Comorbidity of Dominant Drusen and Pellucid Marginal Degeneration

Dominant Druzen ve Pellusid Marjinal Dejenerasyon Birlikteliği

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ABSTRACT

Dominant (familial) drusen (DD) also known as Malattia leventinese (ML) is a rare inherited retinal dystrophy characterized by the presence of numerous drusen in the posterior pole. Pellucid marginal degeneration (PMD) is another rare ectatic corneal disorder which typically affects the inferior peripheral cornea. We report a case of comorbidity of DD and PMD in a 53-year -old male who had complaints about low visual acuity. To the best of our knowledge, this is the first case revealing the association of DD and PMD.

Key Words: Dominant drusen, Pellucid marginal degeneration, retinal dystrophy.

ÖZ

Malattia leventinese (ML) olarak da bilinen Dominant (ailesel) druzen (DD) hastalığı, arka kutupta çok sayıda druzenin olmasıyla karakterize kalıtımsal bir retina distrofisidir. Pellusid marjinal dejenerasyon (PMD) ise tipik olarak alt korneayı etkileyen ve nadir görülen ektatik bir kornea bozukluğudur. Biz bu çalışmada, görme azlığından şikayetçi 53 yaşında erkek bir hastada, DD ve PMD birlikteliğini sunmaktayız. Bildiğimiz kadarıyla, bu iki nadir hastalığın birlikteliği daha önce sunulmamıştır.

Anahtar Kelimeler: Dominant druzen, pellusid marjinal dejenerasyon, retina distrofisi.

INTRODUCTION

Dominant (familial) drusen (DD) is a genetic disorder which has been linked to chromosome 2 and characterized by the appearance of small radial macular and parapapillary drusen.[1] Drusen become quite numerous by the fifth decade, usually beginning from the early adult life.[2] Retinal fluorescein angiography reveals the drusen and atrophy of RPE more extensive than seen by indirect ophthalmoscopy and helps us to put the diagnosis.

Pellucid marginal corneal degeneration (PMD) which is a rare, idiopathic, thinning disease of the inferior peripheral cornea, usually occurs between the second and fifth decades of life similar to DD.[3] The exact aetiology of PMD is unknown and no genetical inheritance pattern has been found yet.[3] Although comorbidity of PMD and several ocular diseases like retinitis pigmentosa, retinal lattice degeneration, keratoconjunctivitis and glaucoma have been reported in literature, there was no report on association of PMD and DD.[3]

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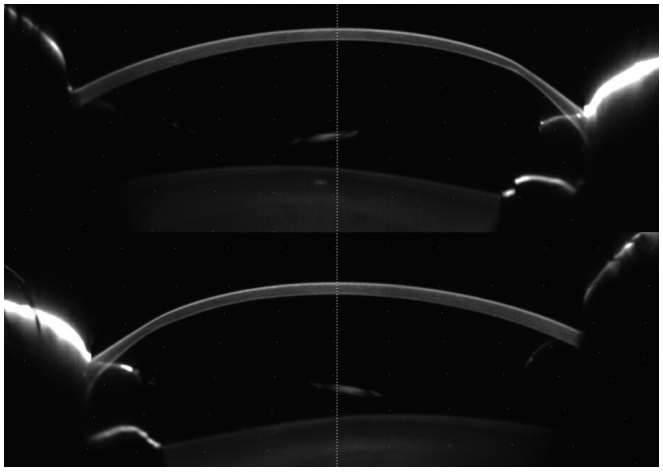

Figure 1: *Pentacam HR scheimpflug anterior segment imaging of the right (above) and left eyes (below) of the patient.*

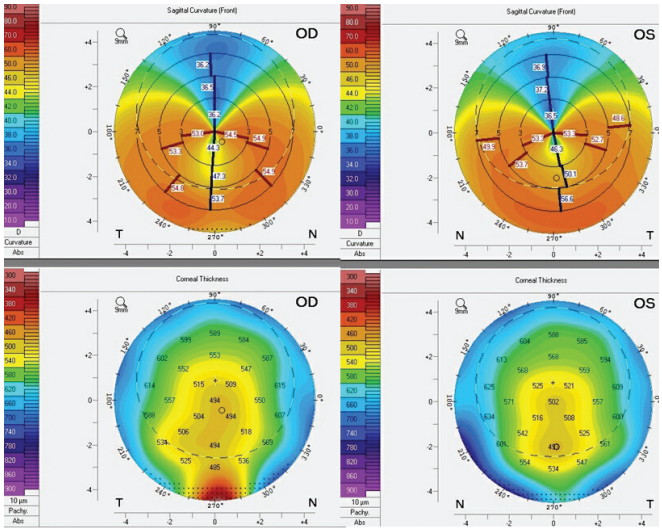

Figure 2: *Corneal topography revealing the classical crab-claw appearance of both eyes.*

CASE REPORT

A 53-year-old man admitted to our clinic complaining of gradual decrease in visual acuity for about two years. There was no ocular surgery or systemic disease in the history of the patient. We asked the patient whether his relatives had similar ocular complaints and he stated he did not notice or know any relative having similar diseases like him. His best corrected visual acuity was 20/100 in the right eye and 20/80 in the left eye. Slit-lamp biomicroscopy and Pentacam HR (Oculus, Wetzlar, Germany) revealed a thinning and ectasia in the inferior half of cornea at about 2mm from the limbus bilaterally (Figure 1). Also characteristic crab-claw pattern of PMD was seen in both corneas of the patient by Pentacam HR (Figure 2). Slit-lamp examination of the right and left lens revealed Grade 3 and Grade 2 nuclear cataract according to LOCS III cataract classification, respectively. The intraocular pressure measured with pneumotonometer was 11 mmHg in the right eye and 10 mmHg in the left eye. In dilated retinal examination, numerous drusen were seen in the posterior pole (Figure 3). Fundus fluorescein angiography (FFA) findings showed the retinal pathology more clearly (Figure 4).

By the help of these examinations, we put the diagnosis of DD. The patient told us that he did not want to use a rigid gas permeable contact lens and he wanted spectacles instead. The patient also declined any surgical procedures. We had the opportunity to examine his daughter and son, but we did not notice any ocular disorders.

DISCUSSION

Dominant (familial) drusen (DD) was first described by Doyne in 1899 (Doyne honeycomb retinal dystrophy).[4] Then, another spectrum of DD was described in patients living in the Leventine valley in Switzerland (Malattia Leventinese).[5] These two diseases share the same phenotypic characteristics and their gene has been localized to chromosome 2.[1] Both the Doyne and ML are similar in the aspect of age at onset of macular drusen in the third decade and progressive accumulation of drusen leading to mild or no decrease in vision; although severe visual loss can occur due to choroidal neovascularization.[1]

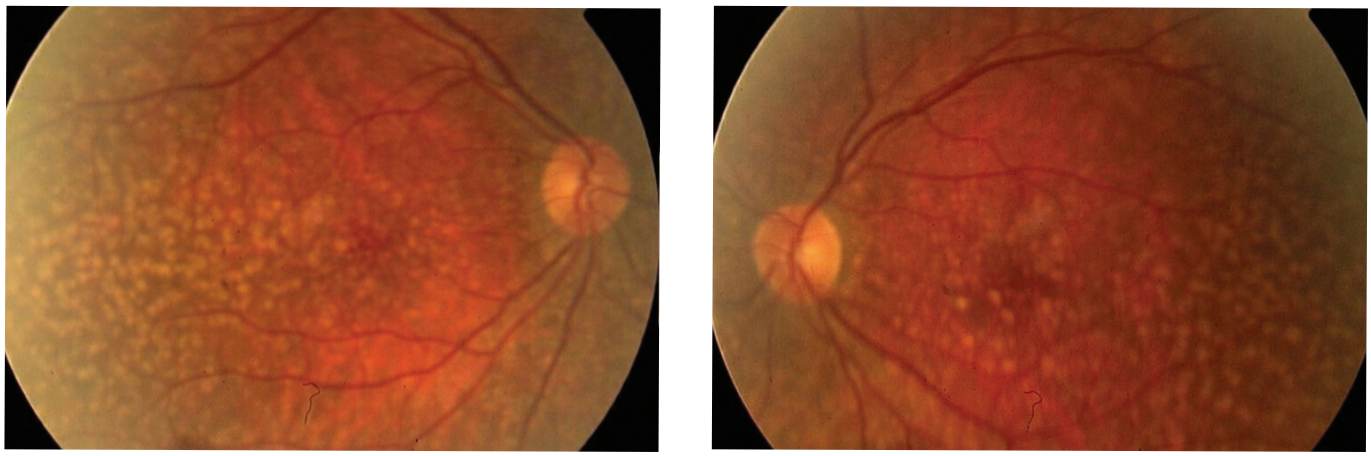

Figure 3: *Retinal photography showing numerous posterior pole drusen bilaterally.*

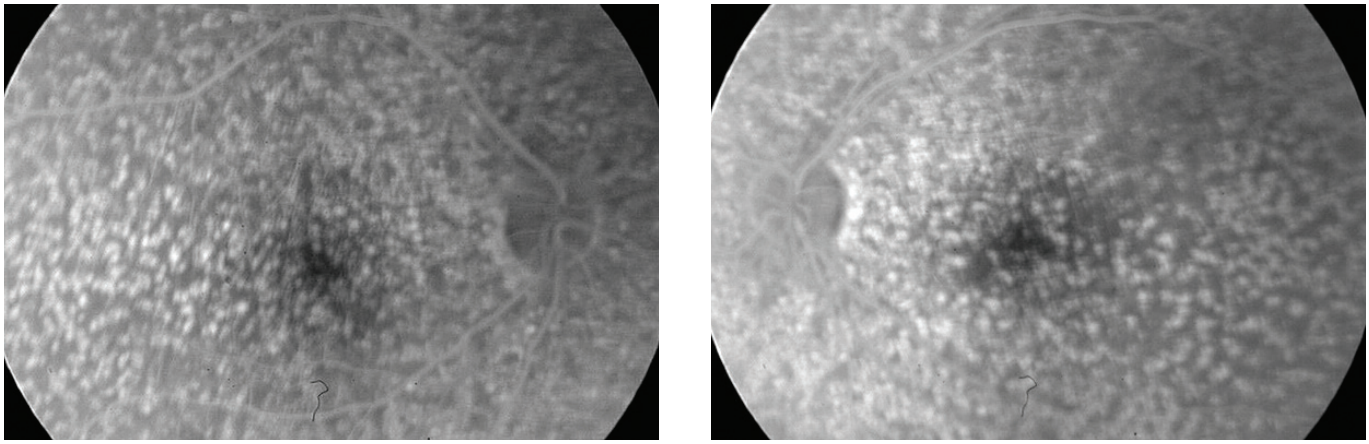

Figure 4: *Fluorescein angiography revealing drusen more extensively.*

Also, there are no systemic or other ocular features specifically associated with these two diseases.[1] In our case, there were no systemic diseases, but there was association with PMD. Possible pathogenic mechanisms of DD disease were considered as inborn error of RPE metabolism and the development of abnormal basement membranes due to the defects in an intercellular matrix protein or a structural protein.[6] Although diffuse drusen are usually accepted as being inherited autosomal dominantly, it is difficult to reveal a family history; because affected individuals usually are not recognized until middle age like our patient, when other potentially affected family members are very old or dead.[6] The most important disease in the differential diagnosis of DD is age related macular degeneration (ARMD). Different from ARMD, drusen distribution extends beyond macula and involve retina nasal to the optic disc.[7] The other diseases that resemble DD are Sorsby's fundus dystrophy and fundus flavimaculatus; but our case does not seem to be related with these disorders, especially in the aspect of fluorescein angiographic photographs. But inability to do genetical analysis to the patient and his relatives is one of the restrictions of this report. There is no definitive treatment strategy for DD, unless choroidal neovascularization occurs.The other ocular pathology in our patient was PMD. Although the exact pathology of PMD is unknown, some authors stated that there is a relationship between keratoconus, keratoglobus and PMD.[8,9] PMD is a rare disease, but there is no exact data about the incidence or prevalence of it.[3] Patients usually complain about reduced visual acuity resulting from irregular astigmatism. PMD is usually located in the inferior cornea extending from the 4 o'clock position to the 8 o'clock position and about 2 mm from the limbus.[8,10] Recommended treatment procedures include spectacle correction, rigid gas-permeable contact lenses, intrastromal corneal ring segments, collagen cross-linking and corneal transplantation.[3] The Pentacam HR examinations helped us to put the diagnosis of PMD to our patient.

Association of these two rare ocular diseases might help us in better understanding the pathology of them. Taking into consideration this comorbidity, it would be possible to assume that some similar mechanisms play a role in the development of both diseases. This case is also unique in revealing an association of an ocular pathology with DD. On the other hand, this association might be a chance event, although the possibility is weak. Third ocular pathology in this patient was nuclear cataract. In our opinion, all of these ocular diseases contributed to the low visual acuity of this patient. Since the patient did not want to undergo any surgical procedures at that time, we prescribed him spectacles.

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