

ORIGINAL ARTICLE

Assessment of demographic features of congenital glaucoma subjects

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ABSTRACT:

Background: Due to increased rates of consanguinity, congenital glaucoma, a potentially blinding pediatric eye condition, is more common in developing nations. The present study was conducted to assess the demographic features of congenital glaucoma subjects. **Materials & Methods:** 120 cases (240 eyes) of congenital glaucoma of both genders were selected. Parameters such as age at first presentation, symptoms at first presentation, laterality of the disease, presence of consanguinity, family history of congenital glaucoma, maturity of the fetus at delivery, and maternal age at conception was recorded. **Results:** Primary congenital glaucoma was seen in 76 males and 74 females and secondary congenital glaucoma in 50 males and 40 females. Among PCG and SCG, positive family history was seen in 76 and 20, consanguinity in 34 and 36 and prematurity in 8 and 12. The mean age at first presentation (days) was 116.2 and 94.2 and mean maternal age at conception (days) was 28.4 and 26.2 respectively. The difference was significant ($P < 0.05$). **Conclusion:** Since early intervention is typically curative, pediatricians should be able to identify this disease and refer patients to a specialist as soon as possible. It is also critical to acknowledge the need for more comprehensive research on the identification of gene abnormalities linked to congenital glaucoma.

Keywords: congenital glaucoma, consanguinity, prematurity

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INTRODUCTION

Due to increased rates of consanguinity, congenital glaucoma, a potentially blinding pediatric eye condition, is more common in developing nations.¹ Congenital glaucoma is defined as a juvenile developmental disorder that is linked to atrophy, globe expansion (buphthalmos), corneal edema, Haab striae, high intraocular pressure, and optic nerve cupping.²

Regarding ocular and systemic relationships or the main ocular anatomic site implicated in the condition, several classification techniques for congenital glaucomas have been proposed.^{3,4} Congenital glaucoma is divided into primary and secondary forms using the previous classification approach. When other ocular and systemic diseases are absent, primary congenital glaucoma, often referred to as solitary trabeculodysgenesis, usually manifests as an isolated idiopathic developmental defect of the trabecular meshwork.⁵ A contributing ocular or systemic pathology is a contributing factor in secondary congenital glaucoma. Although primary congenital glaucoma has been identified as the most prevalent

kind in the past, there are few studies that show the incidence of particular entities.⁶ One of the very few disorders that necessitates rigorous follow-up and requires multiple specialists to monitor the patient throughout their life is congenital glaucoma. Therefore, identifying the potential risk factors may be useful for preventing or detecting this public health issue early.⁷ The present study was conducted to assess the demographic features of congenital glaucoma subjects.

MATERIALS & METHODS

The study was carried out on 120 cases (240 eyes) of congenital glaucoma of both genders. All gave their written consent to participate in the study.

Data such as name, age, gender etc. was recorded. Parameters such as age at first presentation, symptoms at first presentation, laterality of the disease, presence of consanguinity, family history of congenital glaucoma, maturity of the fetus at delivery, and maternal age at conception was recorded. Results thus obtained were subjected to statistical analysis. P value < 0.05 was considered significant.

RESULTS

Table I Distribution of congenital glaucoma

Type	Male	Female	Total
Primary congenital glaucoma	76	74	150
Secondary congenital glaucoma	50	40	90
Total	116	114	240

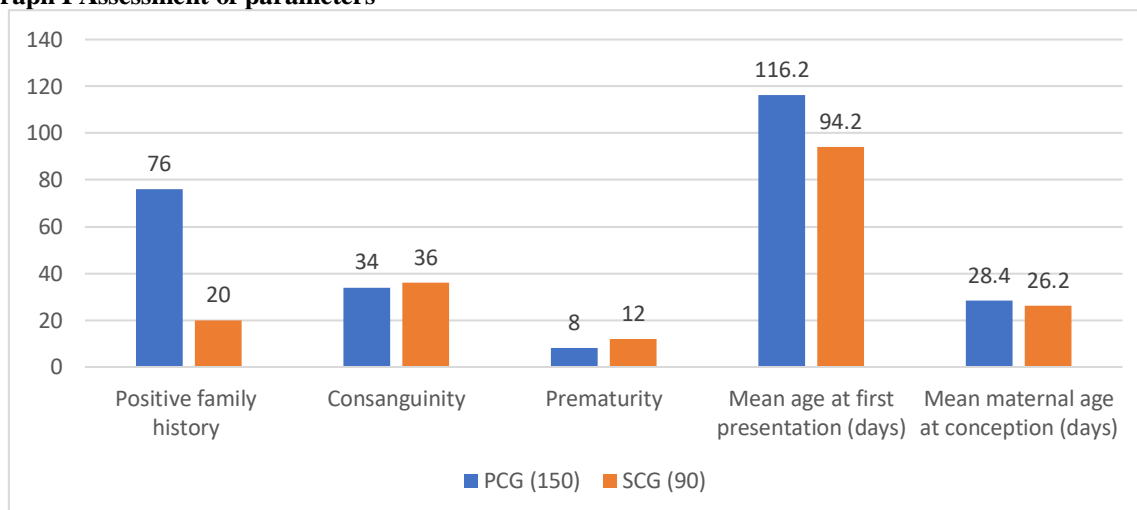
Table I shows that primary congenital glaucoma was seen in 76 males and 74 females and secondary congenital glaucoma in 50 males and 40 females.

Table II Assessment of parameters

Parameters	PCG (150)	SCG (90)	P value
Positive family history	76	20	0.01
Consanguinity	34	36	1
Prematurity	8	12	0.18
Mean age at first presentation (days)	116.2	94.2	0.04
Mean maternal age at conception(days)	28.4	26.2	0.25

Table II, graph I shows that among PCG and SCG, positive family history was seen in 76 and 20, consanguinity in 34 and 36 and prematurity in 8 and 12. The mean age at first presentation (days) was 116.2 and 94.2 and mean maternal age at conception (days) was 28.4 and 26.2 respectively. The difference was significant (P< 0.05).

Graph I Assessment of parameters



DISCUSSION

Congenital glaucoma is a rare yet a preventable cause of blindness which is more frequent in populations where consanguinity is common.^{8,9} Many classification methods have been employed for congenital glaucoma, some of which are more useful for prognostic implications.^{10,11} The present study was conducted to assess the demographic features of congenital glaucoma subjects.

We found that primary congenital glaucoma was seen in 76 males and 74 females and secondary congenital glaucoma in 50 males and 40 females. Tamçelik N et al¹² demonstrated the demographic features of congenital glaucoma subjects. Analyzed data included diagnosis, age at first presentation, symptoms at first presentation, laterality of the disease, sex, presence of consanguinity, family history of congenital glaucoma, maturity of the fetus at delivery, and maternal age at conception. The data of 600 eyes of 311 patients were analyzed. The distribution of primary and secondary congenital glaucoma among the patients were 63.3% (n = 197) and 36.7% (n = 114), respectively. Of the 311 patients, 57.2% (n = 178) were male and 42.8% (n = 133) were female. The overall frequency of bilateral disease was 92.3% (n = 287). Overall rate of consanguinity and positive family history was 45.3% (n = 141) and 21.2% (n = 66), respectively.

We found that among PCG and SCG, positive family history was seen in 76 and 20, consanguinity in 34 and

36 and prematurity in 8 and 12. The mean age at first presentation (days) was 116.2 and 94.2 and mean maternal age at conception (days) was 28.4 and 26.2 respectively. Aponte et al¹³ described the incidence and clinical characteristics of childhood glaucoma. Thirty children were diagnosed as having glaucoma during the 40-year study period. The incidence of childhood glaucoma was 2.29 (95% confidence interval, 1.47-3.12) per 100,000 residents younger than 20 years, with the following types and incidences: 19 acquired (1.46/100,000; 0.80-2.12), 6 secondary (0.45/100,000; 0.08-0.82), and 5 primary glaucoma (0.38/100,000; 0.05-0.72). The birth prevalence of primary congenital glaucoma during the 40-year period was 1 per 68 254 residents younger than 20 years or 1.46 per 100,000 (95% confidence interval, 0.03-8.16). Twenty-four individuals with glaucoma suspect were also identified, yielding an incidence of 1.9 per 100,000 residents younger than 20 years (95% confidence interval, 1.14-2.66).

Vogt et al¹⁴ studied the possible etiological factors of isolated primary congenital glaucoma (IPCG). The study group consisted of 52 cases with IPCG compared to 52 matched control pairs without any defects, and 22,744 malformed controls with non-ocular defects and 37,837 population controls with no defect. Exposure data and family history were collected (i) prospectively by prenatal logbook and other medical records, (ii) retrospectively through a

structured questionnaire completed by mothers, and (iii) from supplementary information obtained by regional nurses visiting the homes of non-respondent mothers. Autosomal recessive inheritance of IPCG was suspected on the basis of sib occurrence and parental consanguinity in 15% of cases. The shorter gestational age (with high proportion of preterm birth), higher birth order, large proportion of births among unmarried women, low socioeconomic status, and high rate of unemployment may be related to Gypsy origin of at least 54% of cases of IPCG. They concluded that the higher incidence of IPCG in the Hungarian Gypsy population is associated with their inbreeding and the possible founder effect of a gene mutation.

The shortcoming of the study is small sample size.

CONCLUSION

Authors found that since early intervention is typically curative, pediatricians should be able to identify this disease and refer patients to a specialist as soon as possible. It is also critical to acknowledge the need for more comprehensive research on the identification of gene abnormalities linked to congenital glaucoma.

REFERENCES

1. Sarfarazi M, Stoilov I. Molecular genetics of primary congenital glaucoma. *Eye (Lond)* 2000;14:422-8.
2. Genčík A. Epidemiology and genetics of primary congenital glaucoma in Slovakia. Description of a form of primary congenital glaucoma in gypsies with autosomal-recessive inheritance and complete penetrance. *Dev Ophthalmol* 1989;16:76-115.
3. Martin SN, Sutherland J, Levin AV, Klose R, Priston M, Héon E. Molecular characterisation of congenital glaucoma in a consanguineous Canadian community: A step towards preventing glaucoma related blindness. *J Med Genet* 2000;37:422-7.
4. Mortemousque B, Amati-Bonneau P, Couture F, Graffan R, Dubois S, Colin J, et al. Axenfeld-Rieger anomaly: A novel mutation in the forkhead box C1 (FOXC1) gene in a 4-generation family. *Arch Ophthalmol* 2004;122:1527-33.
5. Vajsar J, Schachter H. Walker-Warburg syndrome. *Orphanet J Rare Dis* 2006;1:29.
6. Bornemann A, Pfeiffer R, Beinder E, Wenkel H, Schlicker U, Meyermann R, et al. Three siblings with Walker-Warburg Syndrome. *Gen Diagn Pathol* 1996;141:371-5.
7. Berry SA, Peterson C, Mize W, Bloom K, Zachary C, Blasco P, et al. Klippel-Trenaunay syndrome. *Am J Med Genet* 1998;79:319-26.
8. Comi AM. Pathophysiology of Sturge-Weber syndrome. *J Child Neurol* 2003;18:509-16.
9. Vogt G, Horváth-Puhó E, Czeizel AE. A population-based case-control study of isolated primary congenital glaucoma. *Am J Med Genet A* 2006;140:1148-55.
10. Kaur K, Mandal AK, Chakrabarti S. Primary Congenital Glaucoma and the Involvement of CYP1B1. *Middle East Afr J Ophthalmol* 2011;18:7-16.
11. eh R, Child AH, Sarfarazi M. Molecular genetics of primary congenital glaucoma. In: Traboulsi EI, editor. *Genetic Diseases of the Eye*. 2nd ed. New York: Oxford University Press; 2012. p. 295.
12. Tamçelik N, Atalay E, Bolukbasi S, Çapar O, Ozkok A. Demographic features of subjects with congenital glaucoma. *Indian J Ophthalmol* 2014;62:565-9.
13. Aponte EP, Diehl N, Mohny BG. Incidence and clinical characteristics of childhood glaucoma: A population-based study. *Arch Ophthalmol* 2010;128:478-82.
14. Vogt G, Horváth-Puhó E, Czeizel AE. A population-based case-control study of isolated primary congenital glaucoma. *Am J Med Genet A* 2006;140:1148-55.