Case series: Megalopapillae in twins – Congenital or hereditary?

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To the best of the author's knowledge, bilateral megalopapilla are entities with an unknown inheritance pattern, and this is the first case presentation of bilateral megalopapilla in twin siblings. One of the twins presented to the outpatient department with a frontal headache, while the other was asymptomatic. Upon examination of the first family members, the asymptomatic paternal grandfather had a similar presentation of megalopapilla. As a result, this report will help in determining the genetic pattern of development of this optic disc anomaly, as well as its crucial differential diagnosis.

Key words: Megalopapilla, optic disc anomaly, twins

Bock and Franceschetti coined the term "megalopapilla" to describe an optic disc with a surface area of more than 2.5 mm^2 and normal morphology in terms of colour, margin, and blood vessel configuration.[1] There are two phenotypes: type 1 (common), which is bilateral with a large, round or horizontally oval cup, and type 2 (rare). It was proposed by Sampolesi *et al*.[2] as a pseudoglaucomatous disc. Glaucoma has a large vertically oval cup. Type 2 is unilateral and characterized by a superiorly displaced cup that obliterates the neuroretinal rim.

Case Reports

Case 1

A 15-year-old male student presented with a frontal headache for 7 days of mild intensity, dull in character, that lasted 4 to 5 hours and was induced while studying on a laptop. Stable vitals and no signs of raised intracranial pressure.

On ocular examination, 6/6(p) Snellen's visual acuity (AR: -0.25D) both eyes, pupil equal in size and reacting. The pupillary reflex immediately shifted to 'against movement' when dynamic retinoscopy was conducted in both eyes. Corneal diameter (mm) was 11.2 (horizontal) and 11.0 (vertical) on anterior segment examination. A single resident took three readings of intraocular pressure (mm Hg by Goldmann Applanation Tonometer) on two consecutive days, 24 hours apart [Table 1].

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Central corneal thickness (CCT in microns) RE: 500; LE: 490. Gonioscopy: ciliary body band throughout 360 degrees. Axial length RE: 21.50; LE: 21 mm on A-scan.

On Indirect fundoscopy, there was a large disc with well-defined margins, 0.8:1 C:D ratio with a sharp macular reflex in both eyes [Fig. 1].

30-2 SITA-standard field plotting

Blind spot was normally positioned about 12 to 15 degrees temporal to visual axis; blind spot was enlarged with depressed points in surrounding area on both greyscale and pattern deviation. Other findings were normal in both eyes, despite the parametric not taking into account the normative database of this age group.

SD-OCT findings were : horizontal C/D (RE: .93; LE: .91), vertical C/D (RE: .79; LE: .80), Disc Area (RE: 4.48; LE: 4.06), Cup Area (RE:3.31; LE:3.0). In both the eyes, there was no retinal nerve fiber layer (RNFL) defect with an average thickness of 80 microns and a macular thickness (RE: 112; LE: 114 micron). The OCT devices, on the other hand, include an integrated normative database for patients above the age of 18. OCT has been found in studies to be unaffected by disc size and to have a superior sensitivity in distinguishing between healthy and glaucomatous eyes with large discs.[3]

On pattern visual evoked response latency of P100 (RE: 94; LE: 95) and amplitude (RE: 0.53; LE: 0.56), fluorescein angiography showed no leakage of blood vessels around the optic disc. The patient's MRI of the brain and orbits came out normal.

Case 2

Twin brother is asymptomatic and has a normal pupillary reaction. Snellen visual acuity is 6/6 in both eyes. The average IOP measurements were RE:14; LE:12 over three days at 9 a.m., 3 p.m., and 9 p.m. CCT (RE:495; LE:504). Indirect fundoscopy reveals a large disc with well-defined edges, a CDR of 0.8:1 and a prominent macular reflex in both eyes [Fig. 2].

There is no leakage on FFA. The SD-OCT revealed the following: Horizontal C/D (RE:0.91; LE:0.91), Vertical C/D (RE:0.85; LE:0.82), Disc Area (RE:4.30; LE:4.30), Cup Area (RE:3.31; LE:3.22) . In both eyes, there was an average RNFL thickness (RE:78;LE:80), macular thickness (RE:116; LE:116) and no RNFL defect seen.

Case 3

A 72-year-old asymptomatic paternal grandfather of twins presented to us with UCVA of 6/9 in both eyes and BCVA of 6/6 in both eyes by -0.50DS / -0.50DC at 75 degrees with add near of +3DS in both eyes (N/6). Intraocular pressure was RE:14; LE:16mm Hg and CCT (RE:485;LE:490). In both the eyes, on slit light, early lenticular cataractous alterations can be seen, gonioscopy revealed scleral spurs, and on indirect ophthalmoscopy, there was a large disc with defined boundaries, a CDR of 0.9:1, and a slightly dull macular reflex with chorio-retinal degeneration throughout the fundus.

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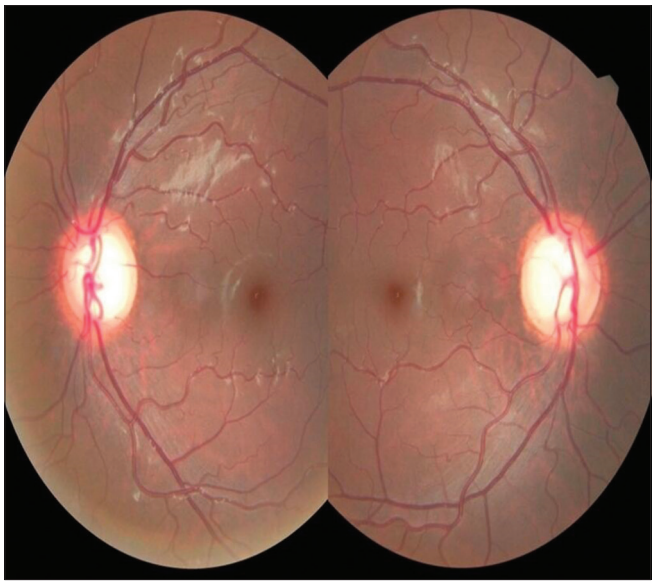

Figure 1: A colored fungus image of Case A showing a large disc with distinct margins , with round 0.9 cups, slightly pale neuroretinal rim with a cilioretinal artery in both the eyes

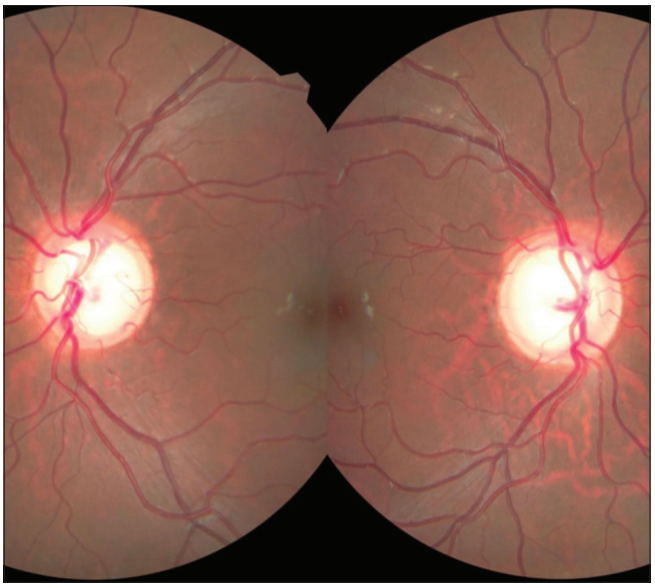

Figure 2: A colored fundus image of Case B showing large optic disc with round cup of 0.9 size, slightly pale neuroretinal rim in both the eyes with 2 cilioretinal arteries in the left eye

Table 1: Showing IOP readings (mm Hg) of case 1

Time/Day	Day 1 (R/L)	Day 2 (R/L)	Day 3 (R/L)
9 am	16/16	16/14	16/14
3 pm	14/16	12/14	14/14
9 pm	14/14	14/16	12/14

The SITA plotting were within acceptable limits. SD-OCT parameters include: C/D horizontal (RE: 0.82; LE: 0.84), C/D vertical (RE: 0.80; LE: 0.80), disc area (RE: 3.70; LE: 3.98), cup area (RE: 2.49; LE: 2.62), and a macular thickness (RE: 232; LE: 230).

The twins' parents and paternal grandmother showed no symptoms. The maternal grandparents are not alive.

Treatment and follow-up

Case 3 received refractive correction, and all three are scheduled for monthly IOP monitoring.

Discussion

The above three cases were excluded from glaucoma because the IOP readings were within normal limits, the optic discs were large (>2.5 mm^2) with round cups; there was no violation of the ISNT rule/nasalization of blood vessels/notching/bayonetting sign/lamellar dots/splinter hemorrhage, and SITA-standard plotting were within normal limits. Patients are being followed up on because studies have shown that large optic discs are associated with an increased risk of glaucoma.[4] Dynamic retinoscopy has ruled out a lack of accommodation in Case A.

Another notable difference in these twins could be Sponsel *et al*.'s[5] Pediatric Eversional Angle Closure with Headache (PEACH) syndrome. Only one of the twins reported a history of accommodation-induced frontal headache, a symptom of PEACH syndrome, but gonioscopy revealed a normal anatomy wide angle.

Due to distinct disc margins in all quadrants, no leakage on FFA, a normal MRI scan, and no evidence of elevated ICT, papilledema was ruled out.

Three genetic loci linked to the optic disc area have a considerable evidence across the genome. TGFBR3 on chromosome 1p22, ATOH7 on chromosome 10q21.3-22.1 (also for VCDR), and SALL1 on chromosome 16q125[6] were the most interesting for the optic disc area, and their association with megalopapilla needs to be investigated.

Conclusion

After differentials were ruled out and parametric values were read, these three cases were diagnosed as Megalopapilla disc anomalies. To our knowledge, this is the first time we've seen megalopapilla in twins. A similar presentation in the paternal grandfather contributes to the understanding that this entity may have a hereditary pattern that can be studied genetically.

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Conflicts of interest

There are no conflicts of interest.

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