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SHORT COMMUNICATION

Phacomatosis Pigmentokeratotica: A Follow-up Report with Fatal Outcome

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Phacomatosis pigmentokeratotica (PPK) was first described in 1996 (1) as a distinct disease characterised by coexistence of a large sebaceous naevus and speckled lentiginous naevus of the papular type. It can be associated with musculoskeletal, neurological and ophthalmological abnormalities (2).

This is a follow-up report of a boy previously reported with grave extracutaneous abnormalities (3) who subsequently died of severe complications.

CASE REPORT

Six years ago, a premature boy was born at 34 weeks of gestation with multiple flesh-coloured verrucous plaques on his limbs, chest, back and both sides of the scalp and face along Blaschko's lines. The large and confluent patches and plaques gradually darkened and became raised with multiple papular speckles (Fig. 1). Two skin biopsies from the aforementioned lesions showed sebaceous naevus and melanocytic naevus, and PPK was diagnosed. After 5 years, these plaques had darkened and become more prominent, and papular speckles increased in number, with few fine vellus hair on his scalp (Fig. 2). He had multiple prematurity complications: retinal haemorrhage, neonatal jaundice and patent ductus arteriosus. Since 5 months of age, he suffered from recalcitrant status epilepticus; only valproic acid and nitrazepam were effective early in the course of epilepsy. Visual impairment was also noticed. An ophthalmological examination conducted by flash visual evoked potential showed normal latency but decreased amplitude over the occipital region, suggesting a bilateral retrochiasmatic problem. A brain echogram showed some ventriculomegaly and relative thinning of the corpus callosum. Mild right-sided hearing loss was confirmed by an audiobrainstem reflex test (ABR). He had severe musculoskeletal deformities, including underdeveloped teeth, scoliosis, low muscle power, and hypophosphataemia that was resistant to vitamin D treatment. The recalcitrant hypophosphataemia made him ricket-like in appearance. Significant retardation in growth and development had also been noted since he was 4 months old (3).

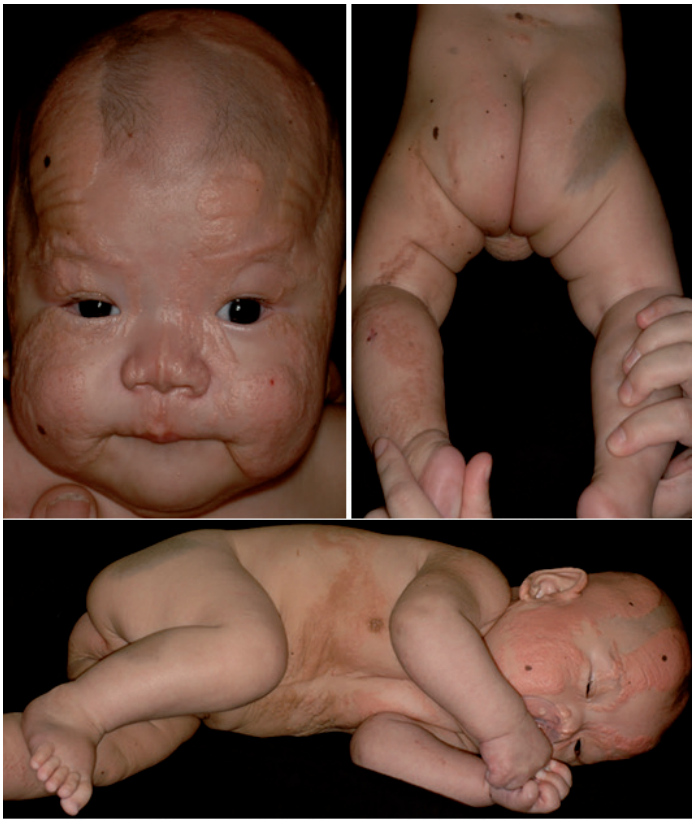

Fig. 1. The photographs were taken when the boy was 4 months old. Multiple flesh-coloured verrucous plaques are on his limbs, chest, back and both sides of the scalp and face along Blaschko's lines. The large and confluent patches and plaques gradually darkened and became raised with multiple papular speckles. Permission to publish this figure, from Dermatologica Sinica, is given.

Malignant tumours from the skin or internal organs were absent. However, he had been hospitalised frequently; he was malnourished since birth by poor digestive function and chronic diarrhoea. At the age of 2, biopsy of the stomach and duodenum indicated chronic inflammation and moderate villous atrophy. His teeth were underdeveloped with gingival enlargement over the right side of his mouth when he was 3 years old. At the age of 6, intractable hypoalbuminaemia (albumin, <3 g/dl), hypokalaemia (potassium, 2.5 mmol/l) and hypophosphataemia (phosphate, 1.3 mg/dl) could not be corrected by nasogastric tube feeding and necessitated total parenteral nutrition. Severe anaemia (haemoglobin, <9 g/dl) persisted, necessitating frequent blood transfusions to maintain cardiopulmonary function.

Subsequently, physical and mental retardation was noted. Since birth, his best consciousness level was E4V2M4 and it worsened during illness. He could not express meaningful words or engage in intellectual activities. Poor nutrition, muscular atrophy and hypotonia, scoliosis and vitamin D resistant hypophosphataemic rickets made him small in stature and susceptible to fracture. The hypophosphataemic rickets were similar to the presentation of previous reports (4, 5). He was mostly bedridden and could hardly turn his body. His physical strength and immunity were poor, and he frequently suffered from severe infection, including infectious colitis, gastroenteritis, pneumonia, urinary tract infection and anal abscess. Recurrent infections resulted in lung atelectasis and impaired lung function. At the age of 6, he needed oxygen supply by biphasic positive airway pressure. The intractable epilepsy was difficult to control even with valproic acid, topiramate and vigabatrin.

On last admission, after total parenteral nutrition and ventilator use, his condition deteriorated. He suffered from seizures, malnutrition (Hgb, 9.5 g/dl; albumin, 2.64 g/dl) and cough with dyspnoea (SpO_2, 80–90%). He experienced

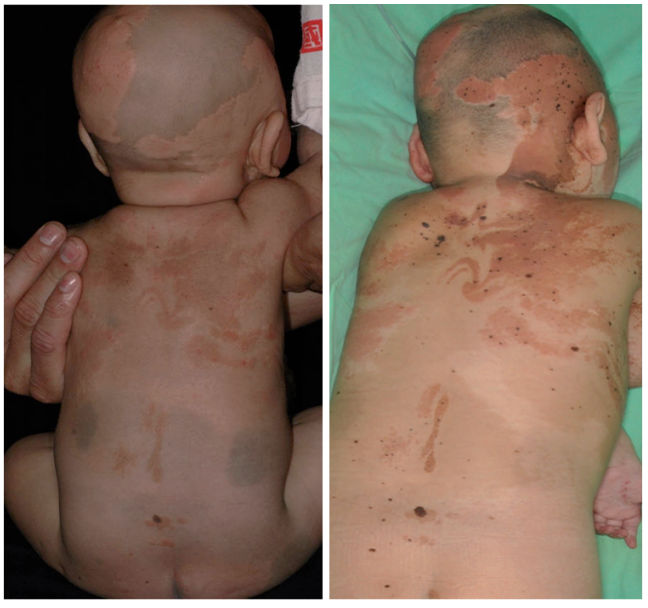

Fig. 2. After 5 years, the plaques had darkened and become more prominent, and papular speckles increased in number, with few fine vellus hair on his scalp. (Left: 4 months old; right: 5 years old).

fever, sepsis (WBC, 15.8 × 10^3/ul; with band form, 15%; C-reactive protein, 41.15 mg/dl) and respiratory acidosis (pH, 7.112; pCO2, 74.9 mmHg). Finally, he died from respiratory failure and cardiac arrest despite all therapeutic measures.

DISCUSSION

Approximately 30 cases of PPK have been reported (6, 7). The phenotype represents an admixture of Schimmelpenning syndrome and papular naevus spilus syndrome (2). The spectrum of extracutaneous anomalies in PPK represents variable features of the aforementioned syndromes (2). We conducted a literature review of PPK cases published during 1997–2012 using Pubmed (Table SI[1]). PPK usually combined with multiple variable anomalies; however, in some cases, no associated extracutaneous abnormalities were found (7). Our case was a male with skin lesions involving both sides. There is a higher incidence in males (approximately 2:1); lesions involve the left side of the trunk twice as often as the right side. Due to the rarity of PPK and limited medical reports, these findings could be incidental, and further studies are needed.

In this case, the patient had PPK complicated with multiple organ anomalies; this devastating outcome has been rare. We present this follow-up report for physicians to recognise this disease and its potential lethal outcome. Formerly, the potential for malignant changes in cutaneous lesions was overstressed. Here we emphasise that when encountering PPK patients with an extensive distribution, complete physical particularly neurologic, ocular and auditory examinations are warranted with electrolyte screening. Still, the potential of bad consequence of the evolution, such as malignant change of the skin lesions and certain tumours occur in other organs should be alert as well, especially in their later life.

The authors declare no conflicts of interest.

[1]https://doi.org/10.2340/00015555-1726

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