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**Abstract**

Pigmentary mosaicism refers to hyper or hypopigmentation due to genetic heterogeneity of cells. Pigmentary mosaicism may follow any of the patterns of cutaneous mosaicism and persist through life. Diagnosis is clinical. Associated extracutaneous manifestations have been reported in as much as 30 % of the cases.

**Keywords**

Pigmentary mosaicism • Blaschko lines • Linear and whorled nevoid hypermelanosis

**Introduction**

- **Pigmentary mosaicism refers to hyper- or hypopigmentation due to genetic heterogeneity of cells.**
- **It has been referred to by many different names such as linear and whorled nevoid hyperpigmentation, segmental pigmentary disorder, nevoid hypermelanosis, or patterned pigmentation.**

Pigmentary mosaicism is a term that has been used to describe variegate or irregular hyperpigmentation or hypopigmentation due to genetic heterogeneity of the cells that compose the skin. In this chapter we are going to focus on mosaic hyperpigmentation only. A mosaic state is the proposed pathogenetic mechanism of this type of hyperpigmentation although it has not been probed in all cases. Therefore some authors prefer the term patterned hyperpigmentation [1]. Pigmentary mosaicism may present conforming to the different patterns of cutaneous mosaicism as described by Happle [2].

**Epidemiology**

- **The exact incidence in dark-skinned individual is not known**

There are no exact figures regarding the frequency of pigmentary hypermelanosis in dark skin. For any race the frequency also depends on the pattern. Phylloid hypermelanosis is the rarer variant with only a few cases published [3, 4].

Pigmentary mosaicism is usually not inherited although a few cases with several affected members in the same family point to epigenetic mosaicism (autosomal monoallelic expression) to explain the familial occurrence [5–7].

**Clinical Features**

- **Pigmentary mosaicism may follow any of the patterns of cutaneous mosaicism**
- **May not be apparent at birth and increase for a few years until it stabilizes.**
- **Many different extracutaneous abnormalities (CNS, ocular, cardiac, skeletal) have been described and investigation should be directed by clinical findings**

Pigmentary mosaicism manifests as hyperpigmented macules and patches that follow different patterns. They are usually present at birth although they may not become apparent until sun exposure during the first summer and may increase in number and size during the first years of life. The color may

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vary from light to dark brown. The patterns that have been described are linear and whorled hypermelanosis in which hyperpigmented, reticulated, streaky, narrow, and whorled macules occur along Blaschko lines (Fig. 8.1); broad band of hyperpigmentation along Blaschko lines; patchy hyperpigmented irregularly shaped macules without midline separation; checkerboard pattern, with alternating squares of



**Fig. 8.1** Linear and whorled nevoid hypermelanosis. A form of pigmentary mosaicism that follows the lines of Blaschko

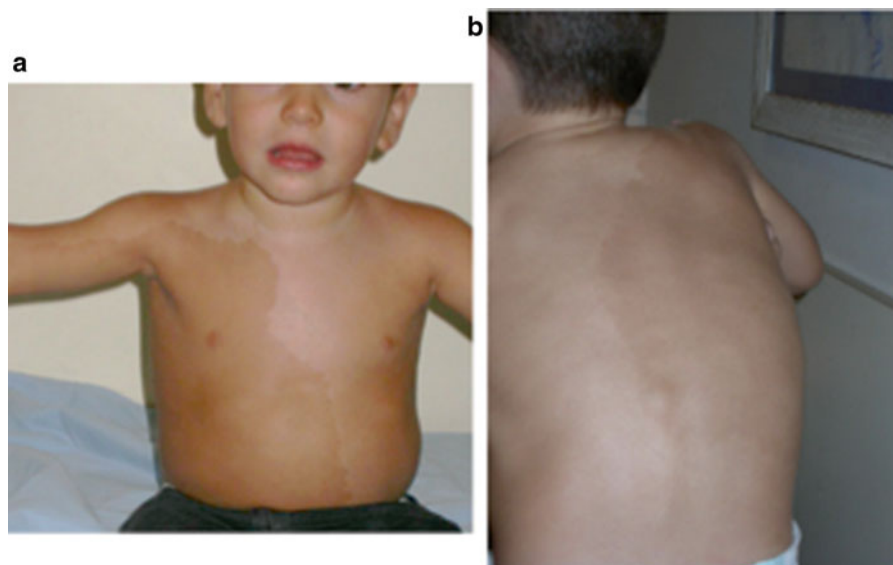
pigmentary anomalies with a sharp midline separation and sometimes with a flag-like distribution (Fig. 8.2a, b) [8]; and lateralization pattern in which hyperpigmented macules affect only one side with midline separation (Table 8.1) [2, 9].

Associated extracutaneous manifestations have been reported in as much as 30 % of the cases, although this figure may reflect a reporting bias [10]. The associated findings include developmental delay, autism, seizures, microcephaly, and ocular, cardiac (ventricular septal defects and tetralogy of Fallot), and skeletal abnormalities (digit abnormalities, hemiatrophy).

The associated anomalies are so diverse that clinical investigation should be directed by clinical findings.

### Pathogenesis

An enormous range of cytogenetic abnormalities has been reported in PM, including polyploidy, aneuploidy, and chromosomal deletions, insertions, and translocations. Mosaic trisomy 20, 7, 14, 18, and X-chromosomal have been reported in linear and whorled nevoid hypermelanosis [11–13]. The cytogenetic abnormalities may be only be found in fibroblast of affected skin. There is no single genotype for each phenotypic expression of pigmentary mosaicism except may be for phylloid hypermelanosis that has been more consistently related to chromosome 13 aberrations or mosaic chromosome 5p tetrasomy [4, 14] In most cases of pigmentary mosaicism a cytogenetic alteration cannot be detected and several hypotheses have been proposed to explain the abnormal pigmentation, such as comigration of genetically different cell populations, X-chromosome functional disomy,



**Fig. 8.2** Pigmentary mosaicism in a lateralization pattern. (a) Front view and (b) back view

**Table 8.1** Patterns of pigmentary mosaicism

Patterns of pigmentary mosaicism	Clinical features
Linear and whorled hypermelanosis	Reticulated, streaky, narrow and whorled macules occur along Blaschko lines
Broad band Hyperpigmentation	Broad band of hyperpigmentation that follow Blaschko lines
Patchy hyperpigmentation	Irregularly shaped macules without midline separation
Checkerboard pattern	Alternating squares of pigmentary anomalies with a sharp midline separation or arranged in a flag-like pattern
Lateralization pattern	Hyperpigmented macules affect only one side with midline separation

“Spreading” of X inactivation to autosomes, transposons, genetic imprinting, and phenotypic reversion [15].

Histology usually shows hyperpigmentation of the basal keratinocytes with normal number of melanocytes.

## Treatment

- **No effective treatment has been described**

There is no effective treatment for pigmentary mosaicism.

## Prognosis

The hyperpigmented macules persist throughout life. The prognosis is marked by the associated extracutaneous findings.

## Conclusion

Pigmentary mosaicism is a common usually sporadic disorder. Many different names have been used to describe it due to the fact that clinically it may manifest in any of the five patterns of cutaneous mosaicism described by Happle [9].

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