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Prevalence of Retinitis Pigmentosa in Kano State, Nigeria

[1]Okpo, E., [2]Nura, Y., S., [3]Nwakuche, P.I. & [4]Muoneke, H.,C.
[1] Department of Optometry, Bayero University, Kano/
Department of Ophthalmology, Aminu Kano Teaching Hospital, Kano.
[2]Department of Optometry, Bayero University, Kano.
Corresponding Author: *[1]Okpo, E.*
Corresponding Email: *doctorokpo@yahoo.co.uk*

Abstract
Background: Retinitis pigmentosa (RP) is a slow degenerative disease of the retina which affects the photoreceptor layer, predominantly rods. **Aim**: A retrospective cross-sectional study aimed to determine the prevalence of RP in Kano State. **Method**: One thousand, five hundred folders were reviewed from 2010-2020 in the Retina and Low Vision Clinics of Makkah Eye Hospital (MSEH) and Aminu Kano Teaching Hospital Kano (AKTH). Clinical presentation, features, history of consanguinity, and degree of visual impairment were studied. Data were analyzed using SPSS's latest version 25.0. **Results:** Sixty patients were diagnosed with RP, representing a prevalence of 4%. The ratio of male to female is 1:1. The age range was 3 to 70 years (mean 31.17 years +/- 17.69 years). Twenty-nine patients had visual acuity of less than 3/60, indicating that half of the patients are blind. The commonest mode of presentation was night blindness in 58.3% of the patients. Bone spicule pigmentation is the most common sign in this study (61.7%), while those with macula edema (18.3%) and normal fundus present with the lowest frequency of 11.7%. The rate of RP correlated with consanguineous history in this study shows a positive correlation(r=0.022). **Conclusion:** The degree of visual loss in Kano state citizens with RP is severe and may be related to inadequate knowledge regarding the condition and late presentation of the patient to the hospital. There is a need for health education of the public and patients about the need for early presentation to the hospital.

Keywords: *Retinitis pigmentosa, Night blindness, consanguineous.*

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Introduction

In 2015, there were an estimated 253 million people with visual impairment worldwide. Of these, 36 million were blind and a further 217 million had moderate to severe visual impairment (MSVI). The prevalence of people that have distance visual impairment is 3.44%, of whom 0.49% are blind and 2.95% have MSVI. (Peter et al., 2017). Although RP is a rare genetic disorder affecting the retina it can lead to legal blindness due to the fact that RP causes tunnel vision in which only the central vision is unaffected while the peripheral vision is lost such patient might be able to read 6/6 VA but are blind because of the constricted visual field. According to some researchers, the prevalence of RP varies from approximately 1:9000 (Na et al., 2017), to as high as 1:750 (Jonas et al., 2012) depending on the geographic location. It is the most frequent cause of blindness during working life in industrialized nations (Malik et al., 2014).

The term RP was first applied in the 19th century when typical pigmentary bone corpuscles were seen in the retina of an affected patient. However, bone corpuscular pigment is a relatively late occurrence and functional retinal abnormalities occur much earlier than these bone corpuscles (Merin, 2005). RP can be defined as a large group of inherited vision disorders characterized by progressive degeneration of the retina, peripheral vision gradually decreases and finally leads to loss of peripheral vision usually tubular vision is one of its main symptoms (Malik ., 2014). It is an inherited

disease that runs in families need utmost care to reduce the burden on the prevalence of blindness as it is one of the non-treatable cause of blindness.
In the late stages of RP, as cones die, people tend to lose more of the visual field, developing tunnel vision. They may have difficulty performing essential tasks of daily living such as reading, driving, walking without assistance, or recognizing faces and objects. Some people retain central vision and a restricted visual field into their 50s, while others experience significant vision loss in early adulthood. Eventually, most individuals with RP will lose most of their sight (Fahimeh ., 2021, Malik ., 2014). Generally, RP can be transmitted as an Autosomal Dominant (AD), Autosomal recessive (AR), and X-linked RP. It can occur in simplex or multiplex form, when a genetic background may not be obvious and defined. Studies have shown that among the three forms of RP, ADRP typically presents with the mildest form of the disease while XLRP presents with the most severe (Hamel, 2006). Despite a large number of RP genes, mutations cannot be identified in 30 to 35% of patients with ADRP (Bowne, et al., 2011). One possible cause of disease in these remaining ADRP patients is that some ‘‘ADRP’’ families without mutations in known ADRP genes may, in fact, have XLRP. Although XLRP is thought to affect male subjects only, many documented cases of RPGR and RP2 mutations cause disease in carrier female subjects, thus giving an impression of occurrence in sequential generations simulating Mendelian dominant transmission (Pomares et al., 2009). With recently developed technology, for example, next-generation sequencing (NGS), 30–80% of mutations in patients can now be identified (Daiger, 2013).

However, families with dominant inheritance patterns and female carriers showing disease symptoms of variable degree (likely due to the dominant nature of some of the mutations or non-random X-inactivation in the affected tissue) have also been described (Berger et al., 2010).

More recently, it has been reported that carrier female subjects exhibit a range of phenotypes that can vary from asymptomatic to a severe retinal disease similar to male subjects (Branham, et al,. 2012). Myopia has been noted to be a feature of XLRP and was a characteristic of the majority of affected family members (Al-Maskari et al., 2009).

No treatment medical or surgical has proved effective for the treatment of diseased retina in RP. However, despite the general lack of effective therapy patient with this condition requires an adequate general medical exam to identify associated RP which has a different treatment, genetic counseling, and evaluation of the possibility of using optical means such as low vision aids (Merin, 2005). Advice patient about general behavior such as limiting night driving, use of sunglasses in bright light, and improving illumination at night. Other surgical therapies include RPE transplant, Photoreceptor transplant, Gene therapy, and Retinal prosthesis which can be implanted either in front (epiretinal) or subretinally. However, almost all these surgical therapies produce little to no improvement in visual functions of significance (Merin, 2005).

RP as a form of retinal inherited dystrophy has consanguineous marriage as one of the key factors because the disease runs in families so family members that undergo such marriage are at risk of having children with the disorder. And consanguineous marriage is common in the northern part of the country a study should be conducted to access the prevalence and shade more knowledge regarding the prevalence of RP in Kano state. In northwestern Nigeria, there is no data on the topic area. Information on RP is needed for the design of health policies in the country.

Materials and Methods

This is a retrospective Cross Sectional Study carried out in Aminu Kano Teaching Hospital (AKTH) and Makkah Specialist Eye Hospital (MSEH). Geographically**,** Kano is located in the northern part of Nigeria**.** Kano State borders Katsina State to the northwest and Jigawa State to the northeast. Kano State lies between latitude 11030'N 50 and longitude80 30'E. The total land area of Kano State is 20,760 km2 with a population of 9,383,682 according to the 2006 National Population and Housing Census (Kano Municipal LGA., 2007). A total of one thousand, five hundred (1500) clinical case folders of patients were reviewed from January 2010- January 2020 seen in the retinal and low vision clinics of the two hospitals. Sixty patients diagnosed with RP were included in the research. The data collection sheet comprises ten columns which include Age, Sex, Presenting complaint, Family history, marital status, Funduscopic (Ophthalmoscopic/Biomicroscopic) findings, Retinal changes, Visual acuity, Refractive error (retinoscopic and subjective Refractions) and treatment options.

Patients whose folders had accurate data and diagnosed with RP, from 2010-2020 with complaints of night blindness found were included. All patients that present with RP, not from 2010-2020 were excluded. Ethical clearance was obtained from the Kano state Ministry of Health with reference number: MOH/OH/797/T.I/2155. Results were presented using tables and charts. Data collected were analyzed using frequencies in software package for social sciences (SPSS) version 25.0 to describe the prevalence of RP in Kano State.

Results

A total of 1500 patients' record folders who attended the Retinal and Low Vision Clinics of Ophthalmology Department of AKTH and MSEH were reviewed. Only 60 patients, presented with RP. A prevalence of 4% of the patients seen in the two clinics over the period under review was established. The age range of the participants ranges from 3-70 years with a mean age of 3.117+/-17.69 years.

Table 1: *Distribution of study participants by gender and age.*

Age (years)	**Male (%)**	**Female (%)**	**N (%)**
3 - 13	8 (13.3)	4 (6.7)	12 (20.0)
14 - 24	3 (5.0)	6 (10.0)	9 (15.0)
25 - 35	9 (15.0)	6 (10.0)	15 (25.0)
36 - 46	4 (6.7)	7 (11.7)	11 (18.3)
47 - 57	3 (5.0)	6 (10.0)	9 (15.0)
58 - 68	1(1.7)	0 (0)	1 (1.7)
>68	2 (3.3)	1(1.7)	3 (5.0)
N	30 (50)	30 (50)	(100.0)

The disease affects females 30 (50.0%) and males 30 (50.0%) equally with a ratio of 1:1. The result indicated that most RP patients presented with the disease at the mid stage of life, with Participants between the 25-35 years (25.0%) having the highest frequency.

Table 2: *Distribution of study participants by presenting complaints*

Presenting complain	**Frequency**	**Percentage%**
Nyctalopia	35	58.3
Poor Vision	21	35.0
Blurry Vision	4	6.7
N	60	100.0

The most presenting complaint or earliest symptom in this study was nyctalopia (58.3%).

Table 3: *Distribution of study participants by consanguineous*

history	Frequency	Percentage %
Consanguineous history	12	20.0
No family history	47	78.3
N	60	100.0

History of consangeinuty was only found amoung 12 (20.0%) of the participants.

Table 4: *Distribution of study participants by funduscopic findings*

Funduscopic findings	Frequency	Percentage%
Bone spicule pigmentation	37	61.7
Normal fundus	7	11.7
Macula edema	8	13.3
Wavy and pale disc	8	13.3
N	60	100.0

Funduscopic findings in these patients are mostly bone spicule pigmentation with the highest frequency of 37(61.7%) and normal fundus with the lowest frequency of 7 (11.7%).

Table 5: *Distribution of study participants by visual acuity*

Visual acuity	Frequency		Percentage (%)	
	OD	OS	OD	OS
≥6/18	15	16	25.9	27.6
<6/18-6/60	9	10	15.5	17.2
<6/60-3/60	5	3	8.6	5.2
<3/60	29	29	50.0	50.0
N	58	58	100.0	100.0

Key: visual acuity measurement according to WHO standard: Normal Vision (VA>/=6/18, Moderate visual impairment (VA<6/18-6/60), Severe visual impairment (VA<6/60-3/60), Blindness (VA<3/60) There was higher right and left eye visual acuity frequency in participants with VA of <3/60 29 (50.0%) each. The findings in both eyes indicate that the visual acuity of both eyes deteriorates almost equally.

Table 6: *Distribution of study participants by refractive errors*

Refractive error	Frequency	Percentage%
Myopia	4	11.8
Hyperopia	6	17.6
Simple Astigmatism	4	11.8
Hyperopic Astigmatism	6	17.6
Myopic Astigmatism	14	41.2
N	34	100.0

Myopic astigmatism 14 (41.2%), has the highest frequency while myopia and simple astigmatism have the same lowest frequency of 4(11.8%).

Table 7: *Distribution of study participants by treatment modalities*

Treatment options	Frequency	Percentage
Counseling	14	23.0
Medication	31	50.8
low vision aids	12	19.7
spectacle correction	4	6.6
N	61	100.0

Treatment of these patients is mostly by prescription of medication such as antioxidants which slows the progression of the disease, with the highest frequency of 30(50.8%), this is followed by counseling of the patient with a frequency of 14(23.0%).

Discussion

This study is a hospital-based prevalence study that focused on the patient that presents to hospitals with RP, the prevalence rate was found to be 4%, this rate would clearly not represent RP in the Kano state population because some patients did not report to the hospital due to socioeconomic factors. In this study, there is equal distribution of the disease across gender, with 50% males and 50% females in a ratio of 1:1, this finding however indicates that the disease indiscriminately affects males and females, which is in contrast with the study by (Eballe .,2010; Ukponmwan .,2004) while studies by (Jitendra *et* al.,2018; Babu .,2017; Prokofyeva ., 2009; Iyamu and Ahmed, 2004; Ukponmwan & Atamah, 2004) disagree with indiscrimination in their studies. The majority of patients in this study (58.3%) present with nyctalopia as the most common presentation, this could be due to early loss of photoreceptors predominantly rods which are responsible for vision in dim illumination and night vision, which is in sharp contrast with the study conducted by Malik et al., (2014). The majority of patients were between the age of 25-35 years indicating that the disease mainly affects the working age group, this is in sharp contrast with study conducted by (Ukponmwan & Atamah, 2004).

The mean age of patient presentation was (31.1167 years +/-17.6 years) this mean finding almost corresponds with a mean age and standard deviation of (Ukponmwan & Atamah, 2004), while researchers like Eballe .,(2010) present with almost the same standard deviation value as this study but with a much higher mean age value, studies by (Thapa .,2018, Ajayi ., 2018, , Onakpoya .,2016, Prokofyeva .,2009) present with much higher mean age and standard deviation value, this variation might be due to age criteria of selecting patient by various researchers. Visual acuity was categorized according to WHO visual acuity classification, half of the patients (50%) presented with VA of <3/60 in both eyes, this is sad as it indicates the majority of the patient were blind at presentation. The almost equal deterioration in VA indicates that the disease occurs bilaterally and almost all reductions in VA occur simultaneously, however, the disease can occur rarely as a unilateral disorder according to a study by Goodwin, ., (2019). The VA percentage corresponds with findings by Ukponmwan & Atamah, (2004). Grover .,(2000) reported a much lower blindness rate of 25% in the US, while a 30% blindness rate was reported in Cameroon.

Family history of consanguinity was documented in 12(20%) of patients in this study while 47(78%) of the patient presented with no family history of the disease or history of consanguinity, the low frequency could be due to poor case history taken by the doctors or the patient being non-complaint or not giving full details as asked by the doctor. RP correlated with consanguineous history in this study shows a positive correlation(r=0.022) indicating they have a direct relationship such that when there is an increase in consanguineous marriage there is an increase in RP meaning that consanguineous marriage predisposes individuals to RP. There is the high rate of consanguineous marriage in this study area which explains why there is positive correlation between RP and Consanguinity.

Bone spicule pigmentation was the earliest sign seen on fundus examination with ophthalmoscope and 37(61.7%) presented with this fundal finding, 8(13.3%) presented with macula edema indicating macular involvement, and the same percentage presented with pale and wavy disc 8(13.3%), only a small number of patient 7(11.7%) present with a normal fundus this finding is in sharp contrast with studies by (Jitendra ,2018,

Ukponmwan & Atamah, 2004) however, since further evaluation to determine the various genetic subtypes of RP were not documented in this study due absence of facilities for routine genetic studies in the state, this findings gave me an insight into thinking that the patient with normal fundus can have Retinitis pigmentosa sine pigmento or the RP might be in its earliest stage and it was diagnosed before fundus background changes.

Treatment of patient in this study is mainly through the prescription of medications (50.8%) daily intake of oral vitamin A helps in retarding the progression of the disease and reduce deterioration of vision while carbonic anhydrase inhibitors improve the visual function in patients with macula oedema (Grover ., 2000) regular use of antioxidants such as ascorbic acid and beta-carotene, are believed to be helpful in slowing the progression of the disease (Ukponmwan & Atamah, 2004).

Conclusion

In conclusion, RP was found to be among the causes of visual impairment and blindness in Kano affecting individuals mostly during mid-decades of life, it affects males and females equally and the majority of patients present with blindness and visual impairment. The most common complaint at presentation is nyctalopia, followed by poor vision, most patients present with no family history of the disease and this could be due to a lack of facilities for genetic testing.

It is sad as the most patients are blind at presentation and due to lack of facilities most patients were given medications to help retard the progression of the disease, some were counseled about their conditions while a few of the patients were given spectacle correction and low vision aids to aid in vision and magnify the patients' vision.

References

Ajayi, I., Omotoye, O.J., Ajite, K., & Omotoso O.O. (2018) Prevalence and Causes of Visual Impairment in Ekiti, Nigeria*: A Hospital Based Study. J Ophthalmology. 3(4): 000158*.

Al-Maskari A, O0Grady A, Pal B, McKibbin M. (2009) Phenotypic progression in X-linkedretinitis pigmentosa secondary to a novel mutation in the RPGR gene. *Eye* 23(3): 519 -521.

Al Merjan, J.I., Pandova, M.G., &Al Ghanim, M. (2005) Registered blindness and low vision in Kuwait. *Ophthalmic Epidemiology.*12:251–257.69

Babu, R. B., & Manjula, P. (2017) Nehakamalini Clinical Study of Prevalence of Retinitis Pigmentosa in Tertiary Care Hospital. *International journal of contemporary medical research. 4(9)*

Berger, W., Kloeckener-Gruissem, B., Neidhardt, J. (**2010**)The molecular basis of human retinal and vitreoretinal diseases. *Prog. Retin. Eye Res.*, *29*(5), 335-375.

Bowne, S.J., Sullivan, L.S., Koboldt, D.C. (2011) Identification of disease-causing mutations in autosomal dominant retinitis pigmentosa (adRP) using next-generation DNA sequencing. Invest Ophthalmol Vis Sci. 52:494–503.

Branham, K., Othman, M., Brumm, M. (2012) Mutations in RPGR and RP2 account for 15% of male subjects with simplex retinal degenerative disease. Invest Ophthalmol Vis Sci. 53: 8232–8237.

Daiger, S. P., Sullivan, L. S., & Bowne, S. J. (2013) Genes and mutations causing retinitispigmentosa. *Clin Genet.*, 84(2), 132-141.

Eballe, A. O., Koki, G., Emche, B. C., Bella, L. A., Kaum, J. M., Melong, J. (2010) Blindness and visual impairment in retinitis pigmentosa: a Cameroonian hospital-based study. Clinical Opthalmology 4:661-665

Fahimeh, B., Mohammad, Y.M., Masoud, R.M., &Hamid R.A. (2020) Study of Patterns of Inheritance in Affected Patients with Retinitis Pigmentosa in Iranian Populations,International

Journal of Medical Laboratory(7(1):9-14)

Goodwin D., Olsen, A.M., Citek, k. (2019) Unilateral retinitis pigmentosa: Case report and review of the literature. *Int J Clin Exp Ophthalmol* 3(2), 013-019.

Grover, S.,Fishman, G.A., Anderson., R.J., & Linderman, M.A (2000) longitudinal study of visual functions in carriers of x-linked recessive retinitis pigmentos *Ophthalmology.(Vol.107, pp.386-396)*

Hamel, C. Retinitis pigmentosa. (2006) *Orphanet J Rare Dis* 1, 40, https://doi.org/10.1186/1750-11721-40.

Iyamu, E., & Ahmed, M.T. (2004) the common causes of blindness in northwestern Nigeria. *Jnoa - vol 11.*

Jitendra K., ArtiKushwaha, N., & Kanhaiya, P. (2018).IOSR *Journal of Dental and Medical Sciences(IOSR-JDMS). (Vol. 17, Issue 3 Ver. 1, pp 01-031)*

Jonas J.B., Nangia V., Kulkarni, Gupta R., Khare A. (2012) Associations of early age-related macular degeneration with ocular and general parameters. Central India eyes and medical study. *Acta ophthalmologica*. 9 (3), e 185-e191.

Kano Municipal L.G.A. Population as per: Federal Republic of Nigeria Official Gazette (15 May 2007). "Legal Notice on Population of the details of the breakdown of the National and State Provisional Total 2006 census" Retrieved 2007 – 15 – 19.

Malik, T., Khalil, M.,Munawar, S., Bhatti, M., Muhammad, M.S,(2014).Clinical patterns of retinitis pigmentosa with special reference to maculopathy .*Pakistan Journal of Medical and Health Sciences*,*8(5), 3-5.*

Merin, S. (2005) Inherited Eye Diseases: Diagnosis and Management, Boca Raton: CRC Press. https://doi.org/10.1201/b14269

Na K.H., Kim H.J., Kim K.H., Han S., Kim P., Hann H.J. (2017) Prevalence, age at diagnosis, mortality, and cause of death in Retinitis pigmentosain Korea-a nationwide population-based study. *American journal of ophthalmology.* 176, 157-165.

Onakpoya, O.H., Olateju, S.O., &Ajayi, I.A. (2008) Retina diseases in a tertiary hospital: the need for establishment of a vitreoretinal care unit. *Journal National Medical Association. (100:1286–1289).*

Peter A., Serge R. & Rupert B. (2017) World blindness and visual impairment: despite many successes, the problem is growing. *Community Eye Health Journal.* 30(100):71-73

Pomares E, Riera M, Castro-Navarro J, Andres-Gutierrez A, Gonzalez-Duarte R, Marfany G. (2009) Identification of an intronicsingle-point mutation in RP2 as the cause of semidominant Xlinked retinitis pigmentosa. *Invest Ophthalmol Vis Sci.* 50:5107–5114.

Prokofyeva, E., Wilke, R., Wilker, L.G., Troeger, E., Strasser, T., & Zrenner, E. (2009).An epidemiological approach for estimation of disease onset in central and peripheral peripheral 76 monogenic retinal dystrophy.*archive for clinical and experimental ophthalmology.247 (7):885-94*.

Ukponmwan, C.U., & Atamah, A. (2004) retinitis pigmentosa in Benin, Nigeria. *West African Medical Journal* 81(5): 77-81