

Two Different Cases with Melanocytoma of the Optic Disc

Optik Disk Melanositomlu İki Farklı Olgu

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ABSTRACT A 45-year-old woman (Case 1) applied for control examination to learn whether there was any growth or not on tumoral lesion in her left eye. Melanocytoma of the optic disc involving the adjacent choroid in the left eye was seen on ophthalmoscopic examination. The appearance of the patient was consistent with achondroplasia. Case 2, a 29-year-old woman, applied with headache complaint. Bilateral optic disc hypoplasia and melanocytoma of the optic disc in the right eye was found on the fundus examination and magnetic resonance imaging revealed an arachnoid cyst in the left temporal fossa. Although melanocytoma of the optic disc has not been proven to have any systemic associations, it is likely to be associated with some of the systemic diseases and other ocular pathologies. Optic disc hypoplasia associated with melanocytoma of the optic disc has been reported previously, however, associations of achondroplasia or arachnoid cyst with melanocytoma of the optic disc have not yet been reported.

Key Words: Arachnoid cysts; optic disk

ÖZET Kırkbeş yaşında kadın hasta (Olgu 1) sol gözündeki tümöral kitlede büyüme olup olmadığının kontrol muayenesi için başvurdu ve oftalmoskopik muayenede sol gözde komşu koroidi tutan optik disk melanositomu görüldü. Hastanın görünümü akondroplazi ile uyumluydu. Olgu 2, 29 yaşında kadın, baş ağrısı şikayeti ile başvurdu. Olgu 2'nin fundus muayenesinde bilateral optik disk hipoplazisi ve sağ gözde optik disk melanositomu tespit edildi ve manyetik rezonans görüntülemeye sol temporal fossada araknoid kist izlendi. Optik disk melanositomunun sistemik bir birlikteliği tanımlanmamış olmakla birlikte, sistemik hastalıkların bazılarıyla ve başka oküler patolojilerle birlikte görülebilir. Optik disk melanositomu ile birlikte optik disk hipoplazisi daha önce bildirilmiştir, ancak optik disk melanositomu ile akondroplazi veya araknoid kistin birliktelikleri bildirilmemiştir.

Anahtar Kelimeler: Araknoid kist; optik disk

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Melanocytoma of the optic disc is a benign tumor that usually occurs as a deeply pigmented lesion of the optic disc and occasionally causes visual loss; it may also lead to afferent pupillary defect and a visual field abnormality. Melanocytoma of the optic disc appears to have a slight predilection for females. The mean age at the diagnosis of melanocytoma of the optic disc is 50 years with a median of 52 and a range of 1-91 years.^{1,2} Melanocytoma of the optic disc is generally an isolated entity with no proven associations with any systemic diseases. Recent studies have shown solitary cases with neurofibromatosis type 2, basal cell carcinoma,

and vitiligo, central nervous system abnormalities such as meningioma and hypopituitarism, optic disc hypoplasia, however they seem to be coincidental.^{3,4}

Associations of melanocytoma of the optic disc with achondroplasia or arachnoid cyst have not been reported so far. To our knowledge, these are the first cases to be reported in the literature.

CASE REPORTS

CASE 1

A 45-year-old woman applied to an eye clinic with presbyopic symptoms, and on her routine examination, a tumoral lesion had been determined on her left optic disc four years earlier. She applied to our clinic as to whether there was any growth in the lesion. She had hypertension and sometimes headache. Cranial computerized tomography (CT) findings were normal. She was 130 cm in height and had macrocephaly, with rhizomelic extremity shortening consistent with the clinical appearance of achondroplasia. On ophthalmologic examination, visual acuity was 20/20 and intraocular pressure was 13 mmHg in both eyes. Slit lamp examination was normal in both eyes. On dilated fundus examination of the left eye, there was a dark brown, finger-like lesion without subretinal fluid and atrophy on inferior part of the optic disc, and the lesion extended to juxtapapillary choroid (Figure 1a). Fluorescein angiography revealed hypofluorescent lesion in inferior part of the optic disc; the lesion became longer, extending to the adjacent choroid as seen in colour picture (Figure 1b). There was no subretinal fluid accumulation, leakage or choroidal neovascularisation. Macula and peripheral retinal areas were normal in both eyes. Optical coherence tomography showed high hyperreflectivity on the optic nerve head with a shadowing behind the lesion (Figure 2). Visual field was normal in the right eye, and minimal enlargement of the blind spot and supero-temporal narrowing of visual field was discovered in the left eye.

CASE 2

A 29-year-old woman applied to neurology department with headache which had been continuing for 6 months; she was referred to our clinic for the

lesion on the optic disc. On her examination, visual acuity was 20/20, and intraocular pressure (IOP) was within normal limits. Slit lamp examination was also normal in both eyes. Dilated fundus examination showed bilateral optic disc hypoplasia and melanocytoma of the optic disc in the right eye (Figure 3a). Fluorescein angiography revealed hypofluorescent lesion, which was extending to adjacent choroid, on the supero-nasal of the optic disc, and there was no neovascularisation or leakage (Figure 3b). Visual field was normal except minimal enlargement of the blind spot. On magnetic resonance imaging, a 1.5 x 1.0 cm arachnoid cyst was determined in the left temporal fossa (Figure 4). Neurological examination was normal, and no other central nervous system abnormalities were found.

Written informed consent was obtained from each patient before study.

DISCUSSION

Up to date, there are no proven systemic associations of melanocytoma of the optic disc. In their study involving 115 cases with ocular melanocytosis, Shields et al reported that only 8% had a significant ocular association.³ They also added that associations such as racial melanosis, optic disc hypoplasia, retinitis pigmentosa, and congenital hypertrophy of the retinal pigment epithelium might be coincidental. In this study, association of optic disc hypoplasia with melanocytoma of the optic disc was reported to be 2%.³ In one of our cases, there was bilateral optic disc hypoplasia with melanocytoma of the optic disc; however, this association could be coincidental as reported by Shield et al.³

Although studies have reported systemic associations of melanocytoma of the optic disc, it is generally indicated that these systemic findings may probably be coincidental. The studies carried out so far consist generally of limited number of case reports, and in these reports, it is not known whether there is any association. In another report, in which melanocytoma of the optic disc in a patient with neurofibromatosis 2 is presented, it is claimed

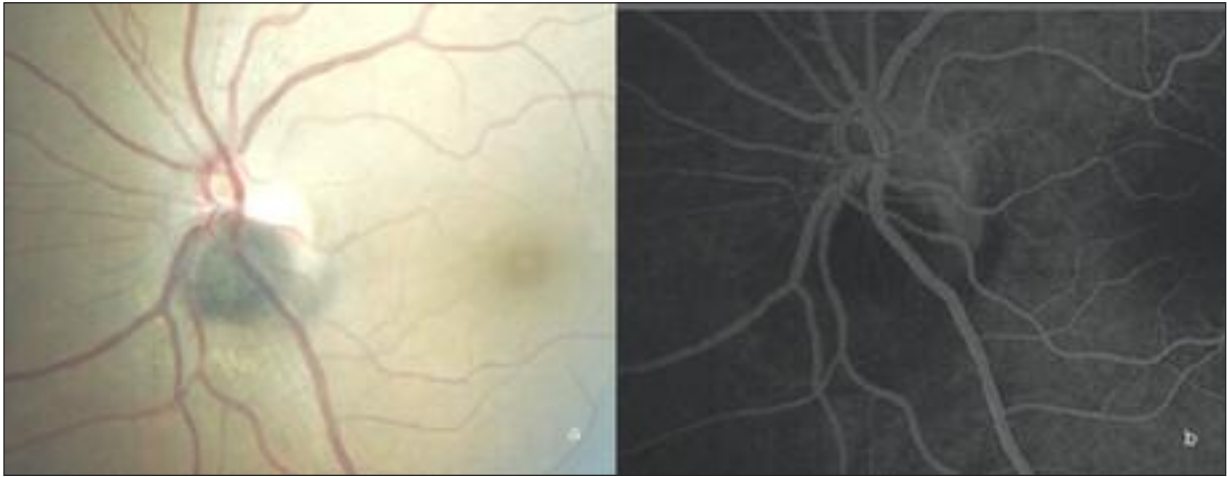


FIGURE 1: a. Dark brown tumoral lesion on inferior part of the optic disc in the left eye of Case 1. b. Fluorescein angiography shows hypofluorescent lesion extending adjacent choroid in Case 1.

that as in other hamartomas associated with neurofibromatosis 1 and 2, melanocytoma is a lesion of neural crest origin, and that melanocytoma in a patient with neurofibromatosis may not be coincidental.⁵ In some other case reports, in which association of intracranial meningioma together with melanocytoma of the optic disc has been reported, authors suggest that melanocytoma of the optic disc may be related with intracranial meningioma in terms of embryologic origin.^{4,6}

In our cases, there was achondroplasia with melanocytoma of the optic disc in Case 1, while there was arachnoid cyst associated with melanocytoma of the optic disc in Case 2. Arachnoid cysts are intra-arachnoid fluid collections that account for about 1% of all intracranial space-occupying lesions, and can be congenital or secondary. The etiology of arachnoid cysts has been a controversial and still remains unclear, however it is claimed that they might arise from a developmental defect in condensation of the mesenchyme or from abnormalities of cerebrospinal flow.⁷ Achondroplasia is the most common hereditary dwarfism. It is characterized by rhizomelic limb shortening, macrocephaly, midface hypoplasia with frontal bossing and short broad hands with a trident finger configuration. The phenotype of achondroplasia is related to a disturbance in endochondral bone formation, due to a mutation in the fibroblast growth factor receptor-3.⁸

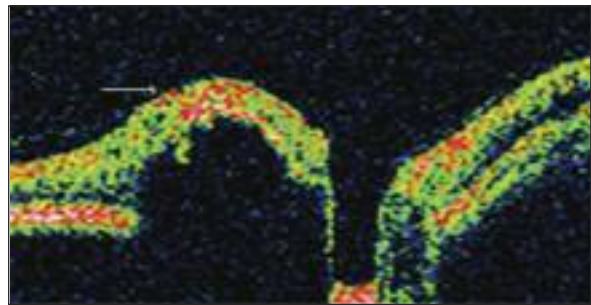


FIGURE 2: In the left eye of Case 1, melanocytoma of the optic disc in optical coherence tomography



FIGURE 3: a. Optic disc hypoplasia and melanocytoma of the optic disc in the right eye of Case 2. b. Fluorescein angiography demonstrates hypofluorescent lesion extending to the adjacent choroid, on the supero-nasal optic disc in the right eye of Case 2.

So far, we have encountered no studies reporting the association of an arachnoid cyst or achondroplasia with melanocytoma of the optic disc in the literature. Any relationship among these disorders and melanocytoma of the optic disc in



FIGURE 4: In Case 2, magnetic resonance imaging shows arachnoid cyst in the left temporal fossa.

terms of origin has not been found. Melanocytoma of the optic disc arise from neurochromaphin cells, but arachnoid cysts originate from mesenchyma. Achondroplasia is mainly an osteochondral disorder.

In conclusion, with regard to the association of melanocytoma of the optic disc with arachnoid cyst or achondroplasia, we couldn't put forward a pathogenetic mechanism. Therefore, further studies are needed as to whether these disorders form a syndrome with melanocytoma of the optic disc although the origin of achondroplasia and arachnoid cyst has not shown any relationship with melanocytoma of the optic disc. However, we suggest that systemic investigations must be performed to reveal any possible associations in patients with melanocytoma of the optic disc.

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