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Case Series

A Case Series of Retintis Pigmentosa with Ocular and Systemic Associations

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ABSTRACT

Background
Retinitis pigmentosa (RP) is a group of rare, inherited retinal disorders characterized by poor night vision, poor peripheral vision and an overall decline in visual acuity due to progressive degeneration of rod photoreceptors followed by cone photoreceptors.

Aim
To study the various manifestations, ocular and systemic associations of retinitis pigmentosa.

Methods
This is a case series of 18 patients of Retinitis Pigmentosa done over a period of 1 year. A brief history was obtained, followed by visual acuity assessment, complete ocular examination, direct and indirect ophthalmoscopy examination, and fundus photography. These patients were evaluated for systemic associations as well. Other specialty evaluations were advised when required.

Results
A total of 18 patients were included out of which 13 patients were males and 5 patients were females. Most of them were in the age group of 15-30 years. 60% of total patients had vision <6/60. Anterior segment findings include posterior subcapsular cataract in 50% and keratoconus in 5% of patients. Posterior segment findings include pigmentary bony spicules involving mid-periphery and beyond in 77%, central and patchy distribution in 23%. Consecutive optic atrophy was identified in 44%, cellophane maculopathy in 33%, cystoid macular edema in 5%. Myopic changes were found in 16%, glaucomatous changes in 11%. Sensorineural hearing loss was identified in 22%, polydactyly, truncal obesity and alopecia were noted in 1 patient each.

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Conclusion
Retinitis Pigmentosa can be found in isolation (Typical RP) or in association with a variety of systemic disease presentation (Syndromic RP)- with Usher's syndrome being the most common syndromic association. Patients present with Nyctalopia, and peripheral vision loss. Visual acuity is affected late in disease but can be affected early due to Posterior Subcapsular Cataract, Cystoid macular edema, foveal atrophy. Electroretinography becomes abnormal very early in the disease and hence aids in diagnosis. Perimetry, Fluorescein angiography, Optical coherence Tomography are other imaging modalities. Low vision aids are useful to these patients. Gene therapy, Stem cell research, and retinal implants are various other treatment modalities available.
Key Words: Retinitis Pigmentosa, Usher's Syndrome.

INTRODUCTION

Retinitis pigmentosa (RP) is a group of rare, inherited retinal disorders characterized by poor night vision, poor peripheral vision and an overall decline in visual acuity due to progressive degeneration of rod photoreceptors followed by cone photoreceptors. The term "retinitis" initially suggested an inflammatory nature, which was presumed to underlie the disease. However, recent understanding states that inflammation is not the primary cause. On the other hand, "pigmentosa" refers to the characteristic accumulation of pigment in the retina due to the degeneration of photoreceptor cells, which release their pigment into the surrounding tissue as they deteriorate. Therefore, retinitis pigmentosa (RP), considered by most to be a misnomer, is a term that describes the combination of the two prominent clinical features of the grossly observed retina in RP: retinal degeneration and pigment accumulation. Fundus abnormalities, often affecting both eyes symmetrically, vary from near normal in early stages to a waxy pallor of the optic nerve head and attenuation of retinal vessels with or without bone spicule pigmentation in the periphery and/or midperiphery in advanced stages. RP has been classified as more common - Non-syndromic (not affecting other organs or tissues), and less common- Syndromic type (affecting other systems). Prevalence of non-syndromic RP is 1-4000[1]

AIMS AND OBJECTIVES

This study was conducted to study the various manifestations, various ocular and systemic associations of retinitis pigmentosa.

MATERIALS AND METHODS

This is a case series of 18 patients of Retinitis Pigmentosa conducted over a period of 1 year at Government General Hospital, Guntur. The study was conducted after obtaining approval from institutional ethics committee and consent was obtained from all patients. Each patient underwent complete ophthalmic examination and additional testing included fundus imaging. Patients were evaluated for systemic associations and other specialty consultations were advised when required.

RESULTS

This is a case series of 18 patients of Retinitis Pigmentosa.

CASE 1:

A 45 year old male patient presented with a chief complaint of diminision of vision during night since childhood. His visual acuity was 6/60 improving to 6/18 in right eye and 6/60 in left eye. Anterior segment revealed a quiet conjunctiva, clear cornea and optically clear anterior chamber. Posterior sub capsular changes are noted in both eyes (Left eye>Right eye).

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Examination of posterior segment revealed optic disc which is normal in shape ,size with distinct margins. Waxy pallor of the disc is noted with no cup disc differentiation. Arteriolar attenuation was noted with A:V ratio of 1:3. Foveal reflex was absent and maculopathy changes are noted. Bony spicules were noted in the mid periphery extending to the central retina .

Image 1

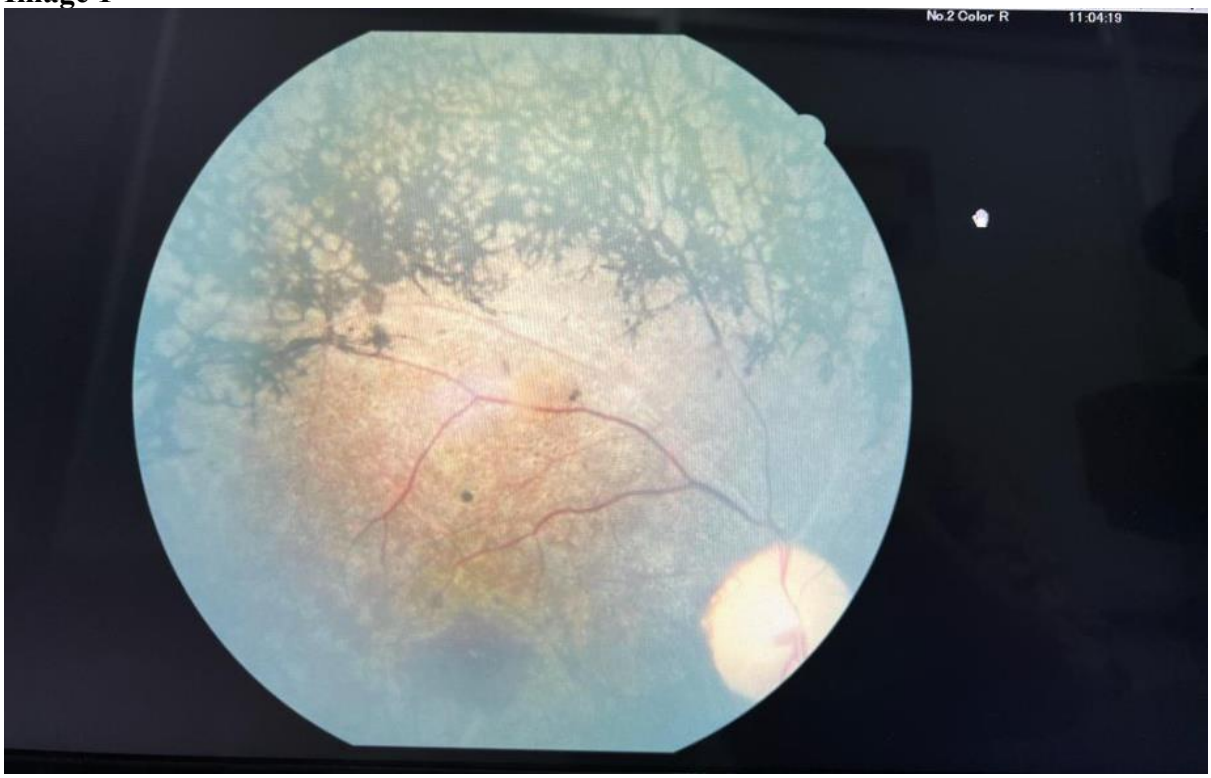
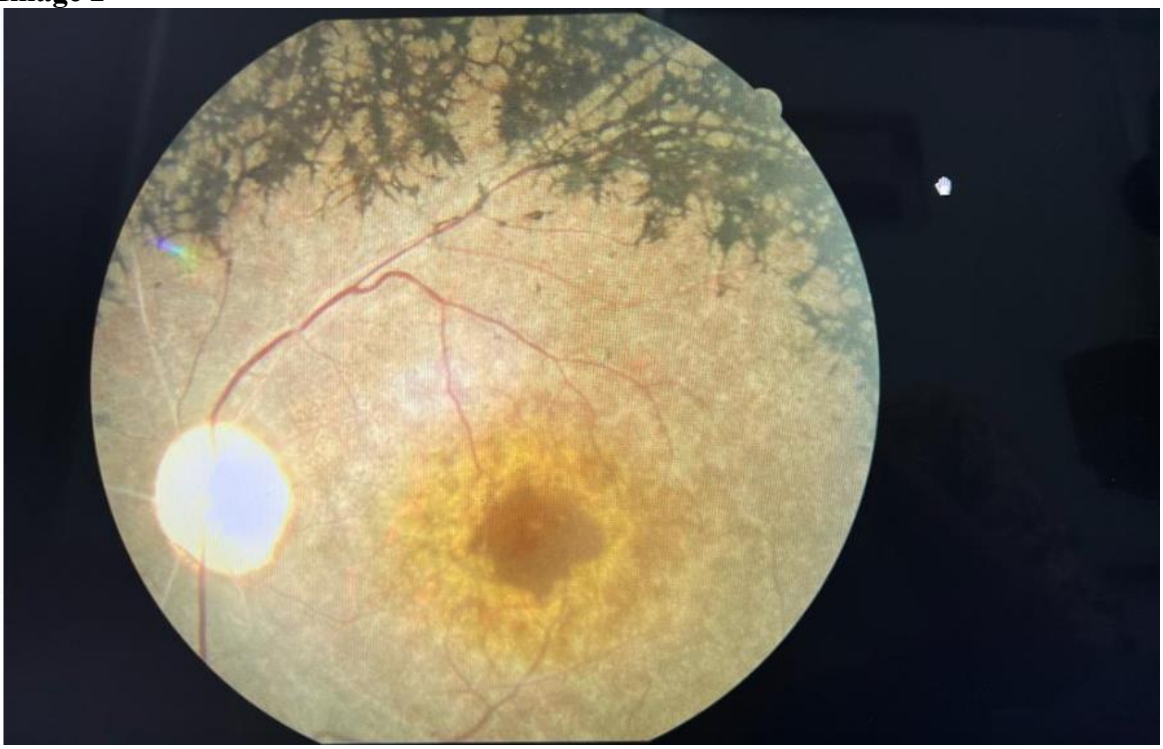


Image 2



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CASE 2:

A 56-year-old male patient presented with a chief complaint of diminution of vision during night since childhood. His visual acuity was counting fingers 3m in both eyes. History of consanguinous marriage was present. Anterior segment findings are normal.Posterior segment findings revealed clear media. Optic disc was normal in size and shape with distinct margins. Waxy pallor of the optic disc was noted with peri papillary atrophy. Arteriolar attenuation was noted with A:V ratio of 1:3. Foveal reflex was dull and background retinal examination revaled chorioretinal degeneration with sparse bony spicules in the mid periphery.

Image 3

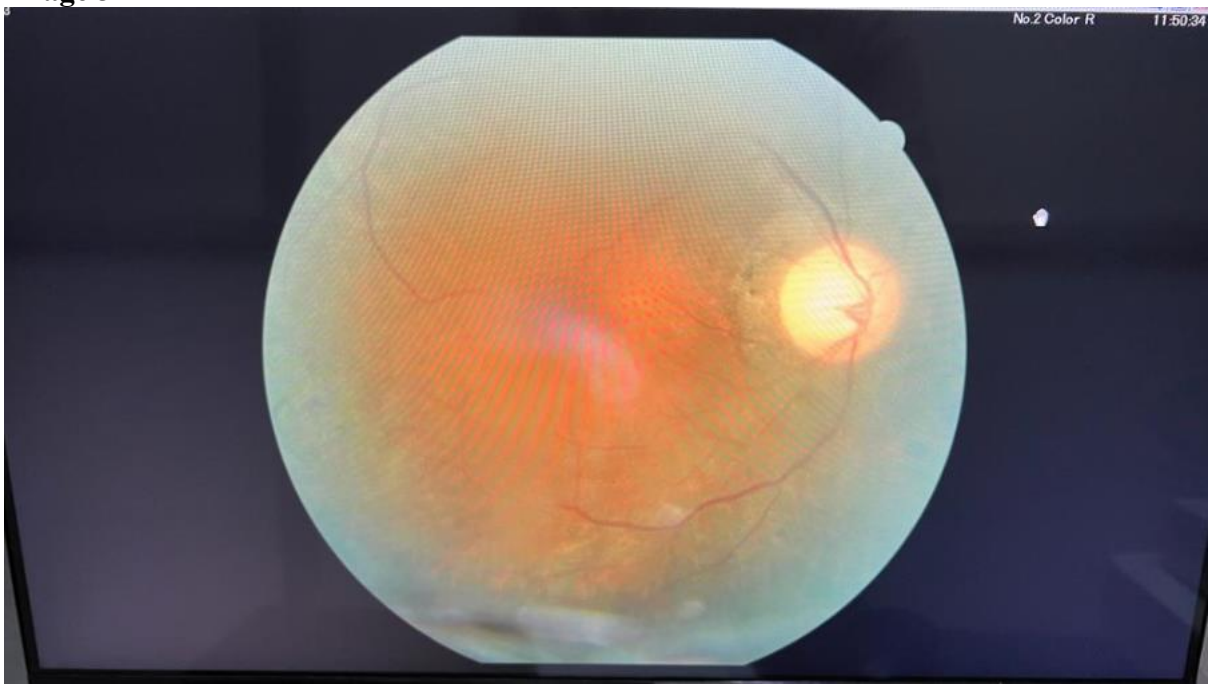
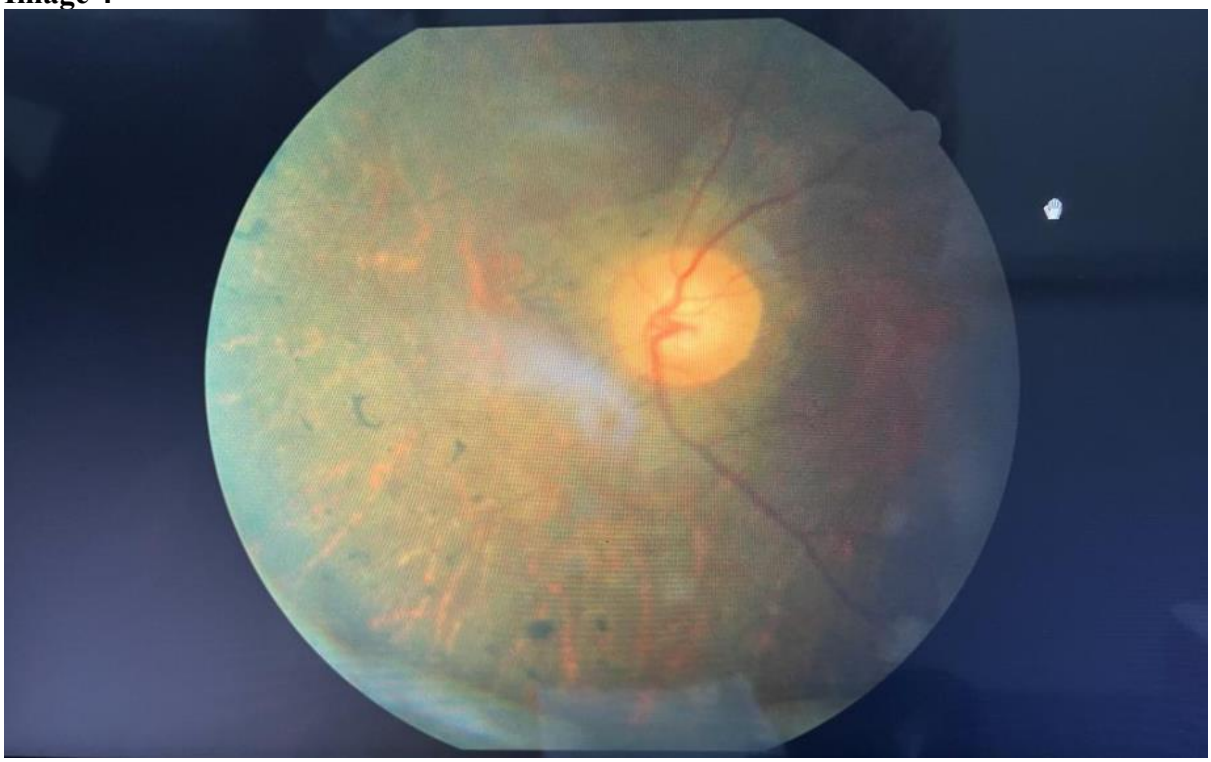


Image 4



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Demographic Features

Table 1: Age Distribution

AGE	FREQUENCY (%)
<15 years	5 (27.7%)
15-30 years	10 (55.5%)
>30 years	3 (16.6%)

Most of them were in the age group of 15-30 years.

Table 2: Gender Distribution

GENDER	FREQUENCY (%)
MALE	13 (72%)
FEMALE	5 (28%)

Table 3: **Vision**

VISUAL ACUITY		*FREQUENCY (%)*
<6/60	***7 (38.8%)***	***-***
>6/60	***11 (61.2%)***	***-***

Anterior Segment Findings

In this study we have found posterior subcapsular cataract in 45% of patients and 20% have already been operated for cataract surgery. We have noted myopic changes in 25% of patients and macular edema in 5% of patients. Other ocular associations we noted were keratoconus and glaucomatous changes.

Table 4: Anterior Segment Findings

FINDING	FREQUENCY (%)
Normal	5 (27.7%)
Posterior subcapsular cataract	8 (44.4%)
Pseudophakia	4 (22.2%)
Keratoconus	1 (5%)

Posterior segment findings

Table 5: Posterior Segment Findings

FINDING	FREQUENCY (%)
Pigmentary bony spicules: - Involving mid periphery and beyond - Central & patchy distribution	14 (77%) 4 (23%)
Consecutive optic atrophy	8 (44%)
Maculopathy	6 (33%)
Cystoid macular edema	1 (5%)
Myopic changes	5 (27.7%)
Glaucomatous changes	2 (11%)
Coloboma	1 (5%)

Systemic Associations

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Table 6: Systemic Associations

ASSOCIATION	FREQUENCY (%)
SENSORINEURAL HEARING LOSS	4 (22%)
HEART BLOCK	1 (5%)
POLYDACTYLY	1 (5%)
TRUNCAL OBESITY	1 (5%)
ALOPECIA	1 (5%)

DISCUSSION

Retinitis Pigmentosa refers to a group of pigmentary retinopathies, a generic name that covers all retinal dystrophies that presented with a loss of photoreceptors and retinal pigment deposits. It is characterized by pigment deposits predominant in the peripheral retina and by a relative sparing of the central retina. In most cases, there is a primary degeneration of rod photoreceptors, with secondary degeneration of cones. Thus, patients initially present with night blindness, and only suffer visual impairment in later life. Retinitis Pigmentosa can be Non-Syndromic Retinitis Pigmentosa and Syndromic Retinitis Pigmentosa.

Many syndromes are associated with pigmentary retinopathies. Frequent syndromes associated include: Usher's Syndrome and Bardet-Biedl Syndrome. Usher's syndrome is the most frequent syndromic form in which Typical RP is associated with neurosensory deafness[2]. About 14% of RP cases are in fact Usher's Syndrome[3]. Deafness, generally congenital and stable may be profound (Type 1) or moderate or medium(Type 2).

Bardet-Beidl Syndrome is less frequent than Usher's Syndrome with a prevalence of 1-1,50,000[4]. The phenotype is characteristic and associated RP with childhood obesity, mental retardation, post axial polydactyly, hypogenitalism and renal anomalies.

Less frequent syndromic associations include Alport's Syndrome, Cockayne Syndrome, Bassen Korntzweig disease, Refsum's Disease and others.

In this study, we have come across 18 cases of Retinitis Pigmentosa of which around 80% of them had ocular associations while only 40% of them had systemic associations.
Natarajan et al has reported that around 50% of patients of RP develop Posterior Subcapsular cataract cause of which has been subjected to be low grade ocular inflammation and that there is a higher prevalence of myopia and astigmatism in RP patients[5].

Juan. D. Arias et al reported prevalence of Macular edema in RP to be around 8%-50%, cause of which has been hypothesised to be breakdown of blood-retinal barrier[6].

In a study done by R P Mills and D M Calver on 17 patients of Retinitis Pigmentosa, they have found 6 patients of Usher's Syndrome- that comprises of RP with Sensorineural deafness[7].

CONCLUSION

In conclusion, our study confirms the retinal associations in previous studies and also adds to the existing literature. Patients of RP present with nyctalopia and progressive constriction of visual fields.

Visual acuity is usually affected late but can be affected early due to Posterior Subcapsular cataract, Cystoid macular edema and foveal atrophy.

Electroretinogram becomes abnormal very early in the disease and hence aids in diagnosis. Perimetry, Fluorescein angiography and Optical Coherence Tomography are other imaging modalities.

Low vision aids are useful to these patients. Gene therapy, Stem cell therapy and retinal implants are other treatment modalities available.

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REFERENCES

1. Ammann F, Klein D, Franceschetti A. Genetic and epidemiological investigation of pigmentary degeneration of the retina and allied disorders in Switzerland. *J Neurol Sci.* 1965;2:183–196. doi: 10.1016/0022-510X(65)90079-1
2. Hamel C. Retinitis pigmentosa. Orphanet J Rare Dis. 2006 Oct 11;1:40. doi: 10.1186/1750-1172-1-40. PMID: 17032466; PMCID: PMC1621055.
3. Boughman JA, Vernon M, Shaver KA. Usher syndrome: definition and estimate of prevalence from two high-risk populations. *J Chronic Dis.* 1983;**36**:595–603. doi: 10.1016/0021-9681(83)90147-9
4. Beales PL, Elcioglu N, Woolf AS, Parker D, Flinter FA. New criteria for improved diagnosis of Bardet-Biedl syndrome: results of a population survey. *J Med Genet.* 1999;**36**:437–446
5. Natarajan S. Retinitis pigmentosa: a brief overview. Indian J Ophthalmol. 2011 Sep-Oct;59(5):343-6. doi: 10.4103/0301-4738.83608. PMID: 21836337; PMCID: PMC3159313.
6. Arias, J.D., Kalaw, F.G.P., Alex, V. *et al.* Investigating the associations of macular edema in retinitis pigmentosa. *Sci Rep* **13**, 14187 (2023).
7. Mills RP, Calver DM. Retinitis pigmentosa and deafness. J R Soc Med. 1987 Jan;80(1):17-20. doi: 10.1177/014107688708000108. PMID: 3560119; PMCID: PMC1290625.