Reviewer's report

Title: Congenital cystic eye associated with a low-grade cerebellar lesion that spontaneously regressed

Version: 1
Date: 14 April 2014

Reviewer: Eleni Papageorgiou

Reviewer's report:

Major compulsory revisions

This is an interesting case report, describing a congenital cystic eye in association with a cerebellar - presumably low grade glioma, which spontaneously regressed. The authors discuss the possible mechanisms for regression of pediatric low-grade gliomas and suggest a wait-and-see policy (instead of biopsy) in selected cases. This presentation is indeed rare, however there are some limitations that should be addressed:

1. Although the authors state that cerebellar lesion of possible glial origin has not been previously reported in association with congenital cystic eye, this is not the first report. Mehta et al. (J Pediatr Ophthalmol Strabismus. 2010 Aug 23;47 Online:e1-4. doi: 10.3928/01913913-20100818-13. Congenital cystic eye: a clinicopathologic study.) also reported ectopic glial tissue in the brain in association with a congenital cystic eye. The authors may consider revising their paper in regard with this publication (especially localization?).

2. Additionally, the suggestion to perform early MRI in children with ocular malformations, as the authors state in their conclusion, is not novel, since it is well-known that MRI is necessary in congenital cystic eye due to the presence of ocular and non-ocular intracranial abnormalities.


4. Do the authors have an explanation for the coincidence of these two clinical entities or they believe that the findings are accidental (which appears to be the case)? This should be briefly discussed in the manuscript.

5. Some language editing would definitely improve the manuscript

Background

6. The background is partially a redundancy of the abstract, maybe the authors could add more information regarding the ocular associations, MRI findings,
unilaterality / bilaterality, treatment and prognosis of congenital cystic eye. The actual clinical and laboratory findings and the clinical course should be moved to the case presentation.

Case presentation

7. When did the parents notice the cystic lesion? At birth or later?

8. The authors performed genomic hybridization testing. Could they please specify briefly what conditions they were looking for / excluded? Karyotype? SOX2 / PAX6 mutations?

9. The lesion was partially excised? Could the authors please briefly explain why a total resection was not performed / not possible?

Discussion

10. In general, the manuscript would benefit by discussing the authors’ findings more in regard to previous published cases, instead of performing a literature review.

11. Some discussion on further differential diagnoses (and their clinical signs), such as microphthalmos with cyst (much more common), cystic teratoma, cephalocele, heterotopic brain tissue, anophthalmos (true anophthalmia is extremely rare, although at birth the cystic eye may resemble anophthalmia)

12. The risks of a brain biopsy could be briefly discussed, as it would support the authors’ suggestion of a wait-and-see policy.

13. Some information on the surgical treatment / options of congenital cystic eye would be interesting. Also recurrence (please check Robb et al., Ophthalmic Genetics 2003)? Mean age at diagnosis?

Figures

14. An image of the pre- and post-operative appearance of the congenital cystic eye would be extremely interesting. Is it available?

Minor essential revisions

Abstract

15. 4th line: “unilateral anophthalmos” is a different diagnosis, please consider correcting

16. 6th line: what do the authors mean by “based on absence”…? Absence of neurological signs? Please specify / rewrite?

17. 7th line: please consider revising “unexpectedly”, see above (regression of glial tumours is not as rare and unexpected, as the authors state later in the manuscript).

Background

18. 5th line: “unilateral anophthalmos” is a different diagnosis, please consider correcting

19. 9th line: “please consider replacing “bioptic procedure” with “biopsy”
Case presentation

20. It is stated that “follow-up MRI, one year after surgery disclosed…”, but the figure number is not included at the end of this sentence. Please correct. Fig 3a-c?

21. Please correct brain “RMI” to “MRI”

Discussion

22. What is the estimated number of reported cases of congenital cystic eye that the authors have reviewed? Guthoff et al. 2004 reported 33 cases.

23. Please correct “hystopathologic” to “histopathologic”.

24. Also, some epidemiology regarding glial tumours of childhood / location / age would help the reader.

Figures

25. An image of the pre- and post-operative appearance of the congenital cystic eye would be extremely interesting. Is it available?

References

26. Please consider including in your list / discussion the following papers
Gupta et al. BMC Ophthalmology 2003, 3:7

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Needs some language corrections before being published

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

Declaration of competing interests:

I declare that I have no competing interests