.
15. Kim YC, Park HJ , Cinn YW. Phakomatosis pigmentovascularis type IIa with generalized vitiligo: Br J Dermatol. 2002 Nov;147(5):1028-9.
16. Du LC, Delaporte E, Catteau B, Destee A , Piette F. Phacomatosis pigmentovascularis type II. Eur J dermatol 1998;8:569-72.
17. Cho S, Choi JH, Sung KJ, Moon KC , Koh JK. Phakomatosis pigmentovascularis type IIB with neurologic abnormalities: Pediatr Dermatol. 2001 May-Jun;18(3):263.

18. Di Landro A, Tadini GL, Marchesi L , Cainelli T. Phakomatosis pigmentovascularis: A new case with renal angiomas and some considerations about the classification. Pediatr dermatol 1999;16:25-30.
19. Huang C , Lee P. Phakomatosis pigmentovascularis IIb with renal anomaly. Clin Exp dermatol 2000;25:51-4.
20. Park JG, Roh KY, Lee HJ, Ha SJ, Lee JY, Yun SS et al. Phakomatosis pigmentovascularis IIb with hypoplasia of the inferior vena cava and the right iliac and femoral veins causing recalcitrant stasis leg ulcers. J Am Acad Dermatol 2003;49:S167-9.
21. Gilliam AC, Ragge NK, Perez MI , Bolognia JL. Phakomatosis pigmentovascularis type IIb with iris mammillations. Arch dermatol 1993;129:340-2.
22. Finklea LB, Mohr MR, Warthan MM, Darrow DH , Williams JV. Two reports of phacomatosis pigmentovascularis type IIb, one in association with Sturge-Weber syndrome and Klippel-Trenaunay syndrome. Pediatr dermatol 2010;27:303-5.
23. Happle R. Loss of heterozygosity in human skin. J Am Acad Dermatol 1999;41:143-61.
24. Ono I , Tateshita T. Phacomatosis pigmentovascularis type IIa successfully treated with two types of laser therapy. Br J dermatol 2000;142:358-61.
25. Kono T, Ercocen AR, Chan HH, Kikuchi Y, Hori K, Uezono S et al. Treatment of phacomatosis pigmentovascularis: a combined multiple laser approach. Dermatol Surg 2003;29:642-6.
26. Kono T, Sakurai H, Groff WF, Chan HH, Takeuchi M, Yamaki T et al. Comparison study of a traditional pulsed dye laser versus a long-pulsed dye laser in the treatment of early childhood hemangiomas. Lasers Surg Med 2006;38:112-5.
27. Kagami S, Asahina A, Watanabe R, Mimura Y, Shirai A, Hattori N et al. Laser treatment of 26 Japanese patients with Mongolian spots. Dermatol Surg 2008;34:1689-94.
28. Mazereeuw-Hautier J, Carel-Caneppele S , Bonafe JL. Cutis marmorata telangiectatica congenita: report of two persistent cases. Pediatr dermatol 2002;19:506-9.