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Olgu Sunumu

Cat Eye syndrome: Case report

Cat Eye sendromu: Olgu sunumu

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ABSTRACT

Cat Eye Syndrome (CES) or Schmid-Fracccaro syndrome is a genetic disorder characterized by mutations in the long arm of chromosome 22, first described by Schachenmann et al. in 1965. Its classic triad consists of iris coloboma, anal malformation and ear anomalies. In this case, a 3-year-old male patient with a 30-second tonic clonic seizure was presented. On physical examination, he had downslanting palpebral fissures, micrognathia and microphthalmia. His family told that he was monitored with mild retardation in cognitive development by the department of pediatric psychiatry. Karyotypic analysis was performed for cat eye syndrome, because of the presence of neuropsychiatric findings and eye / facial anomalies. DNA microarray analysis revealed a gain involving 3 OMIM genes in the 22q11.1 region of about 548 Kb was detected. In this case, there were no major anomalies, but followed by genetic consequence analysis, and diagnosed with Cat-Eye Syndrome. Therefore, genetic analysis should be requested in case of clinical suspicion in atypical cases without classic triad.

Keywords: Neuropsychiatric findings, atypical facial features, seizure, Cat Eye Syndrome

ÖZ

Cat Eye Sendromu (CES) veya Schmid-Fracccaro sendromu ilk kez 1965 yılında Schachenmann ve ark. tarafından tanımlanan, 22. kromozomun uzun kolunun mutasyonları ile oluşan genetik bir hastalıktır. Klasik triadı iris kolobomu, anal malformasyon ve kulak anomalisinden oluşmaktadır. Bu makalede tonik klonik vasıfta 30 saniye süren kasılma ile başvuran üç yaşında bir erkek olgu anlatıldı. Fizik muayenesinde, aşağı yerleşimli palpebral fissürler, mikrognati, mikroftalmisi mevcuttu. Aileden, çocuk psikiyatrisinde bilişsel gelişimde hafif gerilik ile izlendiği öğrenildi. Nöropsikiyatrik bulguları ve göz/yüz anomalileri doğrultusunda genetik sendromlar için karyotip analizi yapıldı. DNA microarray analizinde 22q11.1 bölgesinde yaklaşık 548 Kb büyüklüğünde 3 OMIM geni içeren duplikasyon saptandı. Hastamızda majör anomali bulunmamış, ancak genetik analiz sonucu ile Cat-Eye sendromu tanısı ile takibe alınmıştır. Bu nedenle klasik triad bulgularının olmadığı atipik olgularda da klinik şüphe halinde genetik analiz istenmelidir.

Anahtar kelimeler: Nöropsikiyatrik bulgular, atipik yüz, nöbet, Cat Eye sendromu

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INTRODUCTION

Cat Eye Syndrome (CES) or Schmid-Fracccaro syndrome is a genetic disorder, described by Schachenmann et al. [(1)] in 1965. It is seen at an incidence rate of 1 / 50,000-150,000 among live births [(2)]. CES is most often caused by the proximal duplication of the long arm of chromosome 22 [(3)]. The classic triad is iris coloboma, anal malformation and ear anomalies [(4)]. It is not expected to see all of the findings in every CES case [(3)].

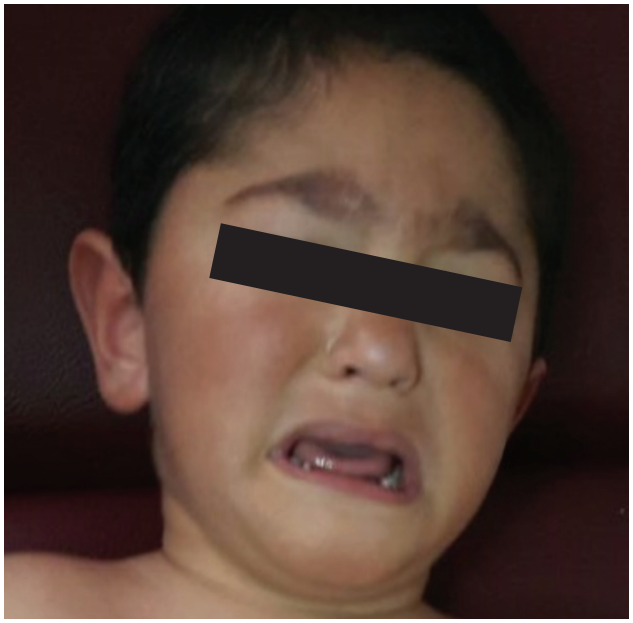

Figure I. Distribution of patients according to admission time.

CASE REPORT

A 3-year-old male patient was admitted to hospital because of the presence of a 30-second tonic clonic seizure. Downslanting palpebral fissures, micrognathia and microphthalmia were detected during physical examination (Figure 1). He did not have an eye contact and demonstrated aggressive behavior. He had been followed by psychiatry due to mild cognitive retardation. Electrolyte and blood glucose levels were within normal limits. Electroencephalography (EEG) demonstrated a suspect focal epilepsy foci; cranial magnetic resonance imaging was evaluated as normal. There was no iris coloboma in the patient, abdominopelvic ultrasonography revealed no renal abnormality and echocardiography was normal. DNA microarray analysis was performed for cat eye syndrome, because of the presence of neuropsychiatric findings, ocular, and facial manifestations. DNA microarray analysis revealed gain involving 3 OMIM genes at a size of 548 Kb in 22q11.1 region. He was diagnosed with cat eye syndrome. CES may come into play with a new mutation or one of the parents may be a carrier or mosaic for the marker chromosome. For this reason, karyotypic examination the of mother and the father was performed.

DISCUSSION

CES was first described by Schenmann et al. [1] in 1965 with iris coloboma and anal atresia. In 1878, Haab revealed the presence of renal anomalies [5]. Gerald et al. [6] reported the presence of coloboma in patients' eyes which resembled the cat's eye, hence the definition of Cat Eye Syndrome was made. Classical triad of CES is seen in about 41% of the cases [4]. CES cases were evaluated and major (anorectal malformations, urogenital malformations, ocular coloboma, congenital heart defects) and minor features (downslanting palpebral fissures, orthopedic malformations, ear anomalies, abdominal malformation, micrognathia, microphthalmia, microcephaly, cleft palate) were determined. Many of the cases do not carry all the features of the syndrome [7]. Only 9 cases with all major features of CES have been reported in the literature [3]. In addition, there are patients who do not have coloboma and anal malformations, and only due to the presence of face and ear anomalies they can get diagnosis of CES diagnose based on genetic analysis [8]. In these cases, mental status can be normal or mental retardation can be seen [3]. CES can be seen in patients with proximal duplication of the long arm of chromosome 22, partial tetrasomy (22pter-22q11), duplication-inversion (22pter-22q11), and partial trisomy (22pter-22q11) [1]. The amount or size of the material that underwent duplication in the 22nd region of the CES critical region (CESCR) is not directly proportional to the occurrence of phenotypic features [3].

Our case was investigated for seizure etiology, and genetic analysis was requested because of his atypical facial appearance. In our patient, there were no major anomalies but minor anomalies as palpebral fissures, micrognathia and microphthalmia were detected. In the literature, 4 out of 68 patients had been reported to have seizures and abnormal EEG findings [4]. In addition, various cerebral anomalies (interhemispheric cyst and corpus callosum agenesis, hypothalamic pituitary acute malformation) can be detected in CES cases [9,10]. Abnormal waveforms

were detected in the EEG of our patient but cranial magnetic resonance imaging findings were within normal limits .

In this case, there were no major anomalies, but downslanting palpebral fissures, micrognathia and microphthalmia were detected as minor anomalies, and he was followed by genetic consequence analysis, and diagnosed as cat eye syndrome. CES has a broad clinical spectrum ranging from mild facial features to heart anomalies, due to genotypic and phenotypic differences, and it is hard to recognize from it's phenotypic characteristics alone. Therefore, genetic analysis should be requested in case of clinical suspicion in atypical cases without any classic triad.

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