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A Case of Ocular Neurofibromatosis

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Ocular involvement of multiple neurofibromatosis is very infrequently reported. Localized tumors occurring along the course of the ciliary nerve and diffuse neurofibromatosis of the choroid have been described along with pigmented tumors of the uveal tract and buphthalmos[1-5]. However, intra-ocular neurofibromatosis has not been described in Korean literature, although there are several cases of neurofibromatosis involving eyelid or other extraocular tissue[6,7]. This is a report of a case of multiple neurofibromatosis which showed characteristic lesions in the eyeball.

Case: This 21 year old female came to the Seoul National University Hospital because of a soft cystic mass on her left face on December 8, 1982. She developed a bean sized left preauricular mass at the age of 7 years. And by 11 years of age her left eye became very poor in vision. No family history of neurofibromatosis was elicited. She underwent a rhytidectomy at Seoul St. Mary Hospital 5 years ago. The biopsy report at that time was neurofibroma. On examination she was found to have multiple cafe-au-lait spots on anterior chest and back. The left eye showed a high myopic tigroid fundus and a normal anterior segment. There was no vision at all. The visual acuity of the right eye was also decreased. The remainder of the physical examination was unremarkable. Laboratory data showed a normal hemogram and blood chemistries. Computed tomography of the skull showed a soft tissue mass in the left face and a double contoured sellar floor. On January 20, 1983, the right eye enucleation with a prosthetic replacement was performed.

Grossly the eyeball measured 2.5 cm in anterior-posterior diameter and 2.3 cm in transverse diameter. The cornea measured 1.0 cm in diameter and was clear. Cut section showed the lens intact and 0.7 cm long, and the anterior chamber was filled with serous fluid (Fig. 1). The vitreous cavity was filled with yellowish gelatinous material. Detached retinal tissue was found to be attached to the posterior margin of the lens. The optic nerve cut at its bulbar attachment was unremarkable. Microscopically, the cornea showed a focal chronic inflammatory reaction. There were capillary proliferation and plasma cell infiltration at limbus portion of the cornea as well. Occasional pigmented cells were encountered along the basement membrane of the corneal epithelium. Corneal staphyloma was seen in one portion. The corneal stroma was scattered with capillaries and a few neutrophils. The iris contained increased amount of melanin and plasma cell infiltration. The ciliary body was enlarged containing ganglion cells, glial cells and proliferated Schwann cells (Fig. 3). The ciliary process is abnormally elongated and interadherent. Focal episcleritis of plasma cell type was seen. The sclera itself was thickened and hyalinized. There were several neurofibromatous growths in surrounding fibrofatty tissue of the sclera. Those were in part of plexiform type. The optic nerve appeared to be thick and was totally replaced by neurofibromatous tissue. The optic nerve sheath was unremarkable. The retina was totally detached

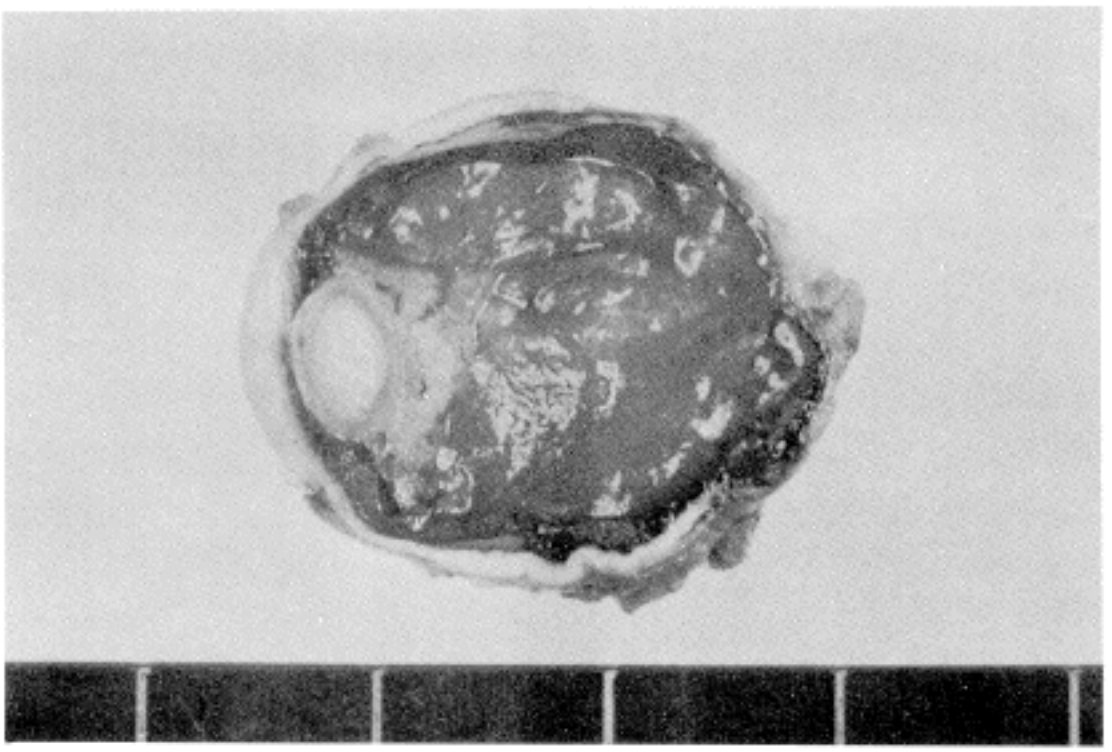

Fig. 1. Mid sagittal section of the eyeball, showing irregular thickenings of the wall. Abnormal proliferation of ciliary body and iris as well as choroid and retina are seen.

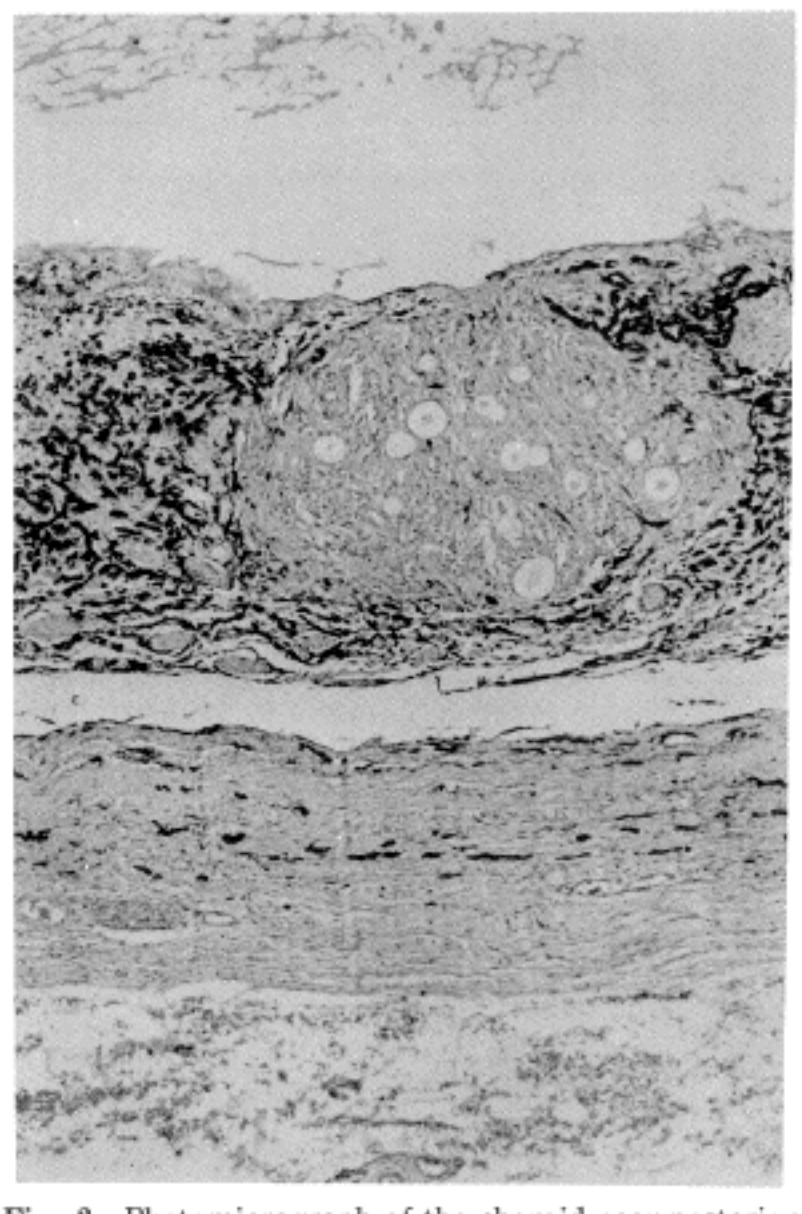

Fig. 2. Photomicrograph of the choroid near posterior pole, showing a nodule composed of hamartomatous ganglioneuromatous proliferation. H & E ×100

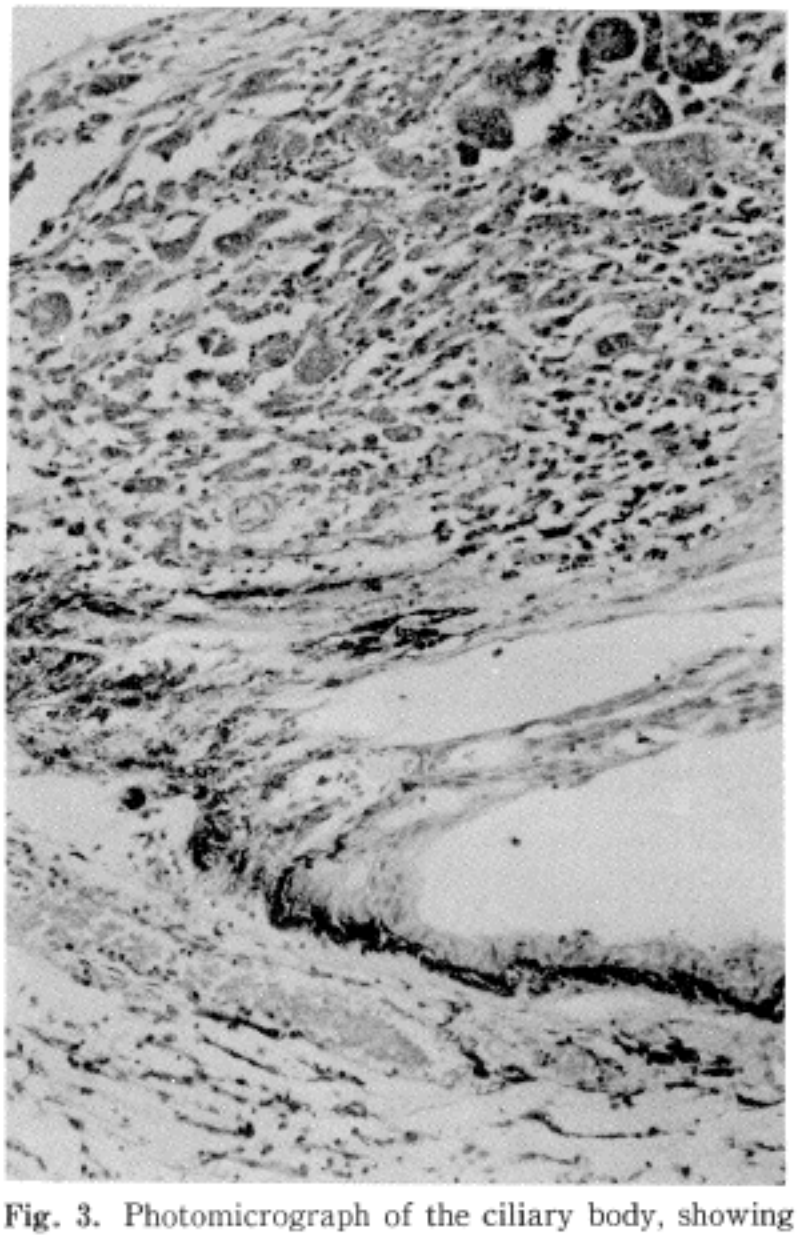

Fig. 3. Photomicrograph of the ciliary body, showing large pigmented neural cells and other proliferating cells. H & E ×250

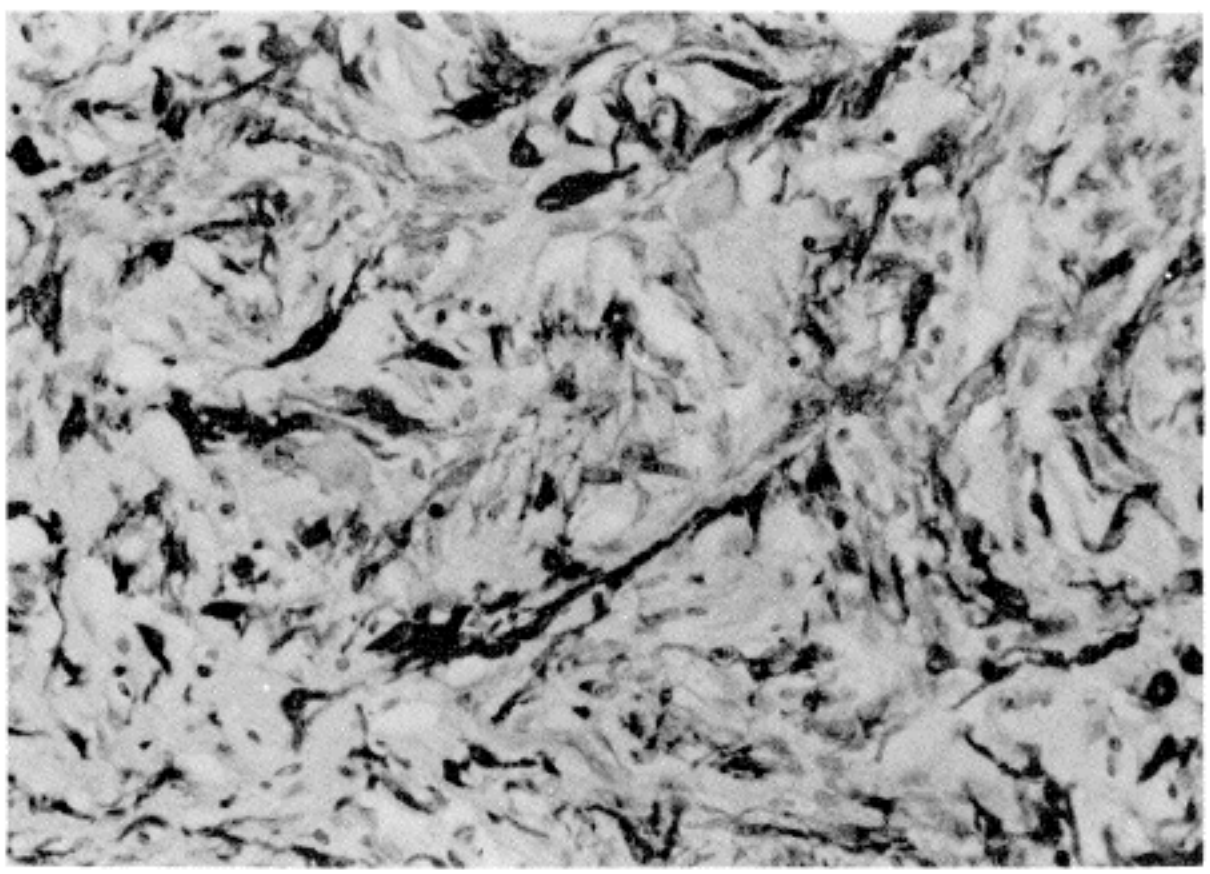

Fig. 4. Photomicrograph of the choroid, showing proliferation of neural cells (choroidal hamartoma) inbetween pigmented cells. H & E ×250

and there was an organized subretinal exudate. The detached retina was conglomerated behind the lens. Tubular epithelial structures mimicking dysplastic retina were seen along with irregular proliferation of neuroglial tissue (Fig. 2). The choroid appeared most unusual showing multiple foci of ganglioneuromatous nodules comprising of distinct large ganglion cells and some satellite cells (Fig. 2). In addition to this numerous nerve plexi proliferative spindle cells were seen in this zone. Heavy pigmentation accompanies this neuromatous growth (Fig. 4). The choroid in general was thick and irregular. The vessels were normally prominent without any specific abnormalities. Collagenous connective tissue was deposited in the interphase between clustered ciliary processes and dysplastic retinal tissue. Hemosiderin deposits were seen around these areas.

On May, 1983 she was again admitted because of left supraorbital and preauricular growing masses. These masses were removed and rhytidectomy was done. The tumor mass received was 6×3.5×1.5 cm in size and showed plexiform neurofibroma.

Comment and summary: Neurofibromatosis is one of the neurocristopathies that involve many system or tissues forming various types of lesion. Almost every tissue or organ can be involved by this disease. However, the eyeball itself is very rarely affected by this process. The findings seen in our case indicate the diversity of lesions in neurofibromatosis, and also suggest hamartomatous nature. Heterotopic ganglion cells and glial cells in uveal tract are not easily understood.

We reported here a case of intra-ocular neurofibromatosis with its characteristic involvement of the uveal tract, in a 21 year old female. Her ocular symptoms began at her age of 11 as poor vision and were slowly progressive together with multiple facial neurofibromas. The involved left eyeball showed many ganglioneuroglial cell nests in iris, ciliary body and retina. Minute plexiform neurofibromas were also seen in small nerve twigs around the eyeball.

Key Words: neurofibromatosis, eyeball, hamartoma, uveal tractanomaly

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=국문초록=

안구 신경섬유종증의 1예

서울대학교 의과대학 병리학교실

지 제 근·박 인 애

21세 여자에서 발생한 안구의 신경섬유종증 1예를 보고 하였다. 본예는 다발성 신경섬유종증 환자로서 11세부터 시력 감소로 시작된 안구 침범 증상이 나타났으며 결국 실명에 이르게 되어 좌측 안구 적출술을 시행 받았다.

안구는 그 내부구조를 거의 모두 침범하고 있는 특징적 병변을 나타내었는데 특히 홍채, 모양체, 맥락 그리고 망막을 미만성으로 침범한 과오종성 신경성장이 눈에 띄었고 그 외에도 안구내외의 신경에도 신경섬유종의 소견을 나타내었다.