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Incontinentia Pigmenti in an Infant Suspected of Retinoblastoma

Seyedeh Maryam Hosseini [1], Ghodsiyeh Zamani [1], Hossein Ghavami [1], * Mehrdad Motamed Shariati [1]

[1] MD, Eye research center, Mashhad University of Medical Sciences, Mashhad, Iran.

Abstract

Purpose: This case study reports on an infant presented with strabismus and poor red reflex, suspected of retinoblastoma, and finally diagnosed as a case of Incontinentia pigmenti (IP).

Case presentation: A 7-month-old infant was referred due to poor fixation and abrupt red reflex of the left eye for further evaluation of retinoblastoma. In anterior segment examinations, we found a micro cornea, a relatively shallow anterior chamber, and hypotonia. Fundus examination revealed a total tractional retinal detachment with vitreous membranes. In her skin examination, we discovered diffuse cutaneous linear pigmentation (lines of Blaschko), consistent with Incontinentia Pigmenti (IP).

Discussion: A wide range of disorders involving the fundus, cause similar signs and symptoms to retinoblastomas, such as leukocoria and poor red reflex, categorized as pseudo retinoblastoma. IP is a dysplastic ectodermal disorder with dominant X-linked inheritance, affecting integumentary, ocular, nervous, and dental tissues, and is responsible for less than 1% of cases of pseudo retinoblastoma. Although rare, achieving early diagnosis for IP is of utmost importance, since not only would it enable clinicians to treat and manage retinal complications, but it would also help them detect potentially fatal neurological issues.

Key Words: Incontinentia pigmenti, Pseudoretinoblastoma, Retinoblastoma.

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Corresponding Author:

Mehrdad Motamed Shariati, MD, Eye research center, Mashhad University of Medical Sciences, Mashhad, Iran. Email: mehrdad_shariati2005@yahoo.com

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1- INTRODUCTION

Incontinentia pigmenti (IP) is an X-linked dominant genetic ectodermal dysplasia defined by characteristic skin pigmentary changes, as well as ocular, neurologic, and dental abnormalities (1). Mutation in the NEMO/IKKgamma gene leads to eosinophil accumulation in tissue through eotaxin activation (2, 3). This mutation is lethal in males. **Table 1**, summarizes the clinical findings in patients with IP.

Table-1: Common abnormalities found in patients with IP

Abnormality	
Skin	A staged rash that starts as erythema, vesicles, and pustules at birth and progresses to verrucous lesions and flat hyperpigmentation over time. Linear, atrophic, hypo-, and hyper-pigmented reticular streaks are the late findings.
Dental	Widely spaced teeth
Neurologic	Mental retardation Corpus callosum hypoplasia Seizures Cerebral atrophy
Ocular	Retinal vascular non-perfusion and neovascularization Tractional retinal detachment Retrolental fibrous tissue Foveal hypoplasia Retinal pigmentary change

Ocular abnormalities in IP are reported in 50-77% of patients. Previous studies have also shown a relationship between ocular findings and neurologic manifestations; and have proposed an eosinophilic vaso occlusive pathogenic mechanism (4). Scarcely, IP may present with retinoblastoma, the most common primary intraocular malignancy in children, with manifestations including leukocoria, retinal detachment, and calcification (5).

In this report, we introduced a seven-month-old girl presented with strabismus and poor red reflex, suspected of retinoblastoma, who was finally diagnosed as a case of IP.

2- CASE PRESENTATION

A 7-month-old girl was referred to Khatam-Al-Anbia eye hospital due to poor fixation, strabismus, and poor red reflex in her left eye (LE), suspected of retinoblastoma. She was born full-term at 39 weeks of gestational age. Because of poor cooperation, we examined her under general anesthesia. The corneal diameter was 11mm for the right eye (RE) and 10mm for the LE. The intraocular pressure (IOP) was ten and 7mmHg for RE and LE, respectively. In the anterior segment examination, we found a relatively shallow anterior chamber (AC) of the LE. The crystalline lens was clear in both eyes. In the fundus examination, there was a total tractional retinal detachment (RD) and vitreous opacities in the LE and normal vasculature of the RE. B-scan ultrasonography confirmed a total tractional RD with no calcification. In her skin examination, we discovered diffuse cutaneous linear pigmentation (lines of Blaschko), consistent with Incontinentia Pigmenti (IP) (**Fig. 1**). There was no suspected family history.

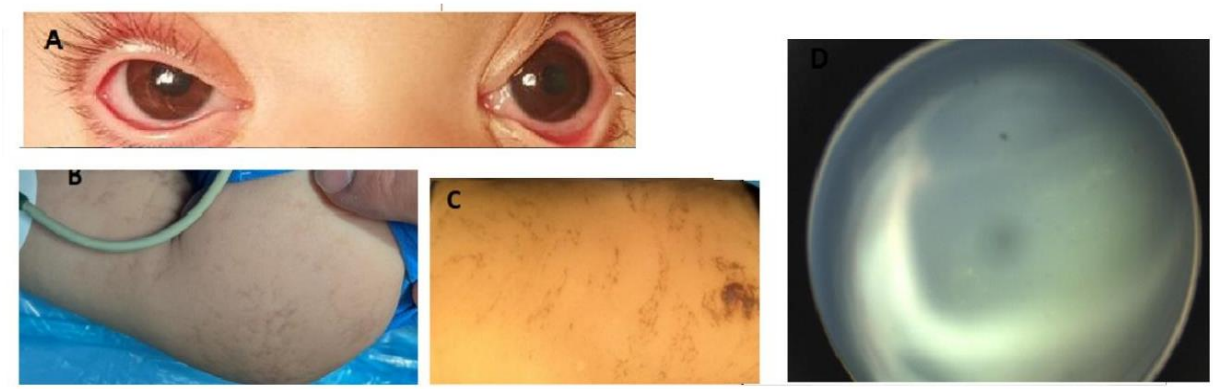

Fig 1: A: Photographs of the eyes show a relatively shallow AC in the LE; B, C: Characteristic skin pigmentary changes; D: Tractional retinal detachment and vitreous haziness

3- DISCUSSION

Pseudoretinoblastoma includes a wide range of disorders involving the fundus, which cause similar signs and symptoms to retinoblastomas, such as leukocoria and poor red reflex (6). Retinoblastoma is the most common intraocular malignancy in children, which is potentially life-threatening. Shields et al., in 2012, reported the conditions simulating retinoblastoma. They showed that Coat's disease and persistent fetal vasculature are the most prevalent among these etiologies, while IP is responsible for less than 1% of cases (5, 7).

IP is a dysplastic ectodermal disorder with dominant X-linked inheritance, affecting integumentary, ocular, nervous, and dental tissues. First observed and very suggestive of the underlying illness are the cutaneous symptoms (**Table 1**). Pegged and widely spaced teeth, as well as anodontia, are among the dental anomalies that are present in 80% of cases. Dental assessment is advised by the age of two years old for those who experience them; such anomalies are often seen at the time of tooth emergence. Analyses of the mother and other female siblings may reveal similar dental problems (8).

Although rare, achieving early diagnosis for IP is of utmost importance, since not only would it enable clinicians to treat and manage retinal complications, but it would also help them detect potentially fatal neurological issues. Previous research has shown correlations between neurological and ophthalmologic manifestations; they, generally, share a common pathogenetic process, known as eosinophilic vaso occlusive disorder (3). Vaso Occlusive events in the retina lead to tissue ischemia, increased vascular endothelial growth factor (VEGF) production, and subsequent retinal neovascularization. Different approaches are used to treat ocular complications to reduce peripheral nonperfusion and the associated neovascularization of the retina, which in turn prevents tractional RD. However, in a study by Chen et al., in 2015, it was shown that in three out of four patients with IP, tractional RD occurred despite undergoing retinal laser photocoagulation(4).

4- CONCLUSION

In this study, we reported an infant suspected of retinoblastoma with characteristic skin pigmentary changes for IP and no sign of retinoblastoma. We showed that retinal manifestations of IP could mimic retinoblastoma.

5- ETHICAL CONSIDERATIONS

Consent for publication was acquired from the legal guardian of the patient. The datasets are published anonymous and are available from the corresponding author upon reasonable request.

6- COMPETE OF INTERESTS

None.

7- REFERENCES

1. Scheuerle AE, Ursini MV. Incontinentia pigmenti. 2017.

2. Poziomczyk CS, Recuero JK, Bringhenti L, Maria FDS, Campos CW, Travi GM, Freitas AM, Maahs MAP, Gazzola Zen PR, Fiegenbaum M, Almeida STd, Bonamigo RR, Kiszewski Bau AE. Incontinentia pigmenti. Anais brasileiros de dermatologia. 2014; 89:26-36.

3. Berlin AL, Paller AS, Chan LS. Incontinentia pigmenti: a review and update on the molecular basis of pathophysiology. Journal of the American Academy of Dermatology. 2002; 47(2):169-90.

4. Chen CJ, Han IC, Tian J, Muñoz B, Goldberg MF. Extended follow-up of treated and untreated retinopathy in incontinentia pigmenti: analysis of peripheral vascular changes and incidence of retinal detachment. JAMA ophthalmology. 2015; 133(5):542-8.

5. Shields CL, Schoenberg E, Kocher K, Shukla SY, Kaliki S, Shields JA. Lesions simulating retinoblastoma (pseudo retinoblastoma) in 604 cases: results based on age at presentation. Ophthalmology. 2013; 120(2):311-6.

6. Holmström G, Thorén K. Ocular manifestations of incontinentia pigmenti. Acta ophthalmologica Scandinavica. 2000; 78(3):348-53.

7. Shields CL, RALPH C EAGLE J, Shah RM, Tabassian A, Shields JA. Multifocal hypopigmented retinal pigment epithelial lesions in incontinentia pigmenti. Retina. 2006; 26(3):328-33.

8. Hadj-Rabia S, Froidevaux D, Bodak N, Hamel-Teillac D, Smahi A, Touil Y, Fraitag S, Prost Yd, Bodemer C. Clinical study of 40 cases of incontinentia pigmenti. Archives of dermatology. 2003; 139(9):1163-70.