LETTER TO THE EDITOR

Genetic mutation in male patients with incontinentia pigmenti

Dear Sir,

Hsiao et al identified the NEMO gene mutations in 14 of 21 (66.7%) Chinese patients with incontinentia pigmenti,1 which is similar to the detection rates in the literature.2 However, Hsiao et al reported that no NEMO mutation was detected in the male patient with typical linear vesicles as a newborn, streaks of hypopigmentation at 2 months of age, typical vesicle histology and normal 46, XY karyotype.

We also had a 14-day-old, full-term baby (38 weeks of gestation) admitted to the National Taiwan University hospital due to multiple erosive skin rashes over the right anterior tibia area at birth and several vesicles developed over the same site in the following days. The karyotype of the newborn was 46, XY. The mother also had multiple hyperpigmentation patches over bilateral lower legs but she denied previous vesicles, ophthalmologic problems, dental problems or neurologic deficit. However, no NEMO mutation was detected by multiplex PCR and sequencing.

Ardelean et al reviewed the literature and only 5 out 42 (12%) male patients with incontinentia pigmenti had evidence of NEMO gene mutation.3 The three mechanisms for survival of males with incontinentia pigmenti were hypomorphic alleles, the 47, XXY karyotype (Klinefelter syndrome) and somatic mosaicism.4 Although somatic mosaicism is possible in these two affected males, there may be some other etiology or causative gene mutation of the incontinentia pigmenti.

References


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