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CASE REPORT / OLGU SUNUMU

Trigeminal nerve involvement in congenital insensitivity of pain with anhidrosis

Doğumsal ağrı duyarsızlığı ve trigeminal sinir tutulumu

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ABSTRACT

Congenital insensitivity to pain with anhidrosis (CIPA) is a rare cause of corneal anesthesia. Here, we report a case of CIPA with clinically prominent features of severe dry eye symptoms with painless corneal epithelial defect, early loss of primary teeth and a painless fissure on her upper lip in association with trigeminal anesthesia, and painless wounds on extremities. *J Clin Exp Invest 2013; 4 (4): 509-511*

Key words: Congenital pain insensitivity, trigeminal involvement, painless corneal epithelial defect

ÖZET

Konjenital ağrı duyarsızlığı ve anhidrosis birlikteliği (CIPA) kornea anestezisinin nadir görülen bir nedenidir. Burada, trigeminal anestezi ile ilişkili olarak ağrısız kornea epitel defekti, ciddi kuru göz belirtileri, süt dişlerinin erken kaybı ve üst dudakta ağrısız fissür ve periferik ağrı duyarsızlığı ile ilişkili olarak ekstremitelerde ağrısız yaraları bulunan bir CIPA olgusunu sunmayı amaçladık.

Anahtar kelimeler: Doğumsal ağrı duyarsızlığı, trigeminal sinir tutulumu, ağrısız kornea epitel defekti

CASE

A four years old girl was brought to our ophthalmology outpatient clinic with white discoloration in her right eye. Despite a large central corneal epithelial defect, she seemed very comfortable and there was no sign of foreign body sensation. The patient was evaluated thoroughly upon a suspicion of corneal anesthesia. According to her family, she had multiple painless lesions on her extremities as well as a previously healed painless fracture on her right arm.

The family was familiar to this event because of their 10 years old son who experienced early loss of deciduous teeth and fractures. On ophthalmological examination, her visual acuity was 20/30 in her right eye and 20/25 in the other eye. In her right eye approximately 2x4 mm fluorescent stained central corneal epithelial defect, superficial punctate keratopathy (SPK), and diminished tear break-up time were found. In her left eye there were only SPK and diminished tear break-up time (Figure 1). Schirmer test was 2 mm in her both eyes. A painless fissure on her upper lip, absence of deciduous teeth (Figure 2), painless ulcers on her right hand and foot, edematous hands, clubbing of the fingers and wound scar of a previous fracture on her right arm were remarkable in her inspection (Figure 3). She was well cooperated, somewhat shy, and mentally normal as documented by a psychiatrist's mental state examination and an IQ test (Wechsler Intelligence Scale for Children (WISC-R). The ulcer in her right eye improved after pressure-patch with antibiotic ointment. For her both eyes preservative-free polyvinyl alcohol eye drop and gel tears were used successfully in the maintenance treatment. Topical treatment was also given for the ulcers on her extremities. After closure of right corneal epithelial defect, the patient was brought with bilateral corneal epithelial defects 1 month later.

The family reported she was rubbing her eyes after use of eye drops. The patient was treated again by using pressure-patch with antibiotic ointment and the family was educated to prevent her from eye rubbing behavior especially after use of eye drops.

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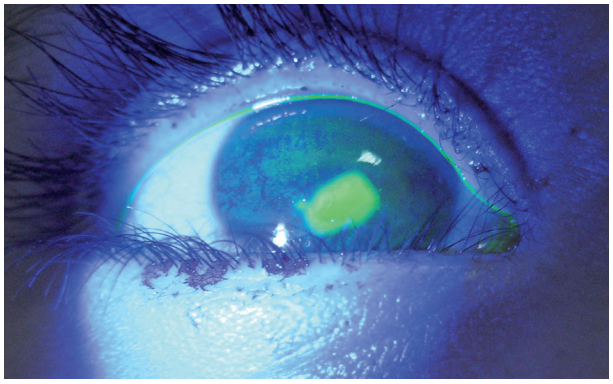

Figure 1. Fluorescence stained cornea showed a corneal epithelial defect and patient is very comfortable without any topical anesthetic usage

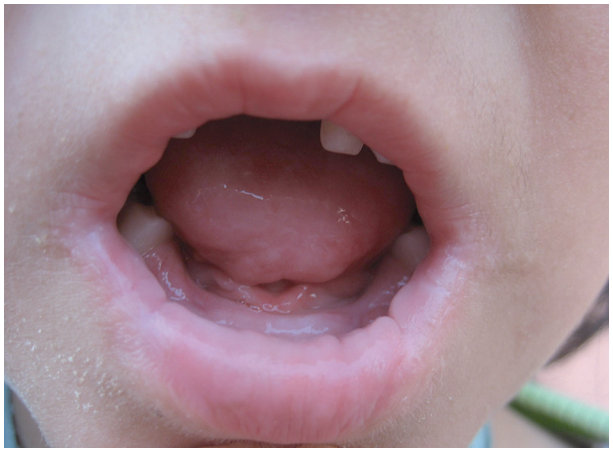

Figure 2. Lack of deciduous teeth

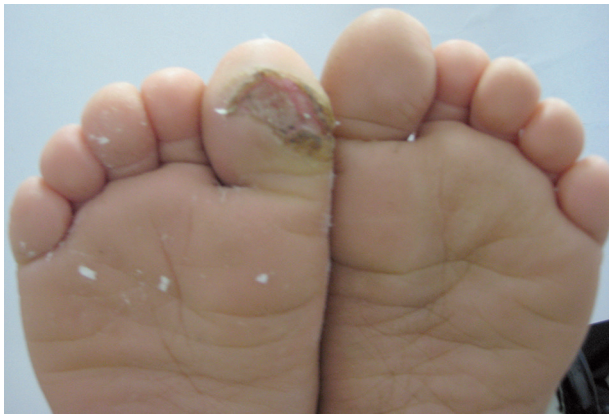

Figure 3. Painless ulcer on plantar surface of her right big toe

DISCUSSION

Congenital insensitivity to pain with anhidrosis (CIPA) is a very rare autosomal-recessive disease of the nervous system which is one of the hereditary sensory and autonomic neuropathies (HSAN) [1,2]. In CIPA, injuries and infections of the extremities and self-mutilation due to lack of pain sensation, fever secondary to anhidrosis or infections, mental retardation, and loss of unmyelinated and diminished small myelinated fibers are known features. Additionally several ocular manifestations such as SPK, diminished tear break-up time, corneal opacity, corneal ulcer, and infectious keratitis have also been reported [3-6].

CIPA may present with a wide variety of different clinical features important to different clinical disciplines. Thus, some associations between these features may be overlooked by the clinicians. For example, among four cases of congenital corneal anesthesia reported by Mathen et al. only one case showed loss of generalized pain sensation who interestingly had also absent dentition like our case [7]. In our case and in her 10 years old mentally retarded brother, who is also a CIPA patient, premature loss of deciduous teeth around 3 years of age and corneal anesthesia were present. These cases make us to think that trigeminal nerve involvement in CIPA takes place as involvement of its all branches (ophthalmic, maxillary and mandibular). In other words there may be association with early loss of primary teeth and corneal anesthesia in CIPA. But it may be overlooked by most of the authors because patients may be presented in different ages and different prominent signs.

Self-mutilation may start with biting behavior including tongue, lip or fingers. Later in life, accidental injuries may cause fractures and infections of the extremities. Schalka and coauthors suggested extraction of all primary teeth as a radical measure to whom, express self-mutilation by biting [8]. Thus, early primary teeth loss sometimes may be protective for self-mutilation. Eye rubbing in our case seems to be an important factor that may facilitate the recurrence of corneal epithelial defect. So we can discuss whether eye rubbing is a type of self-mutilation which is an important character of CIPA or not.

It is worth to note that the reported cases in the current literature with CIPA show similar geographic and ethnic distributions of the disease. The most striking groups are from Middle East, like our case and Asia originated populations and as well as Hispanics [3,4,9].

In most reports relevant departments, as expected, usually handled the cases according to their point of view. As CIPA may affect various organ systems in its course, its management requires a multidisciplinary approach. The ophthalmologist

should remember CIPA in the differential diagnosis of corneal anesthesia in children especially when they come from a suspected ethnic origin. Further investigations should seek if eye rubbing could be considered as a type of self-mutilation in CIPA. Also the patients with loss of pain sensation and early loss of teeth should be consulted to an ophthalmologist to be evaluated for corneal anesthesia as a component of trigeminal nerve involvement.

REFERENCES

1. Sztriha L, Lestringant GG, Hertecant J, Frossard PM, Masouye I. Congenital insensitivity to pain with anhidrosis. Pediatr Neurol 2001;25:63-66.
2. Nagasako EM, Oaklander AL, Dworkin RH. Congenital insensitivity to pain: an update. Pain 2003;101:213-219.
3. Amano S, Fukuoka S, Usui T, et al. Ocular manifestations of congenital insensitivity to pain with anhidrosis. Am J Ophthalmol 2006;141:472-477.
4. Yagev R, Levy J, Shorer Z, Lifshitz T. Congenital insensitivity to pain with anhidrosis: ocular and systemic manifestations. Am J Ophthalmol 1999;127:322-326.
5. Theodorou SD, Klimentopoulou AE, Papalouka E. Congenital insensitivity to pain with anhidrosis. Report of a case and review of the literature. Acta Orthop Belg 2000;66:137-145.
6. Ramaesh K, Stokes J, Henry E, Dutton GN, Dhillon B. Congenital corneal anesthesia. Surv Ophthalmol 2007;52:50-60.
7. Mathen MM, Vishnu S, Prajna NV, Vijayalakshmi P, Srinivasan M. Congenital corneal anesthesia: a series of four case reports. Cornea 2001;20:194-196.
8. Schalka MM, Correa MS, Ciamponi AL. Congenital insensitivity-to-pain with anhidrosis (CIPA): a case report with 4-year follow-up. Oral Surg Oral Med Oral Pathol Oral Radiol Endod 2006;101:769-773.
9. Suriu C, Khayat M, Weiler M, et al. Skoura - a genetic island for congenital insensitivity to pain and anhidrosis among Moroccan Jews, as determined by a novel mutation in the NTRK1 gene. Clin Genet 2009;75:230-236.