# Geleophysic dysplasia associated with bilateral angle closure glaucoma

*Murat Sinan Saricaoglu, Dilek Güven, Ahmet Karakurt, Hikmet Hasiripi*

In this case report, we present occurrence of bilateral angle closure glaucoma in a 9-year-old girl with geleophysic dysplasia. Bilateral YAG laser iridotomy was applied, but intraocular pressure (IOP) remained at high levels, necessitating bilateral trabeculectomy with mitomycin C. On her follow-up examinations for 3 years, IOP remained in the mid-20s with no need for further intervention or antiglaucoma medication. There are few reports describing the ocular findings of geleophysic dysplasia in literature. To our knowledge, this is the first case report describing an application of glaucoma surgery and its results at geleophysic dysplasia.

**Key words:** Geleophysic dysplasia, glaucoma, mitomycin C, trabeculectomy

Geleophysic dysplasia was first reported by Spranger *et al*.[1] as a rare autosomal recessive disorder of generalized lysosomal storage defects. This rare clinical entity is characterized by short stature, brachydactyly, characteristic facial expression, stiffness of hand and foot joints, progressive heart disease and hepatomegaly in some of the cases. Ocular findings were reported as short axial length, thick and steep central corneas, thick crystalline lenses and shallow anterior chambers.[2]

## Case Report

A 9-year-old girl presented with pain in both eyes. Her medical history revealed that she had geleophysic dysplasia and was being followed up in another center for her heart disease. Her parents were first-degree relatives. Her parents and 2 siblings had normal systemic and ocular findings. Her weight was 25 kg (<3rd percentile), and her height was 128 cm (<3rd percentile). At the end of the 3-year follow-up, her weight was 30 kg (<3rd percentile) and her height was 133 cm (<3rd percentile). She had long upper lip with thin vermillion. Her fingers and toes were short. Her skin was thick, and joint mobility was restricted. She had a typical tip-toe posture [Fig. 1]. She was mentally normal. Her cardiologic examination revealed mitral valve anomaly and a low-degree mitral insufficiency. Upper and lower abdominal ultrasonography findings were normal, there was no hepatomegaly.

Ophthalmic examination revealed that best corrected Snellen visual acuities were 6/60 with a correction of –13.00 D, –4.00 D × 150° OD and 6/60 with a correction of –12.00 D, –3.00 D × 10° OS. Goldmann applanation IOP was 50 mmHg and 60 mmHg, respectively. Slit lamp examination disclosed bilateral corneal edema with shallow anterior chamber depth. (Grade I, von Herick classification) Gonioscopy showed bilateral angle closure. (Grade I of Schaffer classification) Cup/disc ratios were 3/10 in the right eye and 5/10 in the left eye. There was a relative macular lamellar hole appearance bilaterally. Optical coherence tomography evaluation revealed pseudohole formation.

Pachymetric corneal thickness measurements were 728µ OD and 740µOS. Ocular dimensions were measured with A-scan biometry [Table 1].

She was hospitalized, and after pediatric and cardiologic consultation, antiglaucoma therapy was initiated as: 75 ml of 20% mannitol intravenously, 250 mg acetazolamide ½ tablets twice daily, betaxolol and brimonidine eye drops twice daily. IOP decreased to 30 mmHg OD and 34 mmHg OS enabling gonioscopic examination, which revealed bilateral total angle closure with anterior synechiae. Bilateral YAG-laser peripheric iridotomies were performed. During a 10-day follow-up period with only topical antiglaucoma medication, IOP changed between 28 - 34 mmHg in the right eye and 34 - 40 mmHg in the left eye. When IOP increased up to 60 mmHg in the left eye in spite of topical therapy, trabeculectomy was planned and performed with 2 minute 0.2 mg/ml mitomycin C (MMC) application. There was no intraoperative complication. IOP was 22 - 24 mmHg during the postoperative 3 days, probably because of residual viscoelastic material left in the anterior chamber. Later, IOP decreased to 16 mmHg and remained stable. Having had good pressure control on the left eye for 5 weeks after

**Table 1: Comparison of case study patient's ocular dimensions with those of normal children**

| | Central corneal thickness (µ) | Axial length (mm) | Anterior chamber depth (mm) | Lens thickness (mm) |
|---|---|---|---|---|
| Normal pediatric values[3,4] (ages 7 - 9) | 540 | 23.4 ± 0.94 | 3.60 ± 0.27 | 3.48 ± 0.18 |
| Case study patient at age 9 years old | | | | |
| OD | 720 | 21.60 | 2.20 | 4.45 |
| OS | 722 | 21.48 | 2.22 | 4.40 |
| Case study patient at age 12 years old | | | | |
| OD | 728 | 21.69 | 2.25 | 4.57 |
| OS | 740 | 21.72 | 2.28 | 4.52 |

Access this article online

Quick Response Code:

Website: www.ijo.in

DOI: 10.4103/0301-4738.104401

Ankara Numune Training and Research Hospital, Eye Clinic Ankara/ Turkey

Correspondence to: Prof. M. Sinan Saricaoglu, Ankara Numune Training and Research Hospital, Eye Clinic Ankara/ Turkey. E-mail: msinansarica@yahoo.com

Manuscript received: 08.11.10; Revision accepted: 05.06.11

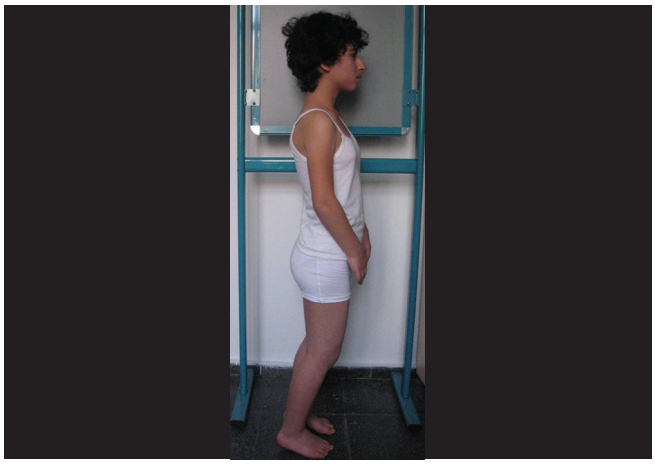

**Figure 1:** Side view. Typical tip-toe posture of geleophysic dysplasia is seen. The patient has small hands and feet

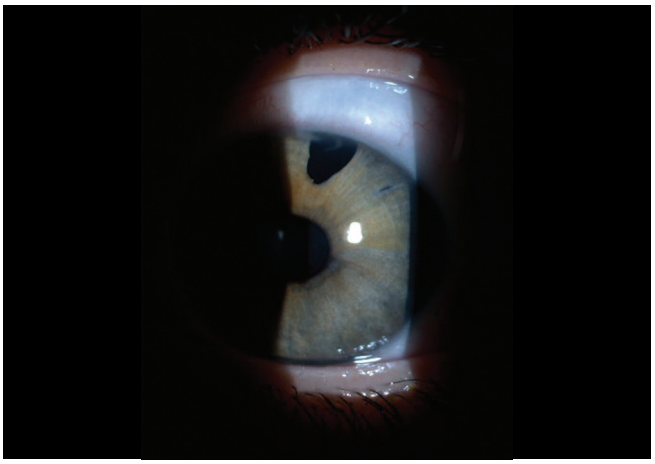

**Figure 2:** Anterior segment of left eye of the patient 3 years after trabeculectomy. There is a partially avascular bleb formation and laser iridotomy site temporal to the peripheric iridectomy

trabeculectomy, trabeculectomy with MMC was performed on the right eye. Although there was no intraoperative complication, hyphema was seen on the first postoperative day, which resolved spontaneously. IOP were 16 mmHg OD and 14 mmHg OS without any antiglaucoma medication. 2 weeks after discharge from the hospital, the patient presented with pain in the right eye. IOP was 30 mmHg OD, and gonioscopy showed closure of the internal ostium by a coagulum. She was hospitalized and an exploration of the trabeculectomy area was made with removal of the coagulum and irrigation of the anterior chamber. IOP was stable at 16 mmHg postoperatively. The patient was followed with 3-month intervals for 3 years. IOP was 16 - 18 mmHg in both eyes without antiglaucoma medication. There was a partially avascular functional bleb formation in both eyes [Fig. 2].

## Discussion

Geleophysic dysplasia is a lysosomal storage disease with a recessive genetic transmission.[1,5] Even though the condition was named after happy-looking or good-natured facial appearance ("gelios" and "physis"), this may not be seen in all patients as is the case with our patient.[6]

In the literature, ocular findings were reported in some of the geleophysic dysplasia patients. To our knowledge, there was only 1 reported glaucoma association.[2] That patient also had an angle closure glaucoma managed by YAG-laser iridotomy and topical antiglaucoma medication. In our patient on the other hand, there was a chronic angle closure glaucoma, which could not be managed by either iridotomy or by antiglaucoma medication. Bilateral trabeculectomy was performed by using low dose MMC with short duration, taking into consideration the age of the patient. Filtration surgery provided intraocular pressure management during the follow-up of 3 years. An alternative management could be a clear lens extraction, which would take care of the high myopia and also the angle closure element, but considering the age of the patient, we preferred keeping the crystalline lens and performing filtration surgery.

Clinically, it may be difficult to differentiate geleophysic dysplasia from Weill-Marchesani syndrome. Since short stature, brachydactyly, joint stiffness are among the clinical manifestations of this syndrome, and its inheritance is either autosomal-dominant or recessive. In contrast to geleophysic dysplasia, in Weill-Marchesani syndrome, there is a dull facial expression with malar and maxillary hypoplasia. Ophthalmoscopically, microspherophakia is a characteristic feature and glaucoma may accompany.[7] Our patient did not present with microphakia.

When compared with normal pediatric biometric values, in our patient, anterior chamber depth was shallower and the lenses were thicker.[3,4] After 3 years, the values were similar without any increase in lens thickness [Table 1]. On the other hand, central corneal thickness was found to be significantly thicker than the normal population as reported by Zhang *et al*.[2] Similar pachymetric and biometric results were obtained in patients with Weill-Marchesani syndrome.[8,9]

Presence of high refractive errors like myopic astigmatism, thick and steep central cornea may cause measurements of artificially higher IOP by applanation.[10] In planning antiglaucoma therapy, aiming for target pressure values and during follow-ups, corrected intraocular pressure values should be considered. Technologies considering corneal hysteresis and biomechanical properties of the cornea are expected to provide more accurate intraocular pressure measurements.[11]

In geleophysic dysplasia, patients' clinical findings may vary from moderate to severe. In severe cases, progressive thickening of the heart valves and upper respiratory stenosis may cause mortality at young ages. Strict control of these patients in cardiology and pediatrics departments is warranted. Our patient will be under ophthalmological follow-up as well as under evaluation of the cardiology department because of mitral valve anomaly and low-degree mitral insufficiency.

## References

1. Spranger JW, Gilbert EF, Tuffli GA, Rossiter FP, Opitz JM. Geleophysic dwarfism: A "focal" mucopolysaccharidosis? Lancet 1971;2:97-8.
2. Zhang X, Boles RG, Law SK, Lin M. Ocular findings in geleophysic dysplasia. J AAPOS 2004;8:198-200.
3. Saw SM, Chua WF, Hong CY, Wu HM, Chia KS, Stone RA, *et al.* Height and its relationship and biometry parameters in Singapore

Chinese children. Invest Ophthalmol Vis Sci 2002;43:1408-13.

4. Doughty MJ, Zaman ML. Human corneal thickness and its impact on intraocular pressure measures: a review and meta-analysis approach. Surv Ophthalmol 2000;44:367-408.
5. Pontz BF, Stoss H, Henschke F, Freisinger P, Karbowski A, Spranger JW. Clinical and ultrastructural findings in three patients with geleopysic dysplasia. Am J Med Genet 1996;63:50-4.
6. Santolaya JM, Groninga LC, Delgado A, Monasterio JL, Camarero C, Bilbao FJ. Patients with geleopysic dysplasia are not always geleophysic. Am J Med Genet 1997;72:85-90.
7. Saricaoglu MS, Sengun A, Karakurt A, Colluoglu Z. Autosomal dominant Weill-Marchesani syndrome and glaucoma management. Saudi Med J 2005;26:1468-9.
8. Razeghinejad MR, Safavian H. Central corneal thickness in patients with Weill-Marchesani syndrome. Am J Ophthalmol 2006;142:507-8.
9. Razeghinejad MR, Hosseini H, Namazi N. Biometric and corneal topographic characteristics in patients with Weill-Marchesani syndrome. J Cataract Refract Surg 2009;35:1026-32.
10. Chihara E. Assessment of true intraocular pressure: the gap between theory and practical data. Surv Ophthalmol 2008;53:203-18.
11. Kotecha A. What biomechanical properties of the cornea are relevant for the clinician? Surv Ophthalmol 2007;52 (Suppl):109-14.

**Cite this article as:** Saricaoglu MS, Güven D, Karakurt A, Hasiripi H. Geleophysic dysplasia associated with bilateral angle closure glaucoma. Indian J Ophthalmol 2013;61:122-4.

**Source of Support:** Nil, **Conflict of Interest:** None declared.