A Rare Case of Retinitis Pigmentosa with Central Areolar Choroidal Dystrophy

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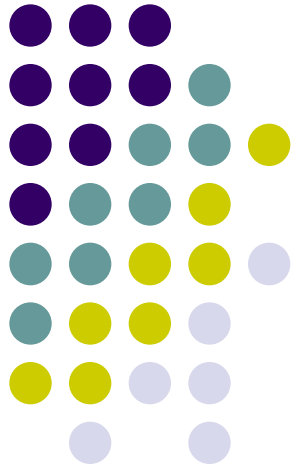


The author has no financial interests in the subject matter of this poster.

Case Report

- A 65yrs old male with defective vision on both eyes for the past 3 years which was progressive in nature was presented to our OPD.
- History of Night blindness for the past 30years.
- No history of ocular surgery, trauma, prolonged drug use was found.
- Family history was negative.

ANTERIOR SEGMENT

Examination	OD	OS
Best corrected visual acuity	2/60 By Snellen Visual Chart	2/60 By Snellen Visual Chart
Intra ocular pressure	12 mm Hg	12 mm Hg

Anterior segment

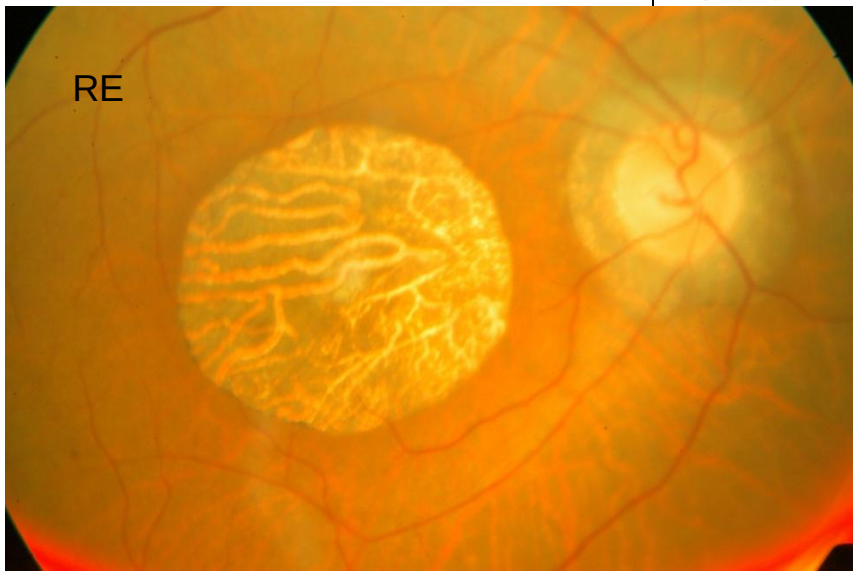
EXAMINATION	OD	OS
Lids	Normal	Normal
Conjunctiva	Normal	Normal
Cornea	Clear	Clear
Anterior chamber	Normal depth	Normal depth
Iris	Normal colour & pattern	Normal colour & pattern
Pupil	3mm & reacting to light	3mm & reacting to light
Lens	Lens changes	Lens changes
EOM	Full	Full

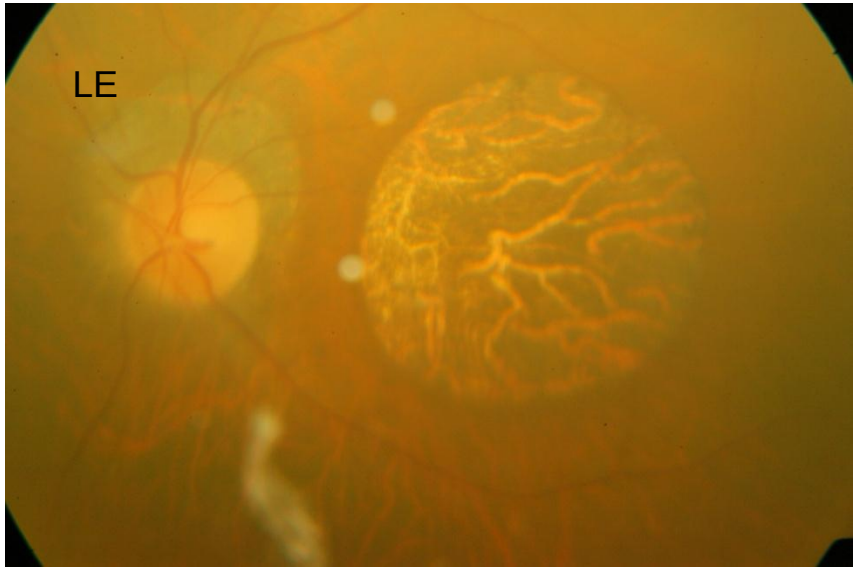
Posterior segment [slit lamp biomicroscopy with 90D lens]

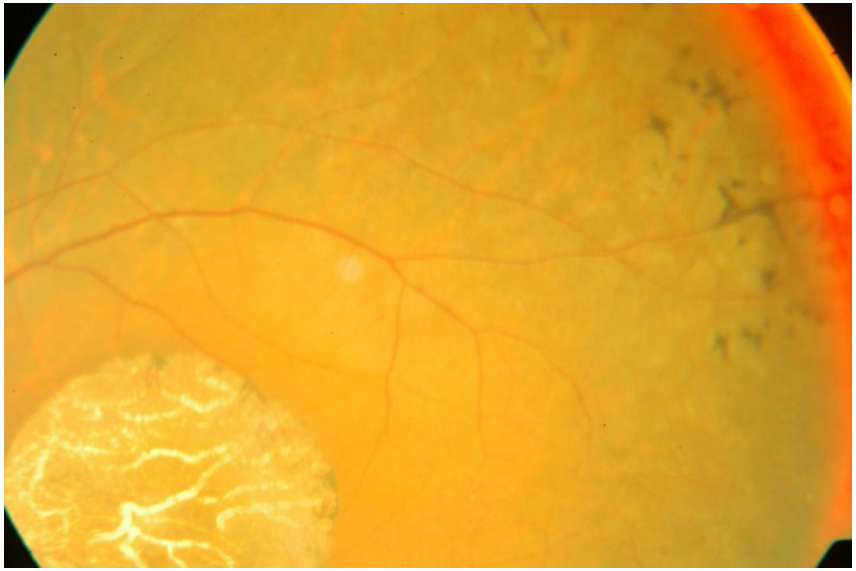
•**Both eyes** showed **waxy pale disc** with **retinal pigmentary changes** in all four quadrants with **reduced arterial calibre** and presence of **bony spicules** at the mid peripheral region of the retina.

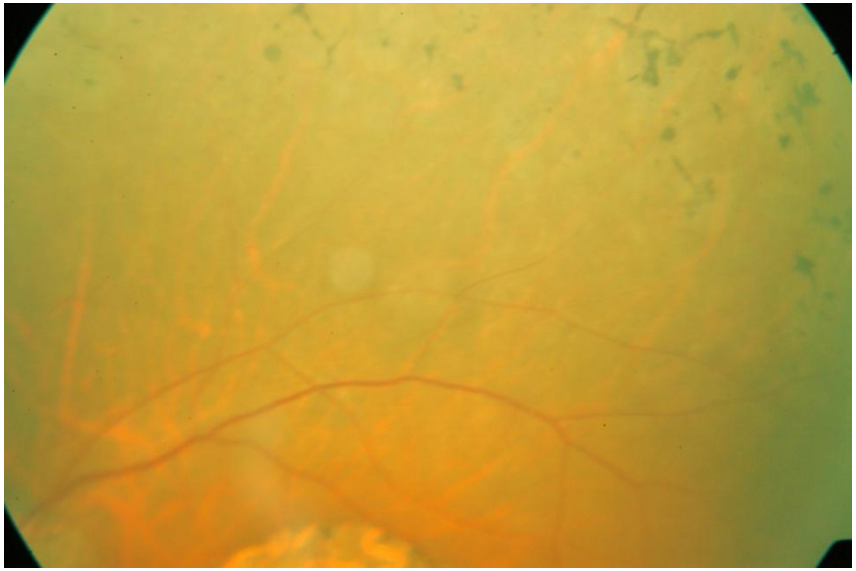
•**At Macula** :

A well defined oval area of **geographic atrophy** of retinal pigment epithelium and choriocapillaris was noted.







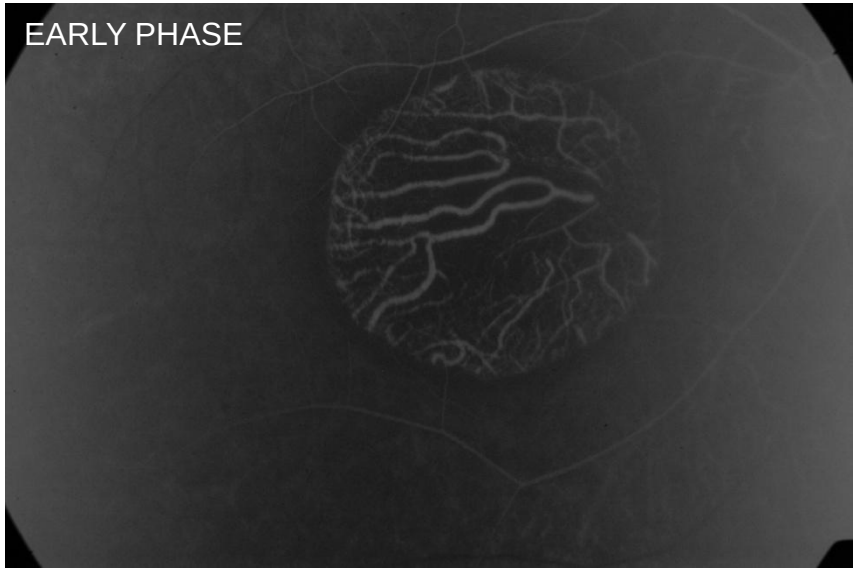


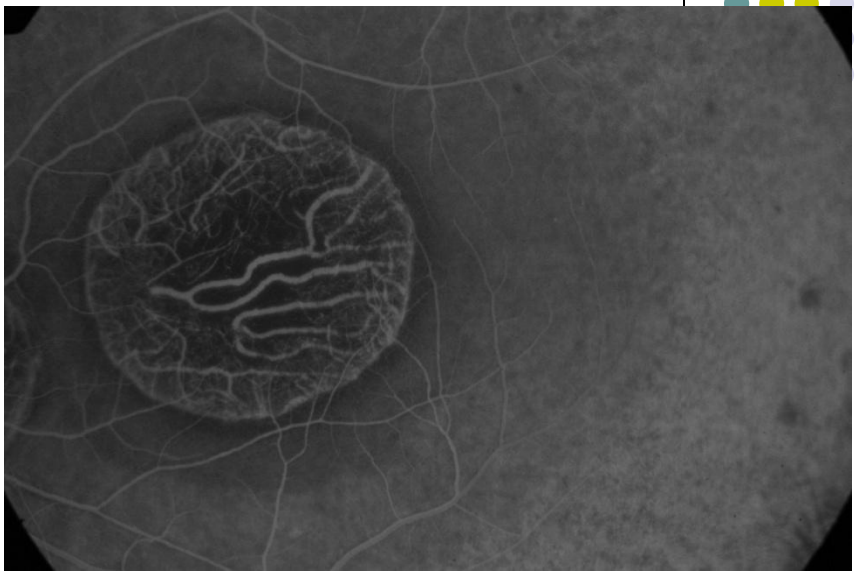
PRESENCE OF BONY SPICULES SEEN IN THE MIDPERIPHERAL REGION

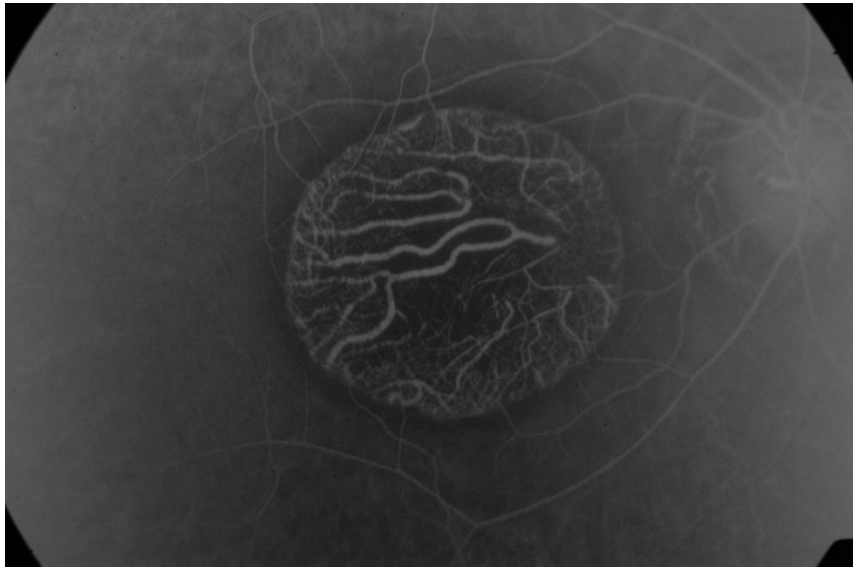
Fundus fluroscencian angiography

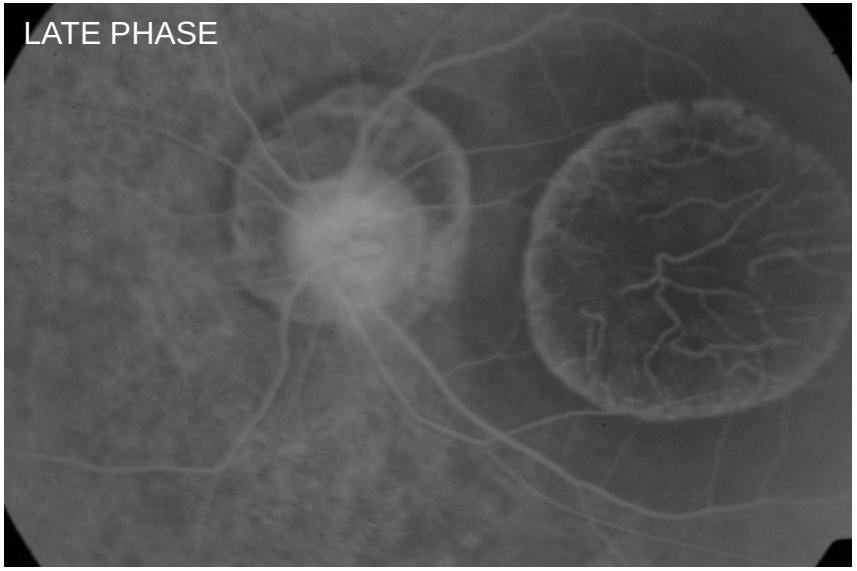
- At early phase window defect and background hyperfluorescence from retinal pigment epithelial atrophy was noted.
- The underlying intermediate and large choroidal vessels were outlined sharply due to the loss of the choriocapillaris.
- At later phase margin of the lesion showed hyperfluorescence due to leakage from choriocapillaris.

FFA









- Thus with the above findings the patient was diagnosed with **“Retinitis pigmentosa with central areolar choroidal dystrophy”**.

- The patient has been advised to use ‘low vision aids’ for better vision and to present himself for regular follow-ups to monitor the progress of cataract every quarter.

Discussion

- To the best of our knowledge the present case report is the first to describe the co-existence of **"Bilateral retinitis pigmentosa with central areolar choroidal dystrophy".**
- Central areolar choroidal dystrophy is a rare autosomal dominant choroidal disease that has been mapped to **chromosome 17p13**.[2,3]
- Interestingly several other inherited retinal diseases have been mapped to **chromosome 17p** [5] which includes

I. Autosomal dominant cone dystrophy,

II. **Retinitis pigmentosa,**

III. Leber's congenital amaurosis.

- A mutation in the peripherin gene on choromosome 6 also has been associated with central areolar dystrophy, linking it with choromosome6[1] retinopathies as well.

- This disease appears to be a primary dystrophy of either choroidal vessels or the pigment epithelium with secondary involvement of choroids.

References

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