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Olgu Sunumu / Case Report

A Case of Ectrodactyly, Ectodermal Dysplasia, Cleft Lip and Palate Syndrome Associated with Hydrocephaly

Hidrosefali ile İlişkili Ektrodakli, Ektodermal Displazi, Yarık Dudak ve Damak Sendromulu Bir Vakanın Değerlendirmesi

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ABSTRACT

Ectrodactyly, ectodermal dysplasia, cleft lip, and palate syndrome (EEC) is a genetic developmental disorder characterized by ectrodactyly, ectodermal dysplasia and orofacial clefts (cleft lip/ palate). A few cases have been reported in literature. The cardinal components of the syndrome are ectrodactyly and syndactyly of the hands and feet, cleft lip with or without cleft palate, and abnormalities ectodermal structures including skin (i.e. hypopigmented and dry skin, hyperkeratosis, skin atrophy), hair (sparse hair and eye brows), teeth (small, absent or dysplastic teeth), nails (nail dystrophy) and exocrine glands (reduction/ absence of sweat, sebaceous and salivary glands). A multidisciplinary approach for treatment is needed which is coordinated by orthopedic, plastic, dental surgeons, ophthalmologist, dermatologists and speech therapists, psychologists. We presented EEC syndrome case with hydrocephaly in the literature.

Key Words: Ectrodactyly, ectodermal dysplasia, cleft palate and lip, hydrocephaly

ÖZET

Ektrodaktili, ektodermal displazi, yarık dudak ve damak sendromu (EEC); ektrodaktili, ektodermal displazi ve orofasiyal yarık ile karakterize gelişimsel bir genetik hastalıktır. Literatürde bu konu ile ilgili çok fazla vaka rapor edilmemektedir. Sendromun esas komponentleri; el ve ayaklarda ektrodaktili ve sindaktili, normal veya yarık damaklı yarık dudak ve deri (hipopigmente ve kuru cilt, hiperkeratinoz, deri atrofisi), kıl (seyrek saç ve kaş), diş (küçük, yok veya displastik diş), tırnak(tırnak distrofisi) ve dış salgı bezleri (terin azalması veya yokluğu, yağlı ve tükürük bezleri) dahil olduğu anormal ektodermal yapılardır. Tedavisi; ortopedi, plastik ve diş cerrahisi, oftalmolojist, dermatolog ve konuşma terapisti ve psikologların birlikte çalışacağı multidisipliner bir yaklaşıma ihtiyaç duymaktadır. Biz literatürde ilk kez rastlanan hidrosefalili EEC sendromlu bir vakayı sunduk.

Anahtar Kelimeler: Ektrodaktili, ektodermal displazi, yarım damak ve dudak, hidrosefali

INTRODUCTION

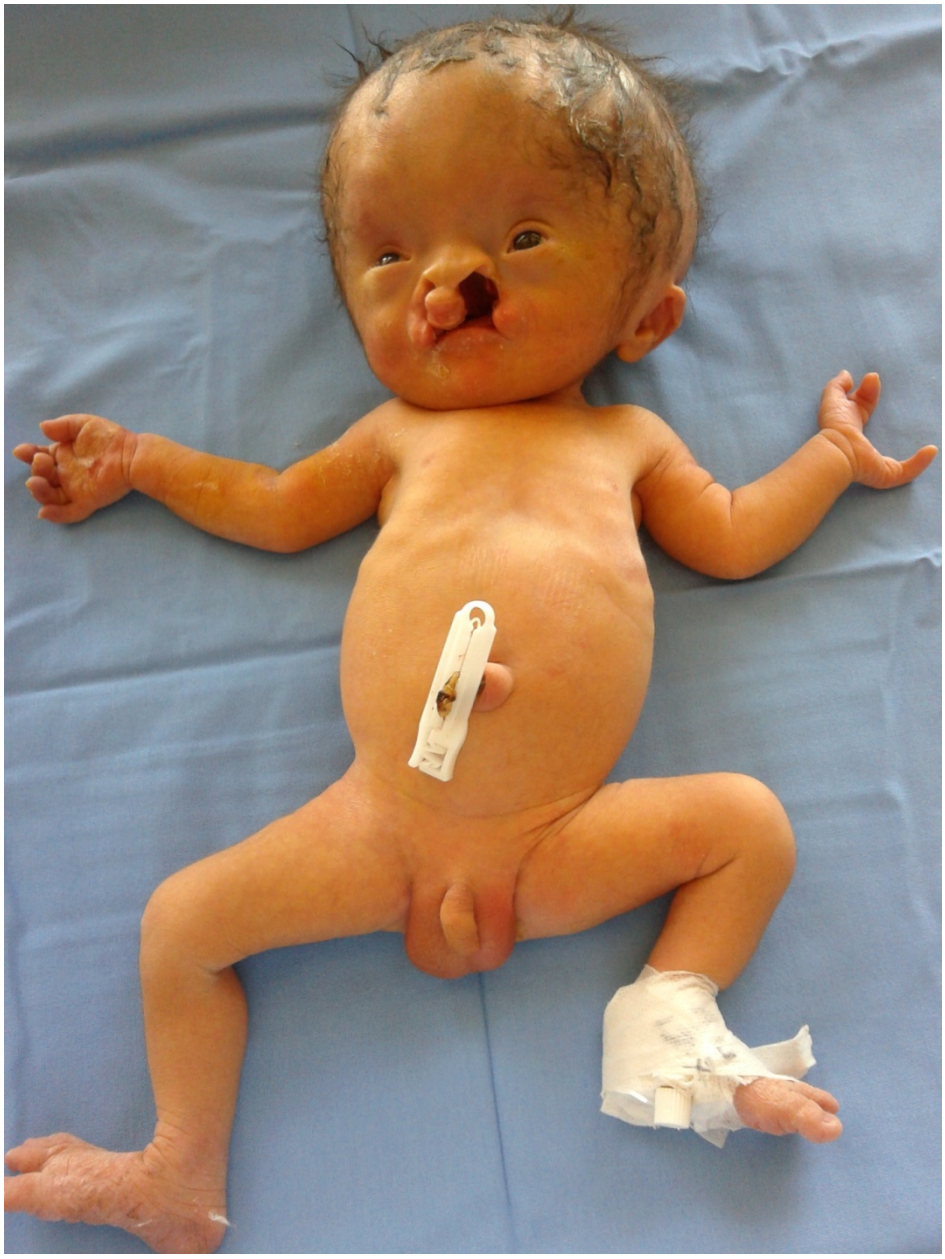
The ectrodactyly-ectodermal dysplasia cleft lip / palate (EEC) syndrome is a rare autosomal dominant genetic syndrome characterized by various degrees of ectrodactyly and syndactyly (hands and feet) cleft lip and /or palate and ectodermal dysplasia[1].

Other less common findings that mental retardation deafness or hearing defects, genitourinary anomalies[2]. The acronym was first used by Rudiger et al[1]. We presented EEC syndrome case with hydrocephaly by the literature.

CASE

A preterm infant (37th week gestational age) was born to a 22 year old mother (4 gravida 4 para). He was born with section due to hydrocephaly. There is no blood relation between parents. His mother has 1,5 and 2,5 month two abort. In post natal third days patient was hospitalizated in our new born intensive care because of hydrocephaly, complet cleft palate and lip. In physical examination weight 2590 gr (25-50% percentile) height 51 cm (50-75% percentile) headcircumference:44 cm (>90% percentile). Patient was seemed to be with pale skin, microphthalmia, hypertelorism, views of the sinking sun eye, anterior fontanelle 4x5 cm and swollen, flattened nasal bridge, low-set ears, complete cleft lip and palate, lobster claw hand (left upper limb), syndactyly (both two foot) (Figure), 1/6 systolic murmur (mezocardiac center), absent suction, moro and capture reflex weaken patient was hypotonic. Laboratory analysis was normal.

Study of patient for midline defects ophthalmic examination, echocardiography were normal. Thyroid function tests were normal. Abdomen ultrasonography right renal cortical cyst and left renal hydronephrosis were detected. Cranial magnetic resonance results observed were; midline fusion defects, inter hemispheric cyst and absent corpus callosum. Due to these defects lateral ventricules extension and hydrocephaly were formed.



DISCUSSION

This syndrome results from a developmental deformation of ectodermal and mesodermal tissues. Each of these defects occurs facial cleft, ectrodactyly, ectodermal dysplasia of hair, teeth, nails and ophthalmologic abnormalities.

The less common findings include abnormalities of the genitourinary system (renal agenesis, atresia, hydronephrosis, vesicourethral reflux), hearing loss, choanal atresia, mammary gland/nipple hypoplasia, ophthalmological findings (blepharitis, conjunctivitis, corneal scarring and

pannus with photophobia, poor lacrimal and meibomian secretion), gland abnormalities (hypothalamic and hypopituitary development defect), malignant lymphomas[3,4,5,6,7].

EEC is due to missense mutations in the sequence of the TP63 gene (3q27) encoding the TP63 transcription factor that is essential for ectoderm and limb development[6]. EEC is diagnosed with antenatal ultrasonography in second trimester of gestation. Genetic testing and molecular analysis by chorionic villus sampling or by amniocentesis may confirm the diagnosis[8]. Management is multidisciplinary require and valuation by orthopedic, plastic, dental surgeons, ophthalmologist, dermatologists and speech therapists, psychologist.

In conclusion, although %5–10 cases were reported microcephaly[9] our case had a hydrocephaly with EEC. It was the first case that we presented in our article. We wanted to indicate EEC syndrome and hydrocephaly association.

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