ORIGINAL ARTICLE

MATERNAL CONSANGUINITY: THE MOST PROBABLE CAUSATIVE FACTOR OF GENETIC EYE DISORDERS

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ABSTRACT

Background: The consanguineous relation between parents could be either maternal or paternal. The objectives of the study were to determine the frequency distribution by type of genetic eye disorders & demographics.

Material & Methods: This cross-sectional study was conducted in Gomal Centre of Biochemistry and Biotechnology, Gomal University, D.I.Khan, Pakistan from June 2014 to September 2014. A sample of 152 cases was selected by consecutive technique. All the children having a congenital eye disorder in one or both eyes were eligible for inclusion in the study. The clinical data and related information was recorded on a structured Performas. Data collection site was Tehsil Kabal, District Swat. Demographic variables were gender, and consanguinity. Research variable was frequency distribution of genetic eye disorders. All data being nominal was analyzed by frequency and relative frequency.

Results: Out of 152 cases, 78(51.3%) were males and 74(48.7%) females. The strabismus was observed in 42 (27.6%) patients, cataract in 39(25.6%), extreme myopia in 32(21.1%), microphthalmia in 13(8.5%), anophthalmia in eight(5.3%), astigmatism in nine(5.9%) and nystagmus in six(3.9%) patients, while keratoconus, glaucoma and retinitis pigmentosa were in single(0.7%) patient each. Out of 42 cases with strabismus, 24 were with maternal and 18 were with paternal consanguinity. Out of 39 cases with cataract, 22 were with maternal and 17 were with paternal consanguinity. Out of 32 cases with extreme myopia, 21 were with maternal and 11 were with paternal consanguinity. Ninety two patients were having maternal & 60 paternal consanguinity.

Conclusion: Strabismus was the most frequent genetic eye disorder in Tehsil Kabal. Maternal consanguinity was the most commonly associated factor with genetic eye disorders. It is suggested that the people should prefer non-consanguineous marriages.

KEY WORDS: Consanguinity; Genetics; Microphthalmia; Data collection.

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INTRODUCTION

It is estimated that globally > 285 million people are visually disabled.[1,2] The WHO 2002 visual impairment survey, found that 90% of ophthalmic impairment cases were in developing countries. Cataract is the most common cause of visual impairment in the majority areas of the world (47%), while glaucoma (12%), age-related macular degeneration (9%), diabetic retinopathy (5%), corneal opacities (5%) and trachoma account for (4%) cases.[3] According to WHO, 1.4 million children less than 15 years of age are blind and most of these cases are suffering from avoidable blindness like cataract and trachoma. The average expenditure on the visual disorders has reached to $35.4 billion in United States.[4] In 1998, WHO aimed to eliminate the avoidable blindness by 2020 termed as Vision 2020 (the Global Initiative for the Elimination of Avoidable Blindness).[5]

Due to high ratio of consanguineous marriages, inherited eye disorders are becoming a serious issue in Khyber Pakhtunkhwa population. The consanguineous relation between parents could be either maternal or paternal. As the consanguineous relation get closer between the parents, the chances of inheriting identical copies of one or more defective genes is increased, which may ultimately

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cause inherited/ genetic disorders including eye conditions. Biologically, consanguineous relation between parents can be either maternal or paternal. Such kinds of marriages are more common in many countries from Middle East and South Asia. This high ratio of consanguineous marriages in Pakistan is the result of prevailing genetic disorders.[6] Due to high ratio of consanguineous marriages, inherited eye disorders are becoming a serious issue in Khyber Pakhtunkhwa population.

Etiologically eye disorders originate either from genetic factors or through environmental injuries. These visual impairment disorders include retinitis pigmentosa, keratoconus, cataract, nystagmus, astigmatism, glaucoma, microphthalmia, anophthalmia, strabismus and myopia. These disorders affect the structure & function of the eye.[7]

According to by Pakistan National Blindness and Visual Impairment Survey, Pakistan has estimated 1,140,000 blind adults.[8] In another study through Pakistan National Blindness and Visual Impairment Survey, Jadoon and his colleague has determined that Pakistan has approximately 570,000 adult persons affected with cataract. Afterwards, the same study group performed an association study of blindness with poverty and determined that blindness occur more severely in poor rural areas.[9]

Ethnically the people of Swat are Pathan who follow their custom of consanguineous marriages, and hence the people are segregating different kind of inherited disorders such as; anemia, allergy, epilepsy, kidney malfunction, thalassemia, cardiac diseases, haemophilia, mental retardation, osteoarthritis and variety of ophthalmic disorders.

The objectives of the study were to determine the frequency distribution by type of genetic eye disorders & demographics.

Table 1: Genetic eye disorders in Tehsil Kabal, Swat (n=152)

S. No	Disease Type	Count	%age
1	Strabismus	42	27.6
2	Cataract	39	25.6
3	Extreme Myopia	32	21.1
4	Microphthalmia	13	8.5
5	Astigmatism	09	5.9
6	Anophthalmia	08	5.3
7	Nystagmus	06	3.9
8	Keratoconus	01	0.7
9	Glaucoma	01	0.7
10	Retinitis Pigmentosa	01	0.7
Total		152	100

MATERIAL AND METHODS

This cross-sectional study was conducted in Gomal Centre of Biochemistry and Biotechnology, Gomal University, D.I.Khan, Pakistan from June 2014 to September 2014. A sample of 152 cases was selected by consecutive, non-probability technique. All the children having a congenital eye disorder in one or both eyes were eligible for inclusion in the study. Detailed history regarding risk factors was sought. The clinical data and related information was recorded on a structured Performa. Data collection site was Tehsil Kabal, District Swat. Demographic variables were gender & consanguinity. Our research variable was frequency distribution by type of genetic eye disorders.

Gender had two attributes of male and female. Consanguinity had two attributes; maternal consanguinity and paternal consanguinity. Types of genetic eye disorders had 10 attributes; Strabismus, cataract, extreme myopia, anophthalmia, microphthalmia, astigmatism and nystagmus, keratoconus, glaucoma and retinitis pigmentosa. Gender, consanguinity, and disease types were all nominal variables.

Data was analyzed by descriptive analysis plan. All data being nominal was analyzed by frequency and relative frequency. Analysis was done through SPSS (version 16.0) (SPSS Inc., Chicago, Illinois).[10]

RESULTS

Out of 152 cases, 78 (51.3%) were males and 74 (48.7%) females with a male to female ratio of 2.1:1.9).

The frequency of strabismus was observed in 42 (27.6%) patients, cataract in 39 (25.6%) patients, extreme myopia in 32 (21.1%) patients 13 (8.5%) persons of microphthalmia, 8 (5.3%) individuals with anophthalmia, 9 (5.9%) patients of astigmatism and 6 (3.9%) patients of nystagmus, while keratoconus, glaucoma and retinitis pigmentosa were observed in single patient (0.7%)each. (Table 1)

Out of 42 cases with strabismus, 24 patients were reported with maternal consanguinity between their parents, while 18 patients were having paternal relation. Out of 39 cases with cataract, 22 were having maternal consanguinity, while 17 were having paternal relation. Out of 32 cases with extreme myopia, 21 were having maternal while 11 were having paternal consanguineous relation. Ninety two patients were having maternal & 60 paternal consanguinity (Table 2).

DISCUSSION

Strabismus was the most widespread disorder (27.6%). However, glaucoma, keratoconus and retinitis pigmentosa were the least common disorders. Whereas in a screening of genetic eye diseases in China, the prevalence of strabismus was 0.58%, degenerative myopia was found as 0.95%,

Table 2: Consanguinous relation between parents of genetic eye disorders in tehsil Kabal, Swat (n=152)

S. No.	Disease Type	Maternal	Paternal	Total
1	Strabismus	24	18	42
2	Cataract	22	17	39
3	Extreme Myopia	21	11	32
4	Microphthalmia	9	4	13
5	Astigmatism	5	4	9
6	Anophthalmia	5	3	8
7	Nystagmus	4	2	6
8	Keratoconus	1	0	1
9	Glaucoma	1	0	1
10	Retinitis Pigmentosa	0	1	1
Total		92	60	152

congenital cataract as 0.037%, retinitis pigmentosa 0.03%, congenital nystagmus 0.025 %, congenital microphthalmos 0.009%, and congenital glaucoma 0.004%.[11]

Maternal consanguinity was considered as the most influential factor. According to study in China the mode of inheritance of strabismus in 425 persons with the familial occurrence rate in first, second, and third degree relatives was 90%, 2.2%, and 1.1% respectively. The heritability of strabismus was calculated to be 18.3%.[11]

CONCLUSION

Strabismus was the most frequent genetic eye disorder in tehsil Kabal. Maternal consanguinity was the most commonly associated factor with genetic eye disorders. It is suggested that the people should prefer non-consanguineous marriages.

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CONFLICT OF INTEREST
Authors declare no conflict of interest.
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AUTHORS' CONTRIBUTION

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