Strabismus in Generations and Among Siblings

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ABSTRACT

Introduction: Strabismus has been observed among family members in involved families. The study was conducted to document manifest strabismus running in generations and among siblings of strabismic patients.

Methods: It was a hospital based prospective study conducted from May to November 2017. Strabismic patients with ocular deviation visiting Department of Paediatric Ophthalmology and strabismus were enrolled using specially designed proforma. Detailed ocular examinations were done and family history obtained regarding the presence of strabismus. Family tree was drawn for those with positive family history.

Results: A total of 78 participants completed the study with 48.7% males. Family history of strabismus was present in 56.4%. Strabismus was present in two generations in 45.45%, in three generations in 18.2%, 1st and 3rd generation in 18.2%, 1st, 2nd and 4th generation in 4.5% and among siblings only in 13.6%. Among those with positive family history, exotropia was present in 75% and esotropia in 25%. Among exotropes, manifest deviation was present in 47.7% and intermittent exotropia in 27.3%. Among esotropes, congenital esotropia was present in 9% and accommodative esotropia in 16%. First degree relatives were affected in 45.45% of strabismic patients among which 55% had manifest exotropia, 20% had intermittent exotropia, 15% had accommodative esotropia and 10% had congenital esotropia.

Conclusions: Strabismus was found in different generations in 56.4% strabismic patients. Two generations were involved in 45.45%, three generations in 18.2%, 1st and 3rd generation in 18.2%, 1st, 2nd and 4th generation in 4.5% and siblings only in 13.6%. Genetic factors appear to be significant in strabismus in Nepalese population and it requires to be substantiated with further larger studies.

Key words: Esodeviation; exodeviation; generations; siblings; strabismus

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INTRODUCTION

Strabismus has been observed among family members in the involved families as genetic factors also play a role in its development. Hippocrates had stated that bald persons had descended from bald parents, blue eyed persons from blue-eyed parents and strabismic children from squinting parents.[1]

About 30% of children born to a strabismic parent will themselves develop strabismus according to large scale studies.[2] At the same time, twin studies reveal a concordance rate for monozygous twin of 73% to 82% and 35% to 47% for dizygotic twins.[3,4] Less than 100% concordance suggests that intrauterine, perinatal or environmental factors alter the expression of the strabismic genotype.

Studies of heritability of strabismus, its running in generations and families are conducted in the western world only. Therefore, we aim to find out how strabismus runs in generations and among siblings in our population.

METHODS

It was a hospital based cross sectional quantitative study. The study duration was of seven months, from May to November 2017. Strabismic patients with manifest deviation with no associated syndromes visiting Department of Paediatric Ophthalmology and Strabismus in a tertiary level eye hospital of Kathmandu during the study period were enrolled. Especially designed proforma was used for documenting patient demographics and the clinical examination findings. Family history of the participants was obtained regarding the presence of strabismus in three generations. Of those strabismic participants with positive family history, family tree was drawn. However, two participants who could recall the positive family history of strabismus even in the fourth generation were also enrolled. Those strabismic patients who cannot give family history of strabismus were excluded from the study. Convenient sampling was done and informed consent was taken from the participant or guardian if minor for enrollment in the study. However, the family members with strabismus were not examined and only manifest deviation of the family members were documented. This is the limitation of the study. Data were entered in excel and analysis was done using SPSS (version 20) and data were expressed in frequency and percentage as per the need.

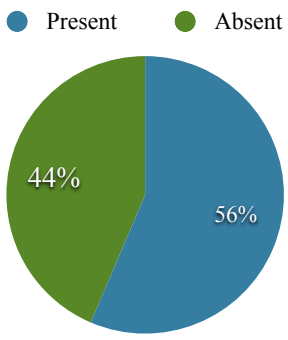

Figure 1. Family History of Strabismus

RESULTS

A total of 78 participants completed the study with 48.7% males and 51.3% females. Among them 52.56% were of Indo-Aryan origin and 47.44% were of Tibeto-Burman origin. Among the participants with positive family history, exodeviation was present in 75% and esodeviation in 25%.

First degree relative of patients with strabismus were affected in 45.45% among which 55% had manifest exotropia, 20% intermittent exotropia, 15% had accommodative esotropia and 10% had congenital esotropia. However, the family members were not examined to document whether they also had the same type of strabismus.

DISCUSSION

Thirteen percent of parents of strabismic probands had strabismus versus a 3% incidence in the general population.[5] Maumenee and associates

Table 1. Strabismus in Generations

Generation	Frequency	Percentage
2 Generations	20	45.45%
3 Generations	8	18.2%
1st and 3rd Generation	8	18.2%
1st, 2nd and 4th Generation	2	4.5%
Siblings only	6	13.6%
Total	44	100%

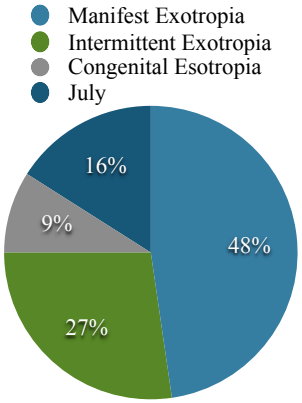

Figure 2. Distribution of Types of Deviation among those with positive family history

analysed the predegrees of 173 families containing probands with esotropia onset before the age of six months in the absence of major refractive error.[6] The results suggested multifactorial or Mendelian codominant inheritance pattern.

The examination of a cohort of 7100 strabismic patients from 12 published family studies had revealed that 2171 strabismic probands (30.6%) had a close relative with strabismus.[3] Families were usually concordant for either esotropia or exotropia. However, families with both forms have been reported. This finding may reflect the presence of two relatively common genes or one gene with variable expressivity.[6,7]

Ocular alignment also depends on several complex sensory and motor pathways in the retina, thalamus, brainstem and visual cortex apart from the development and functioning of extraocular muscles and orbit according to V Parikh et al.[8] The current evidence indicates the inheritance pattern of the common forms of strabismus is complex.[1,6,9] It is also likely that both genes and environment contribute to the occurrence of strabismus.[10] However, the heritability values for strabismus is likely to be underestimated as about 15 - 20% of strabismus is associated with non-ocular conditions like low birth weight, global central nervous system defects[11] and there can also be incomplete detection of phoria, microtropias and monofixation syndromes. This can be the reason why only 56.4% of strabismic patients had strabismus running in generations and among siblings in the present study.

It has been found that esotropia is more common in the white population of United States and Europe[11,12] while exotropia is more frequent in the Asian population and among black populations of United States and Africa.[3,12,13] This can be the reason why exotropes were more common in the present study also.

The advanced maternal age, cigarette smoking during pregnancy and low birth weight (< 1500 gm) also contributed to the risk of strabismus according to Collaborative Perinatal Project.[14] However, when corrected for these environmental risk factors, the odds ratio for heritability of concomitant strabismus remained significant.[15] However, we have not looked for the association between strabismus and the history of smoking and advance maternal age.

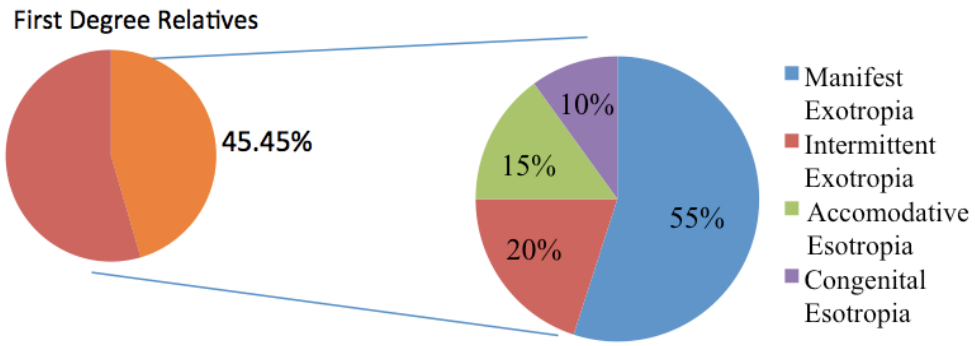

Figure 3. Distribution of types of deviation among those with strabismus in first degree relative

The relative risk for first degree relatives of an affected proband with common strabismus is estimated to be between three and five.[8,14,16,17] In the present study also, first degree relatives were affected in 45.45%.

First degree relatives of patients with hypermetropic accommodative esotropia was affected 26.1%, with infantile esotropia 14.9%, with anisometropic esotropia 12.1% and with exotropia 4% in a study by N G Zikas et al.[18] However, in our study, patients with first degree relatives affected included manifest exotropia in 55%, intermittent exotropia in 20%, accommodative esotropia in 15% and congenital esotropia in 10%.

Sclossmann and Priestley found that 47.5% of their patients with strabismus, 48.9% with esotropia and 36.8% with exotropia, belonged to families with two or more affected members.[7] Aurell et al. found that 17.6% of babies born into families with a first degree relative affected by convergent strabismus developed constant or intermittent esotropia by age of six years.[19] Another study has reported the familial incidence of strabismus to be as high as 65%.[20]

As only the family members with manifest deviation of strabismic participants were studied, the presence of strabismus may be underestimated in the present study. Apart from that, other ocular conditions, central nervous system status, intrauterine, perinatal and environment factors also play a role in the development of strabismus. These can be the reason why only 56.4% of strabismic patients had family history of strabismus in the present study.

CONCLUSIONS

Strabismus was found in different generations in 56.4% strabismic patients among which 75% were exotropes and 25% were esotropes. Two generations were involved in 45.45%, three generations in 18.2%, 1st and 3rd generation in 18.2%, 1st, 2nd and 4th generation in 4.5% and siblings only in 13.6%. Genetic factors do have role in strabismus and our findings support the same in Nepalese population. However, our finding requires to be substantiated with further larger studies.

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Conflict of Interest: None declared

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