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RELATIONSHIP OF GLC3A WITH CONGENITAL GLAUCOMA IN PAKISTAN

[1]Dr. Zagham Hammad, [2]Dr Umer Abdul Rasheed, [3]Dr Abdul Hassan

[1]Medical Officer, District Headquarter Hospital Nankana Sahib

[2]BHU Gojra, Daska Sialkot

[3]Nishtar Medical University & Hospital Multan

Abstract:

Objectives: *The main idea of this research is to know about the role of a gene to initiate the inborn eye disease which leads to blindness due to abnormality in the optic nerve in the residents of Pakistan.*

Methods: *The participants of study were 29 and 3 families. They all were affected from inborn eye diseases due to fault in the gene in 2017. This genetic research was carried out in two different departments of biology departments of GC University, Faisalabad. The models from patients suffering of disease were evaluated for having two same alleles of the same gene for a pair of identical gene. The arrangement of the persons having the same gene was carried out with the use of primers. MLINK along with the marker was used to check the LOD.*

Results: *An eye disease due to the abnormality in the optic nerve which sometimes leads to the blindness is caused by the GLCA3 in most of the cases. Three families were the part of this research. Two families out of three were found with the disease.*

Conclusions: *This research discovered that the gene GLC3A locus on the chromosomes is the essential reason of the eye disease in the people of Pakistan. The organizing the individuals having the same genes in them and arrangement of large number of families would be a landmark in this field for further studies.*

Keywords: *Optic nerve, MLINK, CYP1B1, inborn, biology, glaucoma.*

*** Corresponding author:**

Dr. Zagham Hammad,

Medical Officer, District Headquarter Hospital,
Nankana Sahib

QR code

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INTRODUCTION:

Eye disease due to the abnormality of the optic nerve known as glaucoma is the 2nd most ordinary disease in the world. It is the 3rd essential cause of blindness in n the world. A health study carried out in 2003 discovered that the ratio of loss of eyesight is less than percent in Pakistan. Glaucoma is one of the main reasons in Pakistan which is causing blindness in the people of our country. A study carried out in Britain proved that the occurrence of PCG in the children of Pakistan is much higher than other countries [1].

PCG is an eye abnormality of the infant age. It can be occurred due to the faults of different parts of eye as cornea, eye ball, optic nerve etc. PCG is also known as abnormality of the chromosomes (autosome). Hitherto, mapping of four types of locus has been carried out [2]. These four types are GLC1A to GLC1D and two types of gene has been discovered which are CYP1B1, LTBP2. In many inherited communities of the world, eighty to hundred percent occurrence of this eye diseases are due to the CYP1B1 mutations. In Pakistani communities, there is a very low quantity of information about gene available. This research was carried out to check the role of locus of GCL3A to primary congenital glaucoma.

METHODS:

The section of biology departments of GC University, Faisalabad was taken from the review boards of the departments. The willing was taken from all the participants of the study. The willingness of the parents was taken in case of infant. The PCG affected 3 families and 29 persons were selected for this research from the different areas of the country in 2017. Every participant of the study provided the description of their medical history. Different types of testing ware used for the medical evaluation of the patients in a hospital of Faisalabad. This research was a combined study between the two different educational institutes of biology located in Faisalabad.

Deoxyribos nucleic acid was taken from the models of the blood by using medical procedures. Polymorphic loci in DNA locator's exact location was gained by the use of a special map. Polymorphic loci in DNA locator's polymerase chain reaction carried out with fifty nanograms of deoxyribos nucleic acid in ten milligrams of different primers which are being used worldwide. This mixture contains different amounts of the elements with different percentages [3]. Magnification of this mixture was carried out with the help of a cycler by the use of the polymerase chain reaction code of behaviour. The temperature was higher for the very 1st cycle from the evaluated centigrade of the primers. This hardening temperature was shortening down about one centigrade for each increasing cycle [4]. It was from sixty-five centigrade to fifty-five centigrade after ten complete cycles. The hardening temperature of fifty-five centigrade was applied to the final twenty-five cycles. The organizing of the persons having same gene was carried out with the help of an analyzer specific for the gene. A mapped was used for the exact evaluation of the data.

RESULTS:

The abnormality which was found in two families was closely related to the gene GLC3A. Number one family was a short family which was taking part from Faisalabad. Three members of the family were affected by the diseases as mentioned in figure number one. The medical aspects of the PCG described some abnormal parts of the eye in all the sufferers of the family. The ratio of the abnormality in the eye parts was different in the patients of the family depending upon the age and other medical factors. One patient was found with visual perception on the movement of hand otherwise his both eyes were close to blindness. Treatment of this patient was not carried out from operation but a special medical treatment was prescribed for that patient. One affected child was ten year of age and he was on serious stage of the PCG. She was suffering of the enlargement of the eye ball and swelling of the cornea due to the accumulation of the fluid that causes shortening of one eye. Her visibility was also reduced to the movement with the hand and there was not any hint of the light.

Intraocular pressure in her case was thirty-two millimetres of mercury for the right eye and thirty millimetres mercury for the left eye. The genetic history of the family number one described that all the sufferers were having same characteristics and alleles of the gene. Less than three was the LOD for those persons. The next family which is described in the figure number two belonged to Jhang and it had 4 patients.

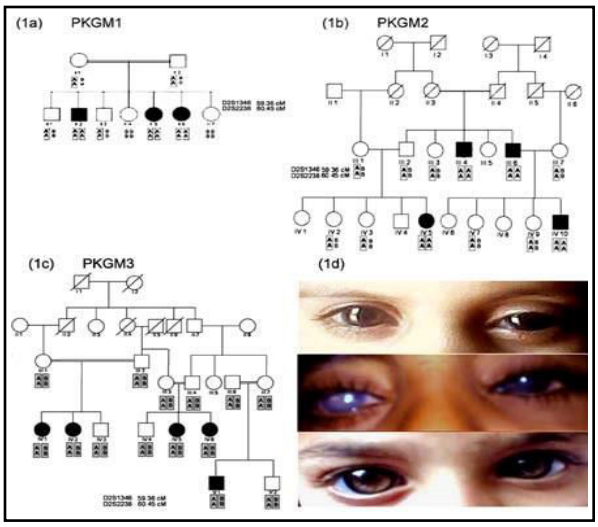

Fig.1: Families PKGM1, PKGM2 and PKGM3 with data of genotyping and images of patients' eyes.

- History of 1st family: A combination of alleles located closely together for the identifiers on the two copies of twenty-one number chromosomes.
- History of the 2nd family: Close allele's information at chromosome number 2 which have a close link with the gene GLC3A.
- History of the 3rd family: The group of organisms with the same gene was not attached with the gene GLCA3 until the markers were dissimilar alleles at loci chromosomes. Marshfield gene mapping was used for the distances of the markers.
- The eye phenotypes of the first family patients were found with enlargement of the eye ball and the swelling of the cornea in each eye. The patients of the second family were less severe in this case.

Some of the patients were unaware about their disease. So, they did not get any proper medical treatment for its rectification. They were suffering from this disease from the time of their birth. One infant patient of one year of age was found with intraocular pressure of twelve by twelve millimetres mercury. This patient had enlargement of the both eye balls.

Another affected patient was found with intraocular pressure of six millimetres mercury for the right eye and twelve millimetres mercury for the left eye [5]. This patient was suffering from swelling of the cornea and had a fear from light. She had to undergo proper treatment to shorten the continuous pain. All these participants were advised to take the anti-glaucoma medicines after the operation of the eye. The organizing of the patients and their healthy family members with the same gene was carried out. D2S2238 and D2S1346 identifiers proved that all the sufferers were homozygous in the identical genes. A high amount of the LOD can be supported by the big family but the affected patients were the reason of the low value of LOD. The 3rd family is the large congenital family which was brought in study from Sheikhupura as described in figure number one. The signs of the PCG were documented at the time of birth in all the participants. All the patients showed the medical aspects related to the PCG as swelling of the cornea, loss of large quantity of tears from eyes all the time, the large size of the eye balls and fear to face the light. Both the identifiers proved that all the members of the family and the patients were heterozygous; therefore, they were taken apart from the gene GLC3A. All the 29 sufferers of the primary congenital glaucoma were selected from the different cities of the Punjab province whose age was from one year to three years. The medical evaluation of the participant confirmed the high value of the intraocular pressure ranges from sixteen millimetres of mercury to thirty millimetres of mercury. Only five members out of twenty-nine were confirmed as homozygous.

Table: Prevalence of congenital glaucoma in relation to GLC3A locus within different populations of the world

Locus	Populations	Contribution to GLC3A locus (CYP1B1)	References
GLC3A	Gypsy	100%	Plasilova et al.[14]
	Indonesia	33%	Sitorus et al.[7]
	Saudi Arabia	80%-100%	Bejjani et al.[16]; Abu-Amero et al.[17]
	Iran	70%	Suri et al.[12]
	Brazil	50%	Stoilov et al.[11]
	India	23%	Chakrabarti et al.[13]
	Pakistan	23%	Firasat et al.[3]
	Pakistan	2/3 Familial cases 17% Sporadic cases	This report

DISCUSSION:

This was the very first study carried out on the genes related to the PCG in our country. The outcome of this research proved that gene GLC3A was playing an important role in the people of this country and CYP1B1mutation is caused by this very gene [6]. CYP1B1 is the main participant to the PCG in various groups of people in Pakistan. CYP1B1 is linked with the inherited communities of the countries such as Arabians, Gypsies and Turkey [7]. CYP1B1 mutation was discovered in 3 families out of 13 or twenty three percent families of Pakistan (related by blood) as described in table number one [8]. This research describes the role of the CYP1B1 in the patients of primary and seventeen percent of these participants were discovered as homozygous [9]. There was a great disparity in the PCG of two communities; one community got this through the inheritance and the other got deafness in addition.

CONCLUSION:

The consistent selection of the Patients to find out the other genetic abnormality for the primary congenital glaucoma will help on the further studies. The arrangement of the deoxyribose nucleic acid will also help in this matter in our country. The early diagnostic of the disease also plays an important role to tackle this problem.

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