

Association of Socioeconomic Status, Consanguinity and Congenital Ophthalmological Anomalies

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ABSTRACT

Congenital ophthalmological anomalies pose a significant health challenge, particularly in developing countries where consanguineous marriages are prevalent. This study investigates the intricate relationship between socioeconomic status, consanguinity, and the incidence of these anomalies in pediatric patients, aiming to identify underlying causal pathways. This cross-sectional analytical study was carried out at a Tertiary Care Institute in Pakistan from September 2020 to January 2021. We included pediatric patients with congenital ophthalmological abnormalities scheduled for surgeries or examinations, born from uncomplicated pregnancies and parturition. Socioeconomic status was determined using government basic pay scales, which provide an estimation of average income. Patients with a history of infertility or complicated pregnancy/parturition were excluded. A total of 184 children with congenital abnormalities were studied, comprising 126 males (68.5%) and 58 females (31.5%). The mean maternal and paternal ages were 23.86 ± 5.4 and 27.07 ± 9.6 years, respectively. The median age of children was 2.0 years, with a 25% inter-quartile range of 1.0. The most common anomaly was congenital cataract (35.9%, 66/184), followed by squint (9.24%, 17/184) and retinoblastoma (6.52%, 12/184). Consanguineous marriages were more frequent among parents from the lower middle-income group (80%) compared to the middle (62%) and upper middle-income groups (20%). Our causal inference analysis demonstrated that lower socioeconomic status influences consanguinity, which, along with age, causes congenital cataracts and retinoblastoma. No significant link was established between glaucoma and consanguinity or socioeconomic status. Congenital malformations were more frequently observed in children of consanguineous parents. This study underscores a significant association between consanguinity, socioeconomic status, and congenital ophthalmological anomalies in the pediatric population of Pakistan. The findings highlight the need for public health interventions, including education, genetic counseling, and improved healthcare access, to mitigate the burden of hereditary eye diseases.

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INTRODUCTION

The term consanguinity refers to the union between couples who share a common ancestor. Offspring of such parents have a higher probability of inheriting identical copies of autosomal recessive genetic mutations, increasing the potential for genetic disorders. On average, these children will receive identical gene copies from each parent, and their progeny will be homozygous at 6.25% of gene loci. Such marriages are more common in communities with lower literacy levels and awareness. (Anwar & Taroni, 2019). Such marriages are more common in communities with lower literacy levels and awareness (Anwar et al., 2020).

Pakistan is one of the countries reporting the highest rate of consanguinity, with 76% reported in Pakistani armed forces personnel's families. (Badr, 1972 This is the second-highest rate ever reported in literature, after the Arab Nubian population of Egypt, with 80.4% being consanguineous (Barakat & Gietel-Basten, 2014). According to data reported by UNICEF, the highest neonatal mortality rate (46 deaths per 1000 live births) is owing to a higher rate of consanguinity, augmenting child mortality and morbidity due to genetic malformations (Ben Halim et al., 2013; Ben Halim et al., 2016). Pre-marital screening tests are mandatory in Syria and Lebanon without which a couple is unable to get an official marriage certificate (Bener & Hussain, 2006). Autosomal recessive disorders are common

in first-degree relatives due to the inheritance of a recessive allele from each parent (Bhinder et al., 2019). Australia and the United States of America have reported only 1% of marriages as consanguineous (Bittles et al., 1993). In Tunisian children, there was a high incidence of non-syndromic recessive deafness in rural areas due to a high degree of consanguineous marriages (Consanguineous Marriages, n.d.; Corry, 2014).

Consanguinity in Pakistan is a serious issue. It is influenced by many societal factors including religion, language (ease of communication), illiteracy as well as economic benefits such as dowry and family wealth (El Sabeh et al., 2021). Most of the world's religions do not encourage inter-family marriages, but is somehow permitted in Judaism, Buddhism and Zoroastrian religion. People tend to marry within the same linguistic group so that they can communicate easily with the other family members. Consanguineous marriages are more common among the areas where the literacy rate is lower which implies that people might not be aware of the genetic consequences of consanguineous marriages. In many communities across the world, consanguineous marriages are performed to keep their royal blood "pure" and/or to retain their family wealth within the family (El Shanti et al., 2015). However, consanguinity causes a higher risk of recessive genetic disorders including congenital ophthalmological abnormalities. Bener and Mohammad reported that there are slightly higher risks of cancer, diabetes mellitus, heart diseases, hearing problems, blood and mental disorders as well as ophthalmological abnormalities (Fareed & Afzal, 2017). Kumaramanickavel reported, in a hospital-based study in south India, that 28.8% of the patients with ocular genetic disorders has a history of consanguineous marriages (Hashmi, 1997). It has been reported that the risk of recessive genetic disorders increased 8 to 9 fold with parental consanguinity (Hug et al., 2019), while a 25 fold increase in xeroderma pigmentosa.

While the pervasive issue of consanguinity in Pakistan and its general link to genetic disorders are well-documented, the specific causal pathways involving socioeconomic status and various congenital ophthalmological anomalies remain less explored. This study aims to fill this gap by evaluating the impact of consanguineous marriages and the socioeconomic status of parents on specific congenital ophthalmological abnormalities in pediatric patients, utilizing a causal inference approach to provide novel insights into these relationships. This investigation will sharpen our understanding of the clinical significance and provide a basis for targeted public health interventions.

METHOD

Study Design and Setting

This descriptive cross-sectional analytical study was carried out at a Tertiary Care Institute in Rawalpindi, Pakistan, over five months, from September 2020 to January 2021. Ethical approval (257/ERC) was obtained prior to commencement.

Sample and Participants

The sample size of 184 participants was calculated considering an 8.9% prevalence of ocular anomalies due to consanguinity, a 95% confidence level, 80% study power, and 10% attrition (Shawky et al., 2013). Informed written consent

was obtained during pre-anesthetic evaluation by the parents of the pediatric patients. All pediatric patients with congenital ophthalmological abnormalities scheduled for ophthalmological surgeries or examinations born with uncomplicated pregnancies and parturition were included in the study.

Exclusion Criteria

Patients were excluded if they had a history of infertility, were on fertility or hormonal treatment during pregnancy, conceived with assisted reproductive techniques, had prolonged complicated labor, premature deliveries, twin births, or prolonged hospital admission immediately after birth.

Since government basic pay scales are fixed, it gives an estimation of average income, therefore, these were employed to determine socioeconomic status. History was obtained by interviewing the child's mother or father whoever accompanied the child to the clinic. Pediatric evaluation and notes were referred to define congenital syndromes associated with ophthalmological abnormality if a couple had a history of infertility and remained on fertility, hormonal treatment during pregnancy, and conceived with any assisted reproductive techniques, they were eliminated from the study. The exclusion criteria included a history of prolonged complicated labor, premature deliveries, twin births, and prolonged hospital admission immediately after birth.

Variable Measurement and Data Collection

1. Socioeconomic Status: Determined using government basic pay scales, which provide an estimation of average income.
2. Data Collection: History was obtained by interviewing the child's mother or father, whoever accompanied the child to the clinic. Pediatric evaluation and notes were referred to define congenital syndromes associated with ophthalmological abnormality.

Bias Control and Team Training

While specific details on bias control and team training are not explicitly mentioned in this study, standardized data collection forms were used, and interviews were conducted by trained personnel to ensure consistency. Instrument validation was not applicable as primary data collection involved structured interviews and review of medical records.

Ethical Considerations

Informed written consent was obtained from the parents of the pediatric patients during pre-anesthetic evaluation. The study was approved by the ethical research committee of Armed Forces Institute of Ophthalmology, Pakistan. Consent for publication of the findings was also obtained from all participants.

Statistical Analysis

Data was analyzed using IBM SPSS software. Descriptive statistics for categorical variables were presented as frequency and percentage, while mean and standard deviation were reported for continuous variables. Categorical groups were compared using the chi-square test, and mean values were compared using independent samples t-test or one-way ANOVA. A p-value of ≤ 0.05 was considered significant. The results were graphically illustrated by using the tidyverse (Wickham et al., 2019), plotly (Plotly Technologies Inc., n.d.) and ggplot2 (Wickham, 2016).

packages of the R programming language (R Core Team, n.d.). Causal inference relationships obtained by the Peter-Clark algorithm from causal learn (Spirtes et al., 1993) package of Python programming language (van Rossum, 1995).

RESULTS OF STUDY

Population Characteristics

The mean maternal and paternal age was recorded to be 23.86±5.4 and 27.07±9.6 respectively. The study included 184 children with congenital abnormalities, with a male-to-female ratio of 126 (68.5%) males and 58 (31.5%) females. The median age in years was reported to be 2.0 years with a 25% IQR of 1.0. The prevalence of consanguinity (inter-family marriage) in the study population was 69.6% (128/184). Demographic characteristics are summarized in Table 1.

Table 1. Demographic characteristics of the study population (n=184)

Demographics	Gender		p-value
	Male (n=126)	Female (n=58)	
Age in years (median/IQR)	2.0±3.0	3.0±4.0	0.059
Age groups in years			
<1	25 (19.8%)	9 (15.5%)	0.431
1 – 5	86 (68.3%)	37 (63.8%)	
6 – 10	9 (7.1%)	8 (13.8%)	
>10	6 (4.8%)	4 (6.9%)	
Socioeconomic Status			
Lower middle	92 (73.0%)	46 (79.3%)	0.001
Middle	21 (16.7%)	-	
Upper middle	13 (10.3%)	12 (20.7%)	
Consanguinity			
Yes	87 (69.0%)	41 (70.7%)	0.822
No	39 (31.0%)	17 (29.3%)	
ASA Status			
Level II	74 (58.7%)	39 (67.2%)	0.271
Level III	52 (41.3%)	19 (32.8%)	

Prevalence of Anomalies

The most common congenital abnormality encountered was congenital cataract, reported in 66/184 (35.9%) of the children. This was followed by squint, present in 17/184 (9.24%) children, and retinoblastoma, reported in 12/184 (6.52%) of children. The detailed distribution of congenital abnormalities is presented in Figure 1. A summary of

different procedures performed on the study group is given in Figure 2.

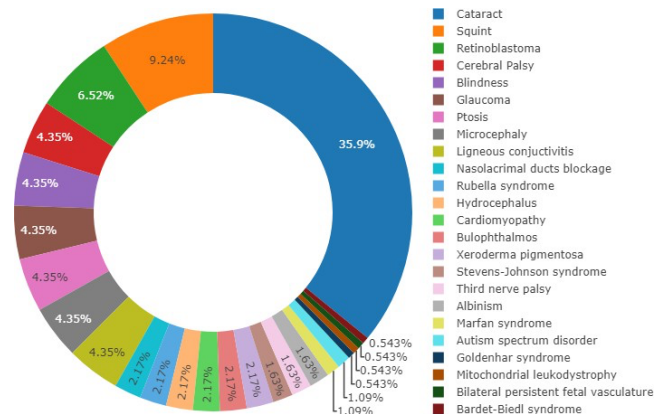


Figure 1. Distribution of congenital anomalies among the study group. Each color represents a specific disease given in the figure legend along with the percentage of patients included in this study.

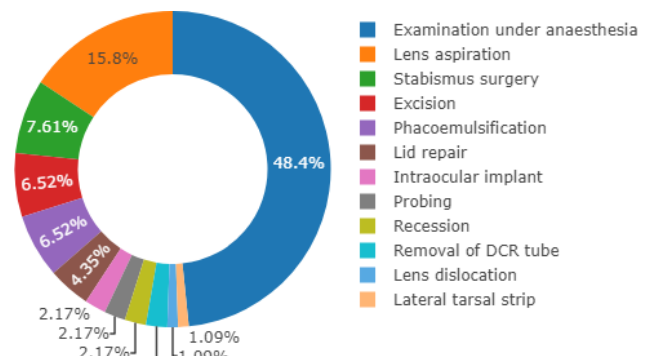


Figure 2. Summary of different procedures performed on the study group. Each color represents a medical procedure (given in the figure legend) along with the percentage.

Association with Consanguinity and Socioeconomic Status

Parents from the lower middle-income group were significantly more likely to have undergone inter-family marriages than those from the middle and upper middle-income groups (85.9% vs 10.2% vs 3.9%, p<0.001). Congenital cataract (p=0.007), congenital blindness (p=0.05), and ligneous conjunctivitis (p=0.05) were found to be significantly associated with consanguinity through univariate analysis, as shown in Table 2.

Table 2. Association of consanguinity with the type of congenital disorder

Congenital disorder	Consanguinity		(OR)	95% CI	p-value
	Yes (n=128)	No (n=56)			
Congenital Cataract					
Yes	54 (42.2%)	12 (21.4%)	1.3	1.1–1.6	0.007
No	74 (57.8%)	44 (78.6%)			
Congenital Blindness					
Yes	8 (6.3%)	0 (0%)	1.4	1.3–1.6	0.05
No	120 (93.8%)	56 (100%)			
Ligneous Conjunctivitis					
Yes	8 (6.3%)	0 (0%)	1.4	1.3–1.6	0.05
No	120 (93.8%)	56 (100%)			

Causal Findings

We observed that individuals of lower socioeconomic status have a higher likelihood of consanguinity, which in turn, along with age, causes congenital disorders such as cataracts and retinoblastoma. To support this observation, we applied the Peter-Clark causal inference discovery algorithm with the Chi-Square independence test to discover causal graphs for congenital cataracts, congenital glaucoma, and retinoblastoma. For each disorder, a separate causal graph was created from socioeconomic status, consanguinity, the disorder itself, and age.

The algorithm indicated that socioeconomic status affects consanguinity ($p < 0.000001$), and consanguinity in turn ($p = 0.006$) together with age ($p < 0.00001$) causes congenital cataracts. For congenital glaucoma, no direct dependence between consanguinity ($p = 0.21$) or age ($p = 0.24$) and the disorder itself was found (Figure 4). Retinoblastoma is caused by age ($p = 0.0009$) and consanguinity ($p = 0.007$), which is in turn caused by socioeconomic status ($p < 0.00001$). All mentioned p-values are from the Chi-Square statistical test for conditional independence of corresponding data. The increased prevalence of consanguineous marriages among individuals of lower socioeconomic status is a particular concern, as it increases the risk of genetic disorders. There is no evidence of causal links between age or consanguinity and glaucoma. Congenital cataracts and retinoblastoma are influenced by both age and consanguinity, which is impacted by socioeconomic status, while squint is regulated only by age. In general, consanguinity is strongly connected to socioeconomic status.

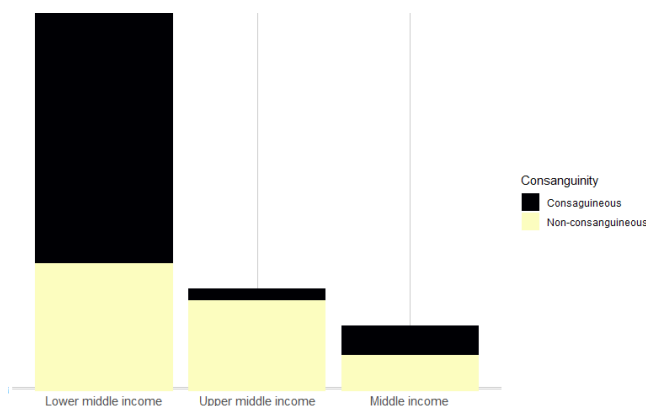


Figure 3. Comparison of consanguinity according to socioeconomic class. The data is distributed according to the sample size from different socio-economic classes (i.e. Lower middle income, Upper middle income and Middle income).

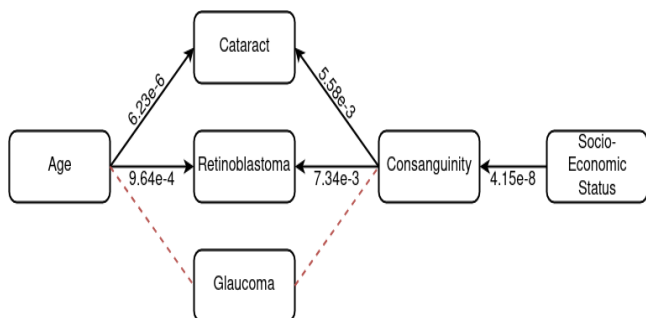


Figure 4. Illustration of combined causal graph for common disorders obtained from concatenating individual graphs for cataract, retinoblastoma and glaucoma. Each edge is accompanied by p-value of Chi-square conditional independence test.

DISCUSSION

A significant number and spectrum of congenital ophthalmological abnormalities and associated disorders and syndromes were observed in first-degree relatives' children. Although not statistically significant, it is pertinent to mention that in certain families, more than one offspring suffered from congenital malformations such as xeroderma pigmentosa, congenital cataract, congenital blindness, and microcephaly. A significant amount of literature is available from third-world countries deriving a substantial correlation between consanguineous marriages and congenital anomalies. Anwar et al. surveyed 7,312 families in Bangladesh, of which 3,694 partners were consanguineously married. The mean prevalence of consanguineous marriages in the studied population was 6.64%. Consanguineous unions were associated with the increased incidence of several multi-factorial diseases and congenital malformations, including bronchial asthma, hearing defects, heart diseases, and sickle cell anemia ($p < 0.05$). Generally, most of the public is unaware of the consequences of consanguineous marriages with similar practices in Pakistan with greater prevalence in lower socioeconomic and literacy groups (Ijaz et al., 2017).

Praveen et al. conducted a cross-sectional epidemiological study in India to gather information regarding the consanguinity of the parents. A total of 10,290 participants underwent a detailed ocular examination. Parental consanguinity was reported in 1,822 (24.7%) rural subjects and 782 (32.9%) urban subjects. Similarly, the parents in the lower middle-income group were more likely to have undergone interfamily marriages than the middle and upper middle income group, respectively (85.9% vs 10.2% vs 3.9%, $p < 0.001$) in our study. Eighty of the 9,757 subjects had an ocular disease with a potential genetic basis. Microcornea and retinitis pigmentosa was significantly associated with the first-cousin relationship between the parents compatible with the subject study. However, the high prevalence of consanguinity did not impact the prevalence of ocular malformations, which contradicts our results (Kumaramanickavel et al., 2002). Education and awareness led to a significant decline in consanguinity rates in industrialized countries, with rare prevalence in the United States of America and European countries (Nirmalan et al., 2006; Oniya et al., 2019). Immigrants from consanguineous communities (Turkey, Pakistan, and the Middle East) increased the proportion of congenital abnormalities and consanguineous marriages in Europe, North America, and Australia (Singer et al., 2020). Bittles et al. conducted a study to determine the prevalence of consanguineous marriages and their repercussions on reproduction and child mortality across 11 cities in the Pakistan province of Punjab between 1979 and 1985; he concluded that the consanguinity related deaths were relatively higher. The 9,520 women interviewed reported 44,474 pregnancies, with data collected on maternal and paternal ages at marriage, abortions/miscarriages, stillbirths, and deaths in the first month, at 2–12 months, and 2–8/10 years. Marriages contracted between spouses related as second cousins or closer accounted for 50.3% of total marriages (Bittles et al., 1993).

Congenital cataract was the most frequently reported disorder, affecting 35.9% of the study population. The statistical evaluation showed a significant correlation between consanguinity and congenital cataract ($p = 0.007$). This finding underscores the hereditary nature of the disease, where recessive genetic mutations from consanguineous

parents elevate the risk of lens opacity in offspring. Given that early-onset cataract can lead to severe visual impairment or blindness, early detection and surgical intervention remain essential. Furthermore, the association with socioeconomic status suggests that limited access to healthcare resources exacerbates the prevalence of this disorder among the lower middle-income group.

Retinoblastoma, a malignant tumor of the retina, was observed in 6.52% of cases. The statistical model employed in the study confirmed that both age ($p=0.0009$) and consanguinity ($p=0.007$) were contributing factors. Given that retinoblastoma is often linked to genetic mutations, its prevalence among children of consanguineous parents suggests an autosomal recessive inheritance pattern exacerbated by inter-family marriages. The findings emphasize the need for genetic counseling and early screening, particularly in populations with a high prevalence of consanguinity.

Although less frequent, congenital blindness and ligneous conjunctivitis were both significantly associated with consanguinity ($p=0.05$). The presence of these rare anomalies further highlights the multifactorial genetic risks imposed by inter-family marriages. Given that congenital blindness is irreversible, preventive strategies, including awareness campaigns and genetic testing, should be implemented to mitigate the impact of these hereditary disorders.

Microcephaly, a condition characterized by an abnormally small head circumference, was also observed among the studied patients. This condition is often linked to developmental delays, intellectual disabilities, and associated ophthalmological abnormalities such as optic nerve hypoplasia, strabismus, and refractive errors. The study suggests that microcephaly has a strong correlation with consanguinity and genetic predisposition. Given its impact on neurodevelopment, children with microcephaly should undergo comprehensive ophthalmological evaluations to detect and manage associated visual impairments at an early stage.

Cardiomyopathy, a disorder affecting heart muscle function, was another significant condition among the patients who underwent ophthalmological evaluations. Genetic forms of cardiomyopathy, particularly those linked to mitochondrial and metabolic disorders, are often associated with ocular manifestations such as pigmentary retinopathy, ptosis, and ophthalmoplegia. The findings from this study indicate that children with cardiomyopathy may present with concurrent ophthalmological abnormalities, necessitating multidisciplinary care for effective management. The presence of both cardiac and ocular defects in these patients further highlights the genetic basis of congenital anomalies seen in consanguineous populations.

Unlike congenital cataract and retinoblastoma, congenital glaucoma did not demonstrate a statistically significant association with consanguinity ($p=0.21$) or age ($p=0.24$). This suggests that congenital glaucoma may be influenced by other genetic or environmental factors rather than inheritance from closely related parents. While the exact etiology remains uncertain, this finding underscores the need for further research to delineate the genetic versus non-genetic contributors to congenital glaucoma.

Novelty Contribution and Sociocultural Mechanisms

A pivotal aspect of this study is the demonstrated link between socioeconomic status and congenital anomalies, specifically establishing socioeconomic status as a mediating pathway for consanguinity. The lower middle-income group

had the highest rate of consanguinity (85.9%), compared to 10.2% in the middle-income group and only 3.9% in the upper middle-income group. This socio-demographic trend indicates that individuals from lower socioeconomic backgrounds are more likely to engage in inter-family marriages, which, in turn, increases the prevalence of hereditary ophthalmological disorders. This is often driven by sociocultural mechanisms such as the preservation of family wealth, maintaining social ties, and a lack of awareness regarding genetic risks due to lower literacy levels. Limited healthcare access, lack of genetic counseling, and cultural practices all contribute to the sustained rise in congenital anomalies in these populations.

The psychological impact on parents dealing with children affected by congenital anomalies is profound. Many parents experience emotional distress, financial burden, and prolonged caregiving responsibilities. The study highlights the need for multidisciplinary care, including psychological support, medical intervention, and social awareness programs, to assist affected families. Additionally, implementing premarital genetic screening policies, similar to those in Syria and Lebanon, may help reduce the incidence of genetic disorders in regions with high consanguinity rates. A study was conducted in Birmingham (England) to compare the pervasiveness of congenital anomalies in consanguineous versus non-consanguineous groups, which turned out to be nine percent and three percent, respectively. The most common consanguinity prevalence was among Pakistanis settled in the UK (Roy et al., 2020). M. A. Hashmi conducted a descriptive study and hospital-based survey on 5,000 families of military personnel in Pakistan. Prevalence of congenital malformations in children of consanguineous parents was 40% (1,530 out of 3,820), particularly first cousins, whereas among non-related parents was 26% (305 out of 1180) ($p<0.01$) (Badr, 1972).

The study's findings provide critical insight into the association between socioeconomic status, consanguinity, and congenital ophthalmological anomalies. A comprehensive evaluation of 184 pediatric patients revealed that congenital cataract was the most common anomaly, followed by squint and retinoblastoma. The analysis of the dataset emphasized the profound impact of consanguinity, with a prevalence rate of 69.6% among affected children. The strong correlation between lower socioeconomic status and higher rates of inter-family marriages further accentuates the inherited nature of these congenital disorders. The study underscores the significant role of consanguinity and socioeconomic status in the prevalence of congenital ophthalmological disorders. The strong association between consanguinity and congenital cataract, retinoblastoma, congenital blindness, ligneous conjunctivitis, microcephaly, and cardiomyopathy indicates the necessity for public health interventions, including education, genetic counseling, and improved healthcare access. Addressing these issues through policy changes and awareness campaigns could help mitigate the burden of hereditary eye diseases in populations with a high prevalence of inter-family marriages. A child with congenital anomalies requires a continuum of care on the part of parents and hospital staff. During the study, families encountered with more than one offspring being affected by a genetic disorder; hence agony of the family was compounded and incomprehensible for the average parents in society. To reduce the risk of genetic disorders, the government should formulate policies like premarital screening, and genetic education, which address such issues and promote awareness about genetically transmitted diseases because of consanguineous marriages. Pediatric

patients with congenital ophthalmological malformations showed a more significant proportion of consanguineous marriages between parents than non-consanguineous marriages. Congenital anomalies in ophthalmological patients are more than we usually realize and are on the rise consistently due to a lack of awareness both in numbers and spectrum. The congenital anomalous patient requires a team effort for management. The psychological impact on the affected child's parents is very high and they need prolonged psychological support to prepare them to fight prolonged illness and care.

Limitations and Future Research

This study's cross-sectional design limits our ability to establish definitive causality, although our causal inference modeling provides strong indications. Furthermore, while the psychological impact on parents was acknowledged, minimal psychosocial data was collected, limiting the depth of understanding of this aspect. Future research should consider longitudinal studies to further solidify causal relationships and include comprehensive psychosocial assessments to better address the holistic needs of affected families. Further research is also needed to delineate the genetic versus non-genetic contributors to congenital glaucoma, given the absence of an association with consanguinity and age in this study.

CONCLUSIONS AND RECOMMENDATION

This study underscores a significant association between consanguinity, socioeconomic status, and congenital ophthalmological anomalies in the pediatric population of Pakistan. The high prevalence of inter-family marriages, particularly among lower middle-income groups, was found to contribute to an increased incidence of congenital disorders such as congenital cataract, retinoblastoma, congenital blindness, and lignant conjunctivitis.

Our findings notably reveal that age and socioeconomic status act as mediating factors, with consanguinity serving as a critical link between genetic vulnerability and disease manifestation, providing a novel and in-depth understanding of these complex interrelationships. The absence of association between consanguinity and congenital glaucoma highlights the need for more nuanced etiological investigations for specific disorders. The psychological and financial burden borne by families affected by such anomalies further emphasizes the need for systemic interventions. These include premarital genetic counseling, targeted awareness campaigns, and improved access to healthcare in underserved populations.

Ultimately, this study reinforces the importance of integrating public health policy with community education to mitigate the hereditary and societal impact of consanguineous marriages and promote early diagnosis and comprehensive care for affected children, with significant potential for implementing these findings in local and regional policies.

DECLARATIONS

Funding: None

Conflicts of interest/Competing interests: None

Ethics approval: The study was approved by the ethical research committee of Armed Forces Institute of Ophthalmology, Pakistan.

Consent to participate: Informed consent was obtained from all participants.

Consent for publication: Consent for publication of the findings was obtained from all participants.

Availability of data and material: The datasets used and/or analyzed during the current study are available from the corresponding author on reasonable request.

Code availability: Not applicable.

Authors' Contributions

Dr. Sana Abbas: Conception of the study, data collection, and initial manuscript drafting.

Dr. Waqar Muzaffar: Critical review and proofreading of the manuscript.

Author Biographies

Dr. Sana Abbas is a Consultant Anaesthetist with over 40 peer-reviewed publications. Her research focuses on perioperative care and patient safety in anaesthesiology.

Dr. Waqar Muzaffar is a Consultant Ophthalmologist and Vitreo-Retinal Surgeon. He has a keen interest in congenital anomalies, particularly those involving ocular abnormalities. His work often explores the systemic implications of ophthalmological defects.

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