

## HEREDITARY ATYPICAL RETINITIS PIGMENTOSA: CASE REPORT

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### Abstract

This report presents four generations of hereditary atypical (pericentric) retinitis pigmentosa in an Itsekiri family of Warri, Delta state of Nigeria. The patients presented with nyctalopia, waxy disc pallor, arteriolar attenuation, pigment deposits around the optic nerve and visual field loss. The cases were typically mild with satisfactory vision beyond the fifth decade of life. The mode of inheritance was most probably autosomal dominant.

**Mots clés :** Pigmentolyse rétinite, héréditaire, atypique

### Résumé

L'objet de ce rapport est de présenter quatre générations de pigmentolyse rétinite (pericentrique) atypique héréditaire chez une famille d'Itsekiri de Warri, État de Delta au Nigéria. Les patients se sont présentés atteints de la nyctalopie, waxy disc pallor, atténuation artériolaire, dépôt du pigment autour de la nerve optique et la perte du champ visuel. Les cas étaient typiquement bénins avec la vision satisfaisante au delà de la cinquième décennie de la vie. La méthode d'héritage était plus probablement autosome dominant.

### Introduction

Retinitis pigmentosa (RP) is the most common form of hereditary retinal degeneration, affecting approximately 1 in 3500 individuals.<sup>1, 2</sup> It affects approximately 1.5 million people worldwide.<sup>3</sup> Classical RP is characterised by progressive night blindness and constriction of the peripheral visual fields, ultimately causing deterioration of the central vision in many patients. These symptoms are accompanied by degenerative and pigmentary changes such as retinal vascular attenuation, optic disc pallor, intraretinal pigment deposition and electroretinographic amplitudes, which are reduced or non-detectable. Abnormalities of electroretinogram are typically present before any detectable retinal change becomes visible to clinical examination.<sup>1, 2, 4</sup>

Retinitis pigmentosa can be transmitted by all inheritance modes, with X linked recessive RP accounting for 10-20% of genetically identifiable cases and being reported among the most severe forms.<sup>1, 2</sup> Molecular genetic research resulting in the localization and mutation analysis of genes responsible for some forms of RP has helped to elucidate the basic pathophysiology of these inherited retinopathies. Mutations in 4 genes have been shown to be responsible for autosomal dominant or recessive forms of non-syndromic RP, that is, rhodopsin,<sup>5, 6</sup>

peripherin/rds,<sup>7</sup> rom 1,<sup>8</sup>  $\beta$  pde,<sup>9</sup> and rod  $\alpha$  cGMP gated channel.<sup>10</sup> This report presents 4 generations of a mild form of retinitis pigmentosa in a Nigerian population.

### Case reports

Six members of one family who attended the consultant outpatient eye clinic of the University of Benin Teaching Hospital in the month of February 2003 were interviewed, examined and the findings recorded.

The index patient (case A) was the first to attend the clinic and was asked to come with all her first-degree relatives on the follow-up visit. The visual acuity was done using the Snellen's chart and near vision chart. When the patient could not see the chart, the ability to count fingers, see hand movement or perceive light was determined. The anterior segment was examined using the pen torch and the Haag-Streit slit lamp biomicroscope. Funduscopy was done using the Keeler's specialist ophthalmoscope. The patients were refracted when necessary to determine the best corrected vision. The pupils were dilated in each case with 10% phenylephrine and 1% tropicamide eye drops to facilitate funduscopy. Visual field analysis was done using the Kowa automatic visual field plotter. Hearing was tested using a tuning fork. Symptoms of systemic disorders including hearing loss, speech defect, neurological and mental defects

were excluded in each of the patients. They also had detailed systemic examination to exclude gross systemic defects.

Case 1 is a 37 years old Itsekiri female from Warri, Delta state, who presented with a history of difficulty reading of 4 years duration and poor night vision since her early teens. She is the third of five children in a monogamous home, two males and three females. Her two sisters also complained of poor night vision but her brothers have no visual complaints. Her father became blind at 80 years and her paternal grandmother also became blind at around 80 years old. Her visual acuity was 6/18 in the right eye and 6/12 in the left eye. Her near vision was N24 both eyes. There was no improvement in her distance vision with pinhole or with refraction but her distance vision improved to N 6 with add 2.75 Dioptre sphere. Fundus examination showed that she had slight waxy disc pallor, vascular attenuation, pigmentary changes around the optic disc, choroidal sclerosis and bronze beaten appearance of the macula. There were no perivascular bone-spicule pigmentary changes. There was also peripheral visual field loss. An impression of atypical retinitis pigmentosa was made and she was requested to bring other members of her family for examination.

Case 2 is an 83 year old retired civil servant. He is the father of case 1. He complained of gradual painless visual loss of about 7 years duration. He also had a history of poor night vision since his early teens. His mother had similar symptoms and became blind at about the age of 80 years. His three daughters all complain of poor night vision but his two sons do not. His visual acuity was hand movement in the right eye and light perception in the left eye. There were lens opacities in both eyes. Funduscopy after dilation of the pupils showed waxy optic disc pallor, vascular attenuation and peripapillary pigmentary changes. There was extensive choroidal sclerosis and marked atrophic changes in the macula. There was no perivascular bone-spicule pigmentation.

Case 3 is the elder sister of case 1. She is a 45 years old female who also developed poor night vision in her early teens. Her visual acuity was 6/18 and N24 in both eyes. She had similar fundus appearance as case 1.

Case 4 is the junior sister of case 1. She is a 35 years old female who also developed poor night vision in her early teens. Her visual acuity was 6/9 and N8 in both eyes. She also had similar fundus changes as case 1. The macular features were however less marked.

Case 5 is the 42 year old elder brother of case 1. He complained of slight difficulty reading small prints of 1 year duration. His visual acuity was 6/6 and N 8 in both eyes. Funduscopy showed normal disc and retina. His vision was improved to N 5 with add 1.00 Ds. Visual field analysis was normal.

Case 6 is the eldest daughter of case 3 who is the elder sister of the index patient. She is 21 years old and complains of slight impairment of vision at night of 2 years duration. Her visual acuity was 6/6 and N5 in both eyes. Funduscopy showed a normal disc and

vessels but with some peripapillary pigment deposit in the retina. Visual field analysis showed minimal peripheral field defects.

## Discussion

The diagnosis of retinitis pigmentosa in this report was based essentially on the symptom of poor night vision (nyctalopia), funduscopy findings of waxy disc pallor, arteriolar attenuation and retinal bone-spicule pigmentation as well as visual field loss. There were no facilities for electroretinography. Typically, the pigmentary changes have a perivascular bone-spicule configuration, which is initially observed in the mid retinal periphery and later extending posteriorly and anteriorly.<sup>11</sup> In contrast the pigmentary changes seen in these patients were in the central retina and had a peripapillary distribution. This finding is compatible with a recognized form of atypical retinitis pigmentosa known as pericentric retinitis pigmentosa.<sup>11</sup>

Retinitis pigmentosa can be associated with a wide variety of systemic disorders and form part of various syndromes.<sup>11, 12</sup> Most of these syndromes are inherited as autosomal recessive trait. Most of the systemic symptoms were excluded by a detailed history and examination of these patients. Investigations that would be of value in excluding these syndromes include detailed audiological assessment, skeletal muscle biopsy, electromyography and electrocardiography.

Molecular genetic studies and linkage analysis would have helped in elucidating the aetiology of this retinitis pigmentosa but facilities for these studies were not available. The usual modes of inheritance of retinitis pigmentosa are autosomal recessive, autosomal dominant and X-linked recessive.<sup>11</sup> Sporadic cases without any family history are the most common group. Some of these are autosomal recessive and some are new autosomal dominant mutations.<sup>11</sup>

This report presents 4 generations of retinitis pigmentosa in an Itsekiri family of Warri, Delta state of Nigeria. Although the grandmother of the index patient was not seen since she had already passed away, her symptoms were so typical that one could safely assume that she had a similar disorder as her son, grand daughters and a great grand daughter who had clinical features of retinitis pigmentosa.

The most likely mode of inheritance in this series is autosomal dominant. This is supported by the fact that this disorder as expressed in consecutive generations even though the patients were married into families where there was no history suggestive of night blindness or retinitis pigmentosa. In addition, this form of retinitis pigmentosa was typically mild allowing satisfactory vision even beyond the fifth decade of life. This is in agreement with the finding that autosomal dominant forms are known to have the best prognosis.<sup>3, 11</sup> Autosomal recessive transmission would require that both spouses should have a family history of the disorder before it can be transmitted to

their offspring. This history was however absent. Autosomal recessive cases are known to be more severe. X-linked recessive retinitis pigmentosa will only manifest clinically in males. Females are carriers. In this report, the disorder was manifest clinically in females. X-linked recessive cases are known to be among the most severe forms of retinitis pigmentosa.<sup>1,2,11</sup> The cases in this report are mild.

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