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BRIEF REPORTS

A case of bilateral corneal opacity

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ABSTRACT

Objective

To describe an extremely rare case of acromesomelic dysplasia with bilateral corneal opacity in a 6-year-old female.

Method

This is a case report.

Results

A 6-year-old female was referred for bilateral corneal opacity. Best corrected visual acuity for both eyes was 6/12. Ocular findings included normal corneal thickness with intact epithelium and diffuse mid to posterior stromal haze. The rest of the eye findings were within normal limits. Corneal topography showed symmetrical bow tie astigmatism of –3.38D for the right eye and –3.00D for the left eye.

Conclusion

Treatment of the ocular findings in acromesomelic dysplasia includes corrective lenses for errors of refraction and polarized lenses for glare. Keratoplasty may be contemplated in cases with total corneal opacification. Follow-up is necessary to assess the progression of the disease and decide on appropriate management of symptoms.

ACROMESOMELIC dysplasia is an extremely rare, progressive, autosomal recessive disorder characterized by premature fusion of the metaphyseal area of certain long bones. This condition is diagnosed on the basis of skeletal and radiologic changes occasionally present at birth, but which become more marked in the first two years of life.[1] A study by Ianakiev et al, localized six consecutive markers on chromosome 9 after gene mapping studies on four affected individuals.[2]

Patients with this condition exhibit unusually short forearms and lower legs and short stature during the first years of life. There may also be some lag in gross-motor performance, but intelligence is normal.[1] Abnormalities include bowed radius, brachydactyly, carpal synostosis, cone-shaped epiphyses of phalanges, irregular end-plates to vertebrae, macrocephaly, mesomelia of legs or hindlimbs, mesomelia of arms or limbs, barrel chest with pectus carinatum, platyspondyly, restriction of supination or pronation, short phalanges, wide metaphysis, and wide phalanges. Other occasional abnormalities are relatively large big toe and corneal clouding.[1, 3, 4]

We report the case of a 6-year-old female born full term via primary low-segment caesarean section for breech presentation to a 28-year-old G2P1 (1-0-0-1) with no fetomaternal complications. At 2 years, she had stunted growth, a prominent chest wall, and gross deformities of limbs, but no developmental delay. Consultation at a local hospital was made with no definitive management.

At 4 years, the patient was brought to the University of the Philippines–Philippine General Hospital (UP–PGH) pediatrics outpatient department where she was assessed to have rickets. Ancillary procedures showed delayed bone maturity and levoscoliosis. Differential diagnoses included a primary skeletal problem such as osteochondrodysplasia, probably acromesomelic dysplasia.

On physical examination, the patient showed stunted growth and short neck; pectus carinatum; enlarged medial and lateral malleoli of lateral, radial, and medial heads of both wrists; bowed radius; short forelimbs and hindlimbs; short and stubby fingers and toes; and genu valgum. Neurologic examination was normal.

Ophthalmologic examination revealed best-corrected visual acuity of 6/12 for both eyes. Pupils were 3 mm and reactive to light. Corneal diameters were 12 mm for both eyes with normal corneal thickness, intact epithelium, and diffuse mid to posterior stromal haze. The anterior chambers were formed with normal iris, round pupil, and clear lens. Intraocular pressure, gonioscopy, and findings on indirect ophthalmoscopy were unremarkable. Corneal topography showed symmetrical bow-tie astigmatism of –3.38D for the right eye and –3.00D for the left eye. Pachymetry was normal. Specular microscopy was unreliable because of poor visibility.

Cloudy cornea is an occasional finding in patients with acromesomelic dysplasia, and the relationship between the two has not been delineated.[1] In a case by Clarke et al., the cornea obtained after lamellar keratoplasty showed replacement of most of the Bowman's membrane and the stroma by fibrous tissue, with accumulation of extracellular acid mucopolysaccharides. They concluded that since the opacities were present at birth, the pathologic process may have occurred in utero and may have included inflammation, trauma, or toxic effects. The biochemical defect in this disease is not known, but may be responsible for

the scarring and the accumulation of mucopolysaccharide material proximal to a possible local enzyme block.

The treatment of acromesomelic dysplasia is directed toward the specific symptom and physical characteristics seen in each patient. Treatment may require the coordinated efforts of a team of specialists such as pediatricians, orthopedists, physical therapists, and ophthalmologists.[1, 3] Ophthalmic treatment includes corrective lenses for errors of refraction and polarized lenses for symptoms of glare. The patient still has good visual acuity and keratoplasty is, therefore, not indicated yet. Follow-up is necessary to assess the progression of the disease and to decide on appropriate management of symptoms.

References

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Frosted-branch angiitis

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ABSTRACT

Objective

To report a case of frosted-branch angiitis.

Method

This is a case report of frosted-branch angiitis seen at the University of the Philippines–Philippine General Hospital.

Results

A 42 year-old male presented with progressive blurring of vision of the left eye. Indirect funduscopy showed dilated retinal veins with perivascular sheathing, giving the appearance of frosted-branches of a tree.

Conclusion

Frosted-branch angiitis is a rare form of retinal vasculitis with various etiologies. Despite the severe retinal appearance, the prognosis is usually good, with rapid recovery of visual acuity after prompt steroid treatment.

FROSTED-BRANCH angiitis is a rare form of retinal vasculitis characterized by white perivascular sheathing of retinal blood vessels. The first case reported in 1976 involved a 6-year-old boy who had severe white sheathing of all retinal vessels presenting an appearance similar to the frosted branches of a tree.[1] Affecting more males (52%) than females (48%), frosted-branch angiitis is mostly seen in children and young adults. It usually affects individuals 6 to 16 years old in Japan and 23 to 29 in other countries. It is typically bilateral although unilateral cases have been reported.

This case involved a 42-year-old male who consulted at the University of the Philippines-Philippine General Hospital (UP-PGH) because of a 4-month history of progressive blurring of vision in the left eye. Visual acuity was 20/20 for the right eye and 20/40, improved to 20/25 on pinhole, for the left eye. Intraocular pressures were within normal limits for both eyes (OU). The anterior segment was normal.

Indirect ophthalmoscopy for the right eye was normal. The left eye, seen through a hazy medium, showed dilated and tortuous retinal veins with perivascular sheathing peripherally. There were some intraretinal foci of inflammation with scattered hemorrhages mostly in the inferior nasal periphery, and numerous vitreous opacities.

Fluorescein angiography (FA) of the left eye showed dilated veins with leakage of dye from the retinal vessels on late phase and multifocal areas of perivenular staining. There were areas of capillary nonperfusion on the inferonasal arcade with foci of hyperfluorescence. Systemic work-up for possible etiology and polymerase chain reaction of the aqueous humor yielded negative results.

It is still unclear whether frosted-branch angiitis is a unique disease entity by itself or a clinical presentation resulting from several causes as reported by Kleiner.[2] Its characteristic features are:[3]

- Severe sheathing of retinal vessels appearing like frosted branches of a tree in one or both eyes;
- Acute visual disturbance associated with signs of anterior-chamber and vitreous inflammation;
- FA demonstrates no occlusion or stasis of sheathed vessels, but late staining and/or leakage along vessels;
- Otherwise healthy patient;
- Prompt response to corticosteroid;
- Typically no recurrence.

In 1998, Kleiner et al.[4] classified the disease into 3 subgroups: idiopathic, those associated with hematologic malignancies like leukemia and lymphoma, and those caused by viral infection or autoimmune disease.

Most cases of frosted-branch angiitis are idiopathic, as in the case of our patient. An immune-mediated mechanism is believed to be the main cause as evidenced by localized ocular vasculitis sparing other organs. This