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Macular hole retinal detachment in extensive myelinated retinal nerve fiber and high myopia with Straatsma syndrome: A case report

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ABSTRACT

Purpose: We present a case of macular hole retinal detachment (MHRD) in a patient with Straatsma syndrome, a rare condition characterized by extensive unilateral myelinated retinal nerve fibers (MRNF), high myopia, and amblyopia. This study aimed to highlight the clinical presentations, diagnostic challenges, and success of surgical interventions.

Observation: A 32-year-old Asian woman with a history of high myopia and poorly corrected vision in her right eye since childhood presented with a sudden loss of vision in the right eye. Examination revealed extensive MRNF and retinal detachment with a macular hole. A standard three-port pars plana vitrectomy was performed, and tight vitreous retinal adhesions were observed. PFCL-assisted inverted internal limiting membrane (ILM) flap technique was performed. Silicone oil was used owing to its tight vitreous retinal adhesion. Postoperatively, the macular hole was closed, the retina was reattached, and partial disappearance of the MRNF was observed.

Conclusion and importance: This case report describes a successful surgical intervention for MHRD associated with Straatsma syndrome. The PFCL-assisted inverted ILM flap technique is effective for managing complicated cases of MHRD. The partial disappearance of MRNF after vitrectomy suggests potential nerve fiber layer damage, possibly due to retinal detachment or the use of silicone oil. This case highlights the unique features of MHRD, a rare disease associated with Straatsma syndrome.

1. Introduction

Myelinated retinal nerve fiber (MRNF) is a developmental anomaly caused by the migration of oligodendrocytes into the retina, which is normally prevented by the lamina cribosa. Straatsma syndrome,[1] first reported in 1979, is a rare disease characterized by extensive unilateral MRNF with myopia and amblyopia.

This report presents a rare case of vitrectomy for macular hole retinal detachment (MHRD) associated with Straatsma syndrome. Our study highlights the clinical presentation and success of surgical intervention for this rare and complex condition.

2. Case report

A 32-year-old Asian woman presented with sudden loss of vision in the right eye. The patient had a history of high myopia in both eyes. Since childhood, the patient had poorly corrected vision in the right eye, measuring 20/600. However, the left eye of the patient maintained normal corrected vision. The disparity in visual acuity has been a consistent issue throughout life; however, the sudden deterioration was a new development. The patient had no history of systemic illnesses. There was no family history of poor vision or known hereditary ocular conditions. At the initial examination, the patient's best-corrected visual acuity (BCVA) was 20/500 in the right eye with a refraction of −17.5 diopter sphere and −2.5 diopter cylinder at 10°, and 20/20 in the left eye with a refraction of −13.0 diopter sphere −1.75 diopter cylinder at 170°. Intraocular pressure (IOP) was 14 mmHg in both eyes. The axial lengths were 27.93 mm and 28.62 mm in the right and left eyes, respectively. Slit-lamp examination (SLE) was within normal limits, with a normal anterior segment and no cataracts.

In fundus examinations, the retina of the right eye displayed a large area of MRNF and macular hole with subretinal fluid, whereas the left eye displayed a normal fundus (Fig. 1a–d). Optical coherence tomography (OCT) showed retinal detachment with a macular hole. OCT also revealed a hyper-reflective retinal nerve fiber layer in the parafoveal area around the superior and inferior arcade vessels (Fig. 1b). We

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diagnosed MHRD associated with Straatsma syndrome.

The patient underwent a standard three-port pars plana vitrectomy using a 27-gauge vitrectomy system. Posterior vitreous detachment (PVD) did not occur; therefore, we attempted to induce PVD. However, the vitreous hyaloid was tightly adhered to the MRNF lesion in the posterior segment and temporal mid-periphery retina, preventing the complete induction of PVD. After fluid-gas exchange, perfluorocarbon (PFCL) was used in the PFCL-assisted inverted internal limiting membrane (ILM) flap technique (Fig. 2). We applied brilliant blue G to visualize the ILM, and the ILM surrounding the macular hole was carefully peeled and inserted into the macular hole (Fig. 2). We observed that the ILM was fragile and tightly adherent, particularly over the MRNF lesion. Owing to concerns regarding traction from the remaining hyaloid, silicone oil was used. After vitrectomy, the macular hole was closed, and the retina was successfully attached (Fig. 3). Corrected visual acuity in the right eye after retinal reattachment was 20/300. Silicone oil was removed 4 months after the initial surgery. We noted partial disappearance of MRNF after vitrectomy, as demonstrated in both fundus photographs and OCT scans (Fig. 4). MRNF disappearance started 3 months after the initial vitrectomy. Even after silicone oil removal, the hyperreflective retinal nerve fiber layer by OCT in the inferior arcade vessels was thinner throughout the course of post-vitrectomy.

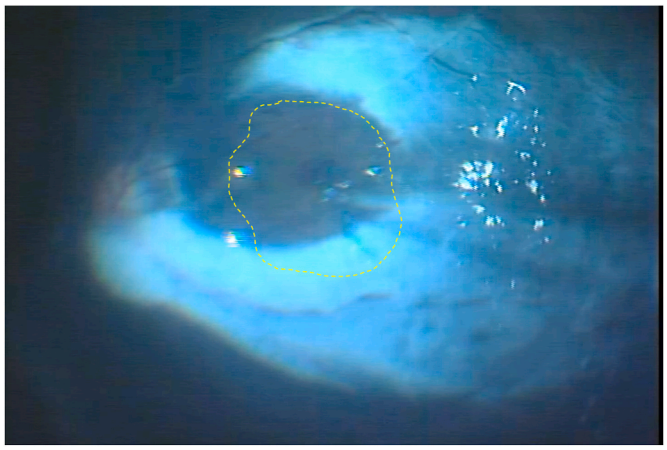

Fig. 2. Perfluorocarbon assisted inverted ILM flap technique in this case. ILM surrounding the macular hole is peeled and inserted into the macular hole. Yellow dash indicates ILM peeled area. ILM on the MRNF lesion of the inferior arcade, which disappeared later is not peeled. (For interpretation of the references to colour in this figure legend, the reader is referred to the Web version of this article.)

3. Discussion

We report the MHRD associated with Straatsma syndrome that underwent vitrectomy. Additionally, we observed unique findings of firm vitreoretinal adhesions in a wide area of the retina and partial disappearance of MRNF after vitrectomy.

MRNF is a congenital thickening of retinal ganglion cell axons that appears as a white patch with a feathery margin in the inner retina, with an estimated prevalence of 0.5–1% of the population.[2–4]

Straatsma syndrome is a rare congenital anomaly marked by extensive unilateral MRNF, high myopia, and amblyopia.[1] The rarity of the condition and the complexity of its manifestations pose significant challenges in diagnosis and management. Here, the patient presented with sudden loss of vision in the right eye, a history of high myopia, and consistent poorly corrected vision in the affected eye since childhood.

MHRD is a severe complication in highly myopic eyes and is relevant to tangential traction by epiretinal vitreoretinal adhesions in high myopia.[5] The surgical approaches for MHRD have been commonly applied in vitrectomy with ILM removal and gas or silicone oil tamponade. Several recent reports have demonstrated that the postoperative MH closure rate, retinal reattachment rate, and postoperative BCVA may yield better outcomes with the inverted ILM flap technique than with ILM peeling for myopic MHRD.[6–11] Recently, the perfluorocarbon liquid–assisted inverted ILM flap technique is found to be effective.[12,13]

Several vitreoretinal diseases associated with MRNF have been reported.[14] These include retinal breaks in the area of the MRNF,[15] retinal detachment,[16,17] macular hole,[18] and vitreomacular traction syndrome.[19] All previous surgical studies indicate strong vitreoretinal

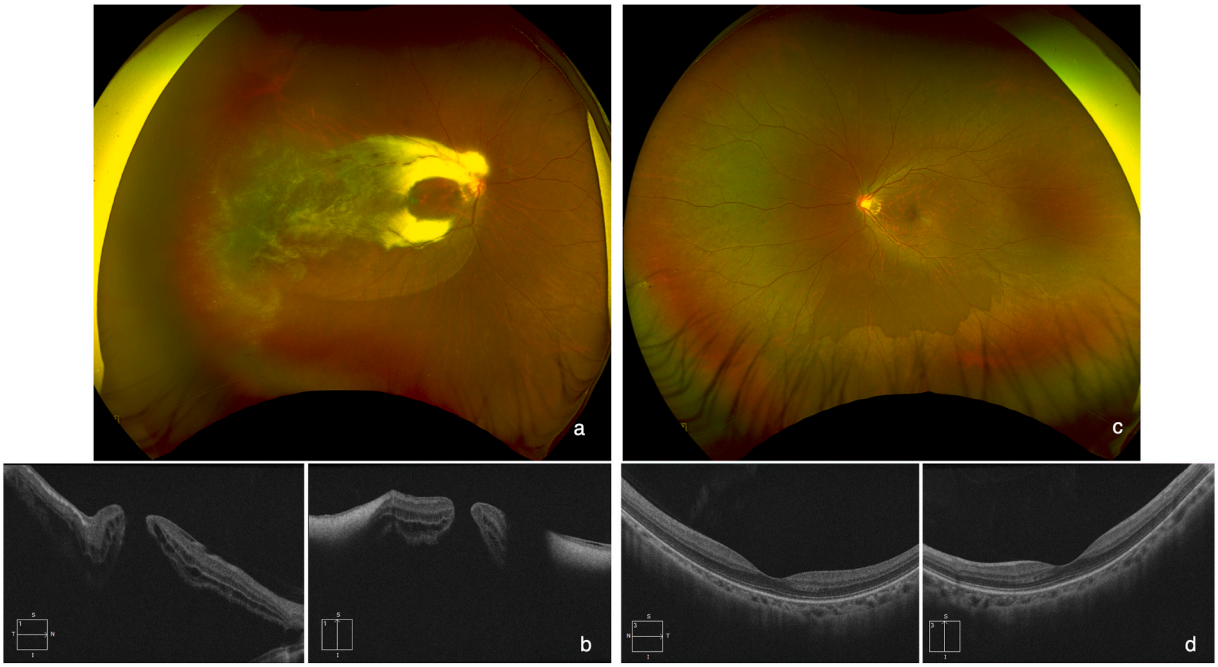

Fig. 1. Fundus examination at the initial visit (a) Fundus photo of the right eye shows extensive myelination of the retinal nerve fiber layer. (b) OCT of the right eye reveals retinal detachment with a macular hole. (c) Normal fundus of the left eye (d) Normal OCT of the left eye.

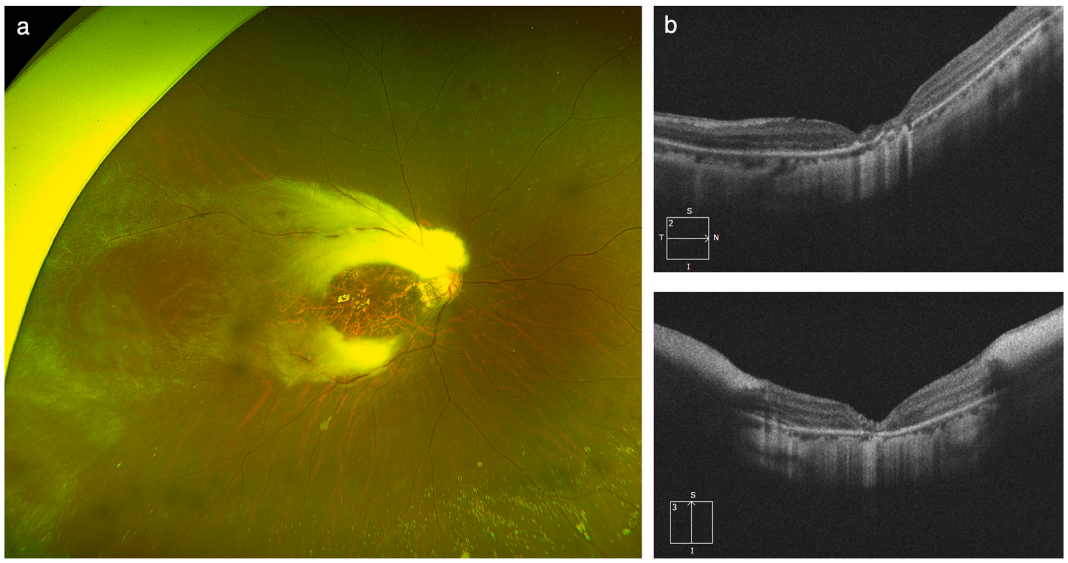

Fig. 3. Fundus examination at postoperative day 7. (a) The fundus photo of the right eye shows retinal reattachment with silicone oil. (b) OCT shows macular hole closure.

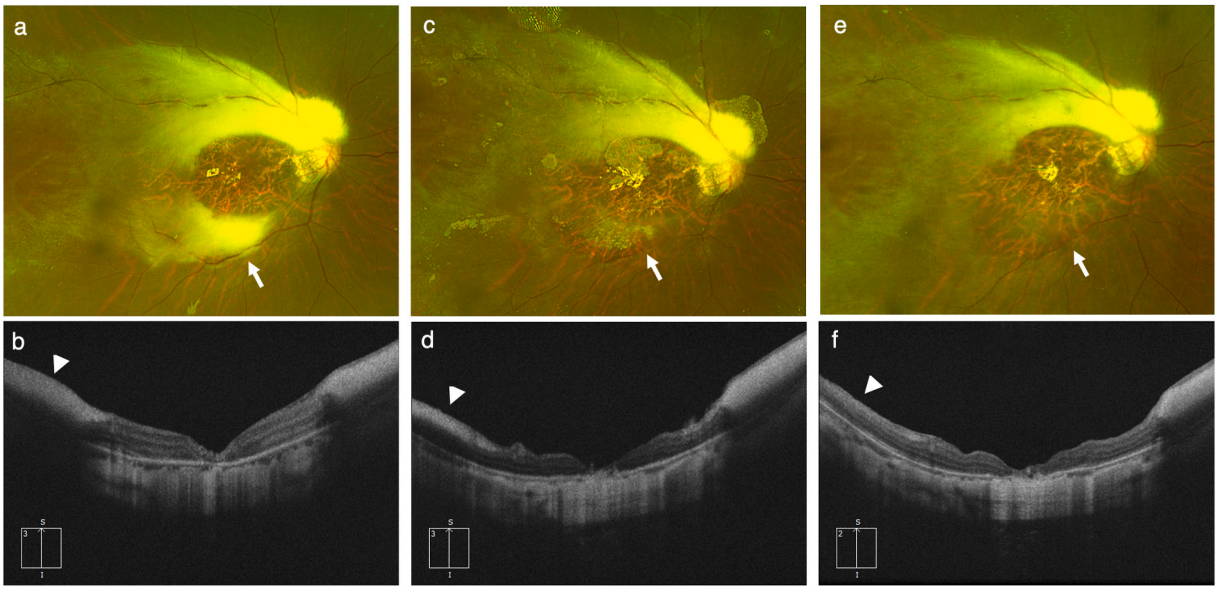

Fig. 4. The time course of partial disappearance of myelinated retinal nerve fiber. (a)(b) postoperative week 1, (c) (d) postoperative month 4, (e)(f) postoperative month 10. The arrows indicate the MRNF lesion of the inferior arcade, and the arrowhead indicates corresponding lesion in the OCT. The disappearance of MRNF in the fundus photos and the thinning of hyperreflective lesion in the OCT is observed starting at postoperative month 4 and through at postoperative month 10.

adhesion in the area of MRNF.[16–18] We found that five cases have been reported describing the development of MHRD in the context of Straatsma syndrome.[20–24] Four cases were treated with vitrectomy and ILM peeling. In one of the five cases, the retina redetached two weeks postoperatively.[23] Yang et al. described that a PVD could not be released from the area of MRNF, and the ILM around the macular hole was left unremoved because it was fragile and tightly adherent.[22] Zhou et al. reported that PVD induction was unsuccessful due to firm vitreoretinal adhesion; although the retina reattached, the macular hole remained unclosed.[24]

In our case, during vitrectomy, induction of PVD was challenging because of the tight adhesion of the vitreous hyaloid to the MRNF lesion and ILM peeling was difficult because it is fragile and tightly adherent. This abnormal vitreoretinal adhesion may necessitate the use of specialized techniques to ensure successful surgical outcomes. The PFCL-assisted inverted ILM flap technique was employed to facilitate macular hole closure, demonstrating its effectiveness in managing complex cases. The use of silicone oil was a strategic decision to mitigate the risk of retinal traction from the remaining hyaloid, thereby ensuring retinal stability postoperatively. Successful closure of the macular hole and reattachment of the retina are significant achievements, highlighting the importance of tailored surgical approaches in managing MHRD associated with Straatsma syndrome.

An intriguing observation in this case was the partial disappearance of the MRNF following vitrectomy, as evidenced by fundus photographs and OCT scans. The disappearance of MRNF is reported in post-vitrectomy for the epiretinal membrane,[25] plaque radiotherapy for choroidal melanoma,[26] central retinal artery occlusion,[27] chronic papilledema,[28] and glaucoma.[29] This phenomenon may be attributed to nerve fiber layer damage caused by retinal detachment or the ILM

peeling process. However, at this time, the ILM in the MRNF lesion of the inferior part was not peeled, and the subretinal fluid exceeded the inferior arcade but not the superior arcade. Furthermore, the use of silicone oil may induce ganglion cell damage.[30–32] Indeed, out of the four cases that reported MHRD associated with Straatsma syndrome, silicone oil was used in three cases.[21–23] In one of these cases, a decrease in the extent of MRNF was mentioned.[21] The exact mechanism underlying this observation remains unclear and warrants further investigation. Continued documentation and analysis of similar cases are essential to advance treatment strategies and enhance patient outcomes owing to the complexity and rarity of this condition.

4. Conclusion

Herein, we report the successful surgical intervention for MHRD in a patient with Straatsma syndrome, a rare condition characterized by extensive unilateral MRNF, myopia, and amblyopia. Our case highlights the complexity of managing MHRD in the presence of MRNF, and emphasizes the need for meticulous surgical planning and technical adaptation to address abnormal vitreoretinal adhesions. The observed partial disappearance of MRNF post-vitrectomy suggests potential nerve fiber layer damage, which may be due to retinal detachment or the use of silicone oil. This report provides valuable insights into the treatment of this rare condition, paving the way for future studies and improving the management strategies for patients with Straatsma syndrome.

Patient consent

Written consent has been obtained from the patient to publish this case report.

Acknowledgment and disclosures

The authors declare no conflicts of interest. All the authors attest that they meet the current ICMJE criteria for authorship.

Submission declaration

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CRediT authorship contribution statement

Ayumi Usui-Ouchi: Writing – original draft, Visualization, Software, Resources, Project administration, Methodology, Investigation, Funding acquisition, Conceptualization. **Nobuyuki Ebihara:** Writing – review & editing, Supervision. **Shintaro Nakao:** Writing – review & editing, Supervision.

Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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