Retinal myelinated nerve fibers associated with macular pseudohole

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Il n’y a aucun lien financier pouvant faire l’objet d’un conflit d’intérêt.

INTRODUCTION

Myelination of the visual system normally begins at 7 months gestational age [1], at the optic tracts, and progresses anteriorly [2]. It is normally complete within 3 months after birth, stopping at the lamina cribrosa. However, it may proceed past this level and be noted as an area of retinal whitening. This process is thought to be complete shortly after birth. Although retinal myelinated nerve fibers are benign lesions, they are rarely associated with retinal abnormalities: retinal telangiectasis, hemorrhages, neovascularization, vascular occlusions [3], cranial-facial lesions, coloboma of the iris, keratoconus, and myopia/strabismus/ambylopyia [4]. A 0.54% incidence of myelinated nerve fibers was found in a large series of eyes examined at autopsy [5]. This has been associated with axial myopia, ambylopyia, strabismus, and nystagmus [5], or multisystem congenital disorders such as Gorlin’s syndrome [6], autosomal dominant vitreoretinopathy with skeletal malformations [7], Down syndrome, neurofibromatosis and various dyscranias [8] and vitreomacular traction syndrome [9].

CASE REPORT

Our observation presents a 24-year-old patient who experienced a sud-
den decrease in visual acuity in the left eye, with unilateral extensive peripapillary myelination of the retinal nerve fiber layer with macular pseudohole and bilateral low myopia. We did the following tests: visual acuity, slit lamp biomicroscopy, Humphrey automated perimetry (Macula 10-2, Sita-fast 30-2 and 120 Screening Points tests), fundus color photography and optical coherence tomography Stratus® OCT (line 0°–90°, fast macular thickness, fast optic disc, and retinal nerve fiber layer tests).

A familial evaluation (father, mother, sister, brother, grandfather, and grandmother) of the ocular fundus was performed, which showed no abnormalities. The young woman, of eutocic childbirth, had no history of any systemic illness or trauma and was on no general or local medication. The visual acuity was 20/30, 20/20 with −0.75 spherical equivalent in the right eye and 20/40, 20/25 with −0.75 spherical equivalent in the left eye. The slit lamp biomicroscopy showed a normal anterior segment. The fundus oculi demonstrated extensive papillary and peripapillary myelination in the left eye (fig. 1): the patch was located at the superior-inferior sectors of the optic nerve head and along the superior-inferior retinal vascular arcades masking the lower vessels. A posterior vitreous detachment was not present: no Weiss ring was seen on biomicroscopy. In the macular region, altered light reflex, donut-shaped yellow ring, approximately 200–300 μm in size, centered on the foveola, darker appearance of the fovea, and steepening of the normal foveal depression was discovered but no myelinated nerve fibers were observed. There was a translucent membrane with fine, radiating folds and very small tortuosity of temporal perimacular vessels. An intraretinal pseudocyst occupied the inner part of the fovea and the foveal floor was elevated: on biomicroscopy, a stage 1B hole was suspected (fig. 1) [10]. The Humphrey tests showed an enlargement of blind spots of varying extent, corresponding to the area of myelination (fig. 2). The OCT Line test (5-mm-long horizontal section) showed superficial diffuse hyperreflectivity along the myelinated nerve fiber area with deep hypodensity and a retinoschisis with a pseudohole aspect in the inner part of the foveola, darker appearance of the fovea, and steepening of the normal foveal depression was discovered but no myelinated nerve fibers were observed. There was a translucent membrane with fine, radiating folds and very small tortuosity of temporal perimacular vessels. An intraretinal pseudocyst occupied the inner part of the fovea and the foveal floor was elevated: on biomicroscopy, a stage 1B hole was suspected (fig. 1) [10]. The Humphrey tests showed an enlargement of blind spots of varying extent, corresponding to the area of myelination (fig. 2). The OCT Line test (5-mm-long horizontal section) showed superficial diffuse hyperreflectivity along the myelinated nerve fiber area with deep hypodensity and a retinoschisis with a pseudohole aspect in the inner part of the fovea and a clear increase in macular thickness (fig. 3). In the macular area, the Line test showed a steepening foveal contour and a reflective epiretinal membrane layer on the surface of the retina: a posterior vitreous detachment was not present. An intrafoveal split or retinoschisis occupied the inner part of the fovea, resulting in foveolar thickening and elevation of the foveal floor. Macular pseudohole was diagnosed (fig. 4). The retinal nerve fiber layer (RNFL) test may not have been reliable in the left eye, although several measurements were taken. The RNFL test of right eye was normal. The Fast Optic Disc test showed superficial diffuse hyperreflectivity along the myelinated nerve fiber area with deep hypodensity and the lack of physiological excavation of the optic nerve head with concavity on the vitreous camera (fig. 5).

DISCUSSION

The histopathologic evidence suggests that the abnormal myelination of retinal nerve fibers was caused by a collection of what we assume to be oligodendrocytes within the inner retina [5] with no concomitant inflammatory process that some authors call choristoma, defined as a congenital overgrowth of microscopically normal tissue in an inappropriate place [2, 5, 11]. The myelinated patches in the human retina contained a mixture of unmyelinated and myelinated axons. These fibers were larger in diameter than fibers found within normal areas of the retina or within the optic nerve [11]: they block light transmission, explaining the visual impairment of the visual field. We could not determine whether these myelin patches were in a dynamic phase of myelination, remyelination, or stability. Intraretinal myelination has been considered a nonprogressive disorder [5]. In addition, other authors have reported progressive cases of myelination [1]. The clinical and pathogenic features between myelination and the macular pseudohole are discussed. The epimacular membrane is an avascular, fibrocellular membrane that proliferates on the surface of the retina: these cells, once in contact and attached to the retina, may proliferate and form sheets of membranes over the surface of the retina. Through their contractile properties, the underlying retina is in turn distorted [12]. Earlier reports proposed that glial cells (primarily fibrous astrocytes) from the inner layers of the neurosensory retina proliferated through breaks in the internal limiting membrane produced after a retinal tear or a posterior vitreous detachment. Therefore we can suggest that myelinated retinal nerve fibers may be the primary cause in the dysfunction of the internal limiting membrane.

CONCLUSION

Retinal myelinated nerve fibers are a benign pathology. However, it requires a periodic check-up when there is wide retinal extension or a decrease in visual acuity and retinal alterations, as described here. The myelination–epiretinal membrane associated with macular pseudohole is not frequent and the relationships between the two pathologies is difficult to explain, therefore requiring observation and further studies. Follow-up will make it possible to evaluate the progression of macular pseudohole and myelination.
Figure 1: Fundus photograph. Myelination of retinal nerve fibers originates on the entire optic nerve head and extends along the superior and inferior vascular arcades with macular pseudohole.

Figure 2: Visual field (Sita-fast 30). Enlarged blind spot of varying extent, corresponding to the area of myelination.

Figure 3: OCT (5-mm-long horizontal section). Epiretinal membrane with superficial diffuse hyperreflectivity along the myelinated nerve fiber area with deep hypodensity (left) and macular pseudohole with evident increase in macular thickness (center).

Figure 4: OCT (5-mm-long vertical section). Retinoschisis in the inner part of the fovea and a clear increase in macular thickness (macular map, 6-mm-diameter area). Note the overlying epiretinal membrane that causes wrinkling of the overlying retina.

Figure 5: Fast Optic Disc test superficial diffuse hyperreflectivity along the myelinated nerve fiber area with deep hypodensity and the lack of physiological excavation of the optic nerve head, with presence of concavity on the vitreous camera.
REFERENCES