Scientific Research Publishing

Open Journal of Ophthalmology, 2024, 14, 229-235
https://www.scirp.org/journal/ojoph
ISSN Online: 2165-7416
ISSN Print: 2165-7408

Late Diagnosis of Congenital Optic Disc Abnormalities

Aïssatou Aw*, Soda Mbaye, Jean Pierre Diagne, Sy El Hadji Malick, Alassane Ba, Aly Mbara Ka, Serigne Sohibou Gaye, Ousmane Ndiaga Senghor, Lamine Ndiaye, Mouhamed Ndiaye, El Hadj Abdellahi Seyid, Papa Amadou Ndiaye

Ophthalmology Department of Abass NDAO Hospital, Cheikh Anta Diop University, Dakar, Senegal
Email: *aichatou18@gmail.com

How to cite this paper: Aw, A., Mbaye, S., Diagne, J.P., El Hadji Malick, S., Ba, A., Ka, A.M., Gaye, S.S., Senghor, O.N., Ndiaye, L., Ndiaye, M., Seyid, E.H.A. and Ndiaye, P.A. (2024) Late Diagnosis of Congenital Optic Disc Abnormalities. *Open Journal of Ophthalmology*, **14**, 229-235.
https://doi.org/10.4236/ojoph.2024.143023

Received: May 2, 2024
Accepted: August 5, 2024
Published: August 8, 2024

Abstract

Purpose: To show epidemiological and imaging aspects of congenital optic disc abnormalities diagnosed late. Method: It was a retrospective study, including all patients with congenital optic disc abnormalities diagnosed at a late age between January 2020 and October 2022 at the eye center of Abass Ndao Hospital. Complete ophthalmological examination was performed with eye imaging according to the cases. Results: 09 patients (10 eyes) were diagnosed with congenital optic disc abnormalities. The mean age was 29 years, with a sex ratio of 0.8. Three patients had consulted for unilateral decreased visual acuity since childhood, two for sudden vision loss and in four cases the diagnosis was fortuitous. Visual acuity was ranged from 1/200 to 20/20. Fundus examination showed myelinated retinal nerve fibers in four eyes, optic disc pit in three eyes including two complicated by maculopathy, two cases of morning glory syndrome and a case of pseudoduplication of the optic disc. Optical coherence tomography, ocular ultrasound B and OCT-Angiography were performed according to the cases. Conclusion: Congenital optic disc abnormalities are often diagnosed late. They are potentially amblyogenic and complications are not rare, worsening the visual prognosis. Their screening should be systematic by ophthalmological examination in newborns.

Keywords

Myelinated Nerves Fibers, Optic Disc Pit, Morning Glory Syndrome, Pseudoduplication Optic Disc

1. Introduction

Congenital optic disc abnormalities include all structural malformations of the optic disc and surrounding tissues, which can cause congenital visual impair-

ment or even blindness [1]. Their clinical aspects are polymorphic and the age at diagnosis might depend on their unilateral or bilateral character. When they are bilateral, they result in low vision behavior leading to early diagnosis while unilateral, they will be discovered fortuitous or in face of a complication. The difficulty may lie in eliminating acquired pathologies that may simulate a congenital abnormality [2]. Their diagnosis is clinical, but the assessment must be accurate, on the one hand because of the association with extraocular anomalies and on the other hand, because there are complications that can threaten the visual prognosis and justify long-term monitoring [3].

In our context, abnormalities in the development of the optic disc are relatively rare and their diagnosis is often late, whereas in the literature cases are reported in school-age children, hence the aim of our study.

2. Method

This was a retrospective and descriptive study conducted from January 2020 to October 2022 at the Eye Center of Abass NDAO Hospital. Were included all patients with transparent ocular media, above age of six years diagnosed with unilateral or bilateral congenital optic disc abnormality (anomalies of size, of myelination, of excavations, or pigmentation). All patients included had a complete ophthalmological examination with retinophotography. Optical coherence tomography, ocular ultrasound, and neuroimaging were performed according to the clinical presentation of the abnormality and the associated signs. All patients underwent a complete somatic examination to look for associated abnormalities.

3. Results

We included 09 patients (10 eyes). The mean age at diagnosis was 29 years with extremes of eight and fifty years. Five patients were women and four patients were men. Three patients consulted for unilateral decreased visual acuity since childhood, two for sudden vision loss and in four cases the discovery of the abnormality was fortuitous. Visual acuity was ranged from 1/200 to 20/20. Ophthalmological examination showed one case of rotational nystagmus and one case of exotropia, with a normal anterior segment in all patients. Fundus examination showed myelinated retinal nerve fibers in four eyes including one case of bilaterality

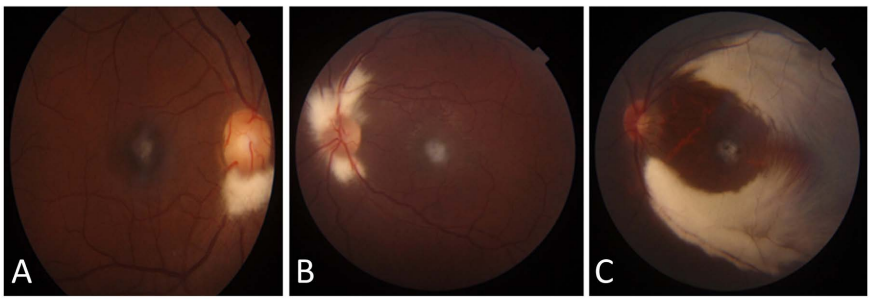

Figure 1. Myelinated retinal nerve fibers with different degree of myelination in three patients.

(Figure 1), optic disc pit in three eyes (Figure 2) including two complicated by maculopathyvisible on optical coherence tomography (OCT) with schisis and serous macular detachment (Figure 3), two cases of morning glory syndrome with an excavation depth visible on OCT (Figure 4) and ocular ultrasound (Figures 5), and a case of pseudoduplication of the optic disc (Figures 6) associated to an occipital arachnoid cyst found on brain scan.

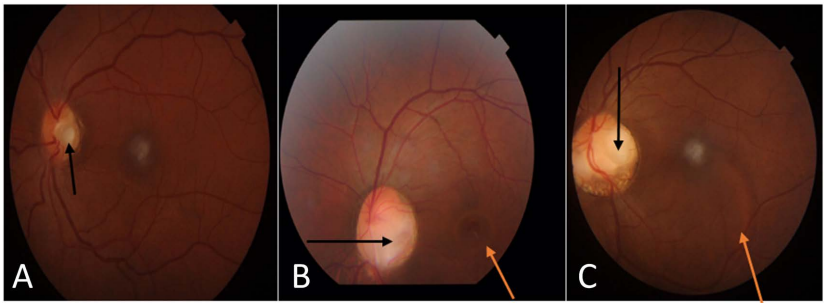

Figure 2. Grayish-white depression corresponding to the optic disc pit (black arrow) with macula detachment in two patients (orange arrow).

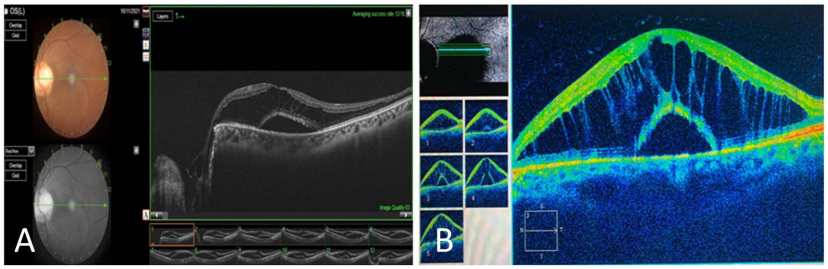

Figure 3. Optic disc pit maculopathy with schisis and serous detachment.

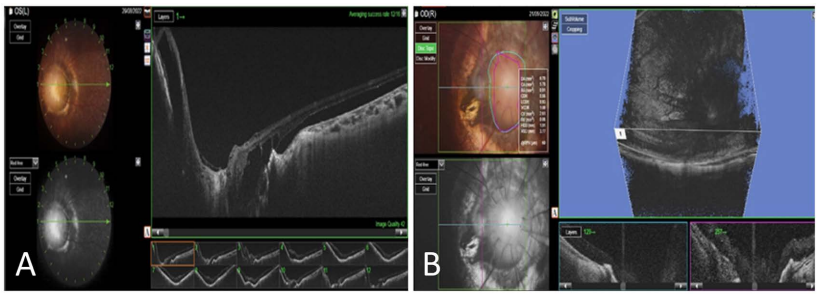

Figure 4. Depth of papillary excavation on radial OCT in two patients.

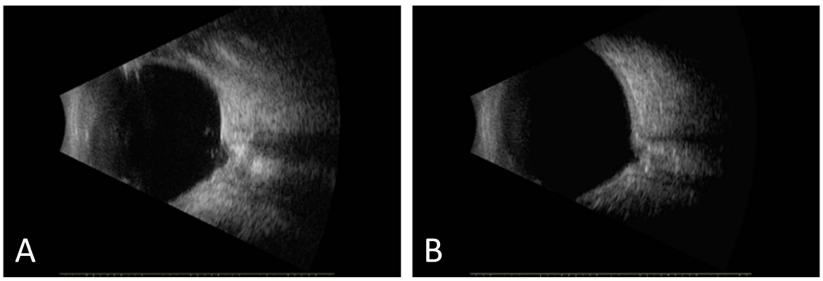

Figure 5. Ocular ultrasound B showing the size and depth of the excavation in two patients.

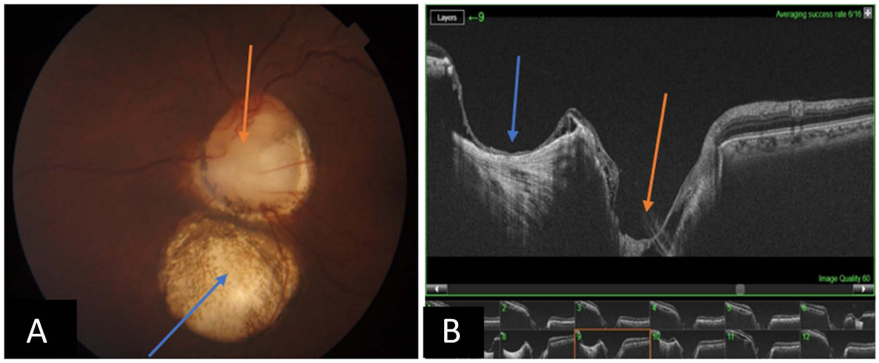

Figure 6. White disc corresponding to a chorioretinal coloboma (blue arrow) below the true optic disc (orange arrow).

4. Discussion

Congenital optic disc abnormalities are a spectrum of developmental pathologies that are not uncommon in daily practice. The age of diagnosis can depend on their unilateral or bilateral character. When it's bilateral, a behavior of visual impairment can guide for early diagnosis. While unilateral, the abnormality will be discovered during systematic screening like in four cases in our study, or later in the face of sensory strabismus or the occurrence of a complication responsible for a decrease visual acuity [1], as was the case with two of our patients. In our study the diagnosis was late in all patients with a mean age of 29 years, with a case of nystagmus and exotropia indicating severe visual impairment.

Myelinated retinal nerve fibers were the most common, observed in four eyes. The patients were aged 21, 22 and 29 years old. It corresponds to myelination of the peripapillary optical fibers secondary to a defect of the cribriform plate, allowing the oligodendrocytes to access the retina [4]. Their incidence is estimated at 0.3% to 1% [5]. It is a unilateral condition in 80% of cases, affecting both men and women, and is thought to be of autosomal dominant transmission [1]. In our study all the patients were male and it was bilateral in one case. The degree of myelination is variable (**Figure 1**), correlated with visual acuity. This is perfectly illustrated by our cases. In the patient with sectoral inferopapillary myelination (**Figure 1(A)**), the VA was 20/20, whereas it was 20/32 in the second bilateral case (**Figure 1(B)**) and 1/100 in the patient with high myelination (**Figure 1(C)**). The main complication of myelinated retinal nerve fibers is vascular occlusion by compression. In our patients, no complications were observed over a two years follow-up.

Pathological congenital excavations were also observed, including three cases of optic disc pit. The origin of optic disc pit is unclear but they seem to come from an abnormal closure of the embryonic fissure. It incidence is estimated at one in 11,000 people. It is usually sporadic and unilateral in 85% of cases [6], like the patients in our study. On the fundus, it appears as a greyish-white, round or oval depression in the optic nerve head with a classic temporal location, but other locations are also described [7]. The optic disc pit was temporal in our

three cases. It remains asymptomatic until the macula detachment occurs. In our three cases, the diagnosis was incidental in the patient with optic disc pit without maculopathy (**Figure 2A**) and the VA was 20/20, unlike the two other patients who consulted for decrease visual acuity secondary to the maculopathy (**Figure 2(B)**, **Figure 2(C)**). This vision loss was sudden with 1/50 VA in the second case (**Figure 2(B)**) and rapdly progressive with 20/40 VA in the third case (**Figure 2(C)**), suggesting that the speed of constitution of maculopathy could impact the severity of decreased visual acuity. The origin of the maculopathy is debated, it generally begins with a schisis, with little or no loss of vision, and subsequently occurs the serous subretinal detachment in 40% to 60% around age of 30 - 40 years [8]. Ours patients with optic disc pit maculopathy were 31 and 37 years old and presented with schisis and serous macular detachment on OCT (**Figure 3**). Vitrectomy with posterior vitreous detachment potentially associated with various other surgical procedures seems to be the technique recommended as first intention in optic disk maculopathy. Self-monitoring and frequent screening are recommended for the patient without optic disc maculopathy for early diagnosis and appropriate management.

The other reported excavation abnormality was the morning glory syndrome observed in an 8-year-old girl and a 27-year-old young woman.

It is a sporadic condition, with no identified genetic risk factor. It preferentially affects women and is most often unilateral as in our study where the two patients were female. The mechanism would be a funnel-shaped widening of the optic gutter due to a lack of differentiation of the sclera, resulting in a deepening of the optic disc [1]. Visual acuity usually ranged from "Counting fingers" to 1 [9] and myopic astigmatism is often found as in our two cases. The visual acuity was 1/200 in the patient with a large excavation with glial material (**Figure 4(A)**), unlike the patient with a less significant excavation and preservation of the neuroretinal ring in whom the visual acuity was 20/25 (**Figure 4(B)**). In ocular ultrasound, the size and depth of the cavitary excavation would be significantly associated with an increased risk of poor vision [10] as in the patient presenting with a larger and deeper excavation with 1/200 of visual acuity. The most common complication is retinal detachment, it can be rhegmatogenous secondary to the traction of fibroglial tissue on the retina or non-rhegmatogenous with poorly understood mechanism (cerebrospinal fluid leakage?) [10]. One patient had serous retinal detachment. The morning glory syndrome is sometimes integrated into a polymalformative association, the clinical examination and a brain scan performed in the two patients did not reveal any associated abnormality.

The last papillary abnormality observed was pseudoduplication of the optic disc diagnosed in a 50-year-old woman. It is defined as the existence of two optic disc, one of them corresponding to a juxtapapillary chorioretinal coloboma with an anastomotic vascular system [11]. It is a rare condition, with no gender predilection that can be unilateral or bilateral. Visual acuity depends essentially on the integrity of the interpapillomacular beam, which was preserved in our patient (**Figure 6**) with 20/32 visual acuity that could justify the delay in diagnosis.

This pseudoduplication is different from the bifurcated optic nerve where there is a duplication of the distal part of the optic nerve in two beams [1]. In that case, the visual field shows two blind spots, ultrasound and Magnetic Resonance Imaging also show a duplication of the optic nerve. In our patient the pseudo-duplication was associated with a homolateral arachnoid cyst. The fact that these are two congenital pathologies and that the lesions are homolateral, made us suspect a possible association of these abnormalities.

At the end of our study, developmental abnormalities of the optic nerve disc are discovered late in our context. This has been reported in other African series [12] [13], justified essentially by accessibility to care. The particularity in late diagnosis is the higher risk of complications and their late discovery which will impact the visual prognosis. Early diagnosis leads to regular monitoring to detect complications and manage them early and also early rehabilitation of amblyopia.

5. Conclusion

The diagnosis of congenital optic disc abnormalities is essentially clinical with heterogeneous presentations. Eye imaging is essential to detect complications. These malformations can be isolated or associated with other ocular and/or extra ocular abnormalities, willingly neurological, thus making neuroimaging a crucial examination. In face of these risks, ophthalmological examination should be systematic in newborns for early diagnosis and appropriate monitoring.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

References

[1] Denis, D., Hugo, J., Beylerian, M., Ramtohul, P., Aziz, A., Matonti, F., *et al.* (2019) Congenital Abnormalities of the Optic Disc. *Journal Français d'Ophtalmologie*, **42**, 778-789. https://doi.org/10.1016/j.jfo.2018.09.011

[2] Brémond-Gignac, D. and Milazzo, S. (2011) Optic Disc Abnormalities in Children. *Les Cahiers d'Ophtalmologie*, **146**, 33-34.

[3] Brodsky, M.C. (1994) Congenital Optic Disk Anomalies. *Survey of Ophthalmology*, **39**, 89-112. https://doi.org/10.1016/0039-6257(94)90155-4

[4] Williams, T.D. (1986) Medullated Retinal Nerve Fibers: Speculations on Their Cause and Presentation of Cases. *American Journal of Optometry and Physiological Optics*, **63**, 142-151. https://doi.org/10.1097/00006324-198602000-00010

[5] Traboulsi, E.I., Lim, J.I., Pyeritz, R., Goldberg, H.K. and Haller, JA. (1993) A New Syndrome of Myelinated Nerve Fibers, Vitreoretinopathy, and Skeletal Malformations. *Archives of Ophthalmology*, **111**, 1543-1545. https://doi.org/10.1001/archopht.1993.01090110109034

[6] Cekić, S., Stanković-Babić, G., Višnjić, Z., Jovanović, I. and Risimić, D. (2010) Optic Disc Abnormalities—Diagnosis, Evolution and Influence on Visual Acuity. *Bosnian Journal of Basic Medical Sciences*, **10**, 125-132.

https://doi.org/10.17305/bjbms.2010.2711

[7] Brown, G.C., Shields, J.A. and Goldberg, R.E. (1980) Congenital Pits of the Optic Nerve Head: II. Clinical Studies in Humans. *Ophthalmology*, **87**, 51-65. https://doi.org/10.1016/S0161-6420(80)35278-0

[8] Irvine, A.R., Crawford, J.B. and Sullivan, J.H. (1986) The Pathogenesis of Retinal Detachment with Morning Glory Disc and Optic Pit. *Retina*, **6**, 146-150. https://doi.org/10.1097/00006982-198600630-00002

[9] Loudot, C., Fogliarini, C., Baeteman, C., Mancini, J., Girard, N. and Denis, D. (2007) Rehabilitation on Functional Amblyopia in Morning Glory Syndrome. *Journal Français d'Ophtalmologie*, **30**, 998-1001. https://doi.org/10.1016/S0181-5512(07)79276-8

[10] Zou, Y., She, K., Hu, Y., Ren, J., Fei, P., Xu, Y., *et al.* (2022) Clinical and Echographic Features of Morning Glory Disc Anomaly in Children: A Retrospective Study of 249 Chinese Patients. *Frontiers in Medicine*, **8**, Article 800623. https://doi.org/10.3389/fmed.2021.800623

[11] Padhi, T.R., Samal, B., Kesarwani, S., Basu, S. and Das, T. (2012) Optic Disc Doubling. *Journal of Neuro-Ophthalmology*, **32**, 238-239. https://doi.org/10.1097/WNO.0b013e3182464d9f

[12] Koman, C.E., Soumahoro, M., Sowagnon, Y.T.C., Kra, A.N.S., Agbohoun, R.P., Kouakou, A.A., *et al.* (2021) Late Discovery Congenital Anomalies of the Papilla. A Report about 5 Cases. *SOAO Review*, No. 2, 15-21.

[13] Diatewa, B.M., Maneh, N., Domingo, A.S., Gnansa, K.E., Ayikoue, Y.F.A., *et al.* (2021) Congenital Ocular Anomalies at the University Hospital Campus in Lome, Togo. *The Pan African Medical Journal*, **38**, Article No. 79.