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Case Report

Pigmentary mosaicism: A report of five cases from a tertiary care hospital in South India

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Abstract Pigmentary mosaicism is a term that describes various patterns of segmental pigmentation disorders based on genetic heterogenicity of skin cells. Pigmentary mosaicism is a collective term, which comprises phenotypes represented by mosaic hypo- and/ or hyperpigmentation in the form of whorls, patchy, streaks or more bizarre skin configurations. Melanocyte precursors are neural crest derived and migrate through the Dorso-lateral pathway during embryogenesis. This group of disorders may be associated with or without systemic abnormalities. In a considerable number of cases, pigmentary mosaicism is observed alongside extracutaneous oddities generally affecting the musculoskeletal system and central nervous system. Pigmentary mosaicism includes a variety of inherited, patterned pigmentary dermatoses which have been described using various terms such as hypo melanosis of Ito, linear and whorled nevoid hypermelanosis, phylloid hypo- and hypermelanoses and pigmentary mosaicism of both hypopigmented and hyperpigmented type. Here, we present a series of 5 cases of pigmentary mosaicism in a single report.

Key words
Hypopigmentation; Hyperpigmentation; Lines of Blaschko; Pigmentary mosaicism.

Introduction

Mosaicism refers to the occurrence of two or more cell populations with different expression of one or more genes, inspite being derived from a single zygote.[1] Pigmentary mosaicism is a spectrum of disorders characterized by dyspigmentation along the lines of blaschko and other patterns such as phylloid, checkerboard and patchy pigmentation without midline separation.[2] This group of disorders may be associated with or without systemic abnormalities.

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Pigmentary mosaicism includes a variety of inherited, patterned pigmentary dermatoses which have been described using various terms such as hypomelanoses of Ito, linear and whorled nevoid hypermelanosis, phylloid hypo- and hypermelanosis and pigmentary mosaicism of both hypopigmented and hyperpigmented type.[3] The lesions may occur in the same individual in any combination.

The seven archetypical patterns include, *Type 1a:* Narrow bands, e.g. observed in hypomelanosis of Ito. *Type 1b:* Broad bands, e.g. observed in McCune Albright Syndrome, *Type 2:* Checkerboard pattern (also called flag-like) characterized by alternating squares of hyperpigmentation with a sharp midline separation, *Type 3:* Phylloid pattern with leaf-like or oblong macules showing a dorsal and ventral midline separation, *Type 4:* Patchy pattern without midline separation, e.g. observed

in cases of giant melanocytic nevi that do not respect the midline, *Type 5:* Lateralization pattern often observed in CHILD Syndrome, *Type 6:* Sash-like pattern.[4]

Here we present a rare series of 5 cases of pigmentary mosaicism belonging to type 1A, Type 1B, Type 3, Type 4 and Type 5 respectively.

Case reports

Case 1 A 27-year-old girl, presented with complaints of dark brown coloured skin lesions localised over upper limbs, mainly over right side. She had no family history of similar skin lesions. These lesions were present since birth and not preceded by any other dermatosis. On physical examination, she had hyperpigmented macules linearly arranged on the extensor aspect of right upper limb along the Blaschko's lines. There was no systemic involvement including limb length discrepancies. This illustrates Type 1A pigmentary mosaicism (thin bands along Blaschko's lines)

Case 2: A 23-year-old boy, presented with complaints of asymmetrical brown coloured spots all over his body, not associated with pruritus, pain or burning sensation and present since birth. There was no history of any pre-existing lesions prior to the pigmentation. Both his parents were healthy without any skin anomalies.

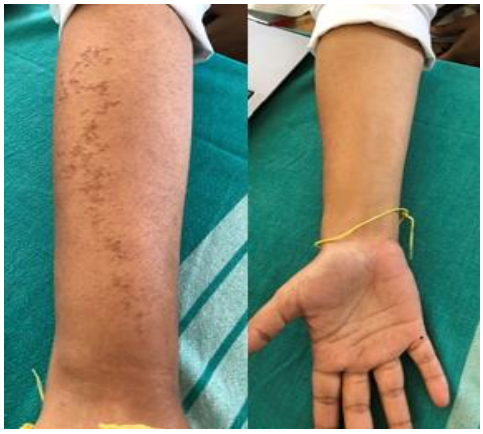

Figure 1 Hyperpigmented macules presenting as spots and thin lines along Blaschko's lines (type 1A).

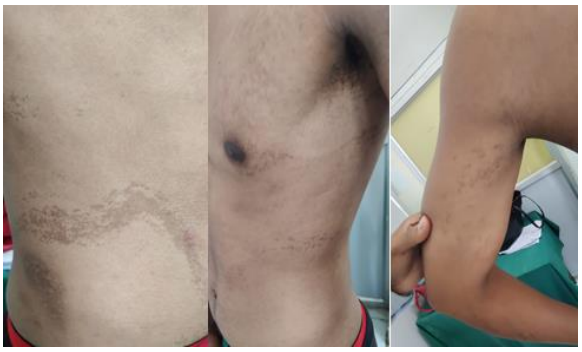

Figure 2 Hyperpigmented macules in streaks and thick bands distributed along Blaschko's lines (type 1B).

On examination, hyperpigmented macules arranged in bands and thin streaks along the Blaschko's lines were observed forming S-shape, mainly in the abdomen, chest and back (specially marked over the right side of body). There were no ophthalmological, hemiface or spine abnormalities, no cognitive delay or history of epilepsy. This is an example of Type 1B pigmentary mosaicism (thick bands along Blaschko's lines).

Case 3: An 8-year-old girl, presented with complaints of dark brown to black coloured flat lesions over her entire body since birth, predominantly over left side of chest, abdomen and back. There was no history of any preceding skin lesions. There were no gait abnormalities, vision complaints or cognitive dysfunction. There was no family history of similar skin lesions. On examination, she had distinctive hyperpigmented macules, most of which had blotchy and leaf like patterns in the trunk, predominantly on left upper limb, thorax, abdomen and back. Linear and curved patterns were also observed along the Blaschko's lines. There was no scaling or ichthyotic lesions. Ophthalmological examination was normal. There were no facial or skeletal abnormalities. Owing to the leaf like hyperpigmented macules, this represents Type 3 pigmentary mosaicism (phylloid type).

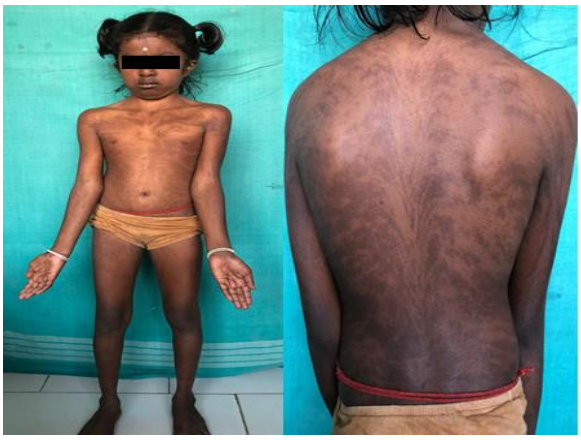

Figure 3 Hyperpigmented macules presenting with leaf-like shapes in upper chest and back. Thick hyperpigmented bands along Blaschko's lines are also observed in lower back, abdomen, and limbs. The lesions have a left-sided predominance (type 3).

Case 4: A 2-year-old boy presented with complaints of a large black raised skin lesion on his lower back and both buttocks since birth. The skin lesion proportionately increased in size with body growth and was associated with increased hair growth and satellite lesions. There was no family history of similar lesions. On examination, he had a large well-defined hyperpigmented black-coloured plaque without midline separation with size more than 20 cm present over lower back and both gluteal regions. Nodularity was noted in the inter-gluteal area. Hypertrichosis was present over the plaque. There were no gait or skeletal deformities. Biopsy from the nodular area revealed features of congenital melanocytic nevus without any atypical or malignant features. Owing to the midline nature of the lesions, this is considered to be Type 4 pigmentary mosaicism (patchy pattern without midline separation).

Case 5: A 4-month-old male baby was brought by his parents with complaints of dark brown coloured flat skin lesions present on his upper parts of the body since birth. The lesions were present only over his left neck, chest, shoulder, arm and back, sparing the right side. There was no history of similar skin lesions in the family. On examination, he had well-defined hyperpigmented brown coloured macules present over upper parts of the body involving the left neck, upper limb, thorax, and back. The macules were present as spots and blotches, with well-defined coast-like margins. There were no lesions anywhere else in the body. There were no obvious skeletal deformities. Owing to the unilateral distribution of hyperpigmentation, this represents Type 5 pigmentary mosaicism (lateralisation pattern).

Discussion

Pigmentary mosaicism is a collective term, which includes phenotypes characterized by mosaic hypo- and/ or hyperpigmentation in the form of streaks, whorls, patchy, or more bizarre skin configurations. Melanocyte precursors are neural crest derived and migrate through the dorso-lateral pathway during embryogenesis. Pigmentary mosaics are thought to illustrate normal or abnormal pathways of melanocyte

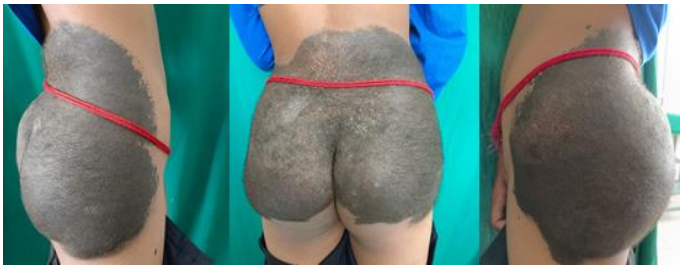

Figure 4 Well-defined black coloured plaque with hypertrichosis involving lower back and buttocks with few satellite lesions (type 4).

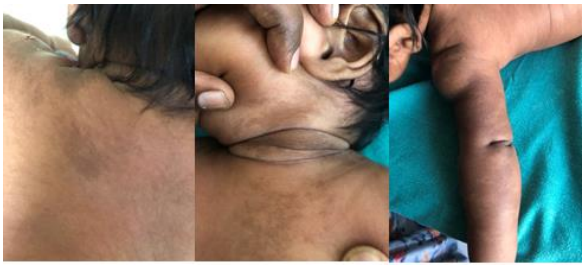

Figure 5 Well-defined hyperpigmented macules with irregular coast-like margins seen in left side of neck, chest, back shoulder and left upper limb (type 5).

precursor migration. Those presenting along Blaschko's lines do not have any genetic abnormalities affecting melanocytes, hence are thought to represent normal pathway of melanocyte migration. Whereas those patterns which do not conform to Blaschko's lines (i.e. phylloid, patchy without midline separation) harbour genetic abnormalities affecting melanocytes and thus represent abnormal melanocyte precursor migration.[5] Generally, the extent of cutaneous mosaicism is greater when the genetic alteration occurs earlier in development and extracutaneous involvement is more likely when it occurs before organogenesis.[6]

A review of literature for pigmentary mosaicism was conducted by Kromann and his colleagues in 2018. They found a combination of hyperpigmentation and hypopigmentation in 7% of a total of 651 published patients.[7] A study by Labadia *et al*; describes the largest series of pigmentary mosaicism, worldwide till date showed hypopigmentation in 19 patients, hyperpigmentation in 43 and combined patterns in 38. Frequent patterns of pigmentation observed in this study was fine and whorled in 95 patients, followed by broad pattern 5 patients.

We have presented here, 5 types of pigmentary mosaicism together in a single report which to the best of our knowledge is the first or its kind in India.

Conclusion

A vigilant approach and careful monitoring of patients with pigmentary mosaicism helps in early management and preventing complications.

Limitation: Standard chromosome analysis of a large number of metaphases or preferable chromosome micro array of uncultured cells could have added more to the literature.

Declaration of patient consent The authors certify that they have obtained all appropriate patient consent.

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Author's contribution

RT, JED: Identification and management of the case, manuscript writing, final approval of the version to be published.

DK, DM, GJ, SKM: Manuscript writing, final approval of the version to be published.

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