Pachyonychia Congenital: A Rare Case Report

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Abstract

We are reporting a rare case of Pachyonychia Congenital in a child who was presented in a specialized eye institute with fleshy growth in super lateral part of ocular surface of both eyes with bloody discharge & recurrent blister like skin lesions and nail deformity in both extremities. There was also a non-healing ulcer in her right cheek. He had a swelling on his both super lateral aspects of orbit just under the upper lid which underwent excision biopsy and histopathology report reveals Pachyonychia Congenital. That swelling was recurrent and we did the repeated biopsy from that site and tissue was sent for histopathology. It usually began within the first few months of life.

Conclusion

Here, we are presenting a rare case, which started at the age of 1 year & 5 months of life. The case is being reported for its rarer occurrence as the patient had conjunctival growth, Corneal Dyskeratosis, Cornea Dystrophy & skin blister which is still uncommon.

Keywords

Conjunctival Growth; Corneal Dyskeratosis; Cornea Dystrophy; Skin Blister; Pachyonychia Congenital.

Introduction

Pachyonychia Congenital is a rare, but well-characterized autosomal dominant disorder of keratinization. Keratinisation is the process by which keratin (scale) is formed and deposited in the outer most layer of the skin. Pachyonychia Congenital occurs when there is a mutation in the genes encoding keratin [1-3].

Pachyonychia congenital occurs when there is a mutation in the genes encoding keratin, K6a, K16, K17, K6b and possibly K6c (listed in decreasing frequency) [4-6]. Nearly 100 mutations have been found [6, 7]. The mutations cause the skin to be more fragile than usual.

Prior to genetic testing, some patients were diagnosed with pachyonychia Congenital without having this disorder. Mutations in other keratin genes can lead to similar skin conditions. The condition is caused by genetic mutations in one of four genes that encode Keratin Protein specific to the epithelial tissues affected in the two forms of the disorder. PC1 is caused by mutations in Keratin 6 A (protein name K6A; gene name KRT6A) or Keratin 16 (protein K16; gene KRT16). The PC2 form is due to mutations in the genes encoding Keratin 6 B (protein name K6B; gene name KRT6B) or Keratin 16 (protein K17; gene KRT17), PC4 results from mutations in the keratins 16 and 17 genes [1, 4, 5]. Pachyonychia Congenital is a rare type of ectodermal dysplasia further classified into 4 types [3].

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Received October 10, 2017; Accepted November 22, 2017; Published November 30, 2017

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• Pachyonychia Congenital type I (also known as "Jadassohn–Lewandowsky syndrome is an autosomal dominant keratoderma that principally involves the plantar surfaces, but also with nails changes that may be evidenced at birth, but more commonly develop within the first few months of life[1,2,3,8].
• Pachyonychia Congenital type II (also known as "Jackson–Lawler pachyonychia Congenital" and "Jackson–Sertoli syndrome") is an autosomal dominant keratoderma presenting with a limited focal plantar keratoderma that may be very minor, with nails changes that may be evidenced at birth, but more commonly develop within the first few months of life[1,3]. Pachyonychia congenital type 3(Schafer –Branuer) has features of corneal leukokeratosis.
• Pachyonychia congenital type 4(PC tarda)-results from mutations in the keratins 16 and 17 genes [1, 3, 4, 5].

Case Report
A male Baby 1 year and 5 months of age, only one child of parents, was presented to us with the complaints of
• Fleshy growth in her both upper lateral part of ocular surface with bloody discharge for 10 months.
• Nails deformity in her both extremities for 14 months.
• Blister like skin lesion for 10 months.
• Rupture of large blister in her right cheek with recurrent self injury and bleeding from that site with poor healing.

Her mother stated that, after 2 weeks of his birth, he found a small blister on her baby’s dorsal aspect of thumb. Then gradually it became turning into pustule and after that it was busted and healed spontaneously. Then after 2-3 weeks, another blister formed on another finger close to nail-bed and spontaneously healed. After one week almost all the nails of his hands and feet became swollen and pus was seen under the nail-bed.

Figure 1-2: Swollen nails and pus under the nail-bed

Then the parents went to local doctor and took medication for him. After some days the pus of nail-bed becomes busted and there was foul smells discharge coming from his nails. It recurrently occurred for 2 to 3 times. At the age of 1 year, the nails of herboth extremities became deformed, friable and recurrent bloody and pus like discharge from there. Parents also gave history of generalized skin blister especially on cheek, face, ear and extremities (thigh) which later formed pus and spontaneously burst. Self injury causes bleeding from those areas. There was soft tiny swelling on her both super lateral aspect of orbit, just below the upper lid, at the age of one year.

Then firstly she took treatment from Dhaka Medical College. They operated it by simple excision. But after some weeks it again occurred.Then he came to the National Institute of Ophthalmology and Hospital for better management.

He (baby-Moon) is the first and only child of his parents. There is no H/O consanguinity of Marriage. She has properly fulfilled his Immunization Schedule

On Inspection
• There is soft conjunctival swelling on his both super lateral aspect of orbit, just below the upper lid.
• Skin blister in his right thigh and face.

Figure 3: Conjunctival swelling super lateral aspect of upper eyelid

Figure 4: Skin blister in the face
• Blister with secondary ulcer on his cheek and ear.
• Thickening and friable nails of feet (Figure. 6)
• Koilonychias on her hands nails.

Investigations

- Previous investigation was histopathology of excised tissue biopsy from Ocular conjunctival mass. And the result was - Pachyonychia Congenital.
- Other Investigations we should do - Genetic analysis of keratin.

Probable Diagnosis

- Pachyonychia Congenital.

Treatment

- Excision of conjunctival mass with histopathology

Discussion

Pachyonychia Congenital (PC) is a group of rare, inherited ectodermal disorder associated with mutations in keratin genes of K6a, K6b, K16 or K17 [4, 5, 8-10]. The most prominent clinical features of PC are nail dystrophy and dyskeratosis of skin and oral mucous membrane [11, 12]. Pachyonychia Congenital is commonly autosomal dominant mode of inheritance; there are also reports of autosomal recessive inheritance as well [13].

Pachyonychia Congenital has been classified into four types, Patients with type 1 Pachyonychia Congenital (also known as "Jadassohn–Lewandowsky syndrome) are characterized by the presence of nail dystrophy since birth accompanied with painful paronychia, hyperkeratosis of palms and soles over the pressure sites, oral leukokeratosis, palmoplantar hyperhidrosis and follicular keratotic papules distributed throughout the body, painful blisters also develop over palms and soles. Another characteristic found is the presence of verrucous lesions over the elbows, knees, popliteal fossae, and ankles [8, 11, 12, 14-17]. In addition hoarseness of voice is also a feature of PC type 1 [14, 18]. It is the more frequent variety [11, 12].

Pachyonychia congenital type II (also known as "Jackson–Lawler pachyonychia Congenital" and "Jackson–Sertoli syndrome"). has the features of type 1 but Oral leukokeratosis and palm planter keratoderma is milder in comparison to type 1PC, additional feature are presence of natal teeth, hair anomalies including pili torti, unruly hair, and bushy eyebrows. Epidermal cysts or steatocysts are the hallmark finding in type 2 PC [8, 11, 12].
Pachyonychia Congenital type 3 (Schafer – Branuer) has features of type 2 and corneal leukokeratosis, cataracts, angular cheilosis [8, 15]. Pachyonychia Congenital type 4 (PC Tarda) – results from mutations in the keratins 16 and 17 genes. Consists of sign symptoms of type I, II, III and laryngeal lesions, hoarseness, mental retardation, hair anomalies, alopecia [9, 10].

The clinical features in our case with characteristic subungual hyperkeratosis and follicular papules over the entire body with painful pachyonychia have similarities with type 1 but palm planter hyperhidrosis and other feature like presence of verrucous lesions over the elbows, knees, popliteal fossae, and ankles are absent in this case. In comparison to type 2 PC natal teeth, hair anomalies, epidermal cysts or steatocysts which are the hallmark finding of type 2 PC are absent. Lachman et al. [12] found corneal dystrophy in type 2 PC. There is corneal dyskeratosis & other clinical finding suggestive of PC type 3 [8, 11, 12]. Corneal dyskeratosis and cataract are hallmark finding of type 3 PC [11]. Lachman et al. [12] found laryngeal lesions, hoarseness, mental retardation, hair anomalies, alopecia are the hallmark finding of type 4 but absent in this case.

This case is very rare as the presence of conjunctival growth which on repeated histopathological finding reveals orthokeratotic hyperkeratosis and acanthosis which confirm our diagnosis as PC, not yet reported in any case [13].

PC with unusual features has been noted [14]. New type of PC with the thickening of all nails in association with severe generalized hypotrichosis has been reported [15]. A case of PC at 6 years of age [16]. Rare cases of pachyonychia congenita tarda have been reported with symptoms developing in the fifth decade of life [17]. Recently, a case of pachyonychia Congenital associated with B-cell lymphoma has been reported [18]. Cases with unusual dental findings have also been reported. An interesting case of PC with wooly hair in a 10 month old patient has been also reported [19, 20]. Cases with isolated involvement of nails have also been described. Apart from the numerous oral manifestations; median rhomboid glossitis in association with PC has been documented [21].

Conclusion

The patient is under periodic follow up. Mechanism of manipulating gene expression, advancement in delivery systems, availability of RNA reduction agents, gene slicing approaches may improve; make the ultimate goal of target gene correction of PC more feasible in future.

References


