A Case of Alport's Syndrome with Nyctalopia

Alport's Sendromlu Bir Olguda Niktalopi

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ABSTRACT

We aimed to present a case with nyctalopia and electrophysiological abnormalities who was diagnosed as Alport's syndrome in childhood. The patient was a 24-year-old woman with a history of nyctalopia for the last one year. Fundus examination revealed normal morphology. On electrophysiological studies, patern visual-evoked potentials (VEP) and electro-oculography (EOG) were normal, but electroretinography (ERG) revealed moderate reduction of rod and cone responses. This case report indicates that subnormal ERG may accompany night vision difficulties which suggests photoreceptor degeneration in patients with Alport syndrome, despite normal visual acuity and fundus appearance.

Key words: Alport's syndrome, nyctalopia

ÖZ

Elektrofizyolojik anormallikler ile seyreden Alport sendromlu 24 yaşında bir hanım olgu sunulmuştur.

Anahtar kelimeler: Alport sendromu

INTRODUCTION

Alport's syndrome is a genetically and clinically heterogeneous disorder characterised by ocular abnormalities, haemorrhagic nephritis, progressive renal failure and sensorineural hearing loss. These clinical findings have been attributed to mutations in the COL4A5 genes these resulting in the loss of the a3(IV)–a5(IV) collagen network from affected basement membranes.[1,2]

A wide range of ocular manifestations have been described including corneal posterior polymorphous dystrophy and anterior lenticonus as well as retinopathies and macular holes. The classic ocular features include a central or perimacular dot and fleck retinopathy, which occurs in 85%of affected adult males, and anterior lenticonus, which presents in 25%.[3]

Some previous studies have reported electrophysiological findings in adult patients with Alport's syndrome.[4,5] These reports showed variable results. We describe a case of Alport's syndrome with ERG abnormality.

CASE REPORT

A 24-year-old woman presented with decreased night vision in both eyes for last one year. She had been diagnosed as Alport's syndrome when she was 14 years old. Visual acuity was 20/20 in both eyes. Intraocular pressures were normal in both eyes. Slit lamp examination of the anterior segment revealed mild anterior lenticonus in both eyes. Dilated fundus examination showed normal retinal appearance (Figure 1, A and B). Autofluorescense images were normal (Figure 2, A and B). Electroretinogram (ERG), visual evoked potential (VEP) and electrooculogram (EOG) tests were performed. In the EOG, the Arden ratios were 2.01 and 2.13 in the right and left eye, respectively (Figure 3). Pattern VEP P100 values were in normal limits in both eyes. Electroretinography (ERG) revealed a moderate reduction of rod and cone responses (Figure 4).

DISCUSSION

Alport's syndrome is an inherited disorder of collagen that affects the kidney, the eye and the cochlea. Approximately

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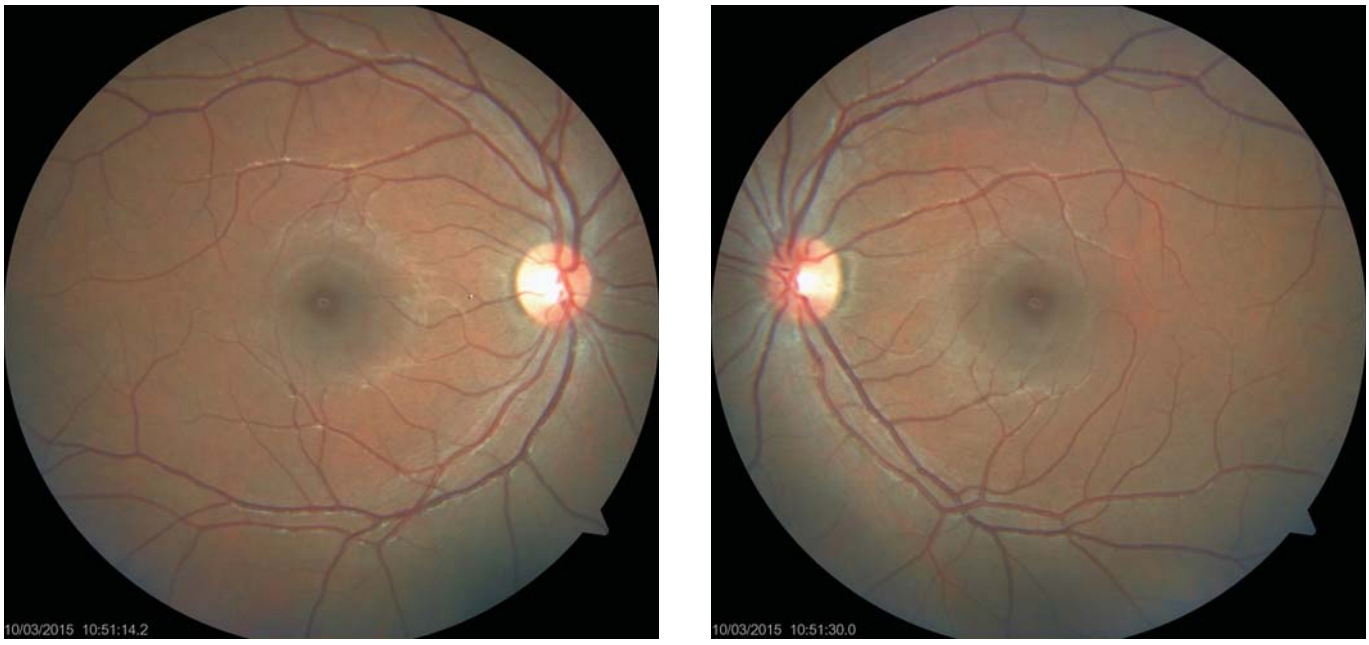

Figure 1 (A and B): normal fundus appearance.

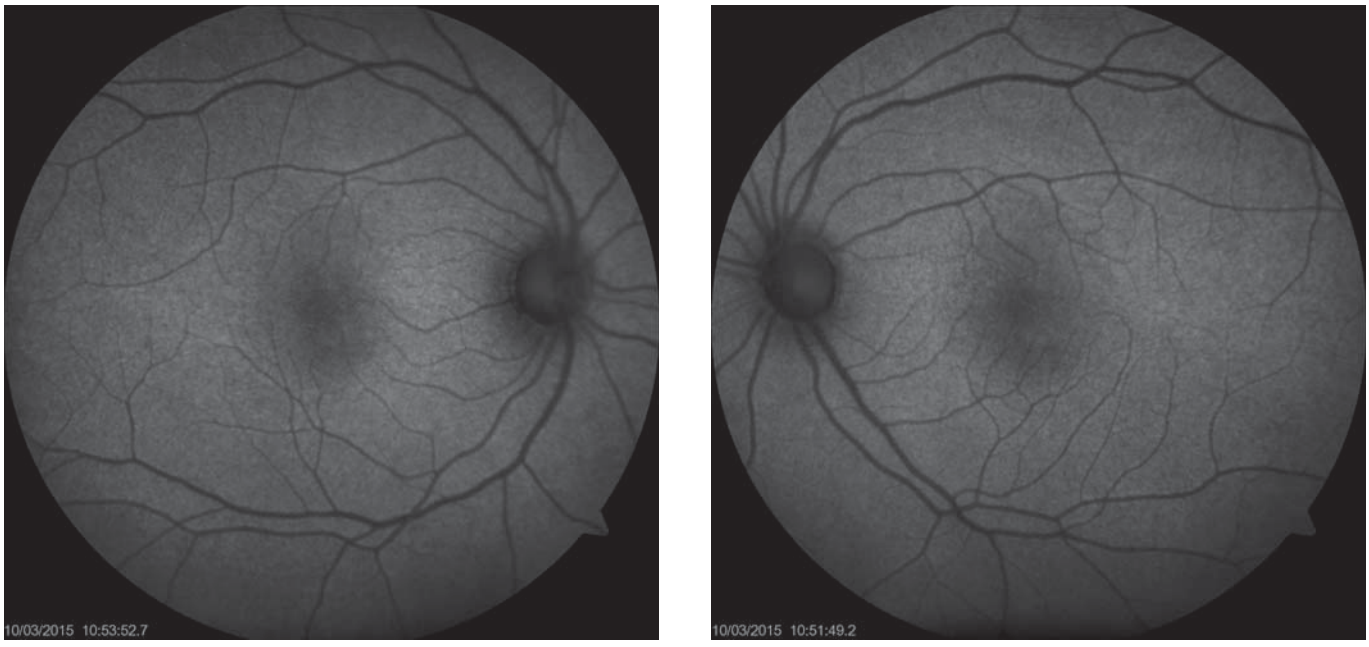

Figure 2 (A and B): normal fundus autofluorescense.

in 80%of patients transmission is X-linked. In about 10%, the transmission is autosomal recessive and exceptionally autosomal dominant. In the remaining 10%, there is no family history.[6] None of the family members of our patient was known to have similar findings.

Electrophysiological studies in Alport's syndrome have not been studied extensively, with most of the reported cases being normal. In 1980, Perrin et al [5] reported one patient with an abnormal ERG and EOG and normal fluorescein angiography. In 1980, Zylbermann et al [7] reported two siblings with Alport's syndrome having retinal changes, decreased ERG and normal EOG. In 1988, Gelişken et al [8] reported one patient with Alport's syndrome, salt and pepperlike retinopathy, diminished scotopic ERG and an elevated threshold on dark adaptation, raising the question of a possible associated retinal degeneration.

Subnormal ERG amplitudes have been reported in patients without Alport's syndrome after chronic renal failure. Sverak et al [9] found that ERGs were generally abnormal after long term dialysis but tended to normalise after successful transplantation. It is possible that renal dysfunction may have a role in producing ERG changes observed in Alport's syndrome. In our case, abnormal ERG results may be a predictive factor for renal dysfunction.

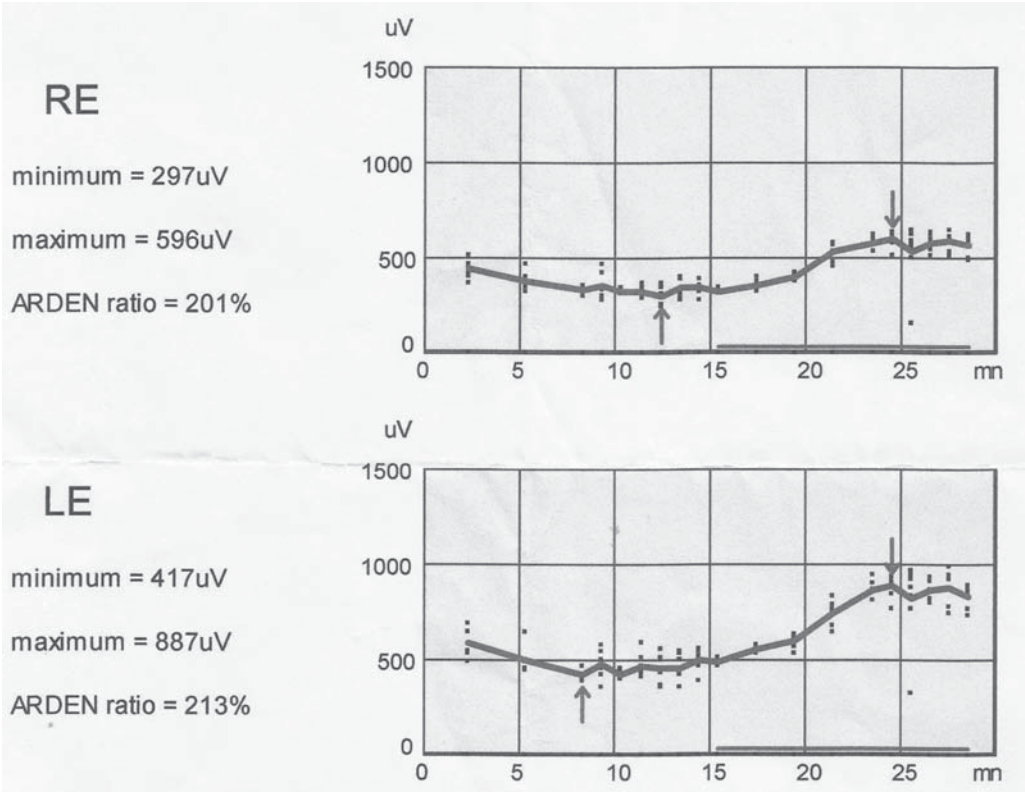

Figure 3: Electro-oculogram, within normal limits.

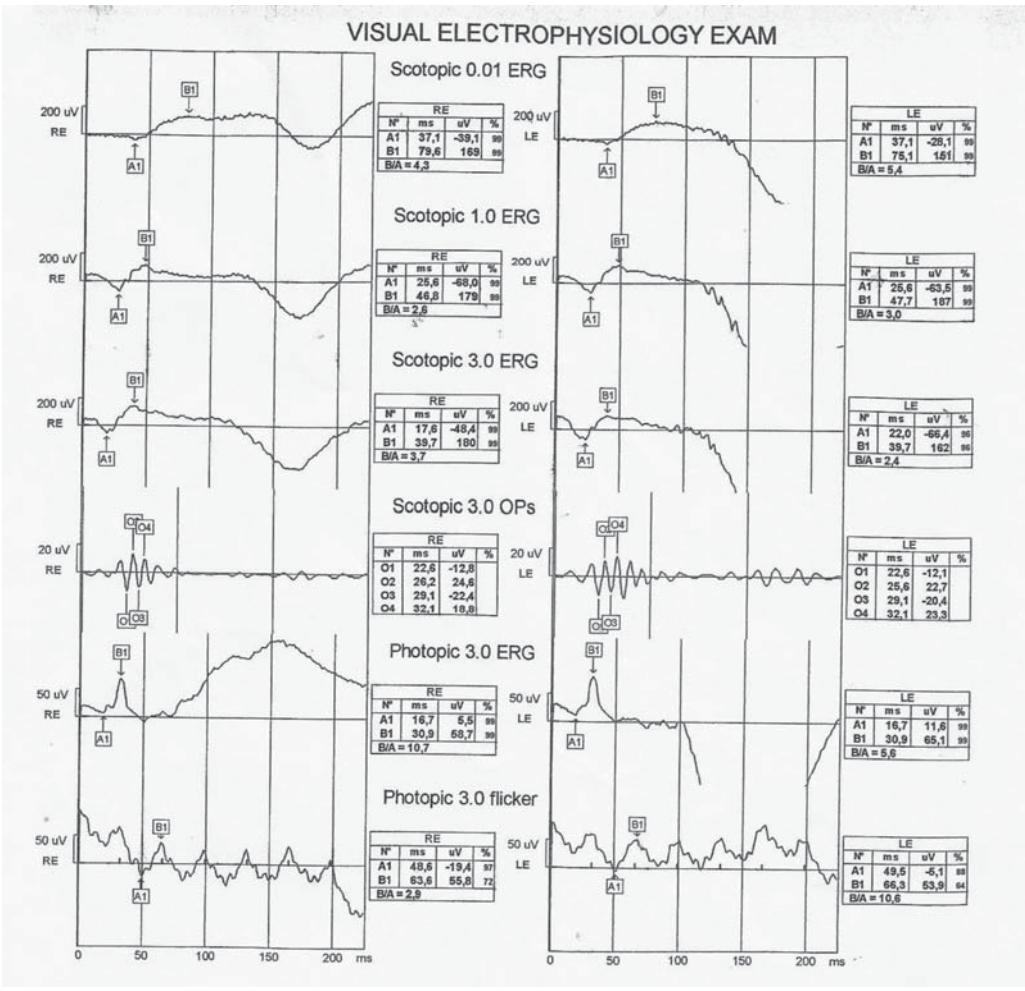

Figure 4: Electroretinogram, moderate reduction of rod and cone responses.

Our normal EOG results are in line with the majority of previous studies.[4,7,8] Nonetheless, Tsilou et al [10] described a case with moderately severe reduction of rod and cone responses in the ERG and a decline in light peak amplitude in the EOG. Their results pointed to a retinal degeneration with generalized involvement and proposed that degeneration of photoreceptors could be primary or secondary to retinal pigment epithelium (RPE) degeneration.

Çıtırık et al [11] reported a case with normal ERG and slightly increased latencies of flash visual evoked potentials. They explained this effect might be due to renal dysfunction. Reduced VEP amplitudes and increased pattern and flash VEP latencies have been reported commonly in patients with renal failure.[4,9]

Randhawa et al [12] described a case with splotchy hypoautofluorescence in the peripheral fundus and abnormal vitreoretinal interface. This pattern was recently described and may be due to retinal pigment epithelium abnormalities. There was no abnormal appearance in the autofluorescence images of our patient.

In conclusion, patients with Alport's syndrome seem to show inconsistent ERG, EOG, or VEP findings. Electrophysiological changes will be required to be further investigated in terms of possible retinal degeneration and renal function abnormalities.

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