ORIGINAL RESEARCH PAPER

Medical Science

KEY WORDS:

A CASE SERIES OF PHYLLOID HYPOMELANOSIS

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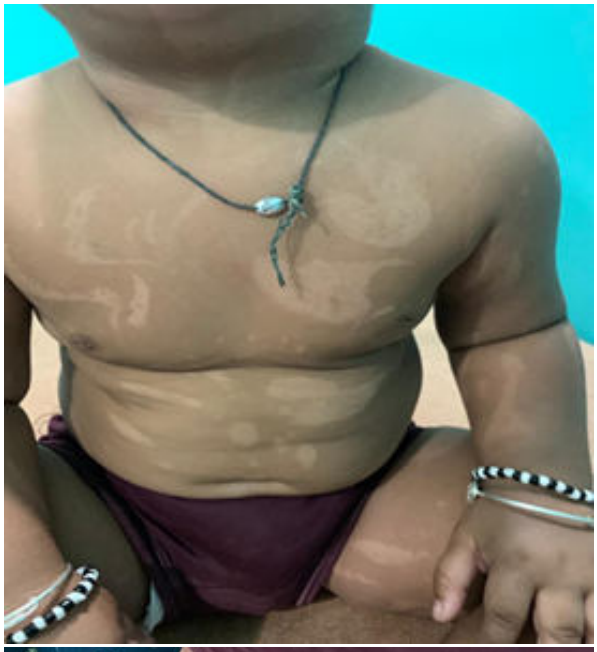
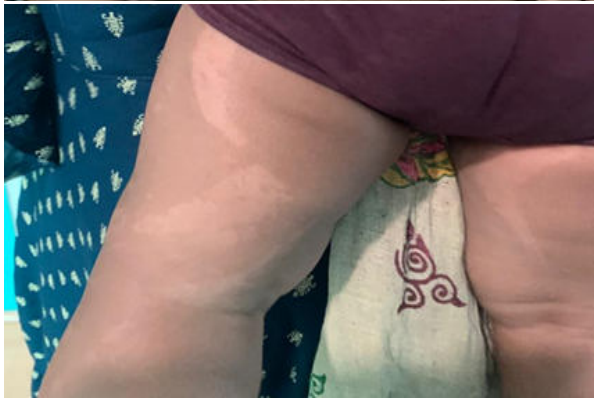
Sir

THE PHYLLOID HYPOMELANOSIS is a defined as a distinct syndrome consisting of achromic phylloid skin lesions in combination with extracutaneous anomalies such as mental retardation , agenesis of corpus callosum , conductive hearing loss , coloboma and various skeletal defects[1].

Herein, we report two cases of phylloid hypomelanosis

Case 1





A 6 months old female baby presented with chief complaints of multiple hypopigmented patches shortly after birth (within 2 to 3 weeks after birth).

On examination there were multiple hypopigmented leaf like , linear S shaped and oval macules and patches over chest , abdomen and extremities .

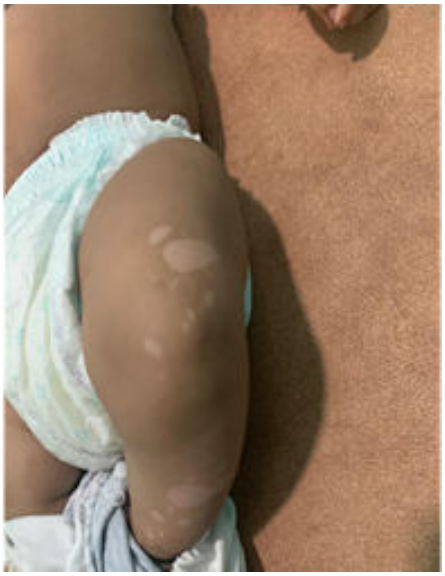
Size of the lesions were , smallest one being 2 x 2 mm and largest one being 5 x 6 cm .

Margins of the lesions were feathery and illdefined .

Skin overlying the lesion was normal .

In contrast to literatures published in the past there was no history suggestive of any neurological , ophthalmological and skeletal abnormalities.

Case 2



A 8 Months old female baby presented with chief complains of multiple hypopigmented patches shortly after birth (within 1 week after birth)

On examination there were multiple oval , round shaped macules and patches over lower limb.

Size of the lesions were 2x2 mm smallest one and 2 x 2 cm largest one.

Margins of the lesions were well defined.

Skin overlying the lesions was normal.

Patient did not give any history suggestive of neurological ,

ophthalmological and skeletal abnormalities.

DISCUSSION –

Pigmentary mosaicism

Mosaicism refers to the occurrence in an individual of two or more cell population that are karyotypically or genotypically different and yet are derived from single zygote.

The pathogenesis of pigmentary mosaicism has been explained by various hypothesis. These include co-migration of genetically different cell populations, functional X-chromosome mosaicism , spreading of X inactivation to autosomes in balanced X ; autosomal translocations , partial activation or silencing of pigmentary genes by transposons , genetic imprinting and phenotypic reversion [Taibjee et al.,2004]

An association between pigmentary anomalies and chromosomal mosaicism has been reported since the 1960s. [Ferrier et al.,1964;Zuelzer et al.,1967;Varela and Sternberg,1969]. Since then several cases reporting association between pigmentary pattern such as linear, whorled or patchy and hypo or hyperpigmented skin and chromosomal mosaicism have been published. Cytogenetic alterations such as mosaicism for trisomy 18, diploidy/triploidy , sex chromosomal aneuploidy, mosaic trisomy 13 and tetrasomy 12p have been reported in literature to be associated with pigmentary anomalies[Happle, 1993,2000;Horn et al.,1997].

The skin changes may be hypo- or hyper-pigmented and the cutaneous pattern follow the line of blaschko , be seen in chequerboard pattern , a patchy pattern without midline separation or arranged in a phylloid (leaflike) pattern[2]. The type 1 mosaic pattern , following the line of blaschko,is the most common mosaic pattern,which can be subclassified into type 1a with narrow bands and type 2b broad bands. The chequerboard or type 2 pattern shows alternating squares of dyschromia with a sharp midline separation . Type 3 shows a phylloid pattern of hypo- or hyperchromic macules with midline separation and type 4 shows a patchy pattern without midline separation[3].

Among the mosaic patterns , there have been only few reports of phylloid presentation in the literatures.

Phylloid hypomelanosis

Phylloid hypomelanosis represents a uniform neurocutaneous phenotype with associated central nervous system defects , conductive hearing loss , choroidal and retinal coloboma , craniofacial and skeletal defects and appears closely linked to abnormalities of chromosome 13,most frequently trisomy 13[4 -10].

Mosaic trisomy 13 is far less frequently observed than nonmosaic trisomy 13 (patau syndrome) , which is a severe birth defect characterized by microcephaly,serious central nervous system anomalies , congenital heart defects , holoprosencephaly , cleft lip or palate ,microphthalmia , polydactyly , scalp defects and numerous other developmental anomalies[11]

The phylloid pattern is described as a peculiar type of pigmentary mosaicism characterized by macules in the form of round and oval spots , patches resembling the asymmetric leaves of the begonia[3].Associated extracutaneous anomalies includes neurological, ocular ,dental and skeletal defects[4].Gonzalez-Ensenant et al[12] indicated that this disorder is most likely related to 13q region.It is proposed that mosaic overexpression of the candidate pigmentary genes , such as endothelin receptor type B (EDNRB) which is related to melanoblast migration , may cause impaired melanoblast migration and melanocyte formation, resulting in phylloid hypomelanosis[10]

Among neurological defects subjects reported uptil now showed various degrees of mental retardation or epileptic seizures (Happle 2000, Schepis et al. 2001, Hansen et al. 2003). Microcephaly is frequently detected (Horn et al. 1997, Pillay et al. 1998, Schepis et al.2001, Ribero Noce et al. 2001).

Karyotyping

Blood sample for karyotyping have been sent.

Results of karyotyping are awaited.

We are expecting mosaic partial trisomy of chromosome number 13 in above patients.

AIM

To create awareness among the treating physicians for this rare entity , its bening nature and genetic association.

Declaration of patient consent

The author certify that they have obtained all appropriate patient consent forms . In the form , the patient has given her consent for her images and other clinical information to be reported in journal . The patient understand that name and initials will not be published and due efforts will be made to conseal identity , but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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