

CASE REPORT

BILATERAL SIMULTANEOUS ANTERIOR AND POSTERIOR LENTICONUS WITHOUT SYSTEMIC SIGNS

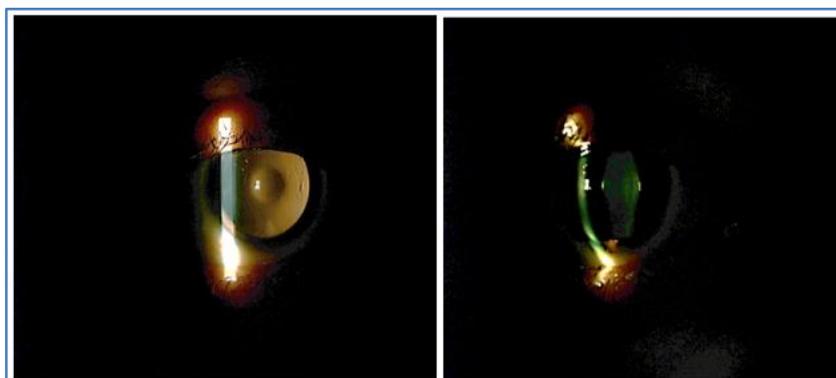
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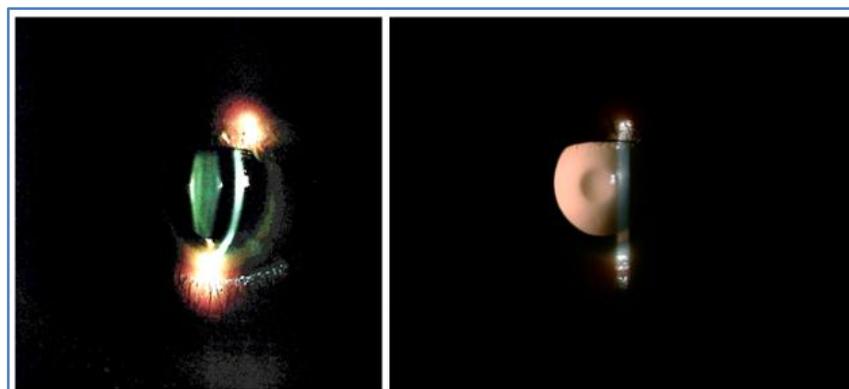
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CASE STUDY: A 21 year male came for eye checkup with a refractive error OD -12.00 sph, -2.25 @ 160° cyl and OS -15.00 sph, -2.50 @ 5° cyl and was referred to department of refractive surgery for opinion His BCVA in OD 6/18, OS 6/ 24. Slit lamp examination under dilation with phenylephrine plus tropic amide revealed the abnormal contour of the anterior and posterior lens capsule. Rest of the findings were normal with clear cornea, Von Herricks grade 3 deep anterior chamber, open angles in Gonioscopy.

Retro illumination showed the typical 'oil droplet configuration' in the lens. Retinal examination with the 90 D lens was otherwise normal except of the peculiar 'OMEGA' shaped curving of the slit beam over the retina which occurred due the difference in the refractive power of the lens in the axial and paraxial zones of the lens.



Right Eye



Left Eye

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Laboratory investigation for renal involvement revealed a normal study and clinically the patient's auditory functions were normal.

A-SCAN reveals an average axial length of 23.5 mm as expected in a case of lenticular myopia. B-scan reveals a normal study.

Patient is advised a refractive clear lens extraction. further due to economic reasons patients referred to a ophthalmic institute at Hyderabad, where he underwent clear lens extraction with Intraocular lens implantation and satisfied with his visual recovery.

CONCLUSION: Though it is documented in literature that Alports syndrome with bilateral lenticonus and renal involvement in the early age of life, it cannot be ruled out there may be delayed renal involvement and early ocular manifestation. Isolated bilateral simultaneous lenticonus without other systemic signs is also possible, though very rare. Nevertheless these patients should be warned and followed for delayed renal and systemic involvement.

DEFINITION OF LENTICONUS: A conical projection of either the anterior or posterior surface of the crystalline lens of the eye, occurring as a rare congenital anomaly. The anterior lenticonus is the most common and most often associated with Alport's syndrome. If the bulging is spherical, instead of conical, the condition is referred to as lentiglobus. It produces a decrease in visual acuity and irregular refraction that cannot be corrected by either spectacle or contact lenses.

Bio microscopically lenticonus is characterized by a transparent, localized, sharply demarcated conical projection of the lens capsule and cortex, usually axial in localization. In an early stage, retro-illumination shows an 'oil droplet' configuration. Using a narrow slit, the image of a conus is observed. Retinoscopically the oil droplet produces a pathognomonic scissors movement of the light reflex. This phenomenon is due to the different refraction in the central and the peripheral area of the lens. Oil drop sign is seen in mild posterior lenticonus. Atoll sign is seen in advanced case of posterior lenticonus with intact posterior capsule on slit lamp examination. The positive atoll sign will have more favorable prognosis since posterior capsule is intact, hence posterior chamber intraocular lens implantation will be more feasible with better visual prognosis as was the case with our patient. Fish tail sign is seen when lenticular cortex is hanging in vitreous cavity after posterior capsular dehiscence.

Ultrasonography also can illustrate the existence of a lenticonus. A-scan ultrasonography may reveal an increased lens thickness and B-scan ultrasonography may show herniated lenticular material, suggestive of a lenticonus. Amblyopia, cataract, strabismus and loss of central fixation may be observed in association with lenticonus posterior. Cataract, flecked retinopathy, posterior polymorphous dystrophy and corneal arcus juvenilis may be encountered in association with lenticonus anterior that occurs as a part of the Alport syndrome.

TYPES:

- Lenticonus anterior; lenticonus anterior is part of the Alport syndrome.
- Lenticonus posterior; lenticonus posterior is more common than lenticonus anterior and is sometimes found in Lowe syndrome.

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Alport syndrome or hereditary nephritis is a genetic disorder characterized by glomerulonephritis, end-stage kidney disease, and hearing loss. Alport syndrome can also affect the eyes, causing eye abnormalities including cataracts, lenticonus, kerataconus, as well as retinal flecks in the macula and mid periphery. Visibly bloody urine and protein in the urine are common features of this condition.

Alport syndrome is caused by mutations in COL4A3, COL4A4, and COL4A5, all genes involved in collagen biosynthesis. Mutations in any of these genes prevent the proper production or assembly of the type IV collagen network, which is an important structural component of basement membranes in the kidney, inner ear, and eye. Basement membranes are thin, sheet-like structures that separate and support cells in many tissues. When mutations prevent the formation of type IV collagen fibers, the basement membranes of the kidneys are not able to filter waste products from the blood and create urine normally, allowing blood and protein into the urine.

The abnormalities of type IV collagen in kidney basement membranes cause gradual scarring of the kidneys, eventually leading to kidney failure in many people with the disease. Progression of the disease leads to basement membrane thickening and gives a "basket-weave" appearance from splitting of the glomerular basement membrane, specifically the lamina densa layer. Single molecule computational studies of type IV collagen molecules have shown changes in the structure and Nan mechanical behavior of mutated molecules. Notably these lead to a bent molecular shape with kinks in the protein at the site of the mutations.

INHERITANCE PATTERNS: Alport syndrome can have different inheritance patterns that are dependent on the genetic mutation.

- In most people with Alport syndrome (about 85%), the condition is inherited in an X-linked dominant pattern, due to mutations in the COL4A5 gene.
- Alport syndrome can be inherited in an autosomal recessive pattern if both copies of the COL4A3 or COL4A4 gene, located on chromosome 2, have been mutated. Most often, the parents of a child with an autosomal recessive disorder are not affected but are carriers of one copy of the altered gene.
- Past descriptions of an autosomal dominant form are now usually categorized as other conditions, though some uses of the term in reference to the COL4A3 and COL4A4 loci have been published. Autosomal dominant transmission is rare and only accounts for 5% of affected patients. The clinical features of autosomal dominant Alport syndrome are similar to those of X-linked disease. However, deterioration of renal function tends to occur more slowly.

DIAGNOSIS: At least four of the following ten criteria must be met to diagnose an individual with Alport syndrome:

- Family history of nephritis or unexplained hematuria in a first degree relative of the index case or in a male relative linked through any numbers of females.
- Persistent hematuria without evidence of another possibly inherited nephropathy such as thin glomerular basement membrane disease, polycystic kidney disease or IgA nephropathy.

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- Bilateral sensor neural hearing loss in the 2000 to 8000 Hz range. The hearing loss develops gradually, is not present in early infancy and commonly presents before the age of 30 years.
- A mutation in COL4An (where n = 3, 4 or 5).
- Immunohistochemical evidence of complete or partial lack of the Alport epitope in glomerular, or epidermal basement membranes, or both.
- Widespread glomerular basement membrane ultrastructural abnormalities, in particular thickening, thinning and splitting.
- Eye lesions including anterior lenticonus, kerataconus, posterior subcapsular cataract, posterior polymorphous dystrophy and retinal flecks.
- Gradual progression to end stage kidney disease in the index case of at least two family members.
- Macro thrombocytopenia or granulocytic inclusions, similar to the May-Haggling anomaly.
- Diffuse leiomyomatosis of esophagus or female genitalia, or both.

The use of eye examinations for screening has been proposed.

IMMUNOHISTOCHEMISTRY: Immunohistochemical (IHC) evidence of the X-linked form Alport syndrome may be obtained from biopsies of either the skin or the renal glomerulus. In this processes, antibodies are used to detect the presence or absence of the alpha3, alpha4, and alpha5 chains of collagen type 4.

TREATMENT: As there is no known cure for the condition, treatments are symptomatic. Patients are advised on how to manage the complications of kidney failure and the spilling of protein in the urine that develops is often treated with ACE inhibitors.

Once kidney failure has developed, patients are given dialysis or can benefit from a kidney transplant, although this can cause problems. The body may reject the new kidney as it contains normal type IV collagen, which may be recognized as foreign by the immune system. Gene therapy as a possible treatment option has been discussed.

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