BLOCH-SULZBERGER SYNDROME (INCONTINENTIA PIGMENTI): A CASE REPORT.

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ABSTRACT

Incontinentia pigmenti (IP) is a X-linked dominant neuro cutaneous syndrome with cutaneous, neurologic, ophthalmologic and dental manifestations mainly in female neonates. Starting from neonatal period, IP passes through stages of vesicular (90%), verrucous (70%), hyper pigmented (98%) and hypopigmented lesions. Authors report a case of IP in 8 months old female child who presented with vesiculo bullous skin eruption and verrucous lesions on hand and feet. The diagnosis was confirmed on histopathology. Variable pattern of lesions presenting over a period of time makes it difficult to diagnose. As disease is incurable, genetic counseling is of paramount importance.

KEYWORDS: Bloch-Sulzberger syndrome in continentia pigmenti, X-linked, Genetic diseases, pigmentation disorders.

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INTRODUCTION

Incontinentia pigmenti (IP) is a rare X-linked dominant neuro cutaneous syndrome with changes in skin, central nervous system, eyes, hair and dental regions. Garrod (1906) report was the first to incontinentia pigmenti as a peculiar pigmentation of the skin in an infant. Bloch and Sulzberger described the pathogenesis of IP in 1926. Mutation of NEMO (NF-Kappa- B essential modulator), gene located on chromosome Xq 28 plays a key role in the pathogenesis of the disease. It effects many systems along with skin expressing clinically with variable features. There were around 800 registered cases of IP with a estimated incidence of one in every 40,000 children as quoted by Gabriela F M, Claudio S T, and Juliana M P. The present case is being reported due to its rarity and variable patterns of presentation during natural course of disease making diagnosis tricky. Various associated affection of different organs like teeth, eyes and nervous system makes management complex.

CASE REPORT

An 8 months old female infant was born at full term by normal vaginal delivery with no history of consanguinity in the parents presented with variable skin lesions when the lesions were first observed at the age of one week after birth. There was no family history of such illness and pregnancy was without any complication. On examination she had vesiculobullous skin eruption with erythematous base on the trunk and dorsum of right little finger and verrucous lesion on dorsum of hand and feet. (Figure 1, 2, 3). The infectious causes were excluded. Tzank test showed no giant cells and test for antibodies for HSV, V2V was negative. Systemic examination including that of eyes, central nervous systems and skeletal system was essentially normal. Complete haemogram, revealed raised eosinophils on differential leucocytes count. VDRL of mother and child was non-reactive.

Figure 1
Photograph showing vesiculobullous skin eruption with erythematous base on the dorsum of right little finger.

Figure 2
Photograph showing vesiculobullous skin eruption with erythematous base on the trunk and dorsum of right little finger and verrucous lesion on dorsum of hand and feet.
A skin biopsy was taken from dorsum of right hand. Histopathology revealed spongiotic intra-epidermal vesicles with a rich eosinophilic infiltrate leucocytes and eosinophils in vesiculo bulbous lesion. There was hyperkeratosis, dyskeratosis, acanthosis and papillomatosis. (Figure 4,5). The epidermis shows moderate hyperkeratosis, acanthosis, mild elongation of rete ridges and presence of few characteristic intraepidermal small, round to oval shape vesicles containing inflammatory cells, each vesicle separated by a thin bridge of keratinocytes. The dermis also shows similar inflammatory infiltrate.

Histopathology of Incontinentia pigmenti lesion in our case Figure 4. The epidermis showing mild hyperkeratosis, acanthosis, elongation of rete ridges, marked spongiosis and few bullae. The dermis showing chronic inflammatory cell infiltration incl. eosinophils.
from vision impairment epidermolysis bullosa simplex herpitiformis and suffered nails sometimes scar-like lesions are found mostly on lower upto adulthood. Pale, hairless patches or streaks, hypo pigmented) occurs in adolescence and persists the form of vertex alopecia, ridged, pitted or dystrophic of the epidermis with the absence of skin appendages in Histopathologically it represents as atrophy or thinning in 90% of patients. It is characterized by a rash of erythematous blisters, which appear in groups along the lines of Blaschko the differential diagnoses at this stage includes Bullous impetigo, Varicella zoster, Epidermolysis bullosa, Herpes simplex, Bullous mastocytosis, Congential bullous pemphigoid, Epidermolytic hyperkeratosis4. Biopsy characteristically exhibits spongiotic dermatitis with massive intraepidermal and dermal eosinophils5. Stage 2 or verrucous stage is observed in 70% cases and clinically presents as eruption of hyperkeratosis verrucous papules and plaques develops over the healing blister. It usually appears within 2 months and disappears within 6 months. Histopathologically hyperkeratosis, dyskeratosis, acanthosis and papillomatosis are present in this stage. Both stages 1 & 2 were observed in the present case. Stage 3 or hyper pigmented is classically the hallmark of IP with overall incidence of 98%. Pigmentation ranges from blue-gray or slate to brown streaks or whorls. It generally develops within the first few months of life and tends to fade by adolescence. Melanophages in the dermis and vacuolization of basal cells is the most common finding 6. Stage 4 (catrophic/hypo pigmented) occurs in adolescence and persists upto adulthood. Pale, hairless patches or streaks, sometimes scar-like lesions are found mostly on lower legs which are permanent signs in adults. Histopathologically it represents as atrophy or thinning of the epidermis with the absence of skin appendages in the form of vertex alopecia, ridged, pitted or dystrophic nails7. Treatment for cutaneous lesion is usually not required, but there is evidence of rapid resolution of the inflammatory stage by the use of topical tacrolimus and corticosteroids8-9. Vesicles should not be punctured and secondary skin infection is checked by topical antibiotics. An extracutaneous manifestation in IP patients varies from 70% to 80%. Dentals lesions being the most common which affect about 80% of patients in the form of delayed dentition, partial anodontia, cone or peg shaped teeth or absence of teeth. Rafatjou et al reported clinical and radiological findings in a case of seven year old child with IP with involvement of teeth and multidisciplinary treatment approach in five years follow up10. Shanker et al also reported a rare case of IP with dental defects which was earlier diagnosed as epidermolysis bullosa simplex herpitiformis and suffered from vision impairment11. It has been reported that herpes simplex should be considered as differential diagnosis of IP in neonates12. Need of oral hygiene and regular visits to dentist cannot be over emphasized.

Dental restoration may be required. There is 30 % to 50% neurologic involvement in the form of seizures, mental retardation, spastic paralysis, ataxia, motor dysfunction and developmental delays 13. Anti convulsants should be used for seizures and occupational therapists should be involved early for assessments of neuro development. Systemic steroids may help in reducing seizure frequency in neonates with encephalopathy.14, 15 Ocular abnormalities are observed in about 47% cases as strabismus, cataract, optic atrophy, retinal dysfunction, ileitis, nystagmus and blindness16. Early ophthalmological evaluation is of paramount importance for early detection of retinal and other ophthalmological problems in first year of life itself Cryosurgery or Xenon laser can treat retinal fibrovascular proliferation17,18. Vitero retinal surgery may be required to treat detachments in retina. Skeletal anomalies have occasionally been reported like somatic asymmetry, skull deformities, spina bifida dwarfism, syndactyly and extra ribs. Sometimes primary pulmonary hypertension and cardiopulmonary failure are also reported by many workers6. Keratotic tumors in late adolescence are occasionally associated with, which sometimes involutes spontaneously19. There are no standardized diagnostic criteria for IP yet, however, diagnostic criteria for incontinentia pigmenti include major and minor criteria. Major criteria are: typical neonatal vesicular rash with eosinophilia, blaschkoid hyperpigmentation on the trunk, fading in adolescence, and linear atrophic hairless lesions. Minor criteria include: dental anomalies, alopecia, wooly hair, and abnormal nails with a definitive family history. The presence of any major criteria strongly supports the diagnosis of incontinentia pigmenti. In the absence of a family history the presence of at least one major criterion is necessary. The presence of minor criteria supports the diagnosis. The diagnosis is mostly based on the characteristic skin lesions and clinical involvement of other systems. Skin biopsy and molecular genetic testing of the NEMO gene confirm the diagnosis. Genetic study was not carried out in present case due to lack of facility. Family history or history of multiple miscarriages also support diagnosis of IP. Rashidghamat et al described a case of IP in a father and daughter20.

CONCLUSION

Disease is rare and variable pattern of lesions presenting over a period makes it difficult to diagnose. There by one should know all patterns of presentation at various stages of life. Genetic counseling is important as disease is incurable.

CONFLICT OF INTEREST

Conflict of interest declared none.
REFERENCES


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