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Rare Case Report on Incontinentia Pigmenti

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1. Introduction

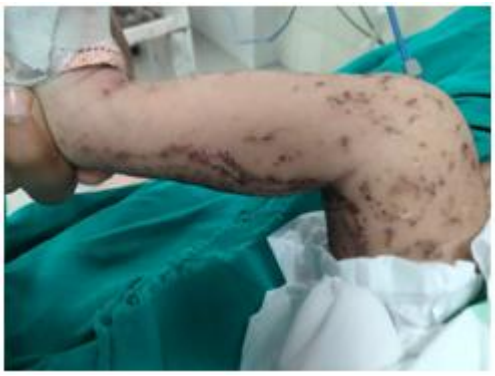
Incognentiapigmenti is a neurocutaneous syndrome. It is a x - linked dominant disorder. It is a rare, inheritable, multisystem ectodermal disorder. Pathogenic IKBKG gene or 47, XXY karyotyping seen in female X - inactivation (45, X) seen. Most commonly involved organ is skin. Other systems involved are CNS, ocular, dental and hair.

2. Case Description

A patient named B/O Savitri / female / day of life 11 was referred to our institute with complaints of skin lesion over arm and leg. Patient also had one episode of seizure (uprolling of eyeballs and tightening of limb). Patient had skin lesion over entire body since birth. On day of life 5, patient became lethargic and had 1 episode of convulsion. At NCHS, patient was started on iv antibiotics andanti epileptic medication. Ophthalmologic evaluation revealed hemorrhagic spot in right eye on slit lamp examination. Skin reference was done and diagnosis of incontinenti pigment was confirmed on the basis of pattern of skin lesions along with other system involved. Also, mother had previous H/O death of previous male fetus in utero.

We would like to report a peculiar finding that was seen in our case. Adrenal hematoma was seen on CT abdomen & pelvis of this patient. Involvement of adrenal glands has previously not been reported in spectrum of multisystem involvement seen in IP. Hence this finding is worth reporting.

All findings were cross checked according to the Major and Minor criteria laid down for diagnosis of IP and the diagnosis was confirmed after consultation with department of Derrmatology of our institute.



3. Discussion

Incognentiapigmenti is a rare X - linked dominant disorders. Most commonly involved organ is skin. Skin lesions have been described in 4 stages according to their onset.

1st stage (Bullous stage) is developed at birth or within a first week of life. It consist of erythematous linear streaks and plaque of vesicles. It is mild, short lived, recurrence of blister.

2nd stage (verrucous stage): - In this stage blisters resolve and become dry, hyperkeratotic and form verrucous plaque.

3rd (pigmentary) stage: - It is the hallmark of incognentiapigmenti. Develops within a week to month. Hyperpigmentation is more on trunk followed by limb, distributed in a macular whorl, reticulated patches, flecks, linear streak follow Blaschko lines. This lesion persist throughout childhood, begins to fade at adolescence and disappear at 16 years of age.

4th stage (Atretic stage): - Lesions are seen mainly on flexor aspect of lower legs. Other systems such as CNS, OPHTHALMOLOGY, DENTAL, HAIR, NAIL are also involved.

Diagnosis primarily depends on clinical grounds. Molecular genetics and skin biopsy are also useful for diagnosis. Following is the diagnostic criteria used for establishing a diagnosis of IncontinentiPigmenti.

Major criteria – Any one of the stages of IP skin lesion as described above.

Minor criteria – Dental anomaly, ocular, CNS, hair, anal, palate, multiple male miscarriage, typical histopathology finding in skin biopsy.

2 major criteria or 1 major + 1 or more minor criteria are required for diagnosis of Incontinentipigmenti.

Genetic confirmation has not been included in diagnostic criteria of IP.

4. Conclusion

A rare case of IncontentiPigmenti was seen at our institute and diagnosis was established based on the following factors:

1) Classical skin lesions, typically stage 3 (pigmentary) as described above, associated with other systemic involvement like seizures and hemorrhagic spots in right eye as seen in this case.
2) History of abortion of male fetus in utero in previous pregnancy.
3) The peculiar finding of adrenal hematoma as seen on CT abdomen of this patient needs further literary review to be included as a diagnostic pointer towards diagnosing Incontentipigmenti.

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