CODEN [USA]: IAJPBB ISSN: 2349-7750

INDO AMERICAN JOURNAL OF
PHARMACEUTICAL SCIENCES

http://doi.org/10.5281/zenodo.1288752

Available online at: http://www.iajps.com Research Article

CONGENITAL ABNORMALITIES AND GENETICAL DISEASES IN A POPULATION OF MAYO HOSPITAL, LAHORE, PAKISTAN

[1]Sidra Ashiq, [2]Madiha Mubarik, [3]Hafsa Zafar

[1]WMO, DHQ Sheikhupura

[2]WMO, BHU Malian Kalan

[3]Services Hospital Lahore

Abstract:

Objectives: *The main idea of this research was to decide about the rate of genes abnormalities and by birth problems in the infants in clinical gene centre and two famous hospitals, in one of the important provinces of Pakistan.*

Methodology: *It was a selective type of research. Thirty-three thousand three hundred and eighty kids from an early stage of childhood to the age of eight years were selected. This research was carried out from 2000 to 2004. The exact information of birth diseases and gene abnormalities were recognized by checking the history of the family background about the same disease. Chromosomal and molecular characteristics of the cells and other important data were collected for further testing.*

Results: *The most common type of by birth abnormalities was instinctive fault in the chemical processes in which animals use food and water to grow and use energy. Seven points eight is the percentage of fault in instinctive metabolism process. The second most common complication in the born children is coronary heart faults. There are six percent patients of coronary heart defects.*

Conclusion: *The role of area from which the population is selected as participants is very important to understand the differences between this research and studies held on other populations. The special types of tests blood and urine are carried out to check the born abnormalities and genes faults. Extreme care is the basic requirement for this kind of testing. The outcomes of these types of studies provide the doctors and gene therapists with the donation of these born diseases to the society and main concern patients should be given priority during treatment.*

Key Words: *Instinctive, abnormalities, energy, coronary, metabolism, chromosome, molecule, cell, genes, congenital.*

Corresponding author:

Sidra Ashiq,

WMO,

DHQ Sheikhupura

QR code

Please cite this article in press Sidra Ashiq et al., ***Congenital Abnormalities and Genetical Diseases in a Population of Mayo Hospital, Lahore, Pakistan,*** *Indo Am. J. P. Sci, 2018; 05(06).*

INTRODUCTION:

Complications in the genes and new born babies are very dangerous. Most of these diseases are difficult to be treated. But some types of these complications are curable [1, 2]. The study of the family background to know about the history of disease, different types of therapies and gene analysis are very essential ingredients in some cases of patients. Malfunctioning of genes and by birth diseases are the cause of a significant number of patients in the whole world [3, 4]. Many innovations in the field of medicine have decreased the diseases which are dangerous for life. But some time later, the by birth diseases would be the imminent cause of deaths of the babies who are in their initial stage of childhood [5, 6]. There is complete non-availability of systematic study by birth and genetic diseases in the country of Pakistan [7]. We interrogated the communities made of different societies about the medical care of child patients to check the by birth and genes disorder rates among our communities.

PATIENTS AND METHODS:

The subject of this study was thirty-three thousand three hundred and eighty patients from new born baby to the age of eight years. In this traditional selective type research, different serious and mild nature disorders were observed in the patients. Both genders were the subject of this study. This research was carried out in children hospital from 2000 to the end of 2004. This medical centre was a major hospital and it was giving the treatments to large populations of a huge district of the province. The data about the changes occur in large group of people over a period of time and medical outcomes at the time of birth were gathered from maternity hospital and from the parents at the time of very first evaluation.

The exact information of the by birth diseases and genes disorders can be interpreted by checking the family history about the disease, different types of tests, the characteristics of chromosomes, the interest and benefits of the interrogator and the role of legacy in family. The results of Ultrasound, electrocardiograph and other important tests were gathered. The categorization of different discovered diseases is managed in 7 different tables and graphs. The patients were divided into the groups of one thousand people to check the occurrence of abnormality and diseases.

Special American made software was used to record the discovered information in the form of charts, graphs and checklists. This information was also checked by this new SPSS ver 11.5 software. The use of this software makes the data arrangement very fast and easy to handle in collecting the information from thousands of patients.

RESULTS:

This research work provides with the information of rate and outcomes of by birth diseases and genetic faults in thirty-three thousand three hundred and eighty children from new born baby to eight years old. 4 years period is covered by this research. Table no 1 to ten are used to describe the medical characteristics found in our participants.

Total discovered cases of congenital anomalies were nine hundred and two. The rate of congenital anomalies in the males was in large quantity than the CA found in females. Five hundred and thirty-nine were males and three hundred and sixty-three were females out of total nine hundred and two. The most common type in our participants was the malfunction in the metabolism of the amino acids; it was seven-point eight percent. It was followed by six percent of the CHD. According to table no 1, faults of chromosomes resulted in Down's syndrome. All the congenital anomalies found in our research with their names and percentages of the patients involved are mentioned in the table no 1 to 10. The findings of our research were matched with the record of the hospitals. So, dual time check of all the information was carried out for valid information.

Table-I: Comparative occurrence of Chromosomal abnormalities

Diagnosis/Disorder	M	F	Cases	%
Mental Retardation	3	3	6	0.20
Fragile X syndrome	1	-	1	0.02
Down's syndrome	21	14	35	1.00
Wolf Hirschhorn	1	-	1	0.20
Total	26	17	44	1.3

Table-II: Comparative occurrence of disorder of in born error of metabolism

Abnormalities	M	F	Cases	%
Hypercalcemia	41	20	61	2.00
Hypocalcemia	38	23	61	2.00
Cholesterolemia	24	3	27	0.90
Galactosemia	2	-	2	0.06
MPS	8	4	12	0.60
Tyrosiemia	1	-	1	0.02
PKU	4	3	7	0.20
Favism(G6PD)	14	10	24	0.70
Hypourecemia	10	7	17	0.50
Acidosis	7	2	9	0.20
Total	149	72	221	7.18

Table-III: Comparative occurrence of Central Nervous System (CNS)

Anomalies	M	F	Cases	%
Meningomyelocele	8	9	17	0.50
Anencephaly	4	3	7	0.20
Omphalocele	13	5	18	0.50
Spinabifida	4	-	4	0.10
Hydrocephaly	13	8	21	0.60
Microcephaly	20	17	37	1.10
Cerebral palsy	8	8	16	0.50
Epilepsy Seizures	5	4	9	0.26
Total	75	54	129	3.76

Table-V: Comparative occurrence of external genital anomalies and disorder of gonadal and internal reproductive ducts

Disorders	M	F	Cases	%
Abnormalities	17	-	17	0.50
in testis Hypospadias	47	-	47	1.40
Cryptorchidism	21	-	21	0.70
Epispadias	2	-	2	0.05
Bladder extrophia	1	-	1	0.02
Renal Urinary	4	-	4	0.01
tract Turner	-	4	4	0.01
syndrome Ambiguous genitalia	6	5	11	0.5
Total	98	9	107	3.3

Table-VI: Comparative occurrence of sensory disorders

	M	F	Case	%
Hereditary	13	11	24	0.70
deafness Blindness	4	5	9	0.30
Total	17	16	33	1.00

Table-VII: Comparative occurrence of thorax, respiratory and abdominal anomalies

Anomalies	**M**	**F**	**Cases**	**%**
Esophageal atresia	16	9	25	0.75
Laryngomalacia	5	3 0	8	0.23
Hirschsprung's disease	17	7	34	1.00
Gastroschisis	3	2	5	0.14
Anal Atresia	6	2	8	0.23
Total	47	23	70	2.35

Table-VIII: Comparative occurrence of haematological disorders

Thalasemia minor	8	6	14	0.40
Thalasemia major	4	2	6	0.20
Hemophilia	6	--	6	0.20
Von-Willebrands disease	1	--	1	0.02

Table-IX: Comparative occurrence of skeletal and muscle, neuromuscular disorders

	M	**F**	**Case**	**%**
Marfan syndrome	1	-	1	0.02
Osteogenesis Imperfecta	2	-	2	0.05
Achondroplasia	1	-	1	0.02
Spondyloepiphysial dysplasia	-	-	1	0.02
Polydactyly	4	2	6	0.05
Ectrodactyly	1	-	1	0.02
Meckel syndrome	1	-	1	0.02
Pierre Robin syndrome	3	1	4	0.05
Amyotrophic lateral sclerosis	1	1	2	0.05
Duchene muscular dystrophy	6	-	6	0.20
Total	20	5	25	0.5

Table-X: Comparative occurrence of miscellaneous

M		***F***	***Case***	***%***
Congenital	90	112	202	6.00
Hearth defects Waardenburg	1	-	1	0.02
Syndrome Tubersclerosis	2	-	2	0.05
Vater Association	1	1	2	0.050
Total	94	113	207	6.12

DISCUSSION:

According to the consequences of this research, the rate of the congenital anomalies was two-point nine percent. The finding of this research is being very much closer to other four studies on congenital anomalies carried out in four different hospitals in Pakistan [8]. Different countries have different percentages of congenital anomalies in their countries.

In this research, the occurrence rate of slow growth and progress of mind and faults of chromosomes was one-point three percent per thousand new births. One ratio one lakh is the occurrence of tyrosinemia in our people (table no 2). Point zero two per thousand is its rate in our research which is much lower than the other studies.

Nervous system abnormalities are point five percent per thousand new born in this research as mentioned in table no 3 which is much less than several studies performed in the same field. The rate of the cut palate and cut lips in USA one percent per seven hundred, China six-point eleven percent per ten thousand, Korea one point eighty-one per thousand, Brazil point nineteen per thousand, Ireland one point fourteen per thousand and point eighty eight percent in Pakistan. In this study, we found cut lips in our participants and their rate is similar to other studies performed for the same abnormality.

The hearing abnormality is discovered in different studies from one to three cases per thousand infants. The occurrence of the hearing abnormality in USA from one point four to three per thousand and one point four to two point one per thousand. In the current study, one thousand and thirty-three patients of hearing abnormality were observed. The data about the deafness is given in table no 6 with twenty-four patients. The disability of the eyes was two-point three percent to eight percent in the infants. The occurrence of blindness is point three percent in this research as mentioned in table no 6. Environment or gene factor can be the cause of this abnormality.

The most frequent type of the congenital disorder is the malfunctioning or deformation of the bones was discovered in different hospitals of Denmark, South Africa, Bahrain, India and Tehran. The ratio of the skeletal disorder was least common against the general contemplation and its percentage was point thirty three percent among the participants. Different studies of CHD in new born babies have been done in various countries of the world. Those countries gave different outcomes which are different from the result of this research which is six percent as mentioned in table no 10. Gene and medical characteristics are involved in the cases of heart failures. There is a little knowledge in our communities about the diseases of heart.

CONCLUSIONS:

The different outcomes which describe the genes disorder and other by birth diseases are interrogated in the country of Pakistan. Some of the diseases are not discovered at the time of infancy and come into appearance in later part of life, proper care, therapies by specialists are some important issues in finding of different results. This non-similarity can be explained by the discovery of serious and mild congenital anomalies; the high amount of cases involved and non-equal ratio of both genders. It is very vital to note that social differences, economic differences, different types of diet are those aspects which cannot be ignored.

REFERENCES:

1. Ahmed, Suhaib, et al. "Screening extended families for genetic hemoglobin disorders in Pakistan." *New England journal of medicine* 347.15 (2002): 1162-1168.
2. Al-Gazali, Lihadh, Hanan Hamamy, and Shaikha Al-Arrayad. "Genetic disorders in the Arab world." *Bmj* 333.7573 (2006): 831-834.
3. Modell, Bernadette, and Aamra Darr. "Genetic counselling and customary consanguineous marriage." *Nature Reviews Genetics* 3.3 (2002): 225.
4. Modell, Bernadette. "Social and genetic implications of customary consanguineous marriage among British Pakistanis. Report of a meeting held at the Ciba Foundation on 15 January 1991." *Journal of Medical Genetics* 28.10 (1991): 720.
5. Ropers, Hans-Hilger. "New perspectives for the elucidation of genetic disorders." *The American Journal of Human Genetics*81.2 (2007): 199-207.
6. Ropers, Hans-Hilger. "New perspectives for the elucidation of genetic disorders." *The American Journal of Human Genetics*81.2 (2007): 199-207.
7. Angastiniotis, Michael, and Bernadette Modell. "Global epidemiology of hemoglobin disorders." *Annals of the New York Academy of Sciences* 850.1 (1998): 251-269.
8. Shafi, T., M. R. Khan, and M. Atiq. "Congenital heart disease and associated malformations in children with cleft lip and palate in Pakistan." *British journal of plastic surgery* 56.2 (2003): 106-109.