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Case Report

Incontinentia pigmenti with ocular, cutaneous and CNS manifestation

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ABSTRACT

Incontinentia Pigmenti (IP) is an uncommon X-linked genodermatosis, with an estimated prevalence at birth of 0.7/100,000, caused by mutations in the NEMO gene. Ectodermic and mesodermic origin of tissue is seen in this systemic disease including cutaneous tissue, teeth, eyes, and the central nervous system. Herein, we present a case of a female newborn with inflammatory vesiculopustular lesions all over the body. This baby also had ocular, and CNS manifestations as well. The importance of a detailed diagnostic workup for the newborns with pustular skin disease has been highlighted in this case. IP is a rare, x-linked dominant genodermatosis with the involvement of multiple organs. Dermatological abnormalities are the most prominent manifestation. The diagnosis is based on the clinical findings of skin lesion brain imaging and biopsy. The skin lesions do not require specific treatment and prognosis depend on other organ involvement.

Keywords: Bloch-Sulzberger syndrome, Incontinentia pigmenti, Newborn, Pustular skin lesions, X-linked, retinal detachment

INTRODUCTION

Incontinentia pigmenti (IP), first described by Garrod in 1906, after that Bloch and Sulzberger defined this disorder in 1926 and 1928, respectively, is a multisystem disorder inherited in an X-linked dominant fashion with lethality in males. The skin lesions are diagnostic and occur in four stages, all of which may not be seen in one patient. IP can have involvement of other systems which include the central nervous system seen as intellectual disability, microcephaly, stroke, and seizures. The ocular changes are ischemia of the peripheral retinal field, retinal dysplasia, retinal detachment, pigment retinopathy, and retrolental dysplasia. The musculoskeletal system can have hemivertebrae, kyphoscoliosis syndactyly, and hemiatrophy. Dental abnormalities include hypodontia, microdontia, and dysplasia. Hair may be coarse, wiry, lusterless with alopecia.[1]

Transmission of the disease in females by a lionization results in functional mosaicism of X-linked genes. Vesicular stages of IP are present at birth or develop in the first few weeks of life in most cases. The characteristic stages are blistering (from birth to about 4 months of age) followed by verrucous plaques (for several months), swirling macular hyperpigmentation (from about 6 months and persist during childhood and usually fade by adolescence) and in later stages linear hypopigmentation (that develops during adolescence and early adulthood which persists indefinitely).[2]

Hair, nail, and dental anomalies are often manifested during infancy. Permanent neurologic (Cognitive delays/mental retardation) and ophthalmologic sequels develop during early infancy so that patients would have retinal vascular abnormalities predisposing to retinal detachment in early childhood but strabismus and cataract are occasionally seen. Breast, skeletal, and structural anomalies are sometimes rarely seen in patients.[2]

CASE REPORT

Female baby born by normal vaginal delivery to Primi mother with third degree consanguineous marriage, no antenatal issues, no history of fever with rash anytime during pregnancy or delivery nor any history of contact with anybody having rash. She presented with a history of rash since day 1 of life. Rash was in the form of fluid-filled vesicles over the upper limb, lower limb, and trunk. Day 5 of life baby developed poor feeding with intermittent sucking and reduced activity. Day 8 of life baby had one episode of seizure in the form of rhythmic contractions of both upper and lower limbs which lasted for few seconds. LP done was normal. Sepsis screen done was negative. Neurosonogram showed cerebral edema. CBC showed eosinophilia and CRP, VDRL, and HSV PCR was negative [Figure 1a-d].

Dermatology consultation was done followed by Punch biopsy which showed mild hyperkeratosis with subcorneal vesicle containing plasma, neutrophils, and eosinophils. The dermis shows perivascular infiltrates of few eosinophils [Figure 2a and b].

MRI brain reported multiple patchy foci of diffusion restriction in the subcortical white matter of both frontal, right parietal regions and deep white matter of right frontal region appearing hyperintense on FLAIR- subacute infarcts. Linear foci of T1 hyperintensity in the subcortical and deep white matter of both cerebral hemisphere-hemorrhages. Few irregular cystic areas in bilateral parietal left frontal subcortical white matter. Diffuse altered signal intensity of white matter. Corpus callosum appears hypoplastic with findings suggestive of IP [Figure 3a and b].

Ophthalmology evaluation of fundus in the early neonatal period showed patchy retinal hemorrhages. On subsequent follow-up at 9 months showed evolving Retinal detachment with secondary glaucoma of the left eye.

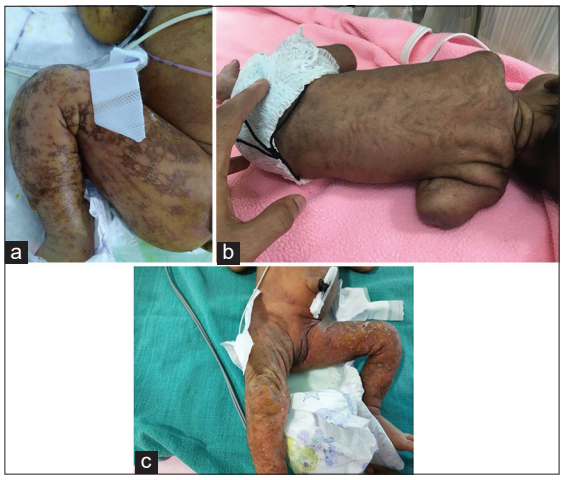

Figure 1: (a) Whorl pattern. (b) Skin lesions along lines of Blaschko. (c) Vesicopustular eruptions.

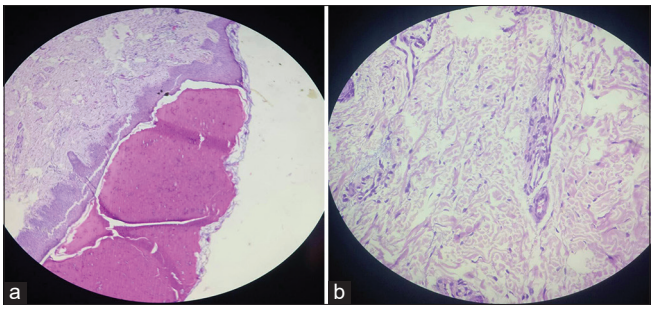

Figure 2: (a and b) Histopathology.

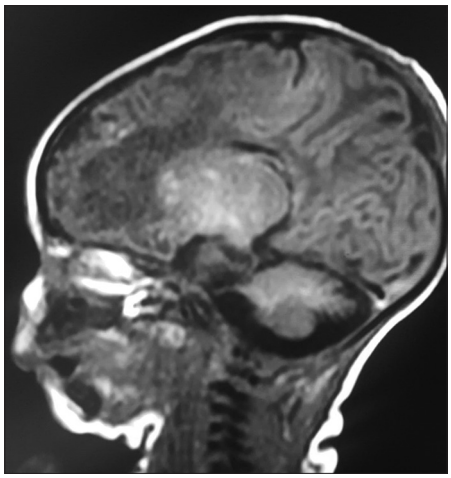

Figure 3: MRI ,corpus callosum agenesis.

DISCUSSION

The IP as an X-linked dominant disorder is lethal in males.

IP is a rare multisystem disease, which is characterized by the abnormalities of the tissues and organs which are embryonically derived from the ectoderm and neuroectoderm.[3] The pigment melanin that normally lies in the melanocytes of the basal epidermal layer is seen in the superficial layer in the IP. The skin changes in IP are the major criteria for diagnosis of this disorder. These variations typically occur at birth or during the 1st weeks of life and continue to adulthood while distributing along Blaschko's lines which were typical in our case.

	System	Evaluation and timing	Expected findings
Neonatal period	Ocular	Fundoscopy	To evaluate the optic nerve, macula, and far periphery
	CNS	MRI brain	Ischemic changes, stroke
	Cardiac	Echo	Pulmonary hypertension
	Skin	Biopsy	Eosinophilia
	Genetics	Genetic testing	x-linked inheritance
Infancy through age 3 years	Ocular	Fundoscopy: Every month until 4 months 4 months-1 year: every 3 months 1 year-3 year: Every 6 months Annually lifetime	Retinal detachment
	Dental &speech	Examination at 6 months	Hypodontia, conical teeth, affected speech
	CNS	Developmental assessment and EEG if seizure occurs	Developmental delay
	Genetics	Evaluated at puberty and prenatally	Each pregnancy will carry a 50% risk of receiving the mutation

The diagnostic criteria for IP was proposed by Landy and Donnai in 1993. In our case, baby was born to parents with third-degree consanguinity, but no family history of any genetic conditions was reported.[4]

The histopathological findings in our patient were compatible with the early phase. The skin biopsy report showed an eosinophilic predominance which is a typical characteristic of IP.

The central nervous system is the most affected system after the skin in IP patients, which is about 10–40% of the cases. Central nervous system involvement can have a major impact on the patient's quality of life, even though the baby had significant changes in MRI she had attained early milestones on time. There has been an early study which showed reversible brain changes in patients with IP.

In 30-70% of the IP patients coular diseases such as strabismus, microphthalmia, and pigmentary retinal changes are seen. Vision loss has been associated with vascular occlusions, secondary extraretinal neovascularization, fractional retinal detachment, and foveal hypoplasia. In our patient routine screening in the neonatal period showed retinal hemorrhage, hence regular follow-up was arranged for fundoscopy wherein early detection of retinal detachment and secondary glaucoma was made.

Hair abnormalities in IP (e.g., alopecia, sparse hair, as well as hypoplasia of eyebrows and eyelashes), had been reported in 28–38% of the patients. Scarring alopecia of the vertex is the most common manifestation of hair involvement.

The IP can also involve nails and lead to such abnormalities as dystrophy and fibromas. Nevertheless, no hair or nail involvement was observed in our case.

The diagnosis of IP is based on the clinical features. Differential diagnosis including neonatal herpes simplex infection, congenital syphilis, impetigo, neonatal bullous dermatoses, and autoimmune blistering were considered. Since the spontaneous resolution of the lesions usually occurs, the skin lesions of IP do not require specific treatment.

As discussed IP had a multisystem involvement, with cutaneous, CNS, and ocular manifestations. Hence a multidisciplinary approach is required for management. It has been hence evident that regular ophthalmologic follow-up is also essential in those cases with IP for timely detection of vascular changes and prompt treatment. Successful use of dexamethasone has been reported in one highlighted case to treat seizures supports the idea that inflammation is a part of the process. Unfortunately, a treatment protocol using dexamethasone in this population is difficult to establish given the very small numbers of infants born with the condition.[5] More of cases reported and research is required in this regard for an early detection, screening, and genetic counseling.

Recommendations for evaluation and follow-up of IP

CONCLUSION

As discussed IP had a multisystem involvement , with cutaneous,CNS and ocular manifestations. Hence a multidisciplinary approach is required for management.It has been hence evident that regular ophthalmologic followup is also essential in those cases with IP for timely detection of vascular changes and prompt treatment . Successful use of dexamethasone has been reported in one highlighted case to treat seizures supports the idea that inflammation is a part of the process.

Declaration of patient consent

Patient's consent not required as patients identity is not disclosed or compromised.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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