Congenital poikiloderma is a dermatological condition characterized in the first few months of life by epidermal atrophy, telangiectasias, and variegated pigmentation (hypo- and hyperpigmentation). Poikiloderma often presents a diagnostic challenge in the first few months of life, with differential diagnoses such as Rothmund-Thomson syndrome (RTS), the eponymous Weary form of hereditary sclerosing poikiloderma, Clericuzio-type poikiloderma with neutropenia, and Kindler syndrome (1-3).

A distinct autosomal dominant form of hereditary fibrosing poikiloderma (HFP) was first described in a South African family in 2006 (3). HFP with tendon contractures, myopathy, and pulmonary fibrosis (abbreviated POIKTMP) is caused by mutations in FAM111B (Homo sapiens family with sequence similarity 111, member B), encoding a trypsin-like cysteine/serine peptidase (4, 5). FAM111B has also been implicated in susceptibility to prostate cancer (6). The main clinicopathological features of POIKTMP comprise early-onset poikiloderma, especially on the face and sun-exposed areas, alopecia, hypohidrosis with heat intolerance, growth retardation, multiple muscle contractures, in particular triceps surae muscle contractures, myopathy, and pulmonary fibrosis (abbreviated POIKTMP) is caused by mutations in FAM111B mis-
sense mutation.

CASE REPORT
We describe here a new case of POIKTMP in a 5-month-old Chinese boy. He was born of non-consanguineous Chinese parents, and had no similar illness history or familial history of atopy. He was the product of a full-term, uncomplicated pregnancy and delivery, with a birth-weight of 3.5 kg, with no immediate perinatal abnormalities.

After 6 days, however, erythema, with desquamation on the cheeks with a recurrent papulovesicular facial eruption was noted. Thereafter the clinical features comprised progressive generalized poikiloderma, telangiectasia, xerosis and innumerable hypo- and hyperpigmentary macules, measuring between 3 and 6 mm, predominantly on the face and in other sun-exposed areas; and worsening non-scarring alopecia affecting the scalp, eyebrows and eyelashes (Fig. 1a–d). The patient also had eczematous lesions on the legs and feet. No lymphoedema of the upper and/or lower extremities was observed.

A biopsy from the leg revealed conspicuous thickening of the spinous layer, blister formation under the epidermis, cellulose-like substance deposition and inflammatory cell infiltration in the blister with scattered eosinophils and neutrophils. In allergen detection, the patient was allergic to worm.

Other normal or negative findings included: weight and height within normal limit; full blood count; immunoglobulins; complement; anti-ds-DNA antibodies, anti-ANA antibodies, anti-CENPB antibodies; TPPA, RPR, anti-TP antibodies; no heat intolerance; no respiratory abnormalities; no liver impairment, no exocrine pancreatic insufficiency; no muscle weakness or wasting; no tendon abnormalities or joint contractures; no nail abnormalities; no cataracts; no recurrent gingivitis; no palmoplantar keratoderma.

Following informed consent, the genomic DNA of the proband was analysed using a gene probe consisting of 541 genetic loci of genodermatoses. This revealed a heterozygous missense mutation, c.A1873C in FAM111B, which results in the mutation p.Thr625Pro (Fig. 2a). No mutation was detected in his non-consanguineous parents (Fig. 2b, c). This mutation converts Thr625 to Pro and was not detected in his non-consanguineous parents (Fig. 2b, c). This mutation was also absent from the public database (NCBI, dbSNP135, the 1000 Genomes Project, and HapMap8) and the deleterious mutation in this patient. In summary, we

Fig. 1. Clinical features of hereditary fibrosing poikiloderma with tendon contractures, myopathy, and pulmonary fibrosis (POIKTMP) in a 5-month-old Chinese boy. (a–d) Aged 5 months, he exhibited generalized poikiloderma, and alopecia with a resolving papulovesicular eruption; eczema-like dermatosis was also present. (e) Haematoxylin and eosin (H&E)-stained sections of proband skin. Scale bar: 200 µm. Written permission to publish these photos are given.
manifest during childhood, and therefore the future clinical course of our patient is uncertain. Indeed, the clinical features of POIKTMP are somewhat variable (4).

In conclusion, we report here another case of POIKTMP, and expand our knowledge of this very rare entity. Further studies are needed to understand the function of FAM111B.

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