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Incontinentia Pigmenti-A rare multisystem disorder

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Abstract

Incontinentia Pigmenti (IP) also called Bloch-Sulzberger syndrome is a rare X linked genodermatosis, affecting the females and lethal in males. The gene affected is NEMO or IKK gamma gene located on Xq28. It is a multisystem disorder affecting ectoderm-derived structures, including skin, teeth, hair, nails, eyes and central nervous systems(CNS).

This 2 year old girl presented with skin lesions, seizures, severe developmental delay and mental retardation, typical of IP. She had lines of Blaschko noticed at birth followed by Ophthalmic, dental and hair abnormalities.

She was treated conservatively with antiepileptic medications, dental and dermatology consultation and rehabilitation.

Keywords: Incontinentia Pigmenti, X Linked genetic disorder.

The Case

Two year-old girl presented with skin lesions, seizures, severe developmental delay and mental retardation. She was born to third degree consanguineous parents at full term. At birth, she had bullous lesions over trunk and upper limbs, which gradually disappeared in 6months, leaving crusts and hyperpigmented scars. Following which she developed linear brownish hyperpigmentation over the trunk and reticular pattern in lower limbs (Fig. A). Child had global developmental delay and seizures of varying semiology.

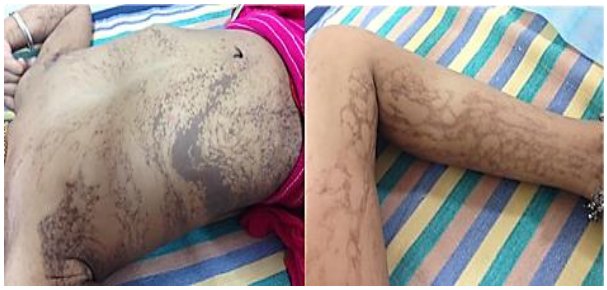

Fig. A

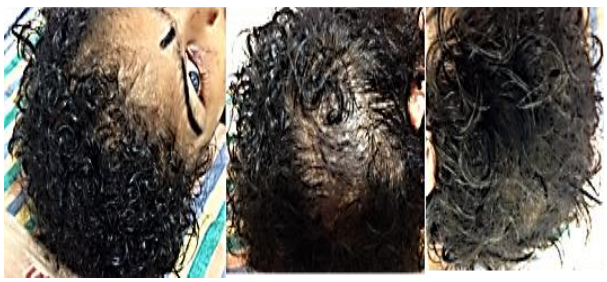

Fig. B

She had corneal opacity in left eye, however optic fundus examination was normal. Hair showed two distinct patterns, curly hair anteriorly and straight hair posteriorly (Fig. B) with areas of alopecia. The clinical manifestation was typical of Incontinentia pigmenti.

Discussion

Incontinentia pigmenti is a rare X- linked dominant condition characterized by abnormalities in ectoderm-derived structures.(2) The clinical manifestations of Incontinentia pigmenti vary considerably even in cases from same family. It ranges from minor cutaneous and dental manifestations to severe ophthalmologic and neurological abnormalities. Classically the disease evolve in 4stages in succession or may occur concurrently. Being an X linked dominant trait, it is usually lethal in males.(3,5) More than 95% of the reported cases are females.(3)

Stages of the disease

First stage: This stage is seen at birth as clear, tense bullae on inflammatory bases in linear groups over the trunk and lower limbs. Inflammatory changes start in utero and never progress after birth. These lesions usually develop on the limbs which tend to follow the lines of Blaschko.(3) They are never found to recur in future. This stage is seen in 90% of the patients.

Second stage: After few weeks, linear verrucous lesions appear similar to the first stage lesion pattern. These lesions also gradually disappear by the age of 6-8 months in 80% of cases.(3)

Third stage: Development of brownish hyperpigmented streaks over trunk and lower limbs. These lesions can develop in areas unrelated to the sites of lesion during the previous stages that follow the lines of Blaschko.(3)

A fraction of patients (14%) exhibit a fourth stage which appears as residual hypo-pigmentation in the areas of previous hyperpigmentation.(3)

Dental abnormalities are the most common non cutaneous manifestation of IP, occurring in more than 80% of the cases. Most common dental abnormalities include alodontia, cone or peg shaped teeth, hypodontia and delayed dentition.(3,6)

Ophthalmic defects can occur in 40% of the IP cases. Asymmetrical involvement is more common.[3] Common defects include strabismus, cataract, conjunctival pigmentation, uveitis, optic atrophy, retinal vascular abnormalities, blue sclerae, corneal opacity (Fig. B), exudative chorioretinitis and microphthalmia.

Although hair abnormalities are not common in IP, cicatrical alopecia is reported in upto 25% (Fig. B).

Central nervous system abnormalities occur in 25% of IP cases, which include seizure, developmental delay, mental retardation, spasticity, cerebral atrophy, hemiparesis and encephalopathy.[3]

Diagnosis of IP is mainly dependent on history and clinical findings. Genetic study is helpful to confirm the diagnosis. Detailed ophthalmological evaluation is essential. MRI or CT scanning of brain, EEG, Dental X- Rays, Bone X-Rays may guide in further management.

Inflammatory lesions in the early days may require antibiotic therapy and other supports.[7] Supportive therapies include antiepileptic and anti-spastic medications. Corrective surgeries for dental, bone and ophthalmic defects (less helpful), may be recommended.[6] Good Physical therapy and nutritional supplementation may be required in majority.

Prognosis of the condition depends on the systems affected and severity. Better outcome is expected, if vision and central nervous system are spared. Survival rate is high in females.

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