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Original Article

Clinical profile of children with pigmentary disorders

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Abstract

Pigmentary disorders are believed to be the commonest group of dermatoses in pediatric age group [6], but, there is a dearth of adequate data regarding the frequency and pattern of different types of pigmentary disorders in children. Any deviation from the normal pattern of pigmentation results in significant concerns in the affected individual. Even, relatively minor pathologic pigmentary changes can cause children to become pariahs in their community. This study was a descriptive study spanning over a period of 23 months. Institute ethics committee clearance was obtained. All children attending the Dermatology out Patient Department (OPD) (6 days in a week) were screened for any cutaneous pigmentary lesions. Children (up to 14 years of age) with pigmentary disorders were included in the study after getting informed consent from the parents/ guardians. Out of 167 children, 53 (31.7%) had hyperpigmentation lesions only, whereas 108 (64.7%) had hypopigmentary lesions only. Six children had presence of both hyper- and hypopigmentary lesions. Five children (2.9%) had overlap of unrelated hyper- and hypopigmentary lesions. Only one child (dyschromatosis universalis hereditaria) had 'true' combination of hyper- and hypopigmentary lesions, the presence of both of were together were required for the diagnosis. The pigmentary disorders in our study had characteristic features in most children, while in some the features noted were rather unique.

Keywords: Clinical profile, children, pigmentary disorders

Introduction

Physiologically, human skin presents a unique color. Skin color varies from individual to individual, and in an individual, variation in the degree of pigmentation occurs in various regions of the body. Normal skin color is dependent on haemoglobin (in the oxygenated and reduced state), carotenoids and melanin pigment. Melanin is the major determinant of skin color, and racial and ethnic differences in skin color are related to the number, size, shape, distribution of melanin-laden organelles called melanosomes. Melanocyte is the sole site of melanin synthesis. Eumelanin and pheomelanin are the two major types of melanin; they impart brown-black and yellow-red color, respectively. Melanin synthesis can be induced by a number of factors; hence skin pigmentation is classified as constitutive and facultative. The constitutive skin color refers to the "baseline" genetically determined color in the absence of sun exposure and other influences. The facultative (inducible) skin color is due to pigmentary darkening after secondary melanin production which can result from sun exposure and endocrine causes. Skin pigmentation is the body's main defence against ultraviolet rays of sunlight [1].

Skin color is an important visible sociocultural characteristic of an individual. Hence any deviation from the normal pattern of pigmentation results in significant concerns in the affected individual. Even, relatively minor pathologic pigmentary changes can cause children to become pariahs in their community. Pigmentary disturbances include decrease or absence or increase in pigmentation which could be either epidermal or dermal. Pigmentary disorders are a group of dermatoses characterized by alteration of the normal pattern of pigmentation in a localized (circumscribed), or generalized distribution, and may be congenital or acquired in origin [2]. Pigmentary disorders may be hypopigmented (hypomelanotic) or hyperpigmented (hypermelanotic) or both hypo- and hyperpigmented. Many of the pigmentary disturbances are present at birth (congenital melanocytic nevus, Mongolian

spots, hypomelanotic macules of tuberous sclerosis complex, cutaneous mosaicism), and parents' of such children usually become very much concerned about the pigmentary disturbances and seek medical advice regarding the future course of the disorder [2, 3].

Pigmentary disorders are believed to be the commonest group of dermatoses in pediatric age group. But, there is a dearth of adequate data regarding the frequency and pattern of different types of pigmentary disorders in children [4]. Hence, this study is being undertaken to find out the pattern of pigmentary disorders in children.

Methodology

This study was a descriptive study spanning over a period of 23 months. Institute ethics committee clearance was obtained. All children attending the Dermatology out Patient Department (OPD) (6 days in a week) were screened for any cutaneous pigmentary lesions. Children (up to 14 years of age) with pigmentary disorders were included in the study after getting informed consent from the parents/ guardians. Children more than 14 years of age, and when parents/guardians (of children less than 14 years of age) are not consenting for participation in the study were excluded. A detailed clinical history was elicited with regard to patients' age, gender, address, age of onset of disorder, type (hypo-/hyper pigmented), site and size of pigmented lesions, familial involvement, associated skin and systemic conditions and the details were recorded on a proforma. Detailed examination of the pigmentary lesion/lesions was done and findings were noted in terms of site, size, shape, morphology of lesions etc. General physical examination was done to see any associated cutaneous or systemic involvement and the relevant findings were recorded.

Results

A total of 34392 children attended the Dermatology OPD during the study period. Thus the frequency of pigmentary disorders among children attending the Dermatology OPD was 4.85 per 1000 children.

Out of 167 children, 53 (31.7%) had hyperpigmentation lesions only, whereas 108 (64.7%) had hypopigmentary lesions only. Six children had presence of both hyper- and hypopigmentary lesions. Five children (2.9%) had overlap of unrelated hyper- and hypopigmentary lesions. Only one child (dyschromatosis universalis hereditaria) had 'true' combination of hyper- and hypopigmentary lesions, the presence of both of were together were required for the diagnosis. Thus, for the ease of analysis, total number of

children with hyperpigmentation lesions was counted as 58, and that with hypopigmentary lesions as 113. Dyschromatosis universalis hereditaria is considered separately.

Age

The mean age of 167 children in the study was 7.1 years, ranging from 3 days to 14 years with a median of 8 years (SD-4.57, SEM-0.35). The mean age of children with hyperpigmentation disorders was 6.89 years, and that in hypopigmentary disorders was 7.2 years.

Gender

There were 93 boys (55.7%) and 74 girls (44.3%) in our study, with a male to female ratio of 1.25:1. The mean age of boys was 7.84 years, with a range of 3 days to 14 years and median of 8 years (SD-4.64, SEM-0.48). The mean age of the girls was 6.35 years, with a range of 45 days to 14 years and median of 6.5 years (SD-4.37, SEM-0.50). Out of 93 boys, 31 (33.3%) had hyperpigmentation disorders and 60 (64.5%) had hypopigmentary disorders, whereas two (2.1%) boys had both. Out of 74 girls, 22 (29.7%) had hyperpigmentation disorders and 48 (64.8%) had hypopigmentary disorders, whereas four (5.4%) girls had both.

Age and Gender Distribution

In our study, most common age group was 11-14 years contributing to 51 out of 167 (30.5%) children, followed by 6-10 years contributing to 49 out of 168 (29.34%). Thirty four out of 93 boys were in the 11-14 years age group, whereas 28 out of 74 girls were in the 6-10 years age group.

Table 1. Age and gender distribution of all children

Age groups (years)	Boys	Girls	Total
<1	13	7	20
1-5	26	22	48
6-10	20	28	49
11-14	34	17	51
Total	93	74	167

Weight and Height

The mean weight of all children was 22.27 kg (SD-12.78, SEM-0.98), ranging from 2.7 kg to 78 kg, and median of 22. The mean height of all children was 110.07 cm (SD-30.93, SEM-2.4), ranging from 44 cm to 168 cm and median of 119 cm.

Table 2: Weight and height characteristics in hyper- and hypopigmentary disorders

Parameters	Pigmentary disorder	No. of children	Mean	Median	Min	Max	SD	SEM
Weight (Kg)	Hyperpigmentation	58	21.179	20.000	3.5	78.0	14.580	1.9145
	Hypopigmentary	113	22.34	22.00	3	55	11.602	1.09
Height (cm)	Hyperpigmentation	58	107.43	112.00	42	168	32.231	4.232
	Hypopigmentary	113	112.48	120.00	44	162	28.61	2.6

There were 58 children (34.7%) with hyperpigmentation disorders including 32 boys (55.2%) and 26 girls (44.8%). The mean age of children with hyperpigmentation disorders was 6.89 years.

Most common hyperpigmentation disorder in our study was café-au-lait macule seen in 12 children (20.6%), followed by post-inflammatory hyperpigmentation (9/58, 15.5%), pigmentary mosaicism (8/58, 13.8%), congenital

melanocytic nevus (7/58, 12%), lichen planus (5/58, 8.6%), Mongolian spots (4/58, 6.8%) and fixed drug eruption (3/58, 5.2%). Two (3.4%) children each had urticaria pigmentosa and Becker's nevus. One child each (1.7%) had Chediak-Higashi syndrome (CHS), discoid lupus erythematosus, incontinentia pigmenti (IP), lentiginos, lichen striatus, nevus of Ota, pityriasis rubra pilaris and speckled lentiginous nevus.

Table 3: Frequency of hyperpigmentation disorders

Type of hyperpigmentation disorders (number = 58)	No. of children	%
Café au lait macule (CALM)	12	20.6
Post-inflammatory hyperpigmentation	9	15.5
Pigmentary mosaicism	8	13.8
Congenital melanocytic nevus (CMN)	7	12
Lichen planus	5	8.6
Mongolian spots (MS)	4	6.8
Fixed drug eruption	3	5.2
Becker's nevus	2	3.4
Urticaria pigmentosa	2	3.4
Chediak-Higashi syndrome (CHS), discoid lupus erythematosus, Incontinentia pigmenti (IP), centrofacial lentiginosis, lichen striatus, nevus of Ota (NO), pityriasis rubra pilaris, speckled lentiginous nevus	1 each	1.7 each

Note: one child had CALM and NO. Another child had CMN and MS.

Most common hypopigmentary disorders in our study was pityriasis Alba which was seen in 28 children (24.7%), followed by vitiligo in 23 (20.4%) children, leprosy in 13 (11.5%), nevus depigmentosus in 11 (10.18%), and tinea versicolor in 7 (6.2%) children. Five (4.42%) children each had hypomelanosis of Ito (HI) and post inflammatory

hypopigmentation. Four (3.5%) children each had pityriasis rosea and steroid induced hypopigmentation. Three (2.65%) children each had lichen sclerosus *et al.* atrophicus, pityriasis lichenoides chronic. Two (1.8%) children each had, lichen striatus, OCA and TSC. One (0.9%) child each had pigmentary mosaicism, Griscelli syndrome.

Table 4: Frequency of hypopigmentary disorders

Diagnosis (Number =113)	Frequency	%
Pityriasis alba	28	24.7
Vitiligo	23	20.4
Leprosy	13	11.5
Nevus depigmentosus	11	10.1
Tinea versicolor	7	6.2
Hypomelanosis of Ito, post-inflammatory hypopigmentation	5 each	4.4 each
Pityriasis rosea, steroid induced hypopigmentation	4 each	3.5 each
Lichen sclerosus et atrophicus, pityriasis lichenoides chronica,	3 each	2.6 each
Lichen striatus, oculocutaneous albinism, Tuberous sclerosis complex	2 each	1.8 each
Hypopigmentary mosaicism, Griscelli syndrome	1 each	0.9 each

Table 5: Sites of involvement in hypopigmentary disorders

Sites	Frequency	Percent
Face	55	28.6
Upper limb	23	11.8
Lower limb	22	11.3
Chest	20	10.3
Back	18	9.2
Abdomen	11	5.6
Loin	8	4.2
Axilla, buttocks, extremities, shoulder	5 each	2.6
Flank, neck, scalp, penis	4 each	2.1
Perianal, scrotum, sacral area	1	0.5
Generalized involvement	3	1.5

Discussion

Variation in skin color, which occurs due to the differences in the melanin content, is one of the most striking human characteristics. Skin color plays a very important role in enhancing one's physical appearance and attractiveness. Differences in skin color also have important evolutionary and physiological implications. Pigmentary differences arise basically from variation in the number, size, composition and distribution of melanosomes, which are lysosome-like granules in the melanocytes in which melanin production takes place. Disorders of pigmentation are one of the most common skin disorders of human beings, and it is difficult to find a person who does not have or never had an alteration in pigmentation anywhere in the skin. Temporary

and localized pigmentary change may not worry many individuals; but, because of easy visibility, such change becomes psychologically distressing when the process is generalized or permanent. Pigmentary disorders in children are little different from those in the adults in terms of etiology (pigmentary alterations due to genetic disorders are more commonly encountered in the children) and heightened parental concerns. Because the pigmentary change in children may be seen in the evolution phase (before the full-fledged manifestations are evident) of the underlying disease, some degree of diagnostic problems may be encountered, and because of the inherited nature of some of the pigmentary disorders, the dermatologists may be faced with significant therapeutic challenges.¹

The prevalence of pigmentary disorders in children varies depending upon the geographical location of the study population. In an Iranian study by Toossi *et al.*, pigmentary disorders were found in 64.2% out of 1143 children (pigmentary disorders were also the commonest group of skin disorders in those children). Osburn *et al.* studied 830 infants in Oklahoma City, USA and found that the frequency of pigmented lesions was 30% in them. In a South Indian study by Karthikeyan *et al.*, the prevalence of pigmentary disorders was 8.6% out of 2100 children. In study by Dogra in Northen India, the point prevalence of pigmentary disorders was 2.6% among 12,586 school children. In another study by Asokan *et al.*, pattern of skin diseases among patients attending tertiary care hospital in

Kerala the frequency of pigmentary disorder was found to be 4.73%. In our study, the prevalence of pigmentary disorders in children attending the dermatology OPD was 4.85% Out of 167 children in our study, hyperpigmentation and hypopigmentary disorders were seen in 31.7% and 64.7% of the children, respectively^[5].

Localized areas of hyperpigmentation are frequently developmental or hereditary in origin, and appear early in childhood. However, pigmented lesions may also be acquired later in childhood.

Café-au-lait macules (CALM) are round to oval, flat macules of light brown color found in association with various genodermatoses as well as in up to 10- 33% of normal children. They are frequently present at birth, or develop soon thereafter, vary in size from 1.5 cm or less in their smallest diameter to much larger lesions that may measure up to 15 to 20 cm or more in diameter. CALMs occur as isolated lesions in most of the affected individuals, but they may be a sign of neurofibromatosis and other systemic disorders. They constitute the major criteria in the diagnosis of neurofibromatosis, if macule were more than six and measuring 0.5 cm or more before puberty and 1.5 cm or more in diameter in adults. The frequency of CALM in Osburn *et al.* study was 2.8% among all the infants included in the study, and 9.2% among infants with pigmented lesions. In a McLeean and Gallagher study (which included freckles, CALM, and other pigmented lesions of school children), prevalence of café-au-lait macule was more than 25% in three ethnic groups. In our study, CALM occurred in 7.2% of all children with all pigmentary disorders in the study and in 20.6% of children with hyperpigmentary lesions^[6]. Eight out of 12 children had the lesions since birth. Only two patients had syndromic associations (TSC and MAS); the remaining 10 children did not have any other abnormality. Father of one child with 20 macules had neurofibromatosis. The size of the macules ranged from 0.3cm x 0.3cm to 20 x 10 cm. One girl had a large CALM (20 x 10) involving the right chest and upper limb, with short stature was diagnosed as McCune-Albright syndrome.

Postinflammatory hyperpigmentation (PIHP) is characterized by an increase in melanin synthesis following cutaneous inflammation occurring due to physical trauma, friction, primary irritants, lichen simplex chronicus, and dermatoses, such as pityriasis rosea, fungal infection, bullous dermatoses, psoriasis, fixed drug eruptions, photodermatitis, and pyoderma^[7].

PIHP in our study was due to tinea infection and chronic bullous disease of childhood. In tinea infections, PIHP is seen in the central portion of the macule in the affected skin. Bullous disorders like chronic bullous disease of childhood lesions heal with hypo or hyperpigmentation. In a study conducted by Aboobaker studied chronic bullous disease of childhood (CBDC) in 30 cases, lesions healed with either with hypopigmentation or hyperpigmentation. In our study, however, CBDC healed with PIHP.

Mosaicism is defined as the existence of two or more genetically different populations of cells originating from one genetically homogenous zygote. Pigmentary mosaicism is a term that has been used to describe variegated patterns of pigmentation (streaks of hyperpigmentation and hypopigmentation) in the skin caused by genetic heterogeneity in the cells that make up the skin. Mosaic skin diseases may show different patterns of clinical involvement

such as^[8]

Type 1 lines represents the of Blaschko (type 1a, displaying narrow bands as seen incontinentia pigmenti, type 1b represents broad bands following Blaschko's lines as seen McCune-Albright syndrome).

Type 2, or checkerboard pattern is characterized by an alternating squares of hyperpigmentation, with sharp midline separation, as observed in systematized speckled lentiginous nevus.

Type 3, or phylloid (leaflike) pattern characterized by an arrangement of pigmentary disturbances reminiscent of floral ornaments or a Jugendstil painting.

Type 4, or patchy pattern without midline separation is observed in cases of giant melanocytic nevus, or neurocutaneous melanosis.

Type 5 a lateralization pattern.

Linear and whorled nevoid hypermelanosis (LWNH) is a sporadic disorder in which multiple streaks of hyperpigmentation are usually noted at birth or within the first 2 years of life. This condition is benign and usually without multisystem involvement (cardiac, neurological defects and mosaicism), however, in some cases various congenital abnormalities have been reported.

In our study, linear and whorled nevoid hypermelanosis was observed in three children, two children had lesions. Linear and bizarre shaped lesions were seen on the lower limb and one child had lesions on the chest and axilla. No extracutaneous abnormalities were noted in the children.

Linear epidermal nevi frequently follow the lines of Blaschko. Epidermal nevi may present in one of two distributions, localized or systematized. Localized lesions (i.e. nevus unis lateris) usually manifest as a single linear unilateral nevus oriented perpendicularly on the extremities or as sworls on the trunk. Widespread linear epidermal nevi, often referred to as systematized, form multiple bilateral verrucoid lines and whorls. They are less common than the localized epidermal nevi. The association of epidermal nevi with systemic anomalies (neurological, skeletal abnormalities, ocular and other organs) comprises the epidermal nevus syndrome.

Linear nevoid hyperpigmentation along the lines of Blaschko was seen in 2 boys, in one child a unilateral distribution involving the chest, abdomen, upper limb and lower limb was seen and in another a linear distribution involving the chest and upper limb was seen. No extracutaneous abnormalities were noted in the children.

Phylloid pattern is delineated as a peculiar type pigmentary mosaicism characterized by macules reminiscent of floral ornaments in the form of round or oval spots, patches resembling the asymmetric leaves of begonia, and oblong lesions. Rieger *et al.* described melanotic macules following Blaschko's line in two McCune-Albright syndrome patients. In their study, macules present over the back in both cases, in a bizarre fashion, and showed typical fountain-like, V-shaped pattern of Blaschko's lines^[9]

In our study, segmental pigmentation in a phylloid (type 4) pattern (linear hyperpigmentation, roughly along the lines of Blaschko) was seen in 3 children, 2 boys, and one girl without the crossing midline. One child had a macule on the left side of face, neck, chest and upper limb (upto wrist) with irregular margins. Another child had a large hyperpigmented macule on the abdomen and in the girl the leaflike hyperpigmented macules distributed over the face and neck was seen in "phylloid" pattern. In all cases, the

lesions were present since birth. No extracutaneous abnormalities were noted in the children. A phylloid pattern of mosaicism involving the face, neck and upper extremities along the Blaschko' line is unique feature in our study^[10].

Pityriasis Alba is a relatively common skin disorder characterized by the presence of asymptomatic, superficial, hypopigmented macules with mild surface scaling, located usually on the face, but can be seen on the neck and shoulders. In a study conducted by Sujatha *et al.* in 200 patients with pityriasis alba in the age ranged from 8 months to 32 years, with a majority (69%) being below 15 years of age. Itching was noted in 17 patients. Personal history of atopy was noted in 34 (17%) patients. In our study, pityriasis Alba occurred in 16.7% of all children in the study. It was the commonest (24.7%) hypopigmentary disorder in our study. The lesions occurred in the age range of 5 months to 14 years (mean of 6.3 years). The macules were distributed over the face only. Itching was present in three children. Three children had atopic diathesis (only one had history of itching).

Vitiligo is a common, acquired, disorder of the skin characterized by well circumscribed, milky white macules. It occurs in about 1% of pediatric patients. Jaisankar *et al.* studied 346 patients with vitiligo, out of which 90 children had vitiligo and incidence of vitiligo in children was found to be 0.72%. Out of 90 children, 35 (38.9%) were boys, and 55 (61.1%) were girls. Age of onset of and the age of presentation of vitiligo was between 4 and 6 years in 35.5%, and between 10 to 12 years in 31.2%. They noticed family history of vitiligo in 3 (3.3%) children. Vitiligo vulgaris was found in 35 (38.8%, 14 boys, 21 girls), focal vitiligo in 18 (20.1%, 8 boys, 10 girls), segmental in 19 (21.1%, 7 boys, 12 girls), acrofacial in 6 (6.7%, 4 boys, 2 girls), and mucosal in 12 (13.3%, 2 boys, 10 girls). In their study segmental vitiligo was found to be more common in children (19, 20.1%) compared to adults (13, 5.0%).

In our study vitiligo was seen in 23 children. Vitiligo vulgaris was seen in 11 (47.8%) cases, segmental vitiligo was seen in 8 children (34.7%), localized vitiligo in 2 girls, lip-tip and lip vitiligo in one girl each. Two children had familial involvement. Leucotrichia was seen in 6 children.

Leprosy in children can be an indicator of disease prevalence in the general population and its detection helps determining the disease transmissibility. Due to its long incubation period, leprosy is rare in children. But, they are at higher risk of developing the disease when living in endemic areas and when exposed to family contacts. Kumar *et al.* studied leprosy data from 1990 to 1999 and diagnosed 1360 new cases of leprosy, of which 61 (4.5%) were children (40 boys and 21 girls; age-0 to 14 years, M: F ratio 1.9:1). In their study, indeterminate leprosy was seen in 6.6%, borderline tuberculoid leprosy in 78.7%, borderline lepromatous leprosy in 8.2%, lepromatous leprosy in 4.9% and pure neuritic leprosy (PNL) in 1.6%. No cases of TT and BB leprosy were noted in their study.

In our study, we observed leprosy in 13 children. The mean age of the children was 10.21 years, ranging from 4 to 14 years. Ten children had borderline tuberculoid (BT) leprosy. Typical tuberculoid (TT) leprosy was seen in 2 children (one boy and one girl), on the face in one child and in the lower limb in the other. Indeterminate (I) leprosy was noted in one 14 year old girl on the left cheek. Ulnar neuritis with claw hand was seen in one child^[11].

Swain *et al.* studied the prevalence of leprosy among

household contacts of leprosy. In their study, 72 cases were detected from 54 families. Out of these, 45 (62.5%) were in the pediatric age group (0-14 years) and of these 45.8% belonged to 6-14 years. Out of 72 cases, 58 were paucibacillary (PNL+I+TT+BT) cases. Risk of secondary case was high when there was a family history of lepromatous leprosy. The attack rate among those exposed to the paucibacillary type was less. Fathers of the children were the source of infection in most of the cases (57%).

In our study, history of contact with lepromatous leprosy patients (father) was present in two children with BT leprosy (paucibacillary), which is consistent with the aforementioned study.

Nevus depigmentosus (ND) is defined as a congenital non-progressive hypopigmented macule or patch that remains stable in relative size and distribution throughout life. In a study by Lee *et al.*, 92.5% of ND patients presented before 3 years of age, among which 19.4% children had lesions at birth. ND was isolated in 59.7 and segmental in 40.3%. The back and buttocks were the most commonly affected sites, followed by chest and abdomen, face, neck, and arms were affected in descending order of frequency. Serrated and irregular margins were seen in 77.4%.

Nevus depigmentosus (ND) was seen in 11 children in our study. Three children had lesions since birth. Chest was the most affected site in our study followed the face and upper limb, abdomen, buttocks and lower limb in a decreasing order of frequency. One child had associated microcephaly.

Tinea versicolor is a mild chronic superficial fungal infection of the stratum corneum caused by *Malassezia* or *pityrosporum*, characterized by patchy and scaly discoloration of the skin. It is the one of the most common pigmentary disorders worldwide. In Jena study of pityriasis versicolor in 271 children, 150 boys and 121 girls, majority of children were aged 8-12 years (31.7%), but 10 infants were also affected. The duration was less than 6 months in all cases. Face was the most affected site (39%) and extensive involvement was seen in 45 (16.6%) children with lesions on the back, shoulder and back^[12].

In our study, tinea versicolor was seen in 7 children, having a mean age of 6.6 years. The age of onset of lesions ranged from 1 month to 13 years of age. The mean duration of lesions was 2.2 months, ranging from 7 days to 6 months. All children had hypopigmented macules. Five children had lesions on the face, remaining two children had on the chest, back and upper limb. Medial canthi of both eyes were involved in two infants, which was a unique finding in our study

Conclusion

- Pigmentary disorders are one of the most common disorders in children. They can appear at birth or develop later in life. These can be either isolated lesions or have syndromic association. Because of easy visibility of the skin lesions, the underlying disease/syndromes can be diagnosed at an early stage.
- Pityriasis Alba is the most common pigmentary disorder in children.
- Vitiligo is common in children, where the proportion of the segmental type is formidable.

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