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Retinitis Pigmentosa in Awka, Nigeria: A Survey Study

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Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

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ABSTRACT

Background: Retinitis pigmentosa which causes irreversible blindness and affects all gender is usually inherited.
Objectives: To describe the incidence and pattern of retinitis pigmentosa at the Eye Unit of Chukwuemeka Odumegwu Ojukwu University Teaching Hospital Awka, Nigeria.
Materials and Methods: The case files of all the new patients seen at the Eye Unit of the Chukwuemeka Odumegwu Ojukwu University Teaching Hospital Awka between January 2014 to December 2021 were reviewed. Those with clinical diagnosis of Retinitis Pigmentosa were selected and information bordering on age, sex, occupation, disease duration. associated morbidity and fundus findings were extracted and analyzed using descriptive statistics.

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Results: Out of the 5461 new patients seen at the Eye Clinic within the study period, 29(0.5%) were diagnosed of Retinitis Pigmentosa. Of the 29 patients 18 (62.1%) were males and 11(37.9%) females, with male to female ratio of 1.6:1. The age range was 7 years to 69 years. Thirteen patients (44.8%) were between 16 years and 44 years. Median age was 33 years while the mean age was 34. 8+15. 2 years. Nine (31.0%) patients were students. The most common complaints of the patients were poor vision, 27(93.1%) and refractive error 8(27.68). Seven patients (24.18) were blind at presentation while retinal pigmentation was the most frequent fundus finding 29 (100%).
Conclusion: Retinitis Pigmentosa is a cause of visual impairment and blindness which reduces the quality of life of affected persons. Early diagnosis and visual rehabilitation should be encouraged.

Keywords: Retinitis pigmentosa; Awka; Nigeria.

1. INTRODUCTION

Retinitis pigmentosa is a group of hereditary disorders characterized by progressive loss of photoreceptor and retinal pigment epithelial (RPE) function [1,2]. It is a primary pigmentary retinal dystrophy predominantly affecting the rods and cones [3]. However, the rods are more affected than the cones [1,2]. The retinal dystrophic lesions of the retinitis pigmentosa is known to cause visual disability in all age groups [4] and retinitis pigmentosa is a common diagnosis among children in school for the visually impaired in Nigeria [5,6]. The prevalence varies between 1 in 4000 and 1 in 5000 of the world population [1,3] It appears in the childhood and progresses slowly, often leading to blindness in advanced middle age [3] More than one million people are affected globally [2].

Retinitis pigmentosa has been reported to be a common cause of blindness during working life in the industrialized nations of the world [7] In Denmark [8] retinitis pigmentosa has been noted to be a leading cause of blindness in people aged 20 – 64years accounting for 29.9%. Also in kuwait [9] retinitis pigmentosa is the leading cause of visual incapacitation in people younger than 60 years of age. In Africa, a study reported retinitis pigmentosa as the most common cause of inherited blindness [10]. A Cameroonian study [11] reported the frequency of retinitis pigmentosa to be 0.16% with bilateral blindness rate of 30%. In Nigeria, retinitis pigmentosa accounted for 0.69% of all new patients seen at the out patients department in Ibadan [12] and 5.2% of all retinal diseases in Ile-Ife [13] all in Western Nigeria.

In Onitsha, southeastern Nigeria, Nwosu [14] reported that retinitis pigmentosa constitutes 11.1% of retinal diseases and that it is an important cause of blindness and low vision. Yet in another study, Nwosu et al. [15] documented the prevalence of retinitis pigmentosa as 0.6%. Retinitis pigmentosa is therefore of worldwide occurrence [4-9,12-15] and no race is known to be exempted or at a higher risk [3].

Retinitis pigmentosa is a genetic disorder of the eye that causes loss of vision [16]. It is one of the most common forms of inherited retinal degeneration [2]. There are multiple genes that when mutated can cause retinitis pigmentosa phenotype [17]. Inheritance patterns of retinitis pigmentosa have been identified as autosomal dominant, autosomal recessive, x-linked and maternally (Mitochondrially) acquired and are dependent on specific retinitis pigmentosa gene mutations present in the parental generation [18]. Missense mutation of rhodopsin gene mutations present in the parental generation [18]. Missense mutation of rhodopsin gene (rhodopsin is a pigment that plays an essential part in the visual transduction cascade, that is vision in low light conditions) most frequently follow autosomal dominant inheritance pattern and accounts for approximately 25% of autosomal dominant form of retinitis pigmentosa [2,19]. The autosomal dominant variant has the best prognosis, of all the inherited retinitis pigmentosa; it has a later onset and runs a milder cause than autosomal recessive. The x- linked recessive disease is least common, has an earlier onset and runs the most severe course resulting in total blindness by the 3rd or 4th decade [20,21]. In rare occasions, a dominant form of the x-linked mutation will affect both males and females equally [22]. Several other studies have reported various codon mutations associated with retinitis pigmentosa [19,23,24,25,26]. Mutation in more than 250 genes has also been linked to retinal dystrophies [20]. There is also sporadic cases of retinitis pigmentosa. Retinitis pigmentosa can also be classified into typical, atypical and syndromic (those associated with rare systemic disorders) types, but the typical variant is the most common [12,20,21,25,27,28]. Symptoms of retinitis pigmentosa include trouble with night vision, decreased peripheral vision and as peripheral vision worsens, the affected may

experience tunnel or tubular vision [16]. The onset of symptoms is generally gradual and often in childhood [16,29]. However, complete blindness is rare [29].

2. MATERIALS AND METHODS

This is a retrospective hospital-based study carried out at the Chukwuemeka Odumegwu Ojukwu University Teaching Hospital Awka, Nigeria. The case files of all the new patients seen at the Eye Unit of Chukwuemeka Odumegwu Ojukwu University Teaching Hospital Awka between January 2014 and December 2021 were reviewed. Those with clinical diagnosis of retinitis pigmentosa were selected and information bordering on visual acuity, age, gender, occupation, disease duration, associated morbidity and fundus appearance were extracted. The data were analyzed using descriptive statistics and presented as frequency Tables.

3. RESULTS

Of the 5461 new patients seen at the Eye clinic of the Chukwuemeka Odumegwu Ojukwu University Teaching Hospital Awka within the study period, 29(0.5%) were diagnosed of retinitis pigmentosa. Eighteen (62.1%) were males and 11(37.9%) females with male to female ratio of 1.6:1. The age range was 7 years to 69 years. Thirteen patients were between the 16-44 years age range. The age range 1 – 15 years and ≥ 65years had four (13.8%) patients each.

The median age was 33 years and the mean age was 34.8+15.2 years. The visual acuity (VA) at the presentation using World Health Organization (WHO) category were as follows: normal vision (27.6%), visual impairment 11(38%) and blindness 10(34.4%). After refraction, some had their visual acuity improved.

Table 3 showed occupational distribution of the patients with retinitis pigmentosa. Nine (31.0%) patients were pupils/students, 7(24.1%) were workers (civil servants/company workers), 6(20.7%) traders, while 2(6.9%) were unclassified. 5(17.2%) were artisans.

The complaints of the patients at presentation were many and varied and some had more than one complaint. The chief complaints were poor vision 27(93.1%), poor night vision 23(79.3%) headache 9(31.0%), eye ache 6(20.6%) and diminished vision which had the highest frequency with all the patients complaining of such.

The associated co-morbidity found among the retinitis pigmentosa patients were refractive error 8(27.6%) of which 4(13.8%) were myopia, 2(6.9%) hypermetropia while myopic astigmatism and hypermetropic astigmatism were 1(3.5%) each. Three (10.5%) patients had bilateral immature cataract of which 2(7.0%) were male and 1(3.5%) female. One male (3.5%) had uniocular aphakia, which was as a result of couching done by some itinerant doctors who came to their village. Glaucoma recorded 5(17.2%) patients of which 3 (10.38) were males and 2(6.9%) females.

The fundus findings of the patient with retinitis pigmentosa were perivascular retinal pigmentation 29(100%), retinal vascular attenuation 22(75.9%) and disc pallor 18(62.1%). Glaucomatous optic disc cupping was seen in 5(17.2%) of retinitis pigmentosa patients.

Table 1. Age and Sex distribution of retinitis pigmentosa patients

Age group (years)	Male	Female	Total
1 – 15yrs	3(10.4%)	1(3.4%)	4(13.8%)
16 – 44yrs	9(31.0%)	4(13.8%)	13(44.8%)
45 – 64yrs	5(17.2%)	3(10.4%)	8(27.6%)
≥65	1(3.4%)	3(10.4%)	4(13.8%)
Total	**18(62%)**	**11(38%)**	**29(100%)**

Male: female ratio = 1.6:1
mean age = 34.8yrs

Table 2. Visual acuity (VA) at presentation and after refraction

Who, Category	VA at presentation	VA after refraction
Normal vision 6/6 – 6/18	8(27.6%)	13(44.8%)
Visual impairment < 6/18 – 3/60	11(38%)	9 (31.1%)
Blindness, VA < 3/60	10(34.4%)	7 (24.1%)
Total	**29(100%)**	**29(100%)**

WHO: World Health Organization
VA: Visual Acuity

Table 3. Occupational and sex distribution of patients with retinitis pigmentosa

Occupation	Frequency	Male	Female
Students	9(31.0%)	6(20.7%)	3(10.3%)
Traders	6(20.7%)	4(13.8%)	2(6.9%)
Artisans	5(17.2%)	3(10.3%)	2(6.9%)
Workers (civil servants/company workers)	7(24.1%)	3(10.3%)	4(13.8%)
Others	2(6.9%)	2(6.9%)	-
Total	**29(100%)**	**18(62%)**	**11(38.0%)**

Table 4. Major complaints of retinitis pigmentosa at presentation

	Complaints	Frequency
1	Diminished vision	27(93.1%)
2	Poor vision at night	23(79.3%)
3	Headache	9(31.0%)
4	Eye ache (Eye pain)	8(27.6%)
5	Photophobia	6(20.6%)
	Total	**73**

Some patients had multiple complaints

Table 5. Co-morbidity with retinitis pigmentosa

Co –morbidity	Frequency
Refractive error	8(27.6%)
Myopia	6(20.7%)
Glaucoma	5(17.2%)
Cataract	3(10.5%)
Hypermetropia	2(6.9%)
Unocular Aphakia	1(3.5%)

Table 6. Fundus finding of the Patients with retinitis pigmentosa

S/N	Fundus Findings	Frequency
1	Bone spicule retinal pigmentation	29 (100%)
2	Retinal vascular attenuation	22 (75.9%)
3	Disc Pallor	18 (62.1%)
4	Disc cupping	5 (17.2%)

4. DISCUSSION

Retinitis Pigmentosa (RP) comprises a large group of inherited vision disorders that cause progressive degeneration of the retina, the light sensitive membrane that coats the inside of the eyes [30]. Peripheral or side vision gradually declines and eventually is lost in most cases. However, central vision is usually preserved until late in these conditions [30]. Retinitis Pigmentosa is generally not a very common disease [31]. The frequency of retinitis pigmentosa in the present study was 0.5% and this is in consonance with earlier studies by Nwosu [15] (0.6%) and Ashaye12 (0.69%).

However, other studies [32,33] carried out on retinitis pigmentosa in same Southern Nigeria as Nwosu15 and Ashaye12 did not give report of frequency on their population of studies. Eballe et al. [11] in Cameroon reported the frequency of 0.16% which is lower than that of the present study and other studies [12,15] The higher mean age in the Cameroonian study may have affected the frequency. More males (62.1%) than females (37.9%) were found to have retinitis pigmentosa in the present review and this is in line with other authors [31,32,33]. The age range mostly affected in this review is 16 – 44years (31.0%) followed by 45 – 64 years (17.2%). Onakpoya et al. [33] in a multicentre study in southwestern Nigeria had reported similar trend in age range affected by the retinitis pigmentosa [33]. This supports the findings of earlier studies [7,8,9] that retinitis pigmentosa is a frequent cause of blindness during active life [34]. This may spell socio-economic doom for the affected, their family and the society in general as the blind

years may be prolonged. The occupational distribution of the patients in this study showed that majority of the patients were in their career and economic pursuits and the attendant visual incapacitation arising from retinitis pigmentosa may likely affect their progression in their chosen fields. Parmegigian [7] and Marmor [35] had earlier reported the economic impact of those affected by retinitis pigmentosa on their families and the society at large. The quality of life and livelihood of the affected individuals are also impacted negatively [36].

Visual impairment (38%) and blindness (34.4%) were elicited in the present study. The blindness rate in the present study is similar to that of Eballa et al. (30%), in Cameroon [11]. However, this differs from that of Onakpoya et all. [33] who reported a higher rate of 41.7% and the difference could be due to the fact that Onakpoya et al. [33] did a multicentre study. Yet while Ukponwan et al. [32] in Benin Nigeria reported blindness rate of 50%, another author [37] in the United States of America (USA) had reported blindness rate of 25%. These differences could generally be attributed to age of presentation of the patients. Onakpoya et al. [33] had earlier reported that older patients had higher rate of blindness at presentation. However, the youngest ,patient, 7 years old, had VA of <3/60 that was not improved by refraction and he dropped out of school. In the same vein, a 30 year old woman in this review also had profound visual loss and was relieved of her duty in her work place. This gives credence to the economic impact of retinitis pigmentosa on the lives of the affected and their families [7,8,9].

A mode of inheritance may also have accounted for the severity of the disease in these patients [3]. However, no positive family history was elicited in this study as opposed to other studies [14,33]. For the fact that many of the diseased patients in this study fell into the visual acuity of economic and social blindness [38], it then becomes necessary to institute the policy of economic and social reorientation and rehabilitation of all the retinitis pigmentosa patients no matter the level of visual acuity at the time of first presentation. Generally, diminished vision (93.1%) was the major symptom that made patients to seek medical help. This finding was similar to the reports of 90% Ukponmwan et al. [32] and Eballe et al. [11], 85% of them were working in Cameroon and Benin city respectively [11,32]. However, the report by Onakpoya et al. [33] in a multicentre study in southwestern Nigeria reported 69.8% which was much lower than the 93.1% recorded in the present survey. Edema et al. [6] and Abah et al. [39] had earlier reported that retinitis Pigmentosa is a common diagnosis among children in schools for the visually impaired in Nigeria. This confirms the fact that retinitis pigmentosa and its co-morbidities are some of the reasons people attend schools for the physically challenged. Poor night vision (79.3%) was the next disturbing symptom in this review and had literally forced those affected to abandon night or dim light activities. Onakpoya et al. [33] and Ukponmwan et al. [32] had reported night blindness (58.3%) and 56.7% respectively which is lower than that of the present study. The differences could have arisen for the fact that Onakpoya's study was a multicentre study with higher participants. Harton et al. [2] had earlier reported that night blindness is usually the initial symptom followed by loss of visual field and visual acuity as the disease progresses. Other complaints noted in this study include headache (31.0%) eye ache (27.6%) and photophobia (20.6%). Onakpoya [33] reported eye ache (10.4%). These headache, eye ache and photophobia could arise as symptoms of refractive error which is a co-morbidity of retinitis pigmentosa. The prevalence of refractive error (27.6%) myopia (20.7%) and hypermetropia (6.9%) were the findings in this review among the retinitis pigmentosa patients.

Onakpoya [33] had reported prevalence of refractive error (37.5%) which was higher than that of the present survey and both studies (present study and Onakpoya's) found myopia to be the most common refractive error (20.7%) and (71.4%) respectively. Myopia is the most common refractive error seen in retinitis pigmentosa patients. In myopia, pupils are somewhat large and a bit sluggishly reacting and may result in photophobia [40]. It had been documented by LU and SU [41] that patients with retinitis pigmentosa may be misdiagnosed and treated simply as myopic cases. Hypermetropia (6.9%) was noted in this study among the retinitis pigmentosa patients – while Onakpoya et al33 in their multicentre study reported 11.1%. Other co-morbidities noted in this study include Glaucoma, uniocular Aphakia (which was due to coaching). Another author [33] had documented prevalence of glaucoma (11%) which was lower than that of the present study (17.2%), but reported lens opacity/pseudophakia (10.4%) among retinitis pigmentosa patients which was similar to that of the present study (10.5%). Other authors [11,32,42,43], had documented association of

these disorders with retinitis pigmentosa patients. Nevertheless, the glaucoma rate in this study is higher than the 7.5% noted in Cameroon [11] and 2.3% in people's Republic of China [43]. The fundus findings of the patients in this study are retinal pigmentation (100%) which is the commonest and this has been collaborated by other authors [3,33].

The typical perivascular pigmentation seems to make the diagnosis of retinitis pigmentosa unequivocal. Other fundus findings were retinal vascular attenuation, disc pallor and is similar to that of other authors [3,33]. Disc cupping, though one of the fundus findings in this study is only secondary to glaucoma which is a co-morbidity of retinitis pigmentosa. No positive family history of retinitis pigmentosa was elicited in this study and this may be attributed to poor family health record among the relatives or the disease has been misdiagnosed in the past [41]. It may also mean that the disease has skipped generations. However, retinitis pigmentosa can also cause psychological effects [44].

5. CONCLUSION

Retinitis pigmentosa is a bilateral degenerative, progressive and inheritable disease of the retina that affects people in their active and productive stage of life. It affects the quality of life and livelihood of the affected with severe economic and social denigration. Early diagnosis, occupational, career and economic re-orientation and rehabilitation should be encouraged. Genetic counseling should also be a high point of health education of the affected.

CONCENT

It is not applicable.

ETHICAL APPROVAL

Ethical approval was sought and granted by the Ethical committee of Chukwuemeka Odumegwu Ojukwu University Teaching Hospital.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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