Pigmentary Mosaicism in Two Infants: Phylloid Hypermelanosis and Linear and Whorled Nevoid Hyperpigmentation

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INTRODUCTION

Pigmentary mosaicism is a term used to describe irregular or variegate hyper or *hypopigmentation* of the skin, due to the genetic heterogeneity of the cells that compose the skin. The more common variant is the *hypomelanosis* variant with the well-described prototype being *hypomelanosis* of Ito.

The true incidence is unknown; however, there are several case reports of the condition in the literature, many of which are associated with extracutaneous manifestations. We report two cases of congenital disorder of skin pigmentation in two male patients seen by the paediatric dermatology unit of the University of Ilorin Teaching Hospital.

CASE REPORT

Two male infants aged 0 – 6 weeks were seen by the paediatric dermatology unit on account of bizarre skin colour noticed at birth.

The first child was delivered via emergency caesarean section to a 43-year-old (P3+0) woman because of foetal macrosomia and breech position. The child had poor Apgar scores at birth and was admitted to the neonatal unit as a case of perinatal asphyxia. Bizarre skin colour was noticed at admission, described as leaf-like hyperpigmented patches and involving the whole body. (Figure 1) There were several extracutaneous abnormalities which included low-set ears, ocular and nipple hypertelorism, clinodactyly, polydactyly, syndactyly, high arched palate and microglossia, suggesting the possibility of underlying Trisomy 7. There was a positive history of similar skin discoloration in the mother; however, this involved only the upper back. Trans fontanelle ultrasounds scan revealed intracranial calcifications in the posterior fossa. An electrocardiogram also revealed right axis deviation. A diagnosis of phylloid hypermelanosis was made. The child was successfully managed and discharged by the neonatal care team but has since been lost to follow-up.

The second child was seen at 6 weeks of life as an outpatient with a complaint of abnormal skin pigmentation which was noticed at birth. However, skin pigmentation consisted of whorls and streaks of hyperpigmented macules and patches following the lines of blaschko.(figure 2) Skin discoloration involved the trunk mainly. There was no history of preceding lesions; lesions had not increased since it was first noticed. There was no family history of the similar disorder. The child also had stridulent breathing that was noticed since birth. No other extracutaneous anomaly was found on examination. Transfontanelle Ultrasound scan done was normal. A diagnosis of Linear and Whorled Nevoid Hypermelanosis (LWNH) was made. Parents were counselled and child is presently on follow-up care.

DISCUSSION

A mosaic is an organism composed of two or more genetically distinct cell populations derived from a genetically homogeneous zygote. Mosaicism can be

expressed in the skin as varying patterns of pigmentation. They are characterised by cutaneous manifestations that are flat, hypo or hyperpigmented lesions. Mosaic Phenotypes can be classified into two major categories according to the underlying genetic mechanism. Functional Mosaicism may result from the Lyon effect of X inactivation and such mosaic phenotypes can be transmitted from a mother to her daughter. By contrast, genomic mosaicism is caused by autosomal mutations and can usually not be inherited. In the 2 cases presented above, both patients were male, and although there was a history of abnormal pigmentation in the mother of the first child, both were apparently the result of genomic mosaicism. The true incidence of pigmentary mosaicism is unknown, there are however pockets of case reports/series in the literature. In 2003, a case report of a 14-year-old girl with focal dermal hypoplasia by Ogunbiyi *et al* described skin pigment abnormalities along the lines of blaschko, although this was not classically labelled a pigmentary mosaicism.

The pathogenesis of pigmentary mosaicism is mainly somatic, with post-zygotic mutation. According to most workers, about one-third of clinically diagnosed cases of pigmentary mosaicism demonstrate chromosomal mosaicism. An enormous range of cytogenetic abnormalities has also been reported in pigmentary mosaicism, including polyploidy, aneuploidy, chromosomal deletions, insertions and translocations. The first child presented in this report had dysmorphic features that could be associated with Trisomy 7, although this was not confirmed with karyotyping due to financial constraints.

Also, up to 30% of them have extracutaneous abnormalities which may involve the brain, the eyes, the musculoskeletal system or the cardiovascular system. Other associations include developmental delays, autism and seizures. Most cases are sporadic with few reports of familial occurrence, although familial occurrence has been reported in the literature as was seen in one of the patients we managed.

There are different clinical patterns described by Happle which include (figure 3):

Type 1 - Most common, follows the lines of blaschko, includes the LWNH

- Type 1a - Narrow bands e.g incontinentia pigmenti
- Type 1b - Broad bands e.g McCune-Albright syndrome

Type 2 - Checkerboard pattern, alternating squares of pigmentary change

Type 3 – Phylloid pattern, arranged in leaf-like or ornamental pattern, Rarer

Type 4 - patchy pattern without midline separation e.g Giant melanocytic naevi

The phylloid hypermelanotic pattern seen in the first case has been less described. Happle *et al* in a case series of 5 patients found extracutaneous abnormalities in all of them. He described certain features such as dysmorphic facie, deafness, mental deficiency as fairly consistent among the reported cases. He went on to postulate that phylloid hypermelanosis may actually be a cutaneous marker of different disorders.

The linear and whorled nevoid hypermelanotic variant is the more common type of pigmentary mosaicism. The second patient had the narrow band subtype. The association of extracutaneous anomalies has not been consistent in this variant. A retrospective study of 16 cases by Vito di Lernia *et al* reported extracutaneous anomalies in only one of the patient. There are however other studies in which other congenital anomalies were identified.

Lesions may be apparent at birth, notably in darker skin, however, it may develop in the first few weeks of life subsequently increasing in size and spread in the first 2 years before stabilising.

Diagnosis is mainly clinical. Histopathology sometimes shows increased pigmentation of the basal layer with prominence of the melanocytes

Management is multidisciplinary and depends on underlying co-morbidities. Psychological support should be given to the patient and the family.

CONCLUSION

Phylloid and LWNH are rare presentations of hyperpigmented mosaicism with pockets of case reports all over the world. They are often associated

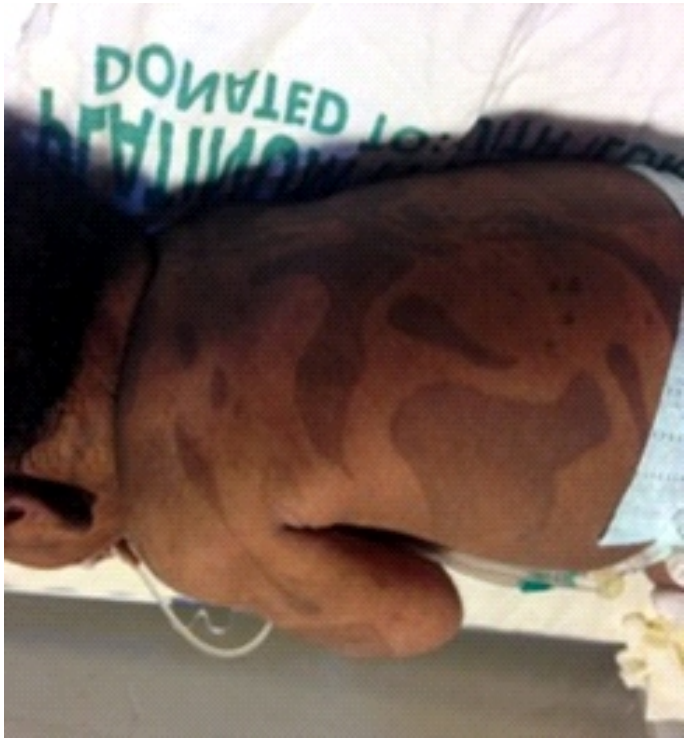

Figure 1: Case seen at birth

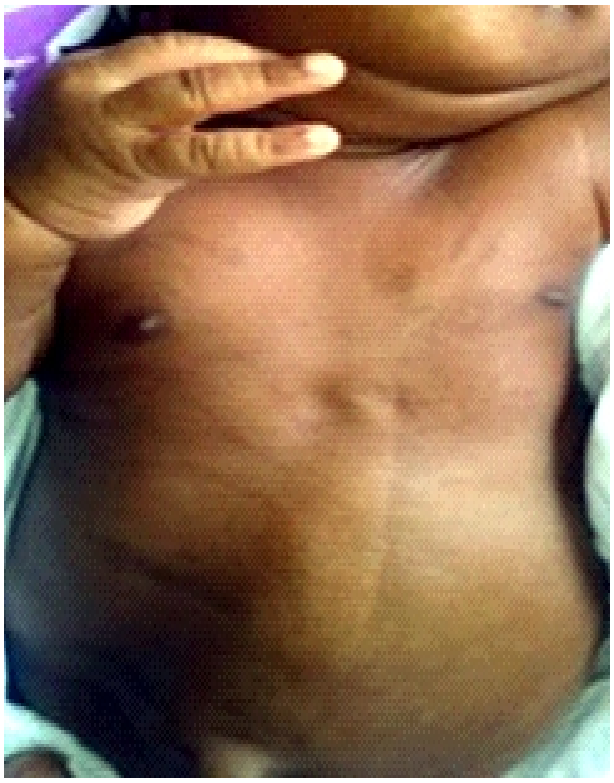

Figure 2: Case seen at age six weeks

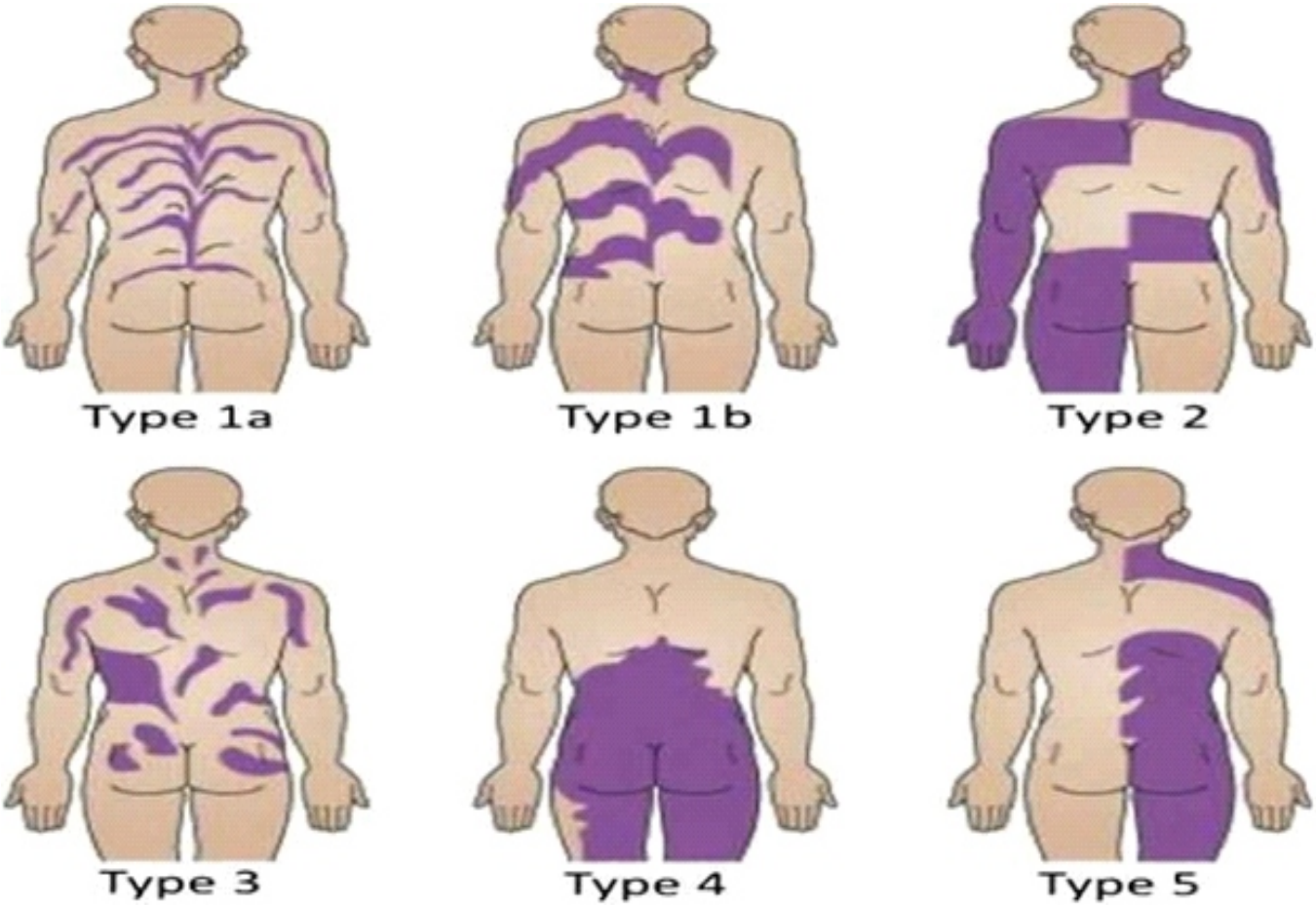

Figure 3

with variable co-morbidities which should be looked for and managed accordingly. In the absence of a definitive treatment, affected children and parents require lots of psychological support.

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