Case Report

A boy with poikiloderm: a possible case of Rothmund-Thomson syndrome

男童的皮膚異色症：疑似先天性皮膚異色症一例

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A two-year boy was noted to have erythema over face at three months old. Subsequently, the rash extended to the limbs. Poikiloderma was found later. Short stature and mild delay in speech development were also found. This is a possible case of Rothmund-Thomson syndrome.

Keywords: Poikiloderma, Rothmund-Thomson syndrome

Case report

A two-year-old Chinese boy was noted to have itchy eruption on face when he was three months old. The erythematous rash affected the cheeks initially. At age of seven months, the rash became more severe. Examination performed at age of 10 months showed post-inflammatory hyperpigmentation over both cheeks and ears and ill defined erythematous papules on both hands and at age of one, the lesions affected both limbs. His past history was unremarkable. There was no consanguinity in the family history and his 11 years old brother enjoyed good health with no abnormal skin features. He was initially treated with topical steroid for atopic dermatitis but no improvement was noted.

Examination performed when the boy was one year and eight months old showed reticular pigmentation over face and ears (Figure 1) and poikilodermatous changes on hands, forearms and shins (Figures 2 & 3). Biopsies of the lesions showed prominent interface change with basal vacuolar degeneration, pigmentary incontinence and epidermal atrophy (Figures 4 & 5) and thus compatible with the clinical findings.