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"Whorled Pigmentation"- Acase Series of Pigmentary Mosaicism along the Blaschko Lines without Systemic Involvement.

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Abstract:
Background*: Whorled pigmentation is defined as streaks of pigmentary changes i.e. either hypo/ hyperpigmentation along the blaschko lines due to heterogenicity of skin cells. It is also called pigmentary mosaicism. It includes Linear and whorled nevoid hypermelanosis, Hypomelanosis of Ito and Incontinentia pigmentii. Most of these are commonly associated with extracutaneous involvement predominantly involving central nervous system, ocular and musculo skeletal system.*
Aims and Objectives :
•To study the clinico-epidemiological aspects of the patients with pigmentary mosaicism attending to our out patient department, Government General Hospital, Kurnool.
•To study the presence of extracutaneous involvement in the patients with pigmentary mosaicism.
Materials and Methods*: This prospective observational study was conducted in tertiary care centre, Government General Hospital, Kurnool, Andhra Pradesh, from July 2022 to December 2022, which included 11 patients with pigmentary mosaicism with uni/ bilateral involvement. Epidemiological data, clinical features, routine investigations and special investigations like Woods lamp examination, Biopsy of the lesion, CT scan, MRI brain, Ophthalmic examination and other needed investigations were done to rule out extra cutaneous involvement.*
Results*: Out of 11 patients with pigmentary mosaicism, 6 were female (54%) and 5 were male(46%), showing slight female preponderance. Seven patients (63%) exhibited hyperpigmentation (Linear and Whorled Nevoid Hypermelanosis), while 4 patients (37%) exhibited hypopigmentation (Hypomelanosis of Ito) along the blaschko lines. Unilateral and bilateral distribution of pigmentation was seen in 8 patients (72%) and (28%) respectively. Majority of the patients (81.81%) manifested skin pigmentation before 2 years of age. Family history is insignificant in all the patients. Extra cutaneous involvement is absent in all the patients.*
Conclusion:- *According to the literature, the disorders of pigmentary mosaicism i.e. LWNH,HI , associate with extracutaneous involvement of CNS, ocular and musculoskeletal system. But the cases included in the study were of pure cutaneous pigmentary mosaicism without extracutaneous manifestations. Through this study, we emphasize that the association of systemic involvement in pigmentary anomalies along the blaschko lines far less often than being reported in the past.*
Key Word*: whorled pigmentation, Linear and Whorled Nevoid Hypermelanosis (LWNH), Hypomelanosis of Ito (HI), pigmentary mosaicism, extracutaneous manifestations, blaschko lines.*

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I. Introduction

Mosaicism is defined as the presence of 2 or more cell population with different expression of 1 or more genes and are derived from single zygote. If the mosaicism is seen in the skin, it is called Pigmentary Mosaicism. Whorled pigmentation is defined as streaks of pigmentary change either hypo/ hyper pigmentation distributed along the blaschko lines. Blaschko lines are the lines that originate due to migration of epidermal cells during embryonic life. They form V shape- over the back, S shape- on the lateral aspect of the trunk, linear – on the extremities etc.Whorled pigmentation is seen in LWNH, HI and Incontinentiapigmentii which are most commonly associated with extracutaneous manifestation involving CNS, Ocular and Musculo skeletal system.

Linear and whorled nevoid hypermelanosis (LWNH):- It is a rare sporadic disorder of pigmentary mosaicism with hyperpigmented macules arranged in a linear / whorled streaky configuration without preceding inflammation or atrophy along the blaschkolines [9] . It is also called "Zebra like hyperpigmentation"," Zosteriform hyperpigmentation" or "Reticulate hyperpigmentation of Iijima". This condition most commonly shows genetic mosaicism of trisomy of 7, 14, 18, 20 and X chromosome. The onset of lesion is few weeks after birth and progress upto 2 years of age before stabilization. Clinical features include reticulate hyperpigmented macules that coalesce to form streaks and whorls over trunk, extremities, back and neck along the blaschko lines sparing palms, soles, mucosa and face. The texture of the skin is normal without any atrophy or hyperkeratotic papules or vesicles. Extracutaneous features include central nervous system, skeletal and ocular abnormalities. Central nervous system anomalies include microcephaly, epilepsy, developmental retardation etc. whereas cardiac anomalies include ventricular septal defects, Tetrology of Fallot etc. Histopathology shows increase in pigmentation of basal cell layer with melanocytes present upto mid epidermis. No focal pigmentary incontinence in the dermis [10] . Dermoscopy shows net like pattern of pigmentation.

Hypomelanosis of Ito (HI):- It is also called Incontinentiapigmentiachromians. Its origin is due to chromosomal mosaicism or translocation or it may be sporadic. It is a neuro cutaneous syndrome most commonly seen in female (M:F= 2.5:1) with frequent CNS and musculo skeletal involvement. The onset of the lesions is at birth or within 2 years of age. Clinical features include streaks and whorls of hypopigmentation along the blaschko lines without the history of inflammation or atrophy [8]. Other cutaneous features are café au lait spots, cutis marmorata, angiomatous nevi, nevus of ota etc. Central nervous system features include mental retardation, seizures, language disorders, motor dysfunction, agenesis of corpus callosum, vascular abnormalities etc. Ocular abnormalities like ptosis, symblepheron, nystagmus, strabismus, cataract corneal opacity etc. Skeletal defects include short stature, scoliosis, syndactyly, polydactyly, brachydactyly, asymmetry of limbs etc. Histopathology shows decrease in melanocytes and few melanosomes without pigmentary incontinence.

II. Aims and Objectives

- To study the clinico-epidemiological aspects of the patients with pigmentary mosaicism attending to our outpatient department, Government General Hospital, Kurnool.
- To study the presence of extracutaneous involvement in the patients with pigmentary mosaicism.

III. Material And Methods

This prospective observational study was conducted in tertiary care centre, Government General Hospital, Kurnool, Andhra Pradesh, from July 2022 to December 2022, which included 11 patients with pigmentary mosaicism with uni/ bilateral involvement.

Study Design: Prospective observational study

Study Location: This was a tertiary care teaching hospital based study done in Department of Dermatology, Venereology and Leprosyof Government General Hospital, Kurnool, Andhra Pradesh, India.

Study Duration: July 2022 to December 2022

Sample size:11 patients.

Inclusion criteria:

Male/ female patient with pigmentary mosaicism of any age who was willing for the study.

Exclusion criteria:

1. Cases who were not willing for the study
2. patients without pigmentary mosaicism
3. patients with history of prior inflammation or any other lesions or presence of atrophy along the pigmentary change.

Procedure methodology:

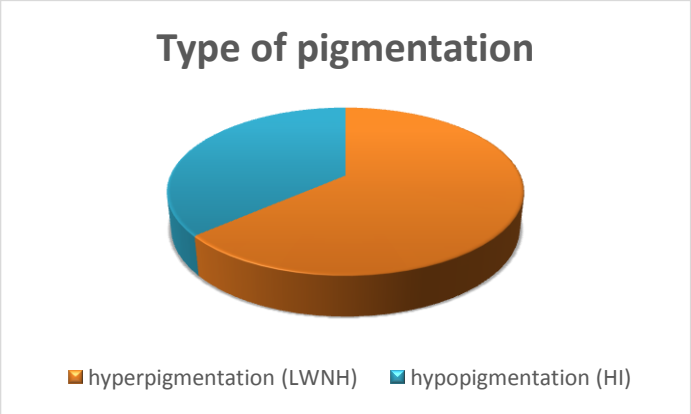
1. After ethical committee clearance, informed written consent was taken.
2. Demographic information of the patient like age, sex, age of onset of the lesions, distribution of involvement, pigmentation type, family history were taken and thorough clinical examination and ophthalmic examination was done to check for extracutaneous manifestations.
3. Investigations done were: Routine investigations like Complete blood picture, Liver function tests, Renal function tests; woods lamp examination of the lesions, biopsy of the lesions, CT brain, MRI brain.

The collected clinico epidemiological data was tabulated and graphs were generated. The results were presented as frequency and percentages.

IV. Result

Sex distribution:- Almost all the patients were under the age of 20 years at the time of presentation. Among the 11 patients, 6 were female (54%) and 5 were male (46%), ie slight female preponderance.

Type of pigmentation:- Two types of pigmentary changes were noted. Out of 11 patients, 7 cases (63%) exhibited hyperpigmentation (LWNH) and 4 cases (37%) exhibited hypopigmentation (HI).

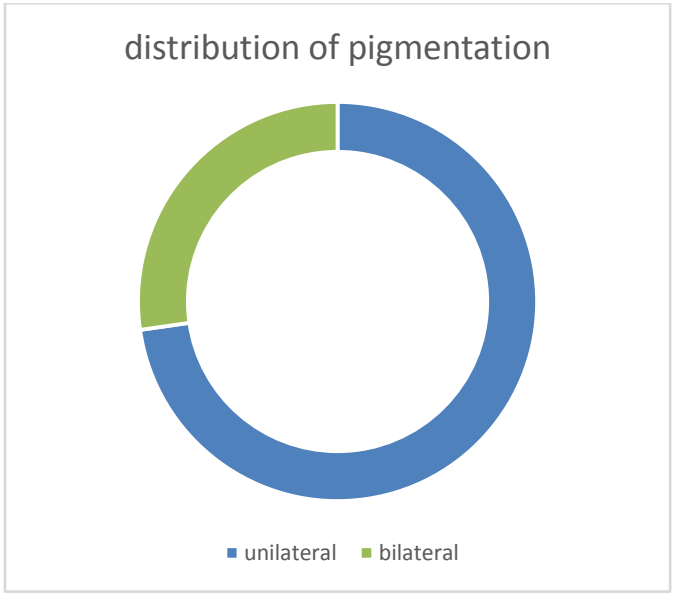


Age of onset:-

Age of onset	No. Of cases with Hyperpigmentation (LWNH)	No. Of cases with Hypopigmentation (HI)
At birth	2	1
< 6 weeks	1	1
6 weeks – 1 year	1	1
1-2 years	1	1
>2 years	2	0
Total	**7**	**4**

Majority of the patients (81.81%) manifested skin pigmentation before 2 years of age.

Distribution of pigmentation:- Unilateral and bilateral distribution of pigmentation was seen in 8 patients (72%) and 3 patients (28%) respectively.



Familial transmission:- family history of similar complaints is insignificant.

Extracutaneous manifestations:- After thorough clinical examination and investigations, no extracutaneous manifestations involving CNS, ocular , musculo skeletal and other systems were detected in all the patients.

Figure 1a.

Figure 1b

Figure 1a & 1b unilateral HYPOMELANOSIS OF ITO hypopigmented macules along the blaschko lines.

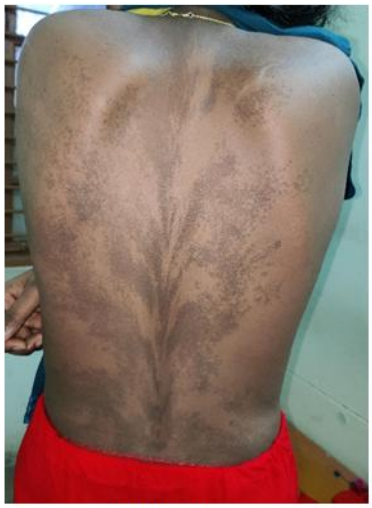


Figure 2a.

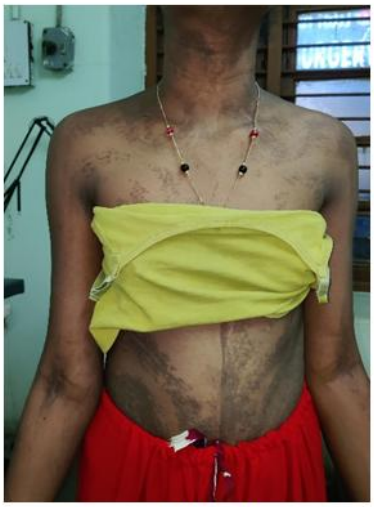


Figure 2b.

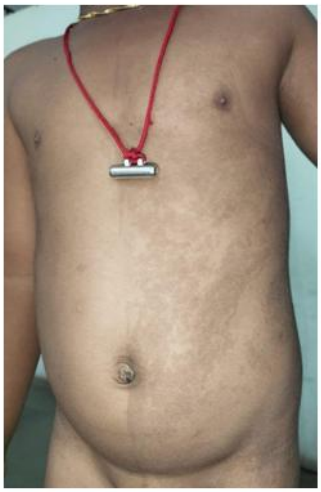


Figure 3

Figure 2a,2b & 3 showing LINEAR AND WHORLED NEVOID HYPERMELANOSIS with hyperpigmented macules along the blaschko lines.

V. Discussion

Whorled pigmentation is characterized as hypoor hyperpigmented streaks along the blaschko line distribution which develop at birth or within 2 years of age. Blaschko lines are those that develop as a result of epidermal cell migration during embryonic life. Whorled pigmentation is seen in LWNH, HI, and Incontinentiapigmentii, which are most frequently accompanied by extracutaneous manifestations affecting the nervous, ocular, and musculoskeletal systems.The present study includes the patients with Linear and Whorled Nevoid Hypermelanosis and Hypomelanosis of ito.

Sex distribution:- In the present study, almost majority of the patients were under 20 years old. Six of the 11 patients (54%) were female, while five (46%) were male, which indicates a slight female preponderance. Castroviejo et al.[3], Kromann et al.[7], Hogeling et al.[5] and Cohen et al.[6] revealed of slight female predominance, whereas Pinhero et al.[1] revealed of slight male dominance (53%)

Type of pigmentation:- In this study, 2 types of pigmentary changes were noted. 7 cases (63%) with hyperpigmentation (LWNH) and 4 cases (37%) with hypopigmentation (HI). This is analogous with the studies by Pinhero et al.[1] (76% hyperpigmented, 15% hypopigmented and 7% mixed pigmented cases) and Hogeling et al.[5] (7% with hyperpigmentation and 23% with hypopigmentation). In contrast, the study by Cohen et al.[6]committed that 58% of the cases with hypopigmented macules and 42% with hyperpigmentation. Kromann et al.[7] showed 43% of hyperpigmeted, 50% of hypopigmented and 7% of mixed pigmented cases in their study.

Age of onset:- In the present study, majority of the patients (81.81%) manifested skin pigmentation before 2 years of age. This is almost identical to the studies by Pinhero et al.[1] (92.3%) and Kromann et al.[7](75%).

Extracutaneous manifestations:- In the present study, no extracutaneous manifestations involving CNS, ocular , musculo skeletal and other systems were detected in the patients. According to the study by Metta AK et al.[2] no systemic involvement was seen, whereas the studies by Castroviejo et al.[3], Nehal et al.[4] (30%), Hogeling et al.[5] (7.6%), Cohen et al.[6] (13.9%) and Kromann et al.[7] (56%) revealed extracutaneous manifestations in the cases of pigmentary mosaicism.

VI. Conclusion

According to the literature, the disorders of pigmentary mosaicism i.e. LWNH,HI associate with extracutaneous involvement of CNS, ocular and musculoskeletal system. But the cases included in the study were of pure cutaneous pigmentary mosaicism without extra cutaneous manifestations. Through this study, we conclude that the association of systemic involvement in pigmentary anomalies along the blaschko lines far less often than being reported in the past. Thorough physical examination and investigations must be done to check for the systemic involvement and to advise regarding corrective measures and to counsel the parents. As the

sample size was small, the results couldn't be generalized. Therefore, a study with large number of cases with pigmentary mosaicism must be done.

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