

Phacomatosis pigmentokeratotic without extracutaneous abnormalities: 12-year follow-up



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Key words: epidermal nevus syndrome; mosaic RASopathy; phacomatosis pigmentokeratotic.

INTRODUCTION

Phacomatosis pigmentokeratotic (PPK) is a rare variant of epidermal nevus syndrome characterized by the combination of a nevus sebaceus and papular speckled lentiginous nevus, usually with associated neurologic, musculoskeletal, and ophthalmologic abnormalities. The nevus sebaceus is arranged along the lines of Blaschko, and the papular speckled lentiginous nevus is typically arranged in a checkerboard pattern. We present a clinically extensive case of cutaneous PPK in a young boy without evidence of extracutaneous abnormalities after more than a decade of follow-up.

CASE REPORT

A 2-year-old white boy was referred to our dermatology department for yellow plaques on the face, chest, groin, and right extremities, as well as pigmented patches on the back. All lesions had been present since birth. He is a dizygotic twin, born at 37 weeks' gestation after in vitro fertilization and an uncomplicated pregnancy. His twin sister does not have any skin findings.

On initial physical examination, the patient was a healthy-appearing boy with growth and development appropriate for his age. He had multiple sebaceous nevi, including a 7-cm skin-colored, waxy, linear, verrucous plaque reaching from the mid-forehead to nasal tip (Fig 1); a similar 1-cm plaque inferior to the medial aspect of the right canthus; and a 7 × 10-cm skin-colored, waxy,

Abbreviation used:

PPK: phacomatosis pigmentokeratotic

alopecic plaque on the occipital aspect of the scalp extending to the right postauricular region. Streaky, blaschkoid, variably corrugated, yellow-brown plaques covered his right pectoral region, infiltrating his right axilla (Fig 2). The right groin had 3 linear plaques, 1 of which extended inferiorly and involved the scrotum. There were additional linear plaques arranged in blaschkoid distribution on the right extremities and across the right foot.

Examination also showed a large, light brown, well-demarcated patch with irregular borders, distributed circumferentially around the abdomen, generally respecting the midline (Fig 3). Numerous superimposed 2- to 4-mm dark brown nevoid papules were present, extending beyond the borders of the patch onto the groin, back, and abdomen. The remainder of the physical examination findings were normal. Extensive evaluation for systemic involvement, including head computed tomography, renal ultrasonography, ophthalmologic examination, and laboratory screenings, did not reveal extracutaneous manifestations.

At the present time, the patient is 14 years old and is followed closely by dermatology specialists. His congenital lesions have increased proportionally with his growth, and his development continues to

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Funding sources: None.

Conflicts of interest: None disclosed.

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JAAD Case Reports 2019;5:1055-7.

2352-5126

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<https://doi.org/10.1016/j.jidcr.2019.07.031>



Fig 1. Phacomatosis pigmentokeratotica without systemic involvement. Linear nevus sebaceus extending from the mid-forehead to nasal tip, with similar, though more hyperkeratotic, sebaceous nevus involvement over his knuckles.



Fig 2. Phacomatosis pigmentokeratotica without systemic involvement. Nevus sebaceus in streaky, blaschkoid arrangement involving the right chest and right axilla.

be normative, without ocular, neurologic, or musculoskeletal abnormalities. He has had numerous benign melanocytic lesions removed from the trunk,



Fig 3. Phacomatosis pigmentokeratotica without systemic involvement. Papular speckled lentiginous nevus distributed circumferentially around the abdomen, generally respecting the midline.

but no malignancies have been detected. Histopathology of several pigmented lesions excised from his flank showed junctional and dermal collections of spindled and epithelioid cells with cell maturation at the base, melanin incontinence, accompanying mild lymphocytic infiltrates, and well-formed Kamino bodies, features consistent with Spitz nevi. Biopsy samples from the inner thigh and neck showed hyperkeratosis, acanthosis, papillomatosis, and variable basilar hyperpigmentation consistent with epidermal nevi. Carbon dioxide laser has been performed twice under general anesthesia for the treatment of intertriginous epidermal nevi, with plans for an additional procedure in the coming year.

DISCUSSION

Epidermal nevus syndrome is an umbrella term used to describe syndromes characterized by an epidermal nevus occurring in conjunction with a variety of other cutaneous and extracutaneous abnormalities. The syndromes are subdivided into organoid nevus and keratinocytic nevus syndromes, depending on the variant of epidermal nevus. Phacomatosis pigmentokeratotica is classified as an organoid nevus syndrome, along with Schimmelpenning syndrome, nevus comedonicus syndrome, angora hair nevus syndrome, and Becker nevus syndrome.¹

Phacomatosis pigmentokeratotica is a mosaic RASopathy in which activating postzygotic mutations in *HRAS* or *KRAS* lead to the syndrome's characteristic cutaneous and extracutaneous findings.^{2,3} Variations in clinical phenotype and the

degree of extracutaneous involvement are thought to be a result of the timing of the mutation during embryogenesis.² The majority of patients with PPK have extracutaneous involvement, most commonly ocular (strabismus and ptosis), neurologic (hemiparesis, hyperhidrosis, dysesthesia), and musculoskeletal (scoliosis, muscular weakness) abnormalities.⁴ Approximately 30 cases of PPK have been reported in the literature, and fewer than 10 of these cases describe patients without extracutaneous involvement.⁵

We report this clinically extensive case of PPK with a 12-year follow-up to add to the body of literature on this exceedingly rare genodermatosis and to highlight an uncommon occurrence of PPK without extracutaneous involvement. Patients with cutaneous PPK require careful monitoring for malignant transformation of sebaceous and papular speckled lentiginous nevi into basal cell carcinoma and melanoma, respectively.^{6,7} As with other RASopathies, PPK has also been associated with nondermatologic malignancies including rhabdomyosarcomas and nephroblastoma (Wilms tumor); however, to our knowledge, no cases of associated internal malignancies in patients with PPK without extracutaneous involvement have been documented. Although no effective medical therapy yet exists for PPK, in our patient, carbon

dioxide laser has offered some benefit to select lesions. Beyond the physical toll of PPK on affected patients, the impact on quality of life by such an extensively visible condition must always be considered.

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