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Case report

Pseudo-duplication of the optic disc with maculo-schisis in a 9-year-old patient

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ABSTRACT

Purpose: To report a unique case of pseudo-duplication of the optic disc with maculo-schisis.
Observations: A 9-year-old girl presented with decreased vision in the left eye. Her measured visual acuity was 20/50. Dilated fundus examination revealed an optic disc-like lesion with a large crater-like depression, pale color and aberrant retinal vasculature arising from its lower region. Optical coherence tomography (OCT) demonstrated maculo-schisis emerging from the optic disc complex and involving the foveal region. Further pathological findings of cellophane maculopathy with retinal striae was observed at the papillo-macular bundle. On MRI scan there were neither signs of doubling of the optic nerve nor any other optic nerve malformations. The patient underwent 25 Gauge Pars Plana vitrectomy (PPV) with posterior hyaloid peel, ILM peel and gas tamponade with SF6. Eight month post-operatively, a significant reduction in intra-retinal fluids and an improvement in the maculo-schisis magnitude were seen.
Conclusions and Importance: To the best of our knowledge, this is the first reported case of a pseudo-duplication of the optic disc with maculo-schisis treated with PPV.

1. Introduction

Duplication of the optic disc is an extremely rare clinical finding described in the literature as one of two entities: true or pseudo-doubling.[1]The diagnosis of Pseudo-duplication of the optic disc is considered when a disc-like lesion with associated vasculature and occasionally surrounding chorioretinal atrophy and profound cupping, arises adjacent to the normal optic disc.[2] Most cases occur unilaterally and may be associated with decreased acuity in the affected eye. Several developmental defects can simulate the appearance of optic disc pseudo-doubling, including optic disc coloboma, peripapillary chorioretinal coloboma, and scarring.[3,4]

In this study, we present the atypical case of a patient with a unilateral doubling of the optic disc and maculo-schsis.

2. Case report

A 9-year-old girl presented with complaints of decreased vision in the left eye for one month. She was generally healthy, born at term appropriate for gestational age, there was no history of maternal drug or alcohol abuse during the pregnancy, no cases of consanguinity in her family and no family history of ocular disease. Notably, she attended a special education program despite lacking an official diagnosis of a developmental disorder. She denied any prior visual complaints and had never undergone prior examination by an ophthalmologist.

Her visual acuity in the right eye was 20/30 and left eye 20/50, with no significant refractive error. Dilated fundus examination of the left eye revealed an optic disc-like lesion, located inferiorly to an apparently normal optic disk, with a large crater-like depression, pale color and aberrant retinal vasculature arising from its lower region (Fig. 1, A). At the papillo-macular bundle area, retinal striae were evident, contributing to the appearance of cellophane maculopathy (Fig. 1, B). Ultrasonographic examination of the left eye showed a broad and perhaps slightly segmented optic nerve shadow (Fig. 1, C). Fundus examination of the right eye revealed a large, irregular, elliptically-shaped optic disc (Fig. 1, D).

Optical coherence tomography exhibited maculo-schisis emerging from the optic disc complex area and extending throughout the papillo-macular bundle and including the fovea region (Fig. 2, A). The exact location from which the schisis seemed to emerge from was within the junction between the superior optic disc and the adjacent “second optic disc” (Fig. 2, B). Retrospectively, we discovered that the patient underwent an MRI of the brain and orbits, two months prior to her admission to our ER, due to early puberty. There were no abnormal

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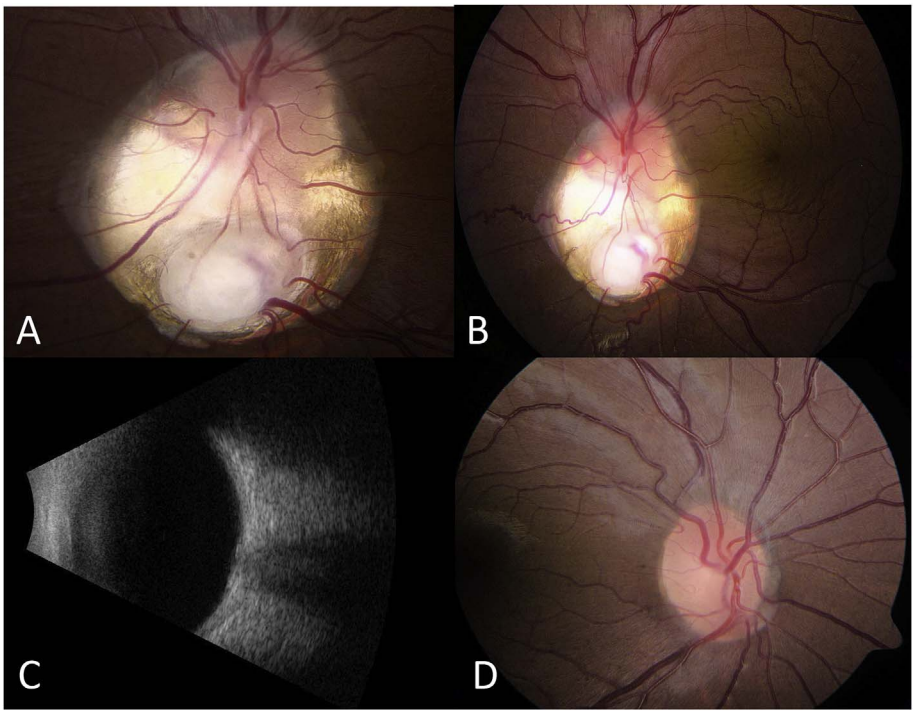

Fig. 1. Color fundus photographs and Ultrasonography at presentation. (For interpretation of the references to color in this figure legend, the reader is referred to the Web version of this article.)
A and B. Color fundus photographs of the left eye, exhibiting doubling of the optic disc and cellophane maculopathy at the central papillo-macular bundle area. C. Ultrasonographic picture showing a broad and perhaps slightly segmented optic nerve shadow. D. Color fundus photographs of the right eye, exhibiting large and irregular optic disc.

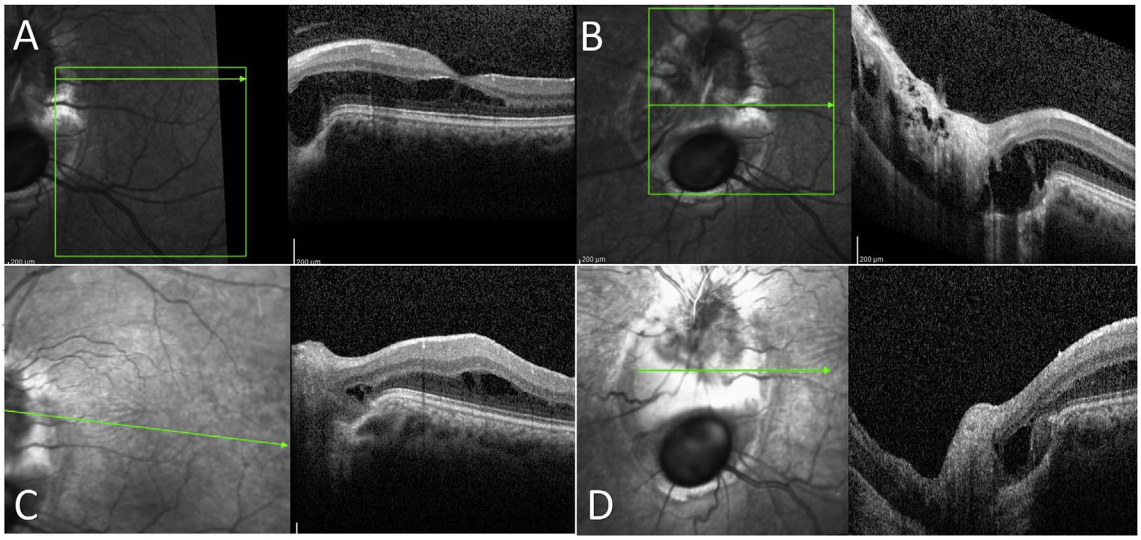

Fig. 2. Optical coherence tomography at presentation and at the 8-months post-op follow up.
A and B. Optical coherence tomography (OCT) of the left eye exhibiting Maculo-Schisis emerging from the optic disc complex and extending throughout the Papillo-Macular bundle and including the Fovea. C and D. At 8-months post-op follow up, the OCT shows a reduction in intra-retinal fluids and an improvement in the maculo-schisis magnitude.

findings in the MRI and no signs of doubling of the optic nerve or any other malformation concerning the optic nerve (Fig. 3).

After consultation with the patient's parents and obtaining an informed consent for a surgical intervention, she underwent 25 Gauge Pars Plana vitrectomy (PPV) with posterior hyaloid peel, ILM peel and gas tamponade with 20% sulfur hexafluoride (SF6). During surgery, the posterior hyaloid was toughly adherent to the retina, however successful induction of hyaloid peeling was performed over the entire macula and also over the pre-papillary area, where a dense membrane was seen and removed. Broad ILM peeling was initiated over the same areas, including the margins of the optic-disc complex. No retinal breaks were documented intraoperatively.

At 8-months post-op follow up, the optical coherence tomography exhibited a significant reduction in intra-retinal fluids and an improvement in the maculo-schisis magnitude (Fig. 2C and D). No change in visual acuity was documented.

3. Discussion

Congenital anomalies of the optic disc are fortunately rare, and in many cases either unilateral or asymmetrical involvement means that functional vision is not significantly impaired.[5] True duplication of the optic disc occurs with separation of the optic nerve into two or more strands, an entity that has yet to be documented in vivo in humans, though common in some lower vertebrates.[1] One case published in 1969 by P.A. Lamba, demonstrated an X-ray image with two optic foraminae in the same orbit, implicating of duplication of the optic nerve.[6] There are several case reports in the literature describing doubling of the optic disc. Padhi et al.[7] reported a case of optic disc duplication with two independent retinal vasculatures in a 41-year-old

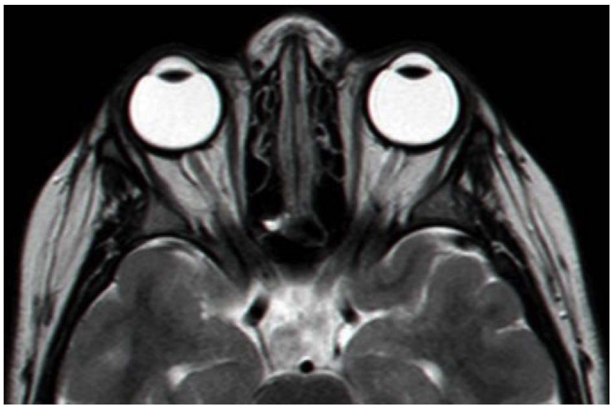

Fig. 3. MRI imaging.
T2-weighted axial MRI image exhibiting normal optic nerve appearance in both eyes.

man. Islam et al.[8] published a retrospective case series of 11 patients with peripapillary colobomatous pseudo-duplication of the optic disc. To note, the visual acuities of patients included in this series were excellent, ranging from 20/17 to 20/30, as opposed to our case. Previous reports documented various concurrent clinical findings accompanying duplication of the optic disc, such as bilateral serpiginous choroiditis, macular congenital hypertrophy of the RPE and bilateral optic disc pits.[2,8,9] Pseudo-duplication of the optic disc is readily distinguished from optic pit as the former comprises an adjacent disc-like lesion and the latter appears as a round depression within the optic disc. Nonetheless, our case resembled in some ways the presentation of optic pit maculopathy, however, the fact that the presumed source of the fluid responsible for the retinal schisis was not located precisely at the "second" optic disc, probably represents a major difference between the two entities in terms of mechanism. To the best of our knowledge this is the first reported case of a pseudo duplication of the optic disc with maculo-schisis, and the first case of a patient with this diagnosis who underwent brain and orbital MRI. The decision to perform surgery as the primary approach for this challenging case was not taken without thorough consideration. The possibility of spontaneous resolution of the maculo-schisis, the known complications of PPV, particularly cataract formation, and lastly the potential complications of general anesthesia were all taken into account. Nevertheless, in our opinion, the evident schisis involving the fovea combined with the clear communication of fluid between the fovea and the optic disc complex, in the context of the patient's young age, could have caused an irreversible retinal damage if left untreated.

The fact that significant improvement in the patient's intra-retinal fluids on the OCT scan was seen at the 8-months post-op follow up, might suggest that surgical treatment with the above-mentioned approach in those complicated cases could be of benefit.

4. Conclusion

Duplication of the optic disc is an extremely rare diagnosis. As demonstrated in our case, additional ocular features such as maculoschisis may appear in conjunction with optic disc doubling, resulting in impairment of vision. Surgical treatment with PPV and ILM peeling seems to be beneficial, but a longer follow up is still needed.

5. Patient consent

An informed consent in Hebrew was provided by the mother of the patient (her legal guardian). The consent was obtained in writing, after a comprehensive explanation of the nature and goals of publishing the details of her daughter medical situation. The informed consent was obtained by Nimrod Dar, the corresponding author.

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Conflicts of interest

The following authors have no financial disclosures: N.D; A.R. Authorship - All authors attest that they meet the current ICMJE criteria for Authorship.

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