Reviewer’s report

Title: Morning Glory Optic Disc Anomaly in Association With Down Syndrome

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Reviewer: Margarita Georgieva Todorova

Reviewer’s report:

Although this is an interesting case describing optic disc findings (morning glory optic disc anomaly) in a patient with Down syndrome, the report should be structured more precisely.

Optic disc- and optic disc vessels anomalies have already been described in association with Down syndrome (see remarks included below).

Major Compulsory Revisions

Title:
1. Unilaterallity of the disc anomaly should be included in the title: even if the anomaly is usually unilateral, billaterality would point toward genetically determined cases.
2. Also, as there is unilaterality in the presented clinical picture, there is more probably no association with the Down condition. Therefore the phrase “association with” should be omitted.

Introduction: The main point of the paper is the abnormality found in a child with Down syndrome; therefore the format of the introduction should be structured as:
1) Description of Down syndrome (including the incidence)
2) Clinical phenotype
3) Ophthalmic features (including the frequency of their presentation)
4) Optic disc findings in Down patients including also the supranumerary retinal vessels emerging from the optic disc
5) Description of the morning glory syndrome
6) The following papers should be cited:
7) Figure 1 should be removed

Case report
Paragraphs 1-2

1) The refraction part needs to be précised: a refractive error of -3.0D sph does not correspond to the axial length given, if the keratometric values are correct, or the other way around. A near vision, if only amertopic myopia is present, would improve if the refractive error is only -3.0D sph. In this regard, it would be helpful to include the information regarding the best corrected near vision.

2) Information about the corneal topography should also be added in the text, here in order to exclude the possibility of keratoconus, that would explain partly a 20/50 distance vision, even if no myopic astigmatisms is measured.

3) Figure 3: please provide fundus photos of both eyes: here to exclude the bilaterality of morning glory anomaly, but also to show the optic disc vascularity a typical sign for Down patients.

4) The possibility of afferent neuropathy due to morning glory anomaly seems to be described as RAPD on the involved side. It would be more precise, if the information about the efferent pupillary reaction and the convergence would be given as well.

5) Ocular alignment description is missing.

Paragraph 3

1) It is not clear, what is the reason to give the data regarding the b-wave ERG (which one ERG). B-wave is stated to be reduced, but it is not clear comparing with the left eye of the same patient or comparing to controls? The refractive myopic error in this case should also be taken into account. This information is not relevant to the case presented and should therefore be omitted.

2) If the authors are presenting the diagnostics findings to rule out neuropathy due to morning glory anomaly, a pattern VEP (or Flash VEP) data would be more informative. The later would exclude a possible anisomyopic amblyopia as well.

Paragraph 4

1) Per definition, morning glory syndrome is a congenital optic disc dysplasia consisting of a conical excavation of the posterior pole including the optic disc, elevated annulus of chorioretinal pigment disturbance and subretinal fibrogial tissue. It includes multiple narrow branches of retinal vessels at the edge of the morning glory disc syndrome. Having had the fundus photograph and OCT imaging of the optic disc provided one can see an enlarged optic disc with enlarged excavation, however, with lacking central tuft of white tissue and the peripapillary subretinal fibrogial tissue is barely seen, a finding which resembles rather an optic nerve coloboma instead.

2) On the picture provided it is difficult to recognize whether the vessels emerge from the ciliary circulation, which would differentiate a morning glory anomaly from a disc coloboma. It is difficult to recognize where the defect is symmetric or focused infero-nasally.

3) Furthermore supranumerary narrow branches optic disc vessels emerging from the optic disc are already described in Down syndrome.
4) Therefore, the strength of this article is the precise presentation and careful interpretation of the B-Scan, OCT and the fluorescein angiography findings. The information of the B-scan ultrasonography, OCT as well as of the fluorescence angiography findings should be presented bilateraly, here also with emphasis to exclude a variant of: optic disc coloboma, peripapillary staphyloma or papillo-renal syndrome due to mutation in PAX2.

5) The following papers are relevant to be sited:


Instead of presenting a coronal OCT scan, please provide a sagittal OCT scan of the optic nerve head which better shows the quantitative assessment of the cup/disk ratio and the neuroretinal rim. Here, it is important to exclude a small tear at the edge of the malformation that may lead to serous retinal detachment. There are also more artifacts in a high myopic eye or in enlarged optic disc, as it seems also to be the cause.

MRI-information of the cranium, sella and orbits to exclude a possible asymmetry of the optic nerve sheaths, abnormalities of the carotid circulation and central nervous system should be included. The spelling of MRI, 3-tesla is not clear.

Again in the Discussion:

1) Discussion part should be shortened to the discussion of the relevant findings and not repeat the data already given in the results section.

2) It is reasonable to start this section briefly discussing all previously reported optic disc findings of Down syndrome and then pointing towards the novel finding…

3) ..and then to go true the presentation of the MGS findings..

In developmental phenotypes, it is often difficult to know what to do with
monocularly-affected patients. Clearly there can be genetic predispositions that can be affected by stochastic processes that result in unilaterality. As the incidence of Down patients worldwide is around 8.3 and 13.66 at 10 000 live births, the possibility to find other pathology associated in a person with Down phenotype is relatively high. Therefore and due to the unilaterality, the MGS, if it is still a cause, seems rather a coincidental finding in the presented child with Down syndrome.

4) MGS is associated with hormonal deficiency and anterior segment dysgenesia in bilateral cases… It is therefore reasonable only briefly to discuss these paragraphs.

5) The Discussion of the DS ocular findings given at the end of the discussion should be placed rather in the introduction section of the manuscript.

Minor Essential Revisions

Abstract:
• The first sentence should be: A 15-year-old-girl with the “clinical phenotype of…” or “with features…”

Key words: please include comas only after the abbreviation.

Case report

Paragraph 1
1) It is not important who is referring the child for examination.

2) Figure 2 is not relevant.

Paragraph 3
3) Information given in regard to audiometry findings is not relevant.

Paragraph 4
4) Visual field examination is not relevant to be shown but should be described in the text, if the OCT scans are provided.

Paragraph 5
5) If the maturity status at birth is to be given, please include the complete data including also the length at birth and gestation age.

Discussion

Last Paragraph
6) “Vision loss” should be replaced with “reduced vision”….if the best corrected vision remained 20/50.

7) The authors conclude: “This case report also emphasizes the importance of funduscopic examination for the patients with the diagnosis of Down syndrome.” It seems to point rather the “the importance of ophthalmic screening-examinations in Down children to rule out any vision relevant pathology”.

Figure legends, Abbreviations, Acknowledgements: Not provided.
References:
The References should be carefully reviewed by the authors for content and style. Briefly checked, there are few mistakes found:
2) I could not find the reference 3 in Pubmed
3) The article sited by Dutton GN. Is published in Eye (Lond).

The English grammar and spelling are unfortunately not of sufficient quality. I have not tried to correct these systematically, but would ask that the authors employ an editor to polish the language, if this paper is accepted for a major review.

Level of interest: An article of importance in its field

Quality of written English: Not suitable for publication unless extensively edited

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:
I declare no competing interests