Influence of Family History Classifications in Cataract Occurrence Rate Among Local Residence of Imo State

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Abstract- ***Cataract is one of leading causes of visual impairments around the globe which is associated with genetic factors. The present study was designed as a descriptive cross-sectional study aimed at assessing the influence of family history classifications on cataract occurrence rate among adult residents in Imo State Nigeria. A multi-stage sampling method was used to sample 792 study participants across the state of study. The presence of cataract was established using Lens Opacity Classification System (LOCS) III system. A slit lamp examination was performed to assess the severity of specific lens opacity. The participants were also assessed of family history of cataract using a study designed self-reported forms. Family history was classified into direct or first-degree relatives and indirect or second-degree relatives. The characteristics of the participants was described using statistical descriptive analysis technique such as frequency distribution table. Chi-square test was used to test for association between family history and occurrence of cataract. Further analysis includes logistic regression method used to compare the significant effects of factors of family history relatives. The results indicates that 200 (25.3%) reported having family history relatives with cataract. Significant association was found between cataract and family history (P=0.0001, $\chi 2=73.42$), with 98% lower odds found among the non-family history reported group (OR =0.022, 95% CI =0.0025 – 0.0883). Interns of classifications of family history relatives, cataract was found highest among the direct or first-degree family relatives. It was also found to be most occurring if the family relative reported was a sister or a brother with up to four time more significant odds compared to non-direct relatives (OR= 4.11, 95% CI =1.040 – 16.331). In conclusion, family history is significantly associated with cataract and contained greater risk among direct relatives such as brothers/ sister relatives. Detailed family history of cataracts classifications should adequately studied to identify the classes of familial cases at greater risk so as to minimise cost and enable the benefit from merits attached to early detection and treatments for the disease.***

Indexed Terms- ***Cataract, Visual Impairment, Family History, Direct Relatives***

I. INTRODUCTION

Cataract is one of the most common causes of visual impairment in the world. According to the World Health Organisation (WHO), cataract is among the leading causes of blindness all over the world, with high prevalence in low- and middle-income countries (World Health Organization, 2023). It is a leading cause of untreated blindness and is responsible for up to 47% of global blindnes [1]. A current systematic review and meta-analysis study on estimated global vision loss burden due to cataract indicated that an increase of 29.7% in cases of cataract blindness has been recorded globally, of which cataract accounts for 39.6% global blindness between 2000 to 2020 with 28.3% among them having moderate to severe vision impairment (MSVI) related blindness [2].

Cataract is defined as partial or complete opacification of the crystalline lens. The crystalline lens has three layers, namely nucleus, cortex and capsule. It occurs to all levels of humans but occurs more among the over forty adults [3]. Part of the consequences of having the disease when not treated early include increased risk of secondary glaucoma high life dependency on others, and economic loss [4, 5]. Other include uveitis and decreased quality of life [6].

The occurrence of cataract has been linked with many sources including association between with genetic factors such as family history [7, 8]. It has been reported that up to 50% of cataract cases are rooted to genetic factors [9, 10]. Also up to 22.3% of childhood cataracts has been associated to genetic or family history of the disease [11]. Family history itself is enlarged circle that required further classification to establish the family relative that are greater risk of cataract. The present study was aimed at investigating the relationship in cataract occurrence at difference classifications of family history. This essence is to encourage the understanding of genetic supports and counseling needed in families for early diagnosis and treatment of cataract so as to facilitate timely intervention and reduction in loss of vision.

II. MATERIALS AND METHODS

This study was designed as a household based cross-sectional study involving different household communities in Imo State Nigeria. The study area and the socio-demographic characteristics of the study group has already been described [12].

Administratively, Imo sate is divided into three geopolitical zones and one Local Government Area (LGA) was randomly selected from each geopolitical zone. The selected LCAs were Nkwere, Okigwe and Aboh Mbaise which represent Imo West (Orlu Zone), Imo North (Okigwe zone) and Imo East (Owerri zone). The study sample size was shared to the selected LGAs in proportion to the number of households in the each LGA.

- Sampling Technique

The multistage sampling technique was used to select the subjects that were studied. The LGAs, wards, villages and households were randomly selected at each stage in the three senatorial zones of Imo State. The sampling involved three stages:

Stage 1: The study LGAs were selected from each zone randomly one LGA was selected from each zone

Stage II: Within the selected LGAs, two wards were randomly selected from each LGAs using computer random number selection technique.

Stage III: Community Selection. One community was selected from each od the identified ward using random sampling technique.

Stage IV: Household/Participants selection. Also, the number of the required households were randomly sampled using numbers assigned to the households in the study communities. The number of cases of Cataract were assesses at the selected households and the household heads or their representatives were interviewed.

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Sample Size

Considering that a previous study in Nigeria, reported a prevalence of 3.9% for cataract [13], Cochrane technique was applied to estimate the sample size of 396 with 10% attrition (Cochrane, 1977). This estimate was doubled (n=792) to cover wider number of people in the study area

Method of Data Collection

Use of Questionnaire

A pre-tested, self-administered, structured questionnaire instrument was developed and used to collect the study data on family history and socio-economic characteristics of the subjects. Family his was described as having any relative that has been diagnosed of cataract. In addition, the family history was classified into fist degree relatives (direct relatives such as parents, brothers and sisters of same parents) and second-degree relatives defined as indirect or extended relatives such as uncles, aunty, nice, nephew, cousins and others. The questionnaire was constructed with English Language but was translated some of the respondents who cannot read or understand English Language. Socio demographic information obtained were age (in years), sex (male/ female). Others include occupation, education and monthly household income in Naira (#).

Slit Lamp Biomicroscopy

Slit lamp biomicroscopy was performed by experienced optometrists to identify abnormalities of the anterior segment. Under topical anesthesia using 0.5% proparacaine, intraocular pressure (IOP) was measured with a handheld Perkins applanation tonometer (HA-2, Kowa Xinghe, Japan) using 0.5% proparacaine and fluorescein staining of the tear film. The right eye was measured first and one reliable measurement was recorded for each eye. The instrument was

Cataract Measurement

The grading of lens opacity was performed by trained optometrists, according to a written standardized protocol, using the Lens Opacity Classification System (LOCS) III system [14]. After dilatation of pupils with tropicamide 1% and phenylephrine hydrochloride 2.5% eye drops (repeated twice if necessary), the participant was examined at a slit lamp (Model BQ 900, Haag-Streit, Bern, Switzerland), and the presence and severity of specific lens opacity were compared and documented according to LOCS III standard photographs.

Method of Data Analysis

Data analysis was performed using IBM-SPSS statistics version 27 (SPSS Inc. Chicago, Illinois USA). Data analysis involved using descriptive and inferential analysis. Descriptive statistics was used to summarize and describe the data through frequency distribution and distributional charts Inferential test performed include chi square test and logistic regression analysis. All tests were performed at 5% significant level. Probability value ($p < 0.05$) was used to explain significance and the odds ratio (OR) was used to explain effect size measures.

Ethical Consideration

Ethical approval was gotten from Imo State University Ethical committee of School of Health Sciences. Oral informed consent was obtained from all participants and participation was made voluntarily.

III. RESULTS

Presence of Family History of cataract among the Study Group

There were in all 792 persons that participated in the study, of which 200 (25.3%) have family history of cataract disease, while the remaining 592 (74.4%) reported not having family history of the disease (Figure 1).

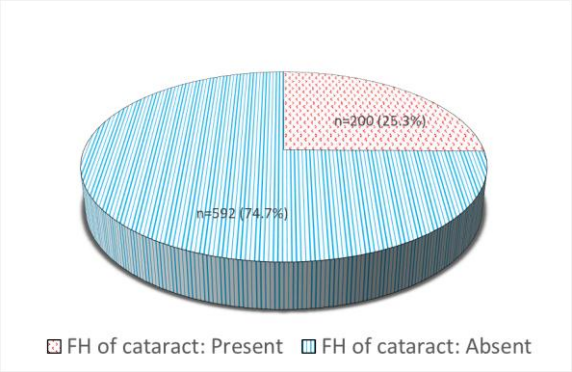

Figure 1: Pie chart showing distribution for Presence and absence of family history among the study group

Sociodemographic Distribution according to family history of Cataract

The Sociodemographic distribution of the participants according to family history status of cataracts is presented in table 1. The table shows that more than half of the study group were between 40 -50 years of age, of which 48% of the age group reported having family history of cataract. The distribution for gender showed close even distribution with family history of cataract reported in 105 (52.5%) females (males: 47.5%). Family history was reported higher among manual workers (23%), farmers (22.5%) and traders (20%), and was also reported high among the less educated (30% in no formal education and 22% in each of the primary and secondary levels of education). Similarly, it was high among the low household income earning groups.

Table 1: Sociodemographic Distribution of the participants according to family history of Cataract

	Presence of Family History of Cataract			
Frequency	Yes (n=200)	(%)	None (n=592)	(%)
Age				
Less than 40	19	9.5	50	8.4
40 – 50	96	48	321	54.2
51 – 60	45	22.5	130	21.9
61-65	40	20.0	91	15.4
Gender				
Male	95	47.5	270	45.6
Female	105	52.5	322	54.4
Occupation				

Farmer	45	22.5	81	13.7
Manual worker (Artisans)	46	23	108	18.2
Civil/ Public servants	31	15.5	130	22.0
House worker	22	11	59	10.0
Others	16	8	39	6.6
Trading/Business	40	20	175	29.6
Education Level				
No formal education	60	30	87	14.7
Primary	45	22.5	111	18.8
Secondary	45	22.5	189	31.9
High school	31	15.5	163	27.5
University and higher institutions	19	9.5	42	7.1
Household Monthly Income in Naira (₦)				
Less than or equal to 30,000	57	28.5	99	16.7
31,000 - 50,000	58	29	188	31.8
51,000-70,000	45	22.5	162	27.4
71,000 - 90,000	35	17.5	108	18.2
91,000 and above	5	2.5	35	5.9

Relationship between family history of cataract and the occurrence in the study Area

The relationship between family history of cataract and the occurrence of the disease is presented in table 2 The table shows that having family history showed significant association with cataract (P=0.0001, χ^2=73.42). Cataract was found present among 13.5% of the group who reported having family history of the disease compared to 0.3% found among non-family history reported group, with 98% lower odds in the no family history (OR =0.022, 95% CI =0.0025 – 0.0883).

However, many of the reported family history relatives that had cataract were of direct or first-degree category of which the largest was either mothers or fathers (44.5%), while sister or brother make up 14.5% of the family history reported.

Among the different categories of family history of cataract, cataract was found to be most occurring if the family relative reported as a sister or a brother. There were up to four time more significant odds of having cataract disease among direct sisters or direct brothers compared to non-direct relatives such as uncles, aunts, nephews and others (OR= 4.11, 95% CI =1.040 – 16.331).

The risk for cataract occurrence was also found to be higher in mother or father compared to second degree relatives (OR= 2.24, 95% CI =0.756 – 7.577), but no evidence of significant difference was established between the two groups (P=0.1094).

Table 2: Family History (FH) in relation to the Prevalence of Cataract in the study area

Family History	Total	Cataract: Absent	Cataract: Present	P	OR	95% CI	
	n (%)	Freq (%)	Freq (%)			Lower	Upper
Presence of FH							
Yes	200 (25.3)	173 (86.5)	27 (13.5)	REFERENCE GROUP			
None	592 (74.7)	590 (99.7)	2 (0.3)	<0.0001	0.022	0.0025	0.0883
FH class							
2nd Degree relatives (Extended relatives)	80 (40.0)	74 (92.5)	6 (7.5)	REFERENCE GROUP			
1st Degree relatives (Mother/ Father)	91 (45.5)	77 (84.6)	14 (15.4)	0.1094	2.24	0.756	7.577
1st Degree relatives (Sister/ Brother)	29 (14.5)	22 (75.9)	7 (24.1)	0.0143	4.11	1.040	16.331

FH= Family history of cataract OR= Odds ratio, 95% CI = 95% confidence interval, P= probability value (significant association found at 5%).

IV. DISCUSSION

The present study was focused of assessing the classifications of family history to establish those who are at greater risk for cataract. The study findings indicated that while up to one quarter of the study group reported having family history of cataract, majority among them were of first-degree relatives. This finding is high considering that greater risk of cataract associating with family history of the disease in the study area. Possibly reason for this result among the study group could be due to poor eye check for cataract as well as poor knowledge associating risk from family history of the disease in the study area. Poor knowledge of cataract has been reported even among those with family history of the disease in Nigeria [15].

Our findings indicates that cataract was found to be higher among those that reported having family history of the disease compared to the none family history group. Lower odds of cataract were found among individuals with no family history of the disease compared to the familial cataract group. Genetic factors are obvious associating factors of cataract, other factors relating to demographics and environment could as well hasten the occurrence of the disease among the hereditary populations [7, 8]. For instance, socio demographics relating to age, gender education level and occupation that are all established significant risk factors of cataract could have influence in the rate of occurrence of cataract among different populations including populations with family history of the disease [12].

Also the first degree relatives were found with higher associating family history risk of cataract than the second degree relatives in this study. Also the significant role of first-degree relatives in the occurrence of cataract disease stood out in some other studies [16, 13]. First-degree relatives have also been reported as an associating risk factor of congenital cataract in a longitudinal study [17].

This is not a surprise finding since the genetic of human cataract has acknowledged that cataract inheritance is likely to follow Mendelian hereditary patterns which recognizes that cataracts can result from mutations at different genetic loci but may have different inheritance patterns [18]. For the present study, family history is a risk factor of cataract but the risk for the disease was more from direct or first-degree relatives.

Further, our findings indicates that having a sister or a brother with cataract signifies higher risk of occurrence of cataract compared to the risk of the disease compared to having an extended family relative (second degree relatives) such as nephew, niece, aunt and uncle with the disease. On the other hand, though the odds ratio for the occurrence of cataract was higher among those who reported having one or both of their parents with cataract compared to those who reported that having extended relatives as relatives with the disease, the occurrence of cataract did not differ significantly in both groups. This appeared as a surprise finding yet it is possible since many parents also have some of the extended family members as their own direct relatives such as brothers abd sisters. The implications of this finding are that direct relatives to individuals with cataracts especially brothers and sisters shared more genetic risk of cataract in this study therefore needed to be closely monitored early once either of the two is diagnosed if cataract.

The present study is limited to the fact that due to cost implications gene testing of the participants was mot performed to establish the extent of their family relations. We relied on self-reported information to assess the family genetics of cataract among the study group. The use of self-reported information could be subjective to reporting bias. However, the study tool was adequately validated to reduce the extent of reported errors.

In conclusion, family history is a significant factor of cataract occurrence and the risk as a result of family history is more among the direct relatives, especially brother and sister relatives. Family history classifications is an important risk factor of cataract that should not be relegated. Detailed family history of cataracts should adequately studied to identify the

classes of familial cases at greater risk so as to minimise cost and enable the benefit from merits attached to early detection and treatments for the disease. This study finding has therefore added to the effectiveness of first degree relative in the occurrence of cataract. It exposes the importance of regular eye check and improvement in quality of genetic counselling about cataract among direct relative.

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