Linear and whorled nevoid hypermelanosis (LWNH) is a rare sporadic disorder, first described by Kalter et al. (1) in 1988 and characterized by hyperpigmented, reticulated, streaky and whorled macules, in the Blaschko’s lines, without atrophy or preceding inflammation. With onset commonly occurring in the first weeks of life, the hyperpigmentation tends to persist throughout life. This is thought to reflect an underlying mosaicism or chimerism, and several cyto genetic alterations have been described. Occasionally, it is associated with systemic abnormalities, including developmental and growth retardation, facial and body asymmetry, and cardiac defects (2, 3). Histology shows hyperpigmentation of the basal epidermal layer and prominent melanocytes, without pigment incontinence.

Axenfeld-Rieger anomaly is a rare autosomal dominant disorder that involves anterior eye structures derived from the periocular mesenchyma (4). There are sporadic cases associated with alcohol or isotretinoin consumption during pregnancy (5). It includes abnormalities of the iridocorneal angle, and is often associated with extraocular disorders (6). In the skin, it can be associated with redundant periumbilical skin and oculocutaneous albinism (7).

**CASE REPORT**

A 35-year-old woman with bilateral neurosensorial hypoacusia (of acute onset at 9 years of age), hypermobility joints, scoliosis (due to a shorter left leg), and amblyopia when she was a child. but neither cardiac disease nor mental retardation. When she was 5 years of age, hyperpigmented asymptomatic macules appeared on her trunk and limbs. These macules had enlarged with body growth until she was 16 years old. The macules had never undergone further modifications. The lesions had never been erythematous or bullous.

Skin examination showed bilateral, symmetrical, whorly and linear bands of mild brown hyperpigmentation involving the upper trunk and the limbs, as well as brown patches in the lumbar zone, sparing the face, palms and soles. In some areas the hyperpigmentation followed a Blaschkooid distribution (Fig. 1). Hair, nails, teeth and maxillar bones were all otherwise normal.

The patient’s mother denied alcohol or other toxic intake during pregnancy. There were several neurological disorders in the father’s family, but the details are lost for follow-up. There was no family history of pigmentary or metabolic disorders or consanguinity. Skin biopsy showed a melanocytic lentiginous pattern, without pigmentary incontinence.

Chromosomal analysis of dermal fibroblasts obtained from biopsies of normal and hyperpigmented skin were performed and revealed a normal karyotype. Fluorescence in situ hybridization was performed in the sample obtained from the hyperpigmented macule in order to detect trisomies of chromosomes 7, 13, 14 and 18. No evidence of these chromosomal alterations was detected.

Ophthalmological examination revealed posterior embryotoxon and a synechiae in the anterior chamber. Optic nerve, lens and intraocular pressure were within normal limits. An ophthalmologist considered the alterations to be an Axenfeld-Rieger anomaly.

**DISCUSSION**

In the last few years there has been increasing evidence that hypo- and hyper-pigmentation along the lines of Blaschko may be the result of an underlying mosaicism or chimerism. Furthermore, Blaschko’s linear skin pattern is thought to reflect the dorsoventral outgrowth of these different cell clones, originated from precursors from the neural crest during embryogenesis (8). There are plenty of genes regulating skin pigmentation, several mechanisms capable of changing the phenotypic expression of melanocytes as well as a wide spectrum of pigmentary alterations (9). All these facts, including cases of coexistence of LWNH and Ito hypomelanosis, and of cutis tricolor, led some authors to create the term “pigmentary mosaicism” to include all of them (10, 11). There are also more pigmentary patterns described in patients with pigmentary mosaicism, such as phylloid...
pattern, patchy pigmentation without midline separation and checkboard pattern (12). Phylloid pattern is characterized by round or oval spots and leaf-like macules. Phylloid hypomelanosis is a clinicogenetic entity related to chromosome 13 (13). Several recently published cases of phylloid hypermelanosis have been reported, one of them related to 5p tetrasomy (14, 15). A degree of pattern overlap can also occur (9), as in our case, with some lesions similar to phylloid pattern and others fitting into Blaschko’s lines.

In LWNH, underlying chromosomal mosaicism and cytogenetic changes have been observed, although in only a few cases (9). Chromosomal abnormalities most frequently observed include trisomy 7(16), 14, 18(17), 20 and X-chromosomal mosaicism (2). Interestingly, all reported patients with LWNH and cytogenetic aberrations had also systemic abnormalities. We could not find any such cytogenetic anomalies in our patient. Furthermore, several ocular abnormalities have been described in patients with pigmentary mosaicism, including coloboma, microphthalmia, abnormalities in retinal pigmentary pattern and optic atrophy (18). However, Axenfeld-Rieger anomaly has not previously been associated with LWNH. The common origin of melanocyte and iridocorneal cells in neural crest can help to explain this association. Thus, we conclude that ophthalmological examination should be included in the diagnostic test for any pigmentary mosaicism, including LWNH.

ACKNOWLEDGEMENT
The authors would like to thank Dr Torrelo for his useful comments on the case.

REFERENCES