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/amlyopia [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, amlyopia, 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 unilater
teral 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 peri-
metry (Macula 10-2, Sita-fast 30-2 and 120 Screening
Points tests), fundus color photography and optical co-
herence tomography Stratus® OCT (line $0^\circ$–$90^\circ$, 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 an-
terior segment. The fundus oculi demonstrated exten-
sive papillary and peripapillary myelination in the left
eye (fig. 1): the patch was located at the superior-infe-
rior sectors of the optic nerve head and along the su-
perior-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, cente-
red on the foveola, darker appearance of the fovea, and
steepening of the normal foveal depression was disco-
overed 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 enlarge-
ment of blind spots of varying extent, corresponding to
the area of myelination (fig. 2). The OCT Line test (5-
mmand horizontal section) showed superficial diffuse
hyperreflectivity along the myelinated nerve fiber area
with deep hypodensity and a retinoschisis with a pseu-
dohole 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 sur-
face of the retina: a posterior vitreous detachment was
not present. An intrafoveal split or retinoschisis occu-
pied the inner part of the foveola, resulting in foveolar
thickening and elevation of the foveal floor. Macular
pseudohole was diagnosed (fig. 4). The retinal nerve fi-
ber 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 hypo-
density 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 abnor-
mal myelination of retinal nerve fibers was caused by
a collection of what we assume to be oligodendrocy-
tes within the inner retina [5] with no concomitant in-
flammatory process that some authors call choristoma,
deined 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 vi-
sual impairment of the visual field. We could not
determine whether these myelin patches were in a dy-
namic phase of myelination, remyelination, or stability.
Intraretinal myelination has been considered a nonpro-
gressive 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 epi-
macular membrane is an avascular, fibrocellular mem-
brane 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 neuro-
sensory retina proliferated through breaks in the inter-
nal 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 limi-
ting 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 pseudo-
hole is not frequent and the relationships between the
two pathologies is difficult to explain, therefore requi-
ing 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