RESEARCH PAPER

Impact Of Parental Consanguinity On Visual Acuity And Color Vision Outcome In Their Offspring

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Abstract Consanguinity refers to offspring of partners who are second cousins or closer. It is associated with a higher risk of congenital anomalies. Visual acuity and color vision are fundamental aspects of vision, critical for daily activities. Aim: To evaluate visual acuity and color vision outcomes in the offspring of consanguineous marriages and compare them with those from non-consanguineous marriages. Materials and Methods: This cross-sectional study included 100 offspring of consanguineous and non-consanguineous couples each. All participants underwent a comprehensive ocular evaluation including visual acuity assessment, refraction,intraocular pressure measurement, and color vision testing. Detailed ocular, systemic,and family history including parental consanguinity was recorded. Results: Reduced visual acuity (<6/9) was observed in two participants (2%) in the consanguineous group, with equal distribution among males and females, while no cases were identified in the non-consanguineous group. Color vision deficiency (CVD) was significantly higher in the consanguineous group (9%) compared to non-consanguineous group (1%) (p <0.05). Males constitutedthe majority of affected individuals in the consanguineous group. Deutan defects were the predominant type of CVD in both groups. In the consanguineous group, 7% exhibited deutan and 2%protan defects, whereas in the non-consanguineous group, only one male participant demonstrated a deutan defect, with no protan defects observed. Conclusion: Offspring of consanguineous marriages show a significantly higher prevalence of color vision deficiency, highlighting the role of genetic factors, while visual acuity remains largely unaffected. Early screening and genetic counseling may help in timely identification and management of inherited visual disorders.

Key words Consanguinity; Color vision deficiency; Visual acuity; Deutan defect; Protan defect; Genetic eye disorders;Optometry; Offspring

How To Cite This Article: Qumar S, Vyas M, Taneja M, Mishra SK. Impact of parental consanguinity on visual acuityand color vision outcome in their offspring. Int J Drug Deliv Technol. 2026;16(8s): 646-651; Doi: 10.25258/Ijddt.16.8s.69.

1. INTRODUCTION:

Consanguineous marriage refers to a union between partners who are second cousins or closer, characterized by an inbreeding coefficient (F) ≥ 0.0156 (Hamamy et al., 2012). This cousin or interfamilial marriage has been practiced globally. They are preferred in some communities, populations, and regions. Globally, one billion people belonging to communities prefer consanguineous marriages (Sandridge at al., 2009).

This cultural propensity towards consanguinity is prevalent globally, with an estimated 1 in 3 marriages in certain regions, with higher prevalence in the Middle East, South Asia, and parts of Africa. where the practice is culturally endorsed (Rajavi et al., 2015)

The prevalence varies within populations and communities. It varies with the culture, ethnicity, religion and geography. In India, the prevalence of consanguineous marriage has been reported to be 9.9%, with the highest prevalence observed in the southern region (23%) and the lowest in the north-eastern region (3.1%) (Sharma at al., 2020).

Consanguinity is associated with a higher risk of congenital anomalies and autosomal recessive diseases, as well as higher chances of miscarriages and under-five mortality in first cousin marriage (Anwar at al., 2020). Studies demonstrate that offspring from consanguineous unions have heightened chances of being affected by autosomal recessive diseases, congenital heart defects, neurological disorders, and metabolic syndromes (Evans et al., 2020; Abdulrazzaq et al., 1997). Additionally, the implications of inbreeding include increased rates of miscarriages and childhood mortality rates. Specifically, in first cousin marriages, the impact on paediatric health is noticeably pronounced, affecting not only physical well-being but also developmental milestones, including cognitive abilities (Anwar at al., 2020). elevated frequencies of congenital abnormalities among children of consanguineous parents, emphasizing a correlation between first-cousin marriages and the prevalence of major malformations (Abdulrazzaq et al., 1997; Majeedet al., 2014; Jaber et al., 1992). Consanguineous marriages are associated with the risk of variety of conditions, including congenital heart defects, neurological disorders, and metabolic syndromes (Bouadil et al., 2022; Temaj et al., 2022).

Visual acuity and color vision are fundamental aspects of vision, critical for daily activities, learning, and occupational performance (Moafa et al., 2021; Male et al., 2023). Vision, a critical component of daily functioning, learning processes, and overall quality of life, is directly influenced by genetic factors inherited from parents. Visual acuity and color vision are crucial aspects of visual health that affect an individual's interaction with their environment (Bocchi et al., 2015). The mechanisms through which these deficiencies manifest involve complex interactions of genetic mutations, particularly in cone photoreceptors responsible for color perception and visual acuity (Hollander et al., 2010). The interplay between genetic inheritance and visual performance is especially relevantin the context of consanguinity. Conditions such as color vision deficiencies (CVD) and visual anomalies have been documented among consanguineous offspring, suggesting that inherited traits significantly impact ocular health (Zafar et al., 2025; Kavitha et al., 2023).This study investigates the association between parental consanguinity and visual performance, focusing on both visual acuity and color vision, including gender-specific differences and the prevalence of deutan defects (absence or anomalous L-opsins in long-wavelength-sensitive cone cells) and protan defects (absence or anomalous M-opsins in medium-wavelength-sensitive cone cells)..

2. AIM:
To evaluate the impact of parental consanguinity on the visual performance of their offspring by assessing visual acuity and color vision, and to determine the prevalence and pattern of color vision deficiencies (CVD) among offspring of consanguineous and non-consanguineous marriages.

3. MATERIAL AND METHODS:
This cross-sectional observational study was conducted on 100 offspring of consanguineous marriages and 100 of non-consanguineous marriages. Patients above the age of 5 years both males and females except those with ocular diseases or systemic conditions affecting vision were enrolled into the study. Ocular complaints were noted, and a detailed history, including ocular, systemic, and family history, was obtained. Visual acuity was assessed, and refraction was performed. The 38 plates pseudoisochromatic Ishihara test was employed to check color vision. The history of parental consanguinity was recorded. Intraocular pressure was measured, and a slit-lamp examination was performed for anterior segment while indirect ophthalmoscope for posterior segments was carried out to rule out any ocular pathology affecting visual acuity or color vision (Fig. 1).

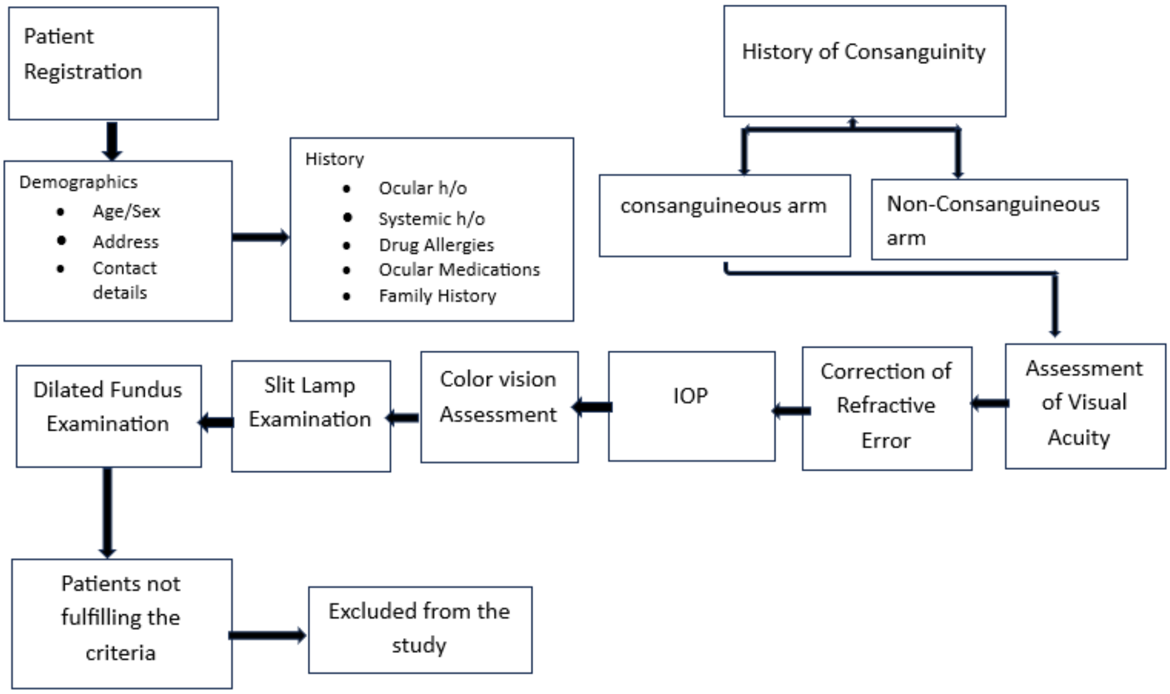

Figure 1. *Flowchart*

2.1. Study Design
The study was designed as a cross-sectional observational study, All the participants were provided with the patient's information sheet and taken a prior informed written consent. The purposive sampling technique was used and consecutive patients visiting the tertiary hospital were included in the study.

2.2 Testing sets
The color vision was assessed using Pseudoisochromatic Ishihara plates (38 plates). The test consists of a series of 38 plates called pseudoisochromatic Ishihara plates. Each plate is made up of a solid circle of dots of different colors and sizes (*Eye Magazine…*, 2022). Dots within the circle are arranged in such a manner that they form either a number or a figure that can be appreciated by a person with normal color vision. There are different types of test plates. The very first plate is a demonstration plate exhibiting the number 12 on it. The plate is included for demonstration purposes only. The number on this plate can be read by all people with normal or deficient color vision. Next to it, transformation plates are there. Individuals with CVD appreciate different numbers or figures as compared to those with unaffected color vision. The test also includes vanishing plates that are detectable only by individuals with normal color vision. Hidden digit plates can be recognized by color-deficient individuals and by those with normal color vision. It further carries diagnostic plates that are designed to rule out the type and severity of color vision deficiency and determine if it is protanopia or deuteranopia. At the end, we have tracing plates where individuals are asked to trace lines across the plates (*Color Vision…*, 2023; *Ishihara…*, 2023).
The test was performed in a well-lit room with fluorescent bulbs to give accurate results and faster recognition speed.
The plates were held at a distance of 50-100 cm, and the patient was given up to 3 seconds to recognize the number on the plate. In order to avoid prior remembrance, the test was done in a random order of plates and the response was noted.
Visual acuity was checked using Snellen chart comprised of eleven rows of block letters called

optotypes arranged with an increasing number of letters and a decreasing letter size. The height and width of each optotype are five times the thickness of the line composing the optotype. The line thickness and interline spacing each subtend an angle of 1 minute of arc, whereas the full optotype subtends an angle of 5 minutes of arc. The standard Snellen chart is designed for a distance of 6 meters, or 20 feet. The smallest line read by a person on the chart was recorded as his or her visual acuity.

2.3. Statistical analysis

Data were compiled and analyzed using the Statistical Package for the Social Sciences (SPSS), version 26. Descriptive statistics were employed to summarize the data. Categorical variables such as gender distribution, presence of visual acuity reduction, and color vision deficiency (CVD) among consanguineous and non-consanguineous offspring presented as frequencies and percentages.

The chi-square test was employed to determine the association between parental consanguinity and visual parameters, including the prevalence of reduced visual acuity and color vision deficiency. Statistical significance was set at $p < 0.05$.

The results indicated a statistically significant association between parental consanguinity and the occurrence of color vision deficiency ($p = 0.0098$), whereas the difference in visual acuity between the two groups was not statistically significant ($p > 0.05$).

3. RESULTS:

Two hundred participants were enrolled in the study, equally divided into offspring of consanguineous (n = 100) and non-consanguineous (n = 100) marriages. Participants had a mean age of 14.2 ± 6.1 years. Gender distribution was balanced in both groups, with 50% males and 50% females in the consanguineous group and non-consanguineous group.

Visual acuity analysis revealed that only 2 individuals, 1 Male and 1 female of consanguineous offspring had reduced visual acuity (<6/9) compared to the non-consanguineous group with no male or female were affected. Among consanguineous offspring, males and females were equally affected (2% vs. 2%), whereas in the non-consanguineous group, the distribution was 0% males and 0% females (Table 1).

Table 1: *Visual Acuity in Offspring of Consanguineous vs Non-Consanguineous Marriages (Visual acuity <6/9)*

Group	Total	Males Affected	Females Affected	Total Affected
Consanguineous	100	1	1	2 (2%)
Non-Consanguineous	100	0	0	0 (0%)

Color vision deficiency (CVD) was observed in 9 offspring of Consanguineous marriages out of which 8 were males while only one was female. 9% of consanguineous offspring versus 1% of non-consanguineous offspring, suggesting a statistically significant difference ($p < 0.05$). Among affected participants in the consanguineous group, males showed the majority (16%) compared to females (2%). In the non-consanguineous group, males with CVD were 2%, and females 0% (Figure 2) (Table 2).

Table 2: Color Vision Deficiency (CVD) in Offspring of Consanguineous vs Non-Consanguineous Marriages

CVD	Consanguineous	Non-Consanguineous	Chi-square test	P – Value
No	92	99		
			6.664	0.001
Yes	8	1		

Analysis of CVD type demonstrated that deutan defects predominated in both groups. In the consanguineous group, 7% of CVD cases were deutan and 2% protan. In the non-consanguineous group, 2% were deutan and 0% protan (Table 3).

Table 3. Comparison of Types of Color Vision Deficiency (Deutan and Protan) between Offspring of Consanguineous and Non-Consanguineous Marriages

Group	Total	Male	Female	Deutan	Protan	Total CVD
Consanguineous	100	8	1	7 (6 Males, 1 Female)	2 (Males)	9 (9%)
Non-Consanguineous	100	1	0	1 (Male)	0	1 (1%)

In the consanguineous group, seven cases were deutan (six males, one female) and two were protan (both males), whereas in the non-consanguineous group, only one deutan case (male) was observed and no protan defects were detected (Figure 3).

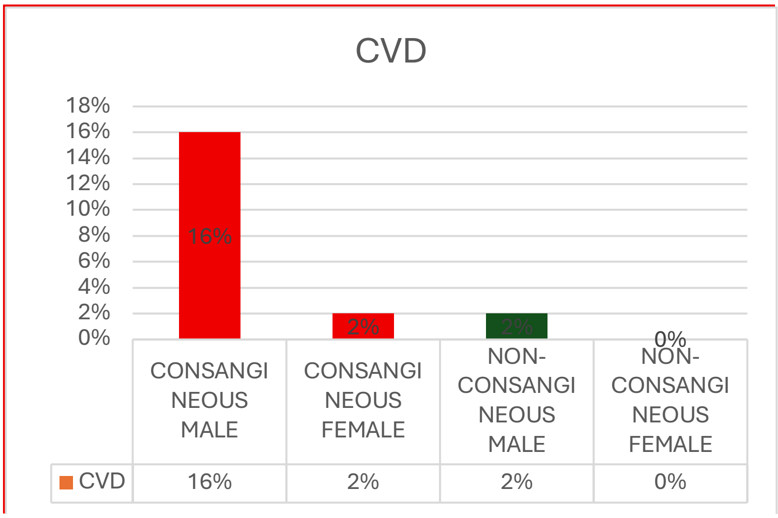

***Figure** Error! No text of specified style in document.**2.** Gender-wise distribution of the Color Vision Deficiency in Offspring of Consanguineous vs Non-Consanguineous Marriages*

Overall, offspring of consanguineous marriages demonstrated higher prevalence of both reduced visual acuity and color vision deficiency, with greater severity and a higher proportion of males affected.

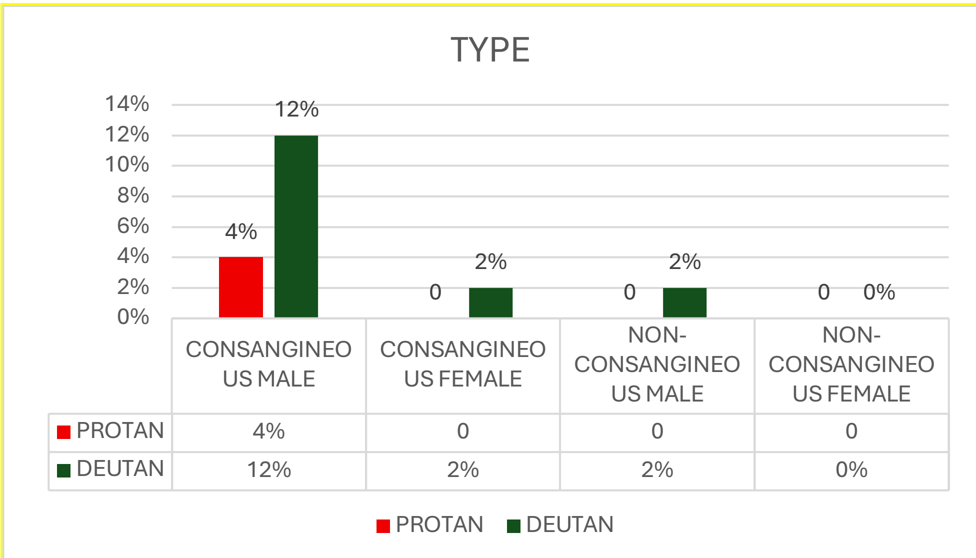

***Figure 3.** Types of Color Vision Deficiency in offspring of consanguineous vs non-consanguineous marriages.*

4. DISCUSSION:

This study demonstrates that parental consanguinity is associated with a higher prevalence of color vision deficiency among offspring, particularly in males, in line with the X-linked inheritance of red–green color vision deficiencies. The predominance of deutan over protan defects aligns with previous observations (Fareed et al., 2015) (Oriowo et al., 2008) (Aprioku et al., 2019). Visual acuity was largely unaffected, suggesting that consanguinity primarily impacts inherited color vision traits rather than visual acuity.

The findings emphasize the importance of early ophthalmic screening and genetic counseling in populations with high rates of consanguinity. Detecting epidemiological CVD early can allow appropriate educational and occupational planning and provide guidance for family planning decisions. Although this study was limited by its cross-sectional design and absence of molecular genetic testing, it offers valuable insight into the epidemiology of visual function in consanguineous populations. Future studies incorporating genetic analysis may further elucidate specific mutations responsible for these defects.

5. CONCLUSION:

The study indicates a notable prevalence of color vision deficits among individuals from consanguineous families, underscoring the potential impact of genetic factor while the vision acuity remained unaffected. These findings underscore the importance of early screening and genetic counseling in at-risk populations to mitigate the effects of inherited visual impairments.

Limitations

The study was limited by a relatively small sample size and a specific geographical setting, which may limit the generalizability of the findings.

Scope of the study
The study focused on evaluating visual acuity and color vision among individuals from consanguineous and non-consanguineous families within a specific regional population. The findings serve as a foundation for future large-scale and multi-regional studies to further explore these associations.

Ethical approval: Ethical approval was obtained from the Institute Ethics Committee.

Conflict of Interests: The authors declare that they have no financial or personal relationships that could be construed as a potential conflict of interest regarding the work presented in this manuscript. The research was conducted independently without any external funding or influence from commercial entities.

Acknowledgement: We would like to thank the staff of the Institute for their assistance in participant recruitment and data collection. Special thanks to the patients who participated in this study, making this research possible.

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