We read with interest the paper by Ballmer-Weber et al. [1] reporting the case of a 16-month-old girl with hypomelanosis of Ito (HI) and genital hypertrichosis without signs of precocious puberty. The authors concluded that focal hypertrichosis may be another phenotypic expression of mosaicism. We had the opportunity to examine 2 patients who may shed some light on that matter.

Patient 1 was a 2,500-gram Emirati girl who was born following a full-term, uncomplicated pregnancy. She was admitted at birth because of absence of sucking and because of dysmorphic features that included microcephaly, large and low-set ears, saddle nose, epicanthic folds, strabismus, microstomia, thin lips, macroglossia, high arched palate, alopecia on the vertex and on the right temple, a right-sided additional nipple, webbing of the anterior fold of the left axilla, clinodactyly of the 5th fingers, a capillary haemangioma 3 cm in size on the left thigh and bilateral hip dislocation. A brain CT scan revealed asymmetry of the ventricles and dysgenesis of the corpus callosum. At 10 weeks she developed achromic patches and streaks disposed along the lines of Blaschko on the trunk, the limbs and the face; we noticed the growth of thick black hair along the spine and on the shoulders, the corners of the upper lip and the forehead. There were also long eyelashes. At the age of 3 years, she underwent surgery for a hepatic solitary cyst. She was profoundly retarded and presented with profuse hypertrichosis on the back, the extensor aspects of the arms, the buttocks, the posterior and external aspects of the lower limbs, an area under the clavicles, the genitals and the face (fig. 1, 2). Silky dark hair, up to 10 cm long over the spine and on the shoulders, the corners of the upper lip and the forehead. There were also long eyelashes. At the age of 3 years, she underwent surgery for a hepatic solitary cyst. She was profoundly retarded and presented with profuse hypertrichosis on the back, the extensor aspects of the arms, the buttocks, the posterior and external aspects of the lower limbs, an area under the clavicles, the genitals and the face (fig. 1, 2). Silky dark hair, up to 10 cm long over the spine, was disposed uniformly on the affected areas; it masked the underlying original pattern of hypo-pigmented streaks and whorls. The parents and 8 siblings were unaffected. The peripheral blood lymphocyte karyotype was normal. The parents refused any skin biopsy procedure on the child.

Patient 2, a girl from Syria, had walked at 21 months of age. At 8 years of age, she was slightly mentally retarded and presented with mild microcephaly, facial asymmetry with deviation of the tip of the nose toward the right, large ears, saddle nose, epicanthus, microstomia, thin lips, decayed teeth, dribbling, small 3rd and 5th toes, short 5th fingers and atactic gait concerning the
right hip. A brain CT scan revealed a prominent right ventricle and partial absence of the corpus callosum. All over the skin, including the lower face, there were hypopigmented patches and streaks arranged along the lines of Blaschko. There was also marked hypertrichosis on the sides of the cheeks and on the back, arms and thighs. Interestingly, hair spread mostly on the whorls and streaks of normally pigmented skin (fig. 3, 4). The rest of the skin, including the genitals, was free of terminal hair. For technical reasons, brain MRI and fibroblast karyo-
Fig. 1. Whorled hypopigmentation, additional nipple and sub-clavicular hair of patient 1. Note the right abdominal scar from surgery for a hepatic solitary cyst.

Fig. 2. Generalized hypertrichosis on the back of patient 1; white streaks are still visible on the side of the chest.

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Fig. 3. Whorled hypertrichosis on pigmented normal skin of patient 2.
Fig. 4. Close-up of the back of patient 2, in whom hypertrichosis follows hyperpigmented whorls.

Types were not performed. The parents and 2 other siblings were normal. Both patients met the diagnostic criteria of HI [2]. HI, however, is no longer taken as an entity but as a cutaneous sign of various forms of mosaicism. HI is a term that is descriptive and non-specific [3], and in many instances patients may show hypopigmentation following the lines of Blaschko without associated abnormal features [4].

Both patients also showed profuse hypertrichosis that was unlikely to be related to HI: in patient 1, despite the presence of genital hair without clinical signs of precocious puberty, hypertrichosis involved equally hypo- and normally pigmented skin. Conversely, in patient 2, hypertrichosis was restricted to streaks and whorls of normally pigmented skin. We can assume that in the latter the process responsible for hypopigmentation was also capable of altering the associated hypotrichosis.

This shows that, in some patients, HI can exert a negative effect on hair growth.

References
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Letters to Dermatology