

Mode of Inheritance and Clinical Based Studies on Hereditary Night Blindness in District Okara, Punjab Pakistan

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Abstract

Background: Reduced vision in low light or at night is a symptom of night blindness, also known as nyctalopia, which occurs when the eye does not adjust quickly enough from bright to dark environments. It can signal a wide variety of inherited and congenital retinal disorders and can also result from vitamin A insufficiency. Night blindness can be present at birth, brought on by an injury, or result from poor nutrition. It is a lack of adjustment to the dark.

Objective: The current study was designed to measure the prevalence, mode of inheritance, and certain clinical aspects of night blindness disorders in selected families from District Okara, Punjab, Pakistan.

Methods: Initially, the data were collected through a survey by visiting schools, colleges, and hospitals to identify affected families with night blindness disorder. After selecting affected families, consent from those families was obtained for the interview. Data on prevalence, mode of inheritance, and clinical symptoms were gathered from those families during interviews. Five families with a history of night blindness (including both consanguineous and non-consanguineous unions) were selected for pedigree analysis.

Results: Recessive, dominant, and sex-linked modes of inheritance were observed. In families OKR 1, OKR 2, OKR 3, OKR 4, and OKR 5, the percentage of family members associated with night blindness was 20.45%, 53.33%, 13.88%, 12.76%, and 6.25%, respectively. It was further noted that most males were affected in all families.

Conclusion: The observed inheritance pattern of night blindness in these families was autosomal dominant, autosomal recessive and X-linked.

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Keywords: Night Blindness, Nyctalopia, Inherited Retinal Disorders, Vitamin A Insufficiency, Prevalence, Mode of Inheritance, Autosomal Dominant, Autosomal Recessive, X-linked Inheritance, Family Pedigrees

Introduction

The majority of genetic disorders can be traced back to either an insufficient amount of genetic material or mutations (dangerous modifications to genes also known as pathogenic variations) [1]. Your genes, which are made up of DNA (deoxyribonucleic acid), include instructions for how your cells should operate and the characteristics that make you unique. DNA mistakes, known as mutations, can alter the genetic material. Reduced vision in low light or at night is a symptom of night blindness, also known as nyctalopia, which occurs when the eye does not adjust quickly enough from bright to dark environments [2]. It is a sign of a

wide variety of inherited and congenital retinal disorders and a consequence of vitamin A insufficiency. When you have night blindness, it is hard, if not impossible, to see in dim conditions. Night blindness from birth, with or without myopia (nearsightedness), is a persistent genetic condition that can be inherited in a dominant, recessive, or sex-linked pattern [3]. Retinitis pigmentosa is an inherited disorder that causes gradual vision loss due to the death of rod cells (visual receptors important for seeing in dim light), which can manifest as sudden-onset night blindness in children or teenagers. A shortage of vitamin A is one of the leading causes of retinitis pigmentosa, a kind of macular degeneration. Progressive degeneration of photoreceptors and the retinal pigment epithelium (RPE) causes RP symptoms such as night blindness, tunnel vision, and a slow loss of central vision. Florent Cunier, in 1838, was the first to describe congenital stationary night blindness (CSNB). Cunier researched the family tree of a 23-year-old child with night blindness and found 56 members of the family who had also been affected by night blindness, going back 11 generations to a man named Jean Nougaret.

The "Ebers Papyrus" No. 351 from around 1500 B.C. describes night blindness in ancient Egypt; this information was likely gleaned from much older medical texts dating back to 2500 B.C. To address this condition, the Egyptians resorted to injecting the juice derived from compressed liver directly into the eye. However, modern studies indicate that the cornea's limited ability to absorb vitamin A from liver extract would not be sufficient to effectively alleviate night blindness [4]. Night blindness is discussed in the Assyrian medical books from around 700 B.C. They believed the moon's beams caused the disease and could be treated by rubbing the ass's liver on the affected eyes. Nettleship continued Cunier's work in the 20th century and confirmed that the disease is strongly heritable but does not worsen over time [5]. Mutations in the RHO, GNAT1, or PDE6B genes cause an autosomal dominant form of stationary night blindness [6]. Mutations in the genes for RHO, GNAT1, and PDE6B alter the normal signaling in rod cells. The inability of the rods to send reliable signals to the brain results in impaired vision in dim light. These genes are essential for healthy vision, especially in dim light. The World Health Organization estimates that 254 million children around the world are deficient in vitamin A [7]. This disorder accounts for the vast majority of cases of blindness in young children. About half of these kids are born in South and Southeast Asian countries. Nyctalopia, retinopathy, xerophthalmia, Bitot spots, keratomalacia, conjunctival and corneal xerosis, conjunctival xerosis, and corneal xerosis are all associated clinical ocular disorders. Children under the age of six in Pakistan's North West Frontier Province (NWFP) are at increased risk for developing night blindness and/or color blindness. About 20% of Karachi's elementary and secondary school students have one of these conditions. Children in the NWFP are at a higher risk for night blindness and color blindness if they are younger than 6 years old. [8]. According to statistics from 2003, there are around 1,140,000 (962,000-1,330,000) blind adults in Pakistan. In Pakistan, cataracts account for over half (48.1%) of all causes of blindness.

The objective of the current study was to provide information on night blindness in the Okara area of Punjab, Pakistan. The prevalence of such genetic disorders in Pakistan is also an important aspect of the present study. This study was designed to record the mode of inheritance, prevalence, and the clinical investigation of night blindness disorder in families of District Okara.

Materials and Methods

Study Area

The current study was conducted in Okara (derived from *Okaan*, which is the name of a type of tree), a city in Punjab, Pakistan. It is the 23rd largest city of Pakistan by population. The city is located southwest of the city of Lahore, and Faisalabad is 100 km away, bypassed by the Ravi River. It is known for its agriculture-based economy and cotton mills. The nearest major city to Okara is Sahiwal, which was formerly known as Montgomery. Pakistan military dairy farms, known for their cheese, are situated in Okara. Wheat, cotton, tobacco, legumes, potato, and oilseeds are the main crops.

Research Design

Mix method research design (epidemiological study and clinical investigation) was applied in the current study.

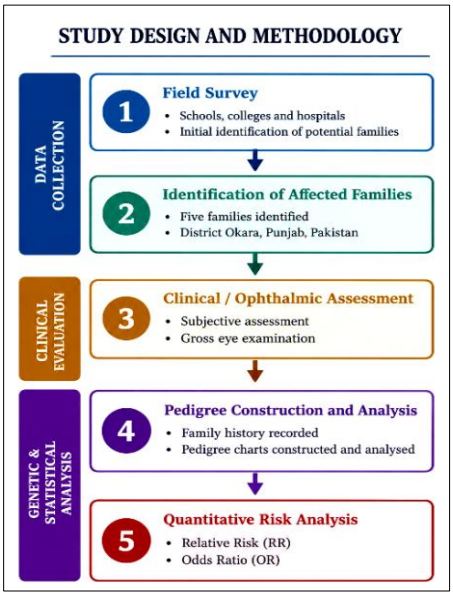


Fig 1: Flowchart illustrating the study design and methodology.

Study Population

The mixed-methods study was carried out on the general population with night blindness disorder in the district of Okara, province of Punjab, Pakistan.

Sample Collection

Both rural and urban areas of District Okara were visited to find families with the disorder. After detailed visits, five families were selected for the study and to construct the pedigree. Pictures of affected individuals were also taken with the consent of each family. The samples were collected from five different families using simple and direct questions. The university's ethical committee granted permission to conduct the current research (Table 1).

Table 1: Sampling Sites

Family	Name of Area	Geographical coordinates
OKARA 1	Chak No. 9/4-L, OKARA	30.7617° N, 73.4283° E
OKARA 2	Raza Block, Street # 01, District OKARA	73° 27' 33.8256" E
OKARA 3	Chak No. 28/4-L, Thana Shahbore, OKARA	30.7059° N, 73.3829° E
OKARA 4	Chak No. 19/1-R, OKARA	30.8920° N, 73.4997° E
OKARA 5	Basti Abdul Sattar, Bangla Gogera, OKARA	28° 41' 45" North, 70° 33' 20" E

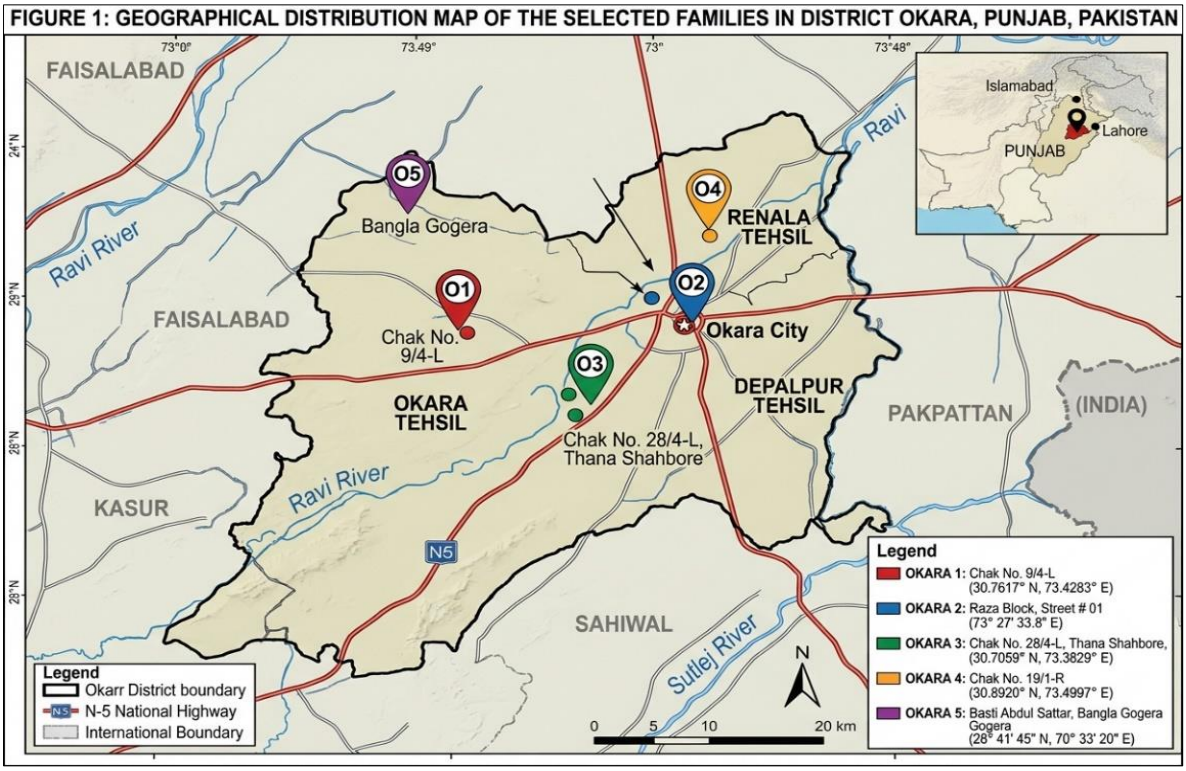


Fig 2: Geographical distribution map of the five selected families across the tehsils of District Okara, Punjab, Pakistan (indicating sampling locations 01 to 05 with exact GPS coordinates).

Data Analysis

Clinical symptoms and cousin marriages are analyzed to determine the number of first-cousin and second-cousin marriages. Males and females are analyzed, either male have more chances of night blindness than females. Rural and urban areas are analyzed, and then we check the epidemiology.

Clinical/ Ophthalmic Assessment

Since, the present study was not a clinical trial, but rather a community based genetic epidemiological study because of the reason was that the full ophthalmic instrumentation was not available in the field. Thus, standard Snellen visual acuity testing or automated refractors could not be used to formally quantify visual acuity. Rather, a detailed subjective assessment of visual function was performed using structured interviews with the affected persons and their family members. Systematically participants were asked about their daytime vision, their ability to see in low-light conditions or at night, the need for physical assistance (e.g. walking stick or someone else) to walk at night, and the presence of other symptoms, such as photophobia or blurring of the distance vision. From these functional descriptions, the severity of the visual impairment in dim light was classified as mild (difficulty seeing at night but walks without assistance), moderate (walks with support or with significant difficulty at night) or severe (walks with assistance and has noticeably reduced vision during the day). Although visual acuity was not actually measured, all affected persons complained of impaired vision in dim light and at night, which is characteristic of nyctalopia.

This study was not a clinical trial with a primary goal of evaluating the treatment of hereditary night blindness, but rather a community-based genetic epidemiological survey, so the focus was to document the mode of inheritance and prevalence of hereditary night blindness, not to be an

ophthalmic case series. Thus, the present fieldwork did not involve advanced retinal imaging and electrophysiological testing. The basic clinical examinations were performed in the field and comprised a standard torch-light examination and a gross external ocular examination. On direct torch-light examination, no anterior segment abnormalities (cataract or corneal opacities), gross fundus abnormalities were seen in affected individuals. Also, all affected people complained of subjective difficulty in dim light and at night, a hallmark feature of nyctalopia. Comprehensive slit-lamp biomicroscopy of the anterior segment, dilated indirect ophthalmoscopy of the posterior segment, Optical Coherence Tomography (OCT), visual field testing and Electroretinography (ERG) were not done. The advanced ophthalmic assessments suggested for future hospital-based studies to fully validate the clinical phenotype and confirm the electrophysiological subtype of Congenital Stationary Night Blindness (CSNB) are very useful, but the present genetic and epidemiological results are also very valuable.

Pedigree Analysis

Five families were selected for pedigree analysis, and all families were interviewed on the spot. The frequency and the mutation of night blindness disorder in each family were recorded through pedigree analysis.

Statistical Analysis

Statistical analysis was performed to check the prevalence and percentage of families with night blindness disorder. Data were analyzed using MS Office software, using descriptive statistics to find the mean, percentage, and prevalence rate of the disease.

Quantitative Risk Analysis of Night Blindness

Theoretical Background

The prevalence of night blindness and the observed correlation between consanguineous marriage and the presence of impaired offspring can both be quantified. Prevalence was employed in this study to measure the percentage of affected people in the entire population under investigation. Additionally, in order to examine the incidence of affected offspring between consanguineous and non-consanguineous marriages, the risk ratio (RR) and odds ratio (OR) were employed as exploratory measures.

Prevalence, which is expressed as a percentage, indicates the percentage of people in the population under study who have a specific ailment. It can be computed by multiplying the result by 100 after dividing the number of affected persons by the total number of people examined.

The ratio of the observed risk in an exposed group to the observed risk in an unexposed group is expressed by the risk ratio, a comparison metric. Consanguineous marriages were the exposure group in this study, whereas non-consanguineous marriages were the comparison group. The odds ratio contrasts two groups' odds of the same result. It offers an extra quantitative indicator of the noted correlation between the type of marriage and the impacted children. RR and OR should be viewed as exploratory estimates rather than conclusive measures of causation because the current research is based on a small number of reported marriages; the impacted child in the marriage was the outcome in this case.

Mathematical Formulation

1. Prevalence Formula

$$P=\frac{N_a}{N}\times 100$$

Where p = prevalence

$$N_a = number\ of\ affected\ individuals$$

N= Total number of individuals

$$P=\frac{31}{190}\times 100$$

P = 16.31%

2. Risk in Consanguineous Group

$$R_c=\frac{A_c}{T_c}$$

$$R_c = risk\ among\ consanguineous\ marriages$$

$$A_c = affected\ offspring\ reported = 3$$

$$T_c = total\ consanguineous\ marriges = 6$$

$$R_c=\frac{3}{6} \qquad R_c = 0.50$$

Observed risk = 50%

3. Risk in Non-Consanguineous Group

$$R_{nc}=\frac{A_{nc}}{T_{nc}}$$

$$R_{nc} = risk\ among\ non-consanguineous\ marriages$$

$$A_{nc} = affected\ offspring\ reported = 2$$

$$T_{nc} = total\ non-consanguineous\ marriges = 27$$

$$R_{nc}=\frac{2}{27}\ R_{nc} = 0.0741$$

Observed risk= 7.41 %

4. Risk Ratio

$$RR=\frac{R_c}{R_{nc}}$$

$$RR=\frac{3/6}{2/27}$$

$$RR=\frac{0.50}{0.0741}$$

RR= 6.75

5. Odds Ratio

$$OR = \frac{ad}{bc}$$

Marriage type	Affected	Non-affected
Consanguineous	a= 3	b=3
Non-Consanguineous	c= 2	d= 25

$$OR = \frac{(3)(25)}{(3)(2)} \quad OR = \frac{75}{6} \qquad OR = 12.50$$

Interpretation

According to the quantitative analysis, 16.31% of the people under study had night blindness. The corresponding odds ratio was 12.50, and the observed risk of affected offspring was 6.75 times higher among reported consanguineous marriages than among non-consanguineous marriages. These results show a significant correlation between the occurrence of affected offspring in the households under study and consanguineous marriage. However, the computed RR and OR should be viewed cautiously as rough exploratory estimates and should not be regarded as proof of a causative association because the research was based on a small number of reported marriages.

Results

This study was to investigate the prevalence and clinical features of night blindness among some selected families of District Okara.

Demographic Characters

In the current study, a total of 5 families participated. These five families were comprising of total 190 members. Of these, there are 103 males and 87 females. Among all individuals, 123 members were alive, and 67 individuals were dead. 15 individuals were residents of urban areas, and 175 were residents of rural areas (Table 2).

Table 2: Demographic characteristics of respondents in the studied population

Participant Characteristics	Frequency	Percentage %
Gender		
Male	103	54.21%
Female	87	45.78%
Age		
≤25 years	38	20%
26-35 years	41	21.57%
36-45 years	71	37.36%
>45 years	40	21.05%
Residence/ Locality		
Urban	15	7.89%
Rural	175	92.10%
Marriage		
Consanguineous	6	18.18%
Non-consanguineous	27	81.81%
Alive		
Male	67	54.47%
Female	56	45.52%
Death		
Male	36	53.73%
Female	31	46.26%

Prevalence of Night Blindness

The present study was planned to record the mode of inheritance of night blindness in selected families of the OKARA, Punjab, Pakistan. Five families with night blindness were selected for the detailed study. Persons with night blindness disorder were identified. The prevalence of this genetic disorder was 16.31% in the studied families. The prevalence was recorded as the number of affected men out of the total number of members of the family. Out of 190 members, 31 were affected with night blindness. (Table 3)

Table 3: Prevalence of Night blindness

Family	Total member	Affected	Percentage%
OKARA 1	44	9	20.45%
OKARA 2	15	8	53.33%
OKARA 3	36	5	13.88%
OKARA 4	47	6	12.76%
OKARA 5	48	3	6.25%
Total	190	31	16.31%

The prevalence of night blindness was different in different families. In family, OKA 1 was observed 20.45%, OKA 2 was 53.33%, OKA 3 was 13.88%, OKA 4 was 12.76%, OKA 5 was 6.25%, in affected members. The prevalence rate of night blindness was higher in males (74.19%), while it was much lower in females (25.80%) (Table 4).

Table 4: Number of Males and Females associated with Night Blindness

Family	Total associated with Night Blindness	Number of Males	Number of Females
OKARA 1	9	4	5
OKARA 2	8	6	2
OKARA 3	5	4	1
OKARA 4	6	6	0
OKARA 5	3	3	0
Total	31	23	8

Clinical Manifestations of Affected Individuals

A thorough clinical evaluation of all affected members in each of the five families showed that the common feature was a defect in vision in low light levels and at night (nyctalopia). However, the severity, length, and ocular symptoms were different amongst families. Each family had a summary of the clinical findings, which is presented below.

Description of Family OKA 1

The Family 1 is from District Okara, Province of Punjab, Pakistan. There were four generations of affected members. The marriages of the majority of parents in this family were non-consanguineous. A total of nine members (4 males and 5 females) were affected by this genetic disorder. The pedigree sketch indicated that there were 5 generations with 44 family members, 1 affected in the 1st generation, 2 affected in the 2nd generation, 3 affected in the 4th generation and 3 affected in the 5th generation. Data from pedigree analysis of affected members (where it was possible to calculate) indicated that the inheritance was probably autosomal recessive. Those affected said that they could not see properly at night, and that their vision in the day had declined over time. They were unable to walk at night without help, like a stick or someone else. The symptoms were evident from childhood and moderate to severe in dim light. During the field visits, no anterior segment defect or other systemic defect was detected. Formal visual acuity testing and dilated fundus examinations were not performed, however. The family's pedigree is given in Fig. 3.

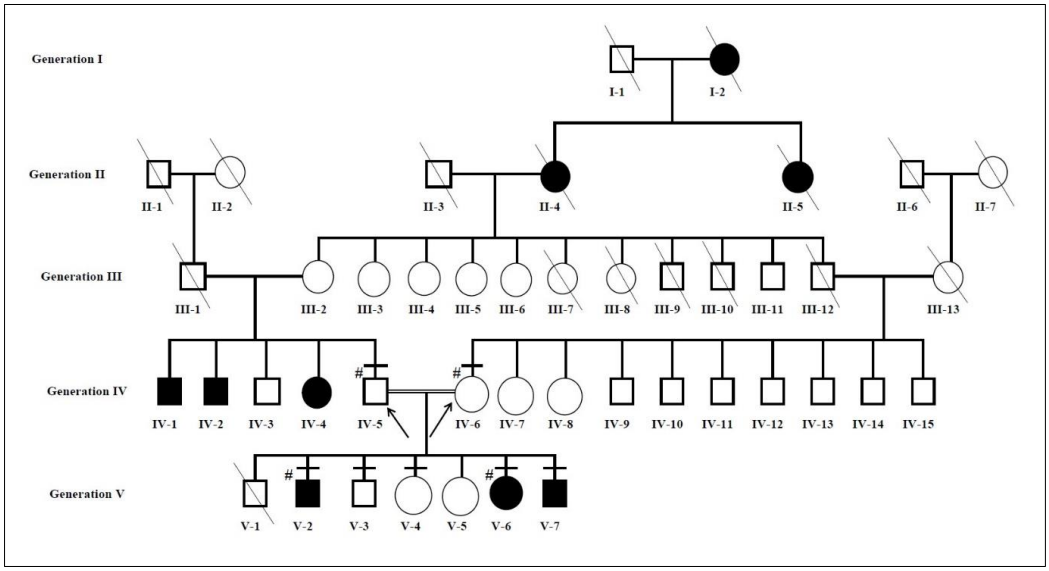


Fig 3: Picture of Pedigree Family 1.

Description of Family OKA 2

Family 2 belongs to District Okara, Province of Punjab, Pakistan. Affected members were present in all three generations. The parents of this family had a non-consanguineous marriage (they were not siblings). A total of eight members (6 males and 2 females) were affected by this genetic disorder. The pedigree sketch showed three generations consisting of 15 family members, with 1 affected member in the 1st generation, 3 affected members in the 2nd generation, and 4 affected members in the 3rd generation. Pedigree analysis of the affected members, whose data could be calculated, suggested an autosomal dominant mode of inheritance. This is corroborated by the direct transmission of the trait from one generation to the next for three generations in a row. In addition, a female affected person (I-2) transmitted the disorder to her male and female children (II-2, II-3, and II-4). Likewise, in the subsequent generation, the affected female (II-2) passed the disorder on to several of her children (III-4, III-7, III-8, and III-9). Since the disease occurs in both sexes and has been found in every generation, an autosomal dominant pattern of inheritance is suggested.

Patients were not able to adequately see at night. They could see during the daytime at the age of 10–12 years, but their eyesight weakened considerably in later years. They walked with support (stick or someone else) at night. Two individuals in the third generation were affected and had severe functional impairment, unable to walk without support. Symptoms were lifelong and progressive deterioration of daytime vision. There were no other gross dysmorphic features or other ocular anomalies observed. The pedigree of the family is shown in Fig. 4.

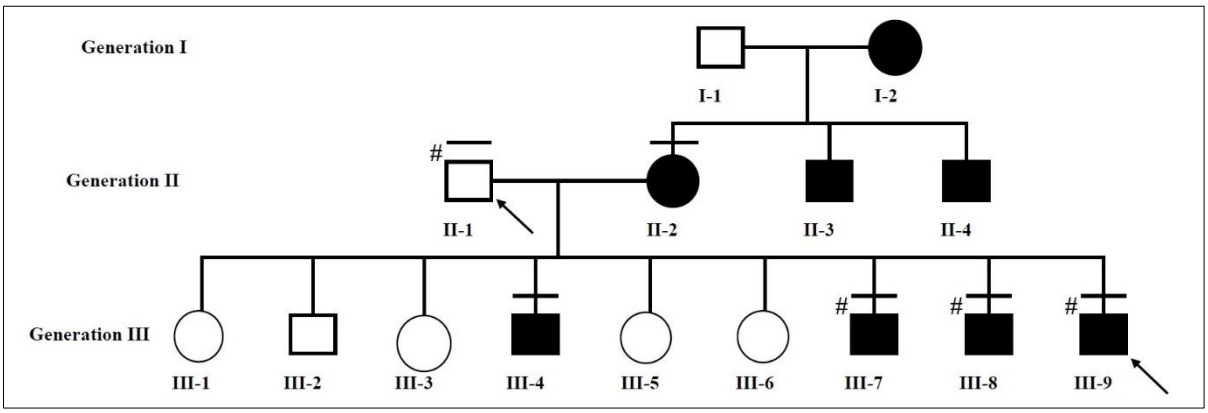


Fig 4: Picture of Pedigree Family 2.

Description of Family OKA 3

Family 3 belongs to District Okara, Province of Punjab, Pakistan. Affected members were present in one generation only. The parents of this family were not related to each other (non-consanguineous marriage). A total of five members (4 males and 1 female) were affected by this genetic disorder. The pedigree sketch showed four generations consisting of 36 family members, with 5 affected members in the 4th generation. The pedigree analysis is very suggestive of an autosomal recessive mode of inheritance. This is apparent from the presence of totally unaffected parents (III-1 and III-2) who gave rise to 5 affected children (IV-1, IV-2, IV-6, IV-10, and IV-11) in Generation IV. The presence of the trait in one sibship of unaffected parents is the typical feature of an autosomal recessive trait, meaning that both parents are obligate carriers. In addition, both affected males and females (IV-11) eliminate the possibility of X-linked inheritance.

Patients had severe photophobia, they were unable to open their eyes during the day with the right amount of sunlight and they had excessive tearing (lacrimation). At night they had poor eyesight. Some members were able to see properly during the day at 10–12 years old, but they became too weak to see later in life. They walked with the aid of a stick at night. The severity of night blindness was moderate to severe and the duration was from early childhood. There was no gross structural ocular anomaly noted. This family's pedigree is presented in Fig. 5.

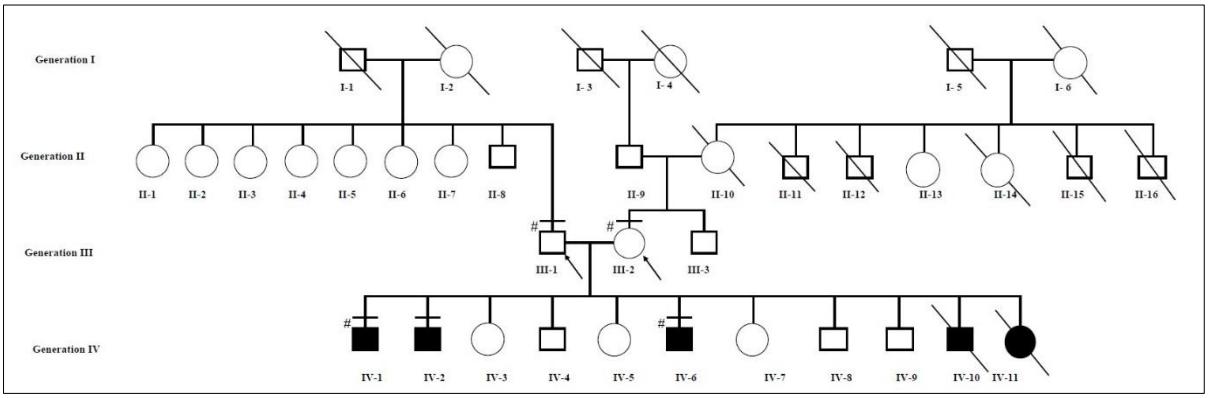


Fig 5: Picture of Pedigree Family 3.

Description of Family OKA 4

Family 4 belongs to District Okara, Province of Punjab, Pakistan. Affected members were present in two generations. Some parents in this family were related to each other (consanguineous), while others were not. A total of six members (all 6 males) were affected by this genetic disorder. The pedigree sketch showed five generations consisting of 47 family members, with 6 affected members: 3 affected members in the 4th generation and 3 affected members in the 5th generation. The pedigree is strongly suggestive of an X-linked recessive mode of inheritance. This is confirmed by the fact that all the males in the pedigree are affected and the transmitting parents are unaffected. For example, generation III (III-3 and III-4) had no affected individuals and were the parents of affected sons in Generation IV and V, meaning that III-3 and III-4 were obligate carriers. In addition, there is no evidence of male-to-male transmission. In addition, there are consanguineous marriages in the pedigree (e.g. IV-6 and IV-7), which further increases the risk of expression of recessive traits.

Affected people were able to see well in the light of day, but could not see things in the distance clearly. They were unable to see clearly at night and only could see the shadows of objects. If they didn't look toward light, their eyesight was fairly good, and they could see colors well. At night, they had difficulty walking anywhere. The symptoms were present since childhood and the severity was moderate. Some of the affected people were reported as being nearsighted (myopic), a known association with some types of congenital stationary night blindness (CSNB). This family's pedigree is given in Fig. 6.

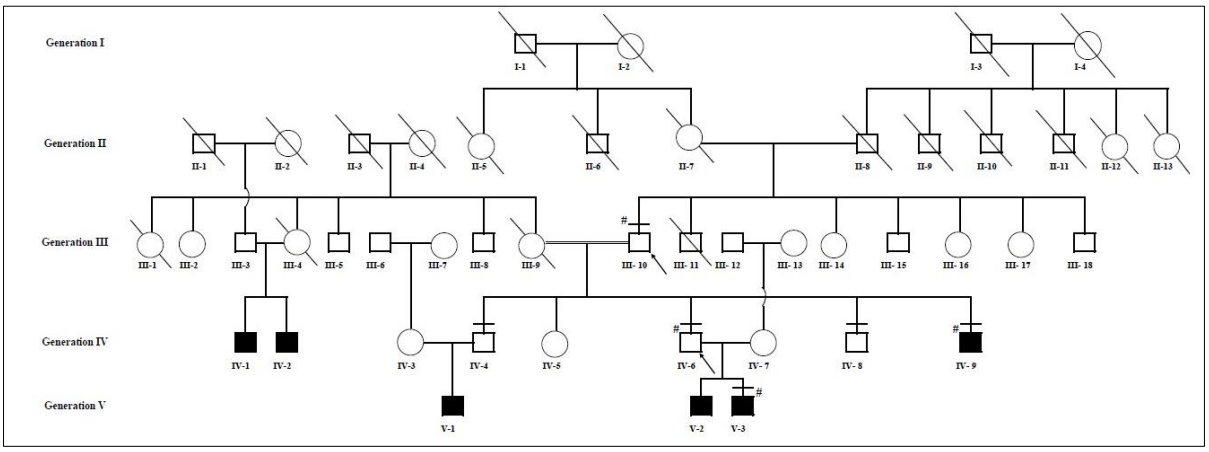


Fig 6: Picture of Pedigree Family 4.

Description of Family OKA 5

Family 5 belongs to Bangla Gogera, Tehsil and District Okara, Province of Punjab, Pakistan. Affected members were present in one generation only. The parents of this family were related to each other (consanguineous marriage). A total of three members (all 3 males) were affected by this genetic disorder. The pedigree sketch showed four generations consisting of 48 family members, with 3 affected members in the 4th generation. Pedigree analysis of the affected members, whose data could be calculated, suggested an X-linked recessive mode of inheritance, as only males were affected and the defect was present from birth in a consanguineous family.

With sunlight, affected persons were able to see well during the day but not clearly at a distance, and vision seemed to be blurred. They were unable to see well at night. The defect occurred at birth. They had developed a low daytime vision over time. At night, they had difficulty walking anywhere. Severity was moderate to severe and the onset was congenital (lifelong). Gross examination did not reveal any nystagmus or strabismus, but a formal ophthalmic examination was not conducted. The pedigree of this family is shown in Fig. 7.

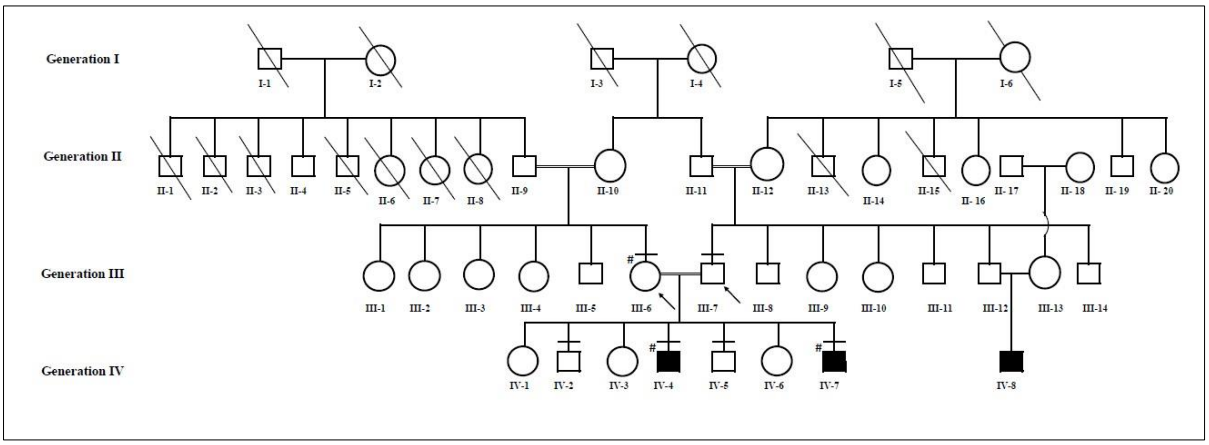


Fig 7: Picture of Pedigree Family 5.

The cardinal symptom was from birth or early childhood, a symptom of nyctalopia, in all five families, in keeping with the congenital stationary nature of the disorder. Other symptoms were photophobia (Family OKA 3), blurred distance vision with possible myopia (Families OKA 4 and OKA 5) and progressive deterioration of the daytime vision in some patients. Gross field examination did not reveal any obvious associated systemic anomalies, nystagmus, or strabismus. This is a limitation of this field-based genetic survey, as a comprehensive slit-lamp examination of the anterior segment, dilated fundus examination, visual acuity testing and electrophysiological evaluation (ERG) were not performed. The electrophysiological type of CSNB is not fully characterized in these assessments, and future studies including these assessments are recommended to confirm the electrophysiological type of CSNB. When asked about the onset of symptoms, structured interviews identified that in most families [5] the onset was gradual and for as long as they could remember, back to early childhood or birth. None of the affected persons experienced a sudden onset of night blindness in adolescence or early adulthood. This is a very important clinical characteristic that is a hallmark of a congenital and stationary disorder.

Summary of Pedigree Analysis and Inheritance Patterns

A brief summary of the genetic analysis, consanguinity status, sex distribution of affected individuals, and proposed mode of inheritance for all five families is given in Table 5. This analysis shows that there is a great amount of genetic heterogeneity within the population studied, with various families having different modes of inheritance (autosomal recessive, autosomal dominant, and X-linked recessive patterns).

Table 5: Pedigree characteristics and suggested modes of inheritance across the five studied families.

Family ID	Consanguinity Status	Total Affected (M/F)	Suggested Inheritance	Key Pedigree Observations
OKA 1	Non-consanguineous	9 (4 M / 5 F)	Autosomal Recessive (AR)	Unaffected parents with affected offspring; both sexes affected.
OKA 2	Non-consanguineous	8 (6 M / 2 F)	Autosomal Dominant (AD)	Vertical transmission across three generations; both sexes affected.
OKA 3	Non-consanguineous	5 (4 M / 1 F)	Autosomal Recessive (AR)	Unaffected parents in Gen III produced 5 affected offspring in Gen IV.
OKA 4	Some consanguineous	6 (6 M / 0 F)	X-linked Recessive (XR)	Only males affected; no male-to-male transmission; unaffected carrier mothers.
OKA 5	Consanguineous	3 (3 M / 0 F)	X-linked Recessive (XR)	Only males affected; born to unaffected consanguineous parents.

Abbreviations: M, Males; F, Females; AR, Autosomal Recessive; AD, Autosomal Dominant; XR, X-linked Recessive.

Discussion

From a physiological standpoint, the development of retinal pigments is essential. Congenital night blindness is a physical condition that can affect humans, dogs, cats, and other animals. (9) The absence of retinal pigments is characteristic of this disorder. Congenital stationary, X-linked recessive, and dominant are three of the many subtypes of this disorder. (10) Night blindness is a catch-all phrase for a group of ophthalmological conditions that diminish one's vision in dim light, or scotoma. (11) This study was designed to study the prevalence rate of night blindness disorder among some selected families of District Okara. Results of the current study showed that the prevalence rate of night blindness among selected families was 16.31%. The reason behind the prevalence rate of night blindness among selected families was due to consanguineous and non- consanguineous marriages. Among 5 families, there were 33 marriages, of which 6 (18.18%) were consanguineous, and 27 (81.81%) were non-consanguineous. Of the 6 consanguineous marriages, 3 were marriages in which their offspring inherited night blindness disorder, and only 2 non-consanguineous marriages had night blindness disorder. This study concluded that the prevalence rate of night blindness varies in each family. The prevalence rate among family OKR 1 was 20.45%, family OKR 2 was 53.33%, family OKR 3 was 13.88%, family OKR 4 was 12.76%, and Family OKR 5 was 6.25%, respectively.

According to the findings of our research, marriage within a family increases the likelihood of contracting an illness when compared to marriage between unrelated individuals. Consanguineous marriages are connected with a high risk for congenital abnormalities and autosomal recessive illnesses, with some instances of higher postnatal mortality in the kids of first-cousin couples as a consequence of this increased risk [12]. The findings of our research indicate a clear association between consanguineous marriages and night blindness disorder in certain families. Consanguinity increases the likelihood of inheriting recessive genetic disorders, including those affecting cognitive function [13]. The higher incidence of night blindness observed in the families studied suggests a potential genetic basis for this condition. Consanguinity leads to an increased risk of offspring inheriting homozygous recessive alleles, which can result in the expression of deleterious genetic mutations associated with night blindness disorder [13].

It is important to note that not all individuals from consanguineous marriages will experience night blindness. The expression of genetic disorders is influenced by various factors, including the specific recessive alleles carried by the parents, the presence of additional genetic modifiers, and the complex interplay between genetics and environmental influences. (14) Pedigree analysis of these families indicates that the mode of inheritance of night blindness exhibits genetic heterogeneity, specifically autosomal dominant, autosomal recessive, and X-linked recessive patterns.

Age of Onset and Clinical Phenotype

One of the important points when studying hereditary night blindness is to differentiate between the age of onset and the age of ascertainment. In our study, the mean age at ascertainment (not at onset) was 25 years or older. The clinical histories obtained from the families clearly show that the nyctalopia was present at birth or from early childhood. This is consistent with the classical presentation of Congenital Stationary Night Blindness (CSNB), which is a congenital (from birth) condition that does not get worse over time. This congenital form should be distinguished from juvenile night blindness, which usually occurs at 18 to 25 years of age, and is often linked to progressive retinal dystrophies such as early onset Retinitis Pigmentosa. Since all the individuals in our study with the disorder reported having symptoms since childhood and in the pedigree, patterns found, it is extremely improbable that these families have a juvenile onset of the disorder. Subsequent molecular diagnosis will be useful to exclude progressive juvenile dystrophies, but the clinical history is very suggestive of a congenital stationary dystrophy.

Limitations of the Study

This study may give us some epidemiological and genetic information about hereditary night blindness in District Okara but some limitations must be noted. As this was a field-based community survey, formal quantification of visual acuity using Snellen charts was not feasible. Another limitation is lacking anterior and posterior segment examination by slit-lamp biomicroscopy and dilated indirect ophthalmoscopy. In the present study, the diagnosis was mainly based on clinical history and pedigree analysis. One of the major limitations is the absence of complete structural and functional assessments of the retina, such as dilated fundus examination, Optical Coherence Tomography (OCT), visual field testing, and Electroretinography (ERG). The clinical appearance of our families, however, with congenital onset of nyctalopia and normal colour vision and absence of any external ocular anomaly fits well with the classical description of congenital stationary night blindness (CSNB) in which the fundus is often normal. The subjective reports of nyctalopia and the pedigree patterns are very suggestive of CSNB, however, no electrophysiological and/or structural evidence exists and progressive retinal dystrophies cannot be

excluded. Future studies including ERG (key diagnostic test for CSNB) and OCT and visual field testing are warranted to confirm the electrophysiological subtype and to definitively exclude progressive retinal dystrophies.

Conclusion

The present study concludes that the hereditary night blindness in the studied families of District Okara is genetically heterogeneous and in different families it is inherited in autosomal recessive, autosomal dominant and X linked recessive manner respectively. The patients complained of night blindness from childhood or at birth, which is typical for congenital stationary nyctalopia. It is important to note that the age of ascertainment of the individuals in this study (25 years or older) is not the same as the age of onset. In addition, the observed risk of affected children was higher in families where both parents were consanguineous (RR = 6.75; OR = 12.50), but non-consanguineous families also had affected children. This means that there are both consanguinity-dependent (recessive) and consanguinity-independent (dominant/X-linked) factors that lead to the disease burden in this population. So, cousin marriage is a significant risk factor for the recessive types, but is by no means the only cause for night blindness in these families.

Conflict of Interest

The authors declare no conflict of interest.

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