TWO SIBLINGS WITH SMALL EYES

HOOYBERGHS P., FOETS B.*, CASTEELS I.**

SAMENVATTING

Posterieure microftalmie is een zeldzame aangeboren bilaterale oogaandoening waarbij het achterste oogsegment abnormaal kleine dimensies vertoont. Bijkomende kenmerken zijn hoge hypermetropie en het frequenter voorkomen van uveale effusie. We beschrijven twee kinderen, broer en zus, met hoge hypermetropie waar de diagnose van posterieure microftalmie werd gesteld. Bij fundusonderzoek was de typische crowding van de oogzenuw met retinale plooien te visualiseren. We bespreken de klinische kenmerken, pathogenese en complicaties van deze aandoening.

ABSTRACT

Posterior microphthalmos is a rare congenital bilateral eye disorder, of which the posterior segment is abnormally small. Additional features include high hypermetropia and a tendency to uveal effusion. We report two siblings who present with high hypermetropia and other features of posterior microphthalmos. Fundus examination revealed the typical crowding of the optic disc and retinal folds. We discuss clinical characteristics, pathogenesis and complications of this disorder.

RESUME

La microphtalmie postérieure est une maladie rare dans laquelle le segment postérieur de l'oeil est anormalement petit. Elle est également caractérisée par une hypermétropie importante et des complications telle que l'effusion uvéale. Nous décrivons le cas de 2 enfants de la même fratrie (fille, garçon) qui présentaient une hypermétropie importante et chez lesquels le diagnostic de microphalmie postérieure a été posé. L'examen du fond d'oeil a mis en évidence la présence de plis caractéristiques de la rétine. Nous discutons des caractéristiques cliniques, de la pathogénie et des complications de cette affection.

KEY WORDS

Posterior microphthalmos, nanophthalmos, hypermetropia, papillomacular fold

MOTS-CLÉS

Microphtalmie postérieure, nanophtalmie, hypermétropie, pli papillomaculaire

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* *Department of Ophthalmology, University Hospitals, Leuven*

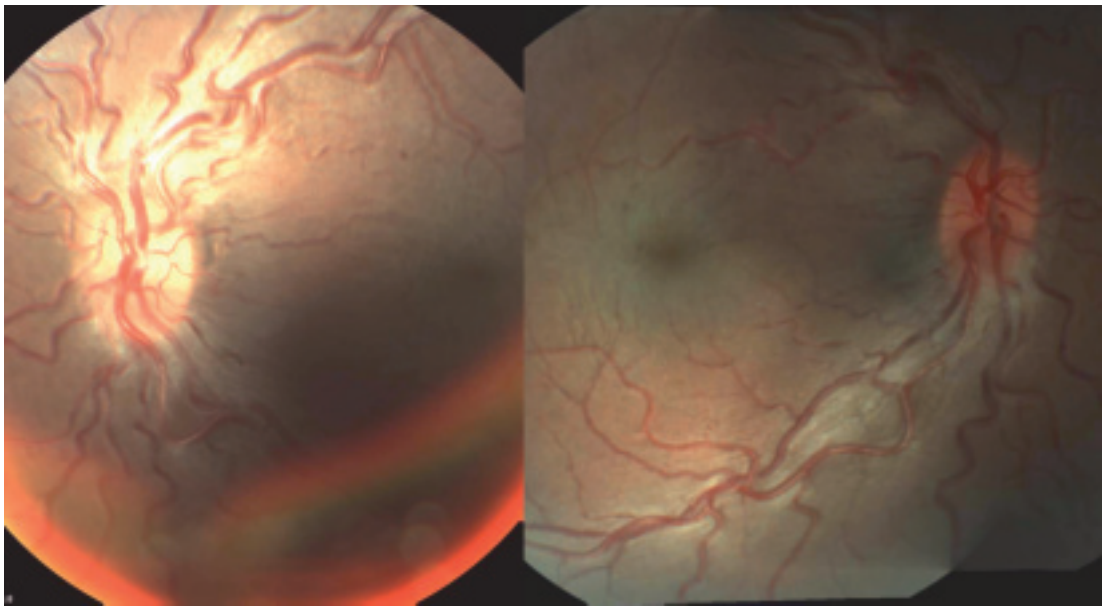

Fig. 1: Fundoscopy of patient 1 with tortuous vessels and crowded discs.

The terminology of microphthalmos in the literature can be confusing.
Microphthalmos is an ocular defect where the overall size of the globe is smaller than normal. More specifically it has been defined as an eye with an axial length more than two standard deviations smaller than the normal for that age group (8). The condition has been classified into simple and complex microphthalmos. Simple microphthalmos is where the globe is small, but otherwise normal. Complex microphthalmos is where the globe has other associated abnormalities (cataract, retinal or vitreous disease, or more complex malformations) (35). Simple microphthalmos can be further subdivided into pure microphthalmos (nanophthalmos) and posterior microphthalmos. Both conditions present with small total axial lengths. In nanophthalmos, this can be attributed to a too small anterior and posterior segment, whereas in posterior microphtalmos, there is a selective reduction of the size of the posterior segment with normal dimensions of the anterior segment (28,30).

CASE REPORTS

A ten year old girl and her six year old brother were seen at our department with visual problems and high hypermetropia. Their parents are of North-African origin and are first cousins.

Case 1

The ten year old girl presented with a best corrected visual acuity of 0.07 on the right and 0.4 on the left eye. Her cycloplegic refraction showed a high hypermetropia of +13.0 D on both eyes. Axial lengths were 15.90 mm and 15.50 mm and horizontal corneal diameters 10.0 mm and 10.5 mm on the right and left eye respectively. Keratometric values showed a steep cornea (50.50 D and 50.25 D on the right and left eye respectively). The anterior chamber depth and central corneal pachymetry were normal. Funduscopy revealed crowded discs, tortuous vessels and macular folds (figure 1). The flash-ERG showed a 30% reduction of the amplitude of the rod response, with normal implicit time of the b-wave.

The lower rod responses were also apparent in the adapto-ERG, recorded at different time spots while dark adapting.

The cone responses were well preserved in amplitude but showed prolonged implicit times (single flash cone response and 30 Hz flicker response).

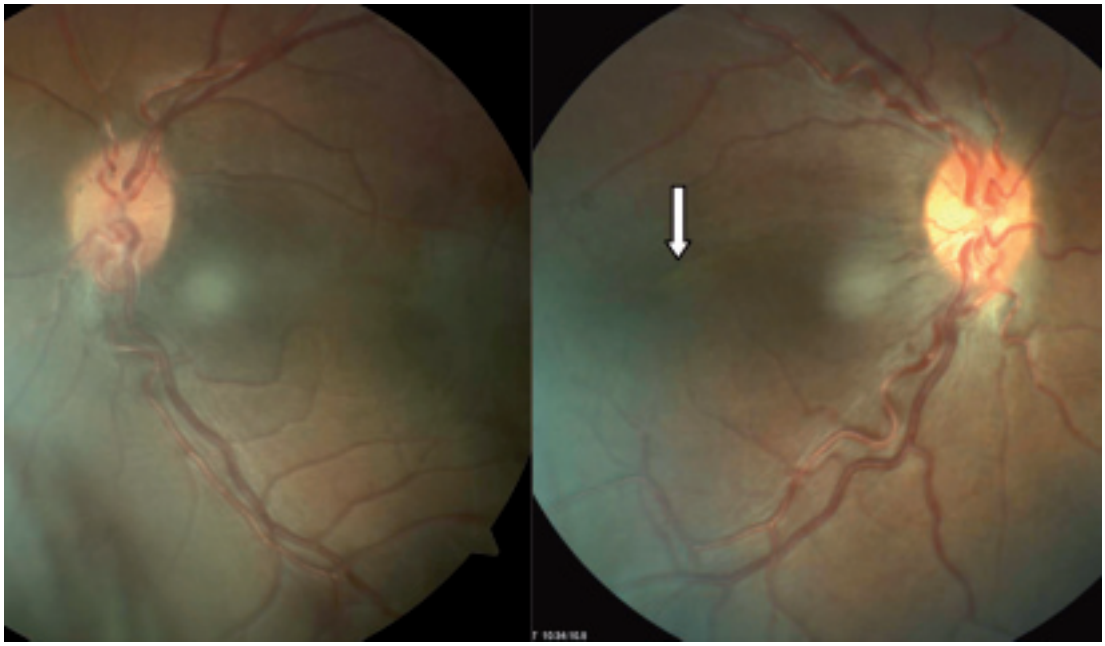

Fig. 2: Fundoscopy of patient 2 with tortuous vessels and crowded discs. Note the horizontal papillomacular fold in the right eye.

The maximal combined response showed normal amplitudes and implicit times.

Case 2

The past medical history in the six year old brother of case 1 revealed premature birth at 28 weeks of gestation with a birth weight of 1400 g. He also presented with poor vision and high hypermetropia. Cycloplegic refraction was +14.0 D on both eyes and best corrected visual acuity was 0.14 and 0.22 on the right and left eye respectively. We found a microcornea (horizontal corneal diameters of 9.0 mm on both sides) with high keratometric readings (52.75 D and 52.00 D on the right and left eye respectively). We measured a total axial length of 15.15 mm right and 15.22 mm left. The anterior chamber depth and central corneal pachymetry were within normal limits. Funduscopy showed crowded discs, tortuous vessels and papillomacular folds (figure 2), of which one could be visualized on optical coherence tomography imaging (figure 3). The flash-ERG showed a similar response to case 1.

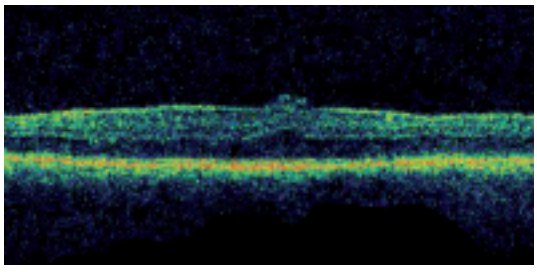

Fig. 3: Optical coherence tomography scan of the right macula of patient 2. Note the fold of the neural retina

DISCUSSION

We describe two highly hypermetropic siblings with eyes that have a small total axial length. Differential diagnosis is within the category of microphthalmos. Taking into account the normal anterior chamber depth and the fact that there are no associated ocular anomalies, we would suggest the term simple microphthalmos in those children, and more specifically posterior microphthalmos although the microcornea in both children could suggest nanophthalmos.

In posterior microphthalmos, the anterior segment has normal dimensions. The axial length is reduced due to a reduction of the size of the

posterior segment. Additional described features are high hypermetropia, an elevated papillomacular retinal fold and less frequently pigmentary retinopathy, chorioretinal folds, uveal effusion and crowded optic discs. (14) Nanophthalmos is generally described as a too small eye with high hypermetropia, axial lengths less than 20 mm, small corneal diameters, a general crowding of the anterior chamber with normal sized lenses, a thickened sclera with abnormal collagen and a tendency to closed angle glaucoma and uveal effusion. (28,25) In 1975, Boynton et al proposed that the combination of anomalies in microphthalmic eyes was the result of a failure of growth of the posterior outer coats of the eye. (2) The suggestion that the neural retina develops independently from the pigment epithelial layer (especially in the macular region), was later confirmed by OCT examination, which showed that the pigment epithelial layer is not involved in the frequently seen papillomacular fold. (1,16,29) These findings are confirmed by the OCT imaging in one of our patients: the neural retina is folded and elevated as a whole without intraretinal edema or cysts, whilst the pigment epithelial layer is unaffected. (29)
Examination of scleras in nanophthalmic patients shows abnormal deposits of glycosaminoglycans and elevated levels of fibronectin, which leads to abnormal growth of the sclera with scleral thickening. (27,32,39,40) The neural retina, as already mentioned, appears to develop independently from the other outer layers and is thrown in folds when the sclera develops inadequately. (7) The abnormal sclera becomes an increased resistance to transscleral protein movement and venous outflow, with possible complications. (23) The heredity in these cases is most likely autosomal recessive, considering the consanguinity of the childrens' parents.

Visual problems in children with microphthalmos can be caused by refractive amblyopia and structural problems in the posterior pole, eg the already mentioned papillomacular fold. (20) In our patients, best corrected visual acuities were 0.07 and 0.4 for the girl and 0.14 and 0.22 for the boy. Visual problems in adults with microphthalmos are mainly due to glaucoma and complications following intraocular surgery. (38)
The relationship between short axial length and hypermetropia is known. If the total axial length equals 15 mm, there will be an excess of 21 D compared with normal values (presuming that 1 mm difference in axial length changes refraction by 3 D). This "excess" can be compensated for in different ways. A steeper cornea can add refractive power to an eye, which means it has an emmetropizing influence. Other possible compensating factors are: lenspower, corneal thickness and foveal thickness. (10,29) These can be seen as compensatory mechanisms to preserve emmetropia, but it is possible that increased corneal power is just a reflection of a smaller corneal diameter with a decreased radius of curvature and that increased lens power is also caused by an increased lens sphericity resulting from underdevelopment or decreased diameter of the suspensory zonular annulus. (35) Several complications can occur in microphthalmic eyes: (11,19)

1) Uveal effusion

Case reports of both nanophthalmos and posterior microphthalmos mention spontaneous or postoperative uveal effusion as a potentially important complication in microphthalmic eyes. (4,14) The abnormal thickened sclera gives rise to a higher resistance to protein drainage from the suprachoroidal space and venous outflow. This leads to choroidal thickening and congestion, resulting in choroidal folds and uveal effusion. (12) Sudden decompression of the eye during surgery can trigger uveal effusion, which may lead to secondary retinal detachment, intraocular hemorrhage and malignant glaucoma. Uveal effusion can be successfully treated with sclerotomies. (9,14,18)

2) Glaucoma

Microphthalmos/nanophthalmos is often associated with a chronic (angle-closure) glaucoma at later age, triggered by normal lens growth in an eye that is already too small. (38) Both the anatomical abnormalities of the eye and uveal effusion, which lead to an anterior displacement of the iris and ciliary body, may cause the angle closure. (22) Peripheral anterior synechiae can be formed secondarily.
Response to medical treatment is poor and miotics can even aggravate the condition by re-

laxing the lens zonules. Results after iridotomy or iridoplasty are variable, but both treatments can be beneficial in an early stage. Glaucoma filtering surgery may be required in the later stages (eg after development of peripheral anterior synechiae). Good intraocular pressure (IOP) control has been achieved in nanophthalmic eyes after trabeculectomy with the use of mitomycin and sclerotomy, although uveal effusion remained a major problem. (38)

3) Retinal folds and other fundus abnormalities

A characteristic elevated papillomacular retinal fold is often seen in microphthalmic eyes, as demonstrated in one of our patients. The fold is thought to be caused by the independent development of neural retina and pigment epithelial layer, which is most evident in the macular region. OCT-imaging and fluoangiography show the stable condition of this fold, without any underlying leakage or abnormality of the pigment epithelial layer. (1,2,14-17,23,30) Other fundoscopic changes seen in microphthalmic patients are chorioretinal folds, pigmentary retinopathy, absent or reduction of the capillary-free zone, crowded optic discs and sclerochoroidal thickening. (14,21,25,31) The absent or rudimentary foveal avascular zone can be an additional explanation for the often poor visual acuity seen in nanophthalmos and posterior microphthalmos. (34)

4) Complications of cataract extraction

Cataract extraction in microphthalmic eyes has been reported to have a high incidence of intraoperative and postoperative complications. (3,6) As mentioned earlier, intraocular surgery in microphthalmic eyes can be associated with choroidal effusion, nonrhegmatogenous retinal detachment and malignant glaucoma, probably at least partially due to sudden changes in IOP. It seems though that there are only small IOP fluctuations in phaco-emulsification through a small incision. Therefore there may be less need for prophylactic sclerotomies. (26,37) In eyes with a history of uveal effusion, it is prudent to consider prophylactic procedures to enhance uveoscleral flow such as lamellar scleral resection. In eyes without a history of uveal effusion, phaco-emulsification without prophylactic measurements is safe. (37)
Additional problems in cataract surgery in microphthalmic eyes are lens power calculation and intraocular lens selection. Calculations of the implant power should be done with the Holladay II or Hoffer Q formula both of which seem the more accurate in highly hypermetropic eyes. Moreover, in microphthalmic eyes the IOL power needed is often greater than that commercially available. (5) The implantation of two or more intraocular lenses, so called piggyback IOL implantation or polypseudophakia, can be used to provide adequate IOL power in patients with microphthalmos. (5,13)

5) Strabismus

The frequent esotropia in microphthalmic eyes is usually of the nonaccommodative refractive kind. Medial rectus recession can be a therapeutic option, making sure not to overcorrect to prevent adduction deficit and convergence insuffiency. (24)

CONCLUSION

Posterior microphtalmos is a rare congenital disorder with a reduced axial length due to a selective reduction of the length of the posterior segment. More insight has recently been gained into the pathogenesis and complications thanks to information provided by OCT and ultrasonography. The two cases presented here illustrate some characteristics of patients with posterior microphthalmos. A thorough examination and follow-up is required to prevent potential major complications.

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Reprint requests:
Pieter Hooyberghs
Department of Ophthalmology, University Hospitals
Capucijnenvoer, 33, 3000 Leuven, Belgium
Fax: +32 16 332367
Tel: +32 16 332385
Email: Pieter.Hooyberghs@uzleuven.be