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Series of cases of Retinitis pigmentosa in the family

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Abstract

Retinitis pigmentosa (RP) is a group of inherited disorders affecting 1 in 3000-7000 people and characterized by abnormalities of the photoreceptors (rods and cones) or the retinal pigment epithelium of the retina involving progressive degeneration of the retina, typically starting in the mid-periphery and advancing toward the macula and fovea.

RP can be inherited in an autosomal dominant, autosomal recessive or X-linked manner.

The course of clinical and ERG changes is more aggressive in the X-linked form than in autosomal dominant retinitis pigmentosa disease.

Typical symptoms include night blindness followed by decreasing visual fields, leading to tunnel vision and eventually complete blindness in many cases.

The most common early symptom of RP is loss of night vision usually starting in childhood. Parents may notice that children with RP have trouble moving around in the dark or adjusting to dim light.

The classic clinical triad of RP is arteriolar attenuation, retinal pigmentary changes (could be either hypopigmentation and/or hyperpigmentation in form of bone-spicules), and waxy disc pallor.

We studied cases from Two-generations of family with retinitis pigmentosa , showing different stages of disease.

A Family of 5 members (father- 45 years,mother-35 years) with children born by second degree consanguineous marriage, (3 Girls -14 years ,11 years ,9 years) presenting with symptoms of nyctalopia. On Examination, Fundus pictures shows signs of Retinitis Pigmentosa in All the family members.

Keywords: Retinitis pigmentosa (RP), ERG-Electroretinography, Near Visual Acuity - N notation chart

Introduction

Retinitis pigmentosa (RP) represents a collection of genetic disorders, affecting roughly 1 in every 3000 to 7000 individuals.

These disorders are marked by progressive degeneration of the retina, starting usually in the outer areas and gradually moving towards the central retina (macula and fovea).

This deterioration primarily affects the photoreceptors in the retina, which are the rods and cones.

RP is passed down through families can be autosomal dominant, autosomal recessive, or X-linked genetic patterns.

Common symptoms of RP begin with difficulty seeing in low light or night blindness, followed by a progressive loss of peripheral vision, often leading to tunnel vision and, in many cases, eventually results in complete loss of vision.

We studied cases from two-generations of family with retinitis pigmentosa showing different stages of disease.

A family of 5 members (father- 45 years,mother-41 years) with children born by third degree consanguineous marriage, (3 Girls -14 years ,12 years,9 year) presenting with symptoms of nyctalopia.

On Examination, fundus pictures show signs of Retinitis Pigmentosa in all the family members.

Aim and Objectives

Aim: To examine the clinical manifestations of retinitis pigmentosa within a family in which the parents are having consanguineous marriage.

Objectives

1. To analyze the anterior segment findings in affected family members.
2. To assess visual field abnormalities through perimetry.
3. To conduct detailed fundus examinations to identify characteristic features of retinitis pigmentosa.

Materials and Methods

Study Design: It is a cross-sectional observational study to investigate retinitis pigmentosa (RP) within a family, involving a 45-year-old father, a 35-year-old mother, with three daughters.

Inclusion Criteria

Family members willing to participate in the study.

Methodology:

Patient Selection

All family members were included the father, mother, and three daughters.

Informed Consent: Informed consent from each participant, for their involvement in the study, and the use of their clinical data for research purposes was taken.

Clinical Examination: Clinical examination of each family member, focusing on anterior segment findings using slit-lamp bio-microscopy was done.

Perimetry Testing: Visual field testing using perimetry for all family members was done to analyze the results for patterns of visual field constriction.

Fundus examination and Photography:

Fundus examination of each participant was done with indirect ophthalmoscopy which was followed by high-resolution fundus photography.

Interpretation and Discussion

Interpretation of the results of anterior segment findings, retinal changes, and visual field patterns was done.

Genetic Counseling

Personalized genetic counseling session for the family and discussion about hereditary nature of Retinitis Pigmentosa was done with guidance for family planning.

Ethical Considerations

This study adhered to the principles outlined in the Declaration of Helsinki. Informed consent was obtained from all participants, and efforts were made to ensure the confidentiality and privacy of their personal information.

Case 1

A 45-year-old male presented with complaints of decreased vision at night since past 5 years. Best corrected visual acuity was finger counting at 1/2 meter in right eye and finger counting 1 meter in left eye.

Anterior segment examination revealed normal findings, with intraocular pressure measured as 12 mmHg in both eyes. Near vision was N12 in the right eye and N10 in the left eye, and color vision was within normal limits.

Fundus examination showed extensive bony spicules in both eyes involving macula with pale optic disc and attenuated blood vessels.

Perimetry showed a small island of vision (tunnel vision) confirming the advanced peripheral visual field loss.

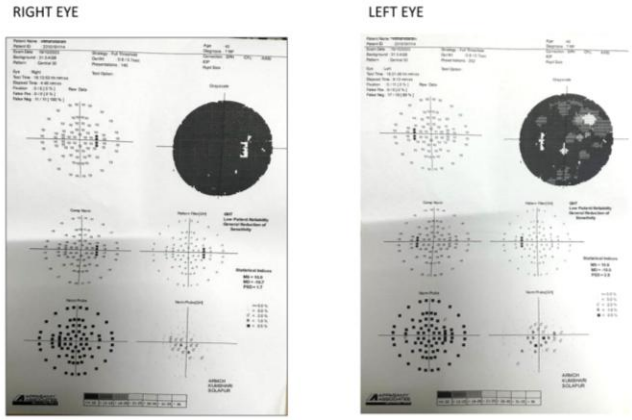

Figure 1

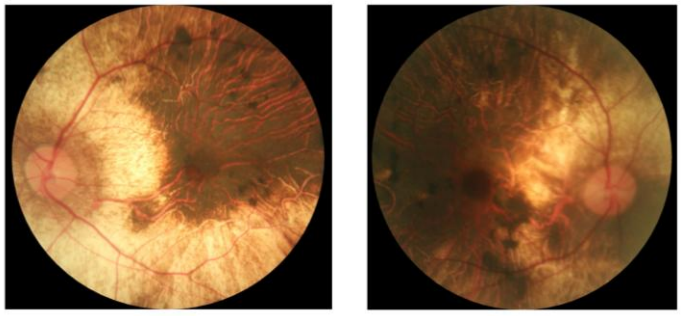

Figure 2

Case 2

A 41-year-old female presented with visual impairment, primarily at night. Best corrected visual acuity was 6/24 in right eye and 6/12 in left eye.

Anterior segment was within normal limits, with intraocular pressure measured as 10 mmHg in the right eye and 12 mmHg in the left eye. Near vision was N12 in both eyes, and color vision was within normal limits.

Fundus examination showed occasional bony spicules in the mid-peripheral retina with brownish pigments. The optic disc and blood vessels appeared to be normal.

Perimetry revealed peripheral visual field loss, consistent with the reported night vision difficulties, indicating progression.

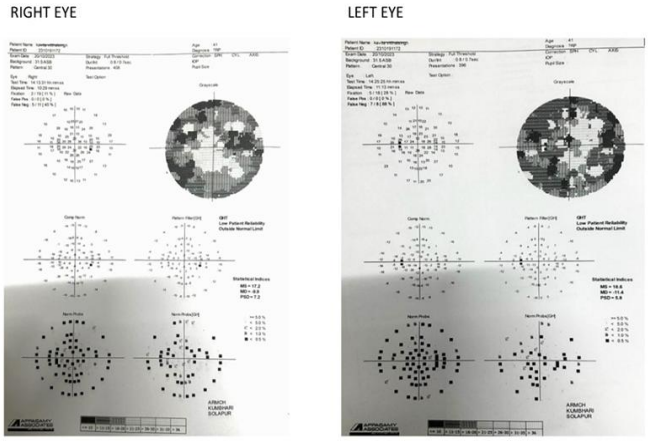

Figure 3

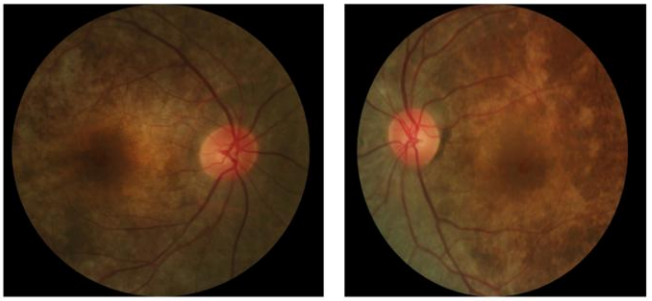

Figure 4

Case 3

The 14-year-old girl presented with complaints of decreased vision at night for the past two years.

Visual acuity was measured at 6/9 in both eyes, with a refractive error of (-5.75 diopters spherical and -2.25 diopters cylindrical @180 degree) in the right eye, and (-7.25 diopters spherical and -1.00 diopter cylindrical @170 degree) in the left eye.

Anterior Segment Findings: Anterior segment examination revealed normal limits, and intraocular pressure was measured at 10 mmHg in both eyes. Near vision was N10 in both eyes, and color vision was within normal limits.

Fundus Examination: Fundus examination showed occasional bone spicules in the mid-peripheral retina, Pale optic disc with myelinated nerve fibers superior and inferior to the disc and attenuated blood vessels.

Perimetry Results: Perimetry revealed a small island of remaining vision, confirming the presence of peripheral visual field loss, a characteristic feature of retinitis pigmentosa.

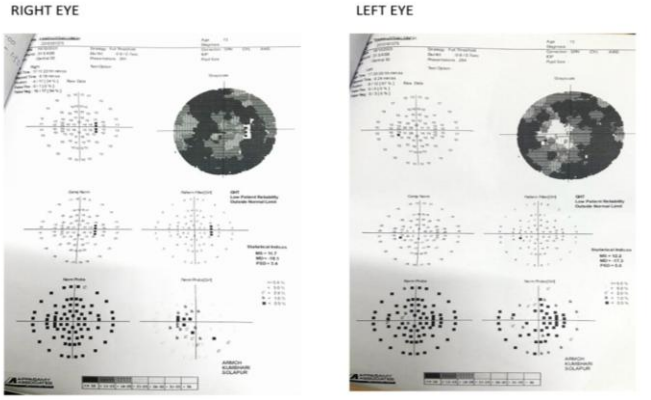

Figure 5

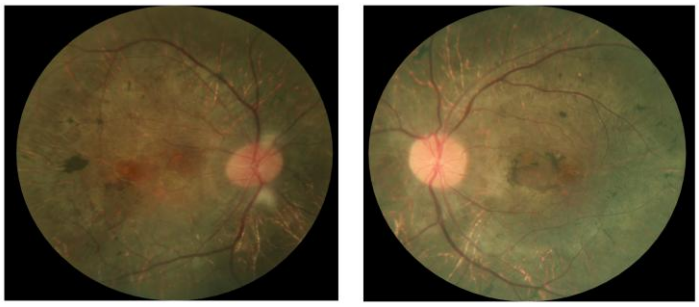

Figure 6

Case 4

A 12-year-old girl, younger sister of the 14-year-old, had not reported any vision complaints. Her visual acuity was 6/6 in both eyes.

Anterior segment examination showed normal findings, with intraocular pressure measured as 10 mmHg in both eyes.

Near vision was N6 in both eyes, and color vision was within normal limits.

Fundus examination revealed a normal appearance of the optic disc and pigmentation present around macula, with occasional bone spicules in the mid-peripheral retina.

No signs of atrophy or abnormalities were noted.

Perimetry was not done as patient was not cooperative.

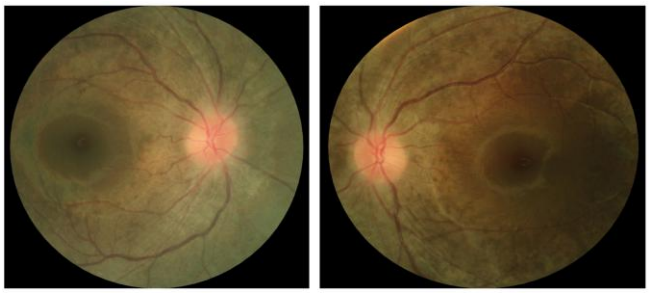

Figure 7

Case 5

A 9-year-old girl, the youngest sibling, had not reported any vision complaints. Her visual acuity was measured at 6/6 in both eyes.

Anterior segment examination revealed no abnormalities, with intraocular pressure measured as 12 mmHg in both eyes. Near vision was N6 in both eyes, and color vision was within normal limits.

Fundus examination showed a normal appearance of the optic disc, with occasional bone spicules and presence of pigmentation around macula observed in the mid-peripheral retina. Vascular architecture and overall retina was within normal limits.

Perimetry was not done as patient was not cooperative.

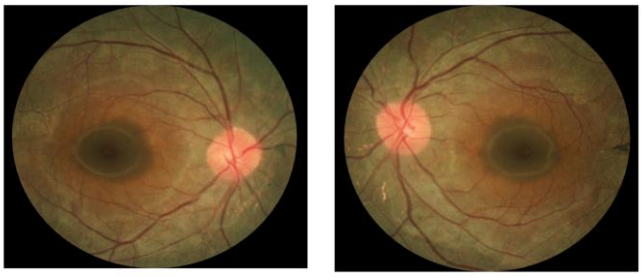

Figure 8

Discussion

Retinitis pigmentosa (RP) involves a complex interplay of genetic factors, resulting in the gradual loss of rod photoreceptor cells in the retina.

More than 60 genes have been associated with RP, with mutations in rhodopsin.

As rod cells deteriorate, patients experience night blindness (nyctalopia).

The most common early symptom of RP is loss of night vision usually starting in early adulthood. Parents may notice that children with RP have trouble moving around in the dark or adjusting to dim light.

The retinal lesions are characterized by intraretinal pigment migration, mainly in the peripheral retina, in addition to vascular attenuation and disc pallor.

Limitations

- Sample size was smaller.
- Collaboration with genetic specialists to perform genetic testing for each family member to identify specific mutations associated with Retinitis Pigmentosa was not done.

Conclusion

This cross-sectional observational study provides a comprehensive overview of retinitis pigmentosa within a family.

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