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INCIDENCE OF CONGENITAL DISEASES IN PATIENTS PRESENTING TO PAEDIATRIC DEPARTMENT OF MAYO HOSPITAL, LAHORE

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Abstract:

Objective: *The study aims to find the incidence of congenital abnormalities among less than 8 years paediatrics population.*

Methodology: *3380 children belonging to <8 years were included in this study. Study was conducted from 2010 to 2015. The information related to gene related diseases was taken by detailed history. Family history was also taken in detail about disease. Investigations were performed to find out the chromosomal abnormalities related to the disease.*

Results: *Thee most common chromosomal abnormality related to diseases was instinctive fault in chemical processes. 7.8 percent had this abnormality. Second most common cause was coronary heart related, making 6% of total population. Conclusion: The incidence of congenital diseases is variable among different populations, so understudy population was selected carefully. Investigations were performed for confirmation of disease and special care was taken while drawing sample and performing tests. Patients with main disease concerns were prioritized in study for treatment.*

Keywords: *congenital diseases, incidence, population, abnormalities.*

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INTRODUCTION:

Congenital disorders are defined as those disorders which are present by birth, transferred from generation to generation due to genetic mutations. Reduced rate of cousin marriages can help to prevent the occurrence of such diseases [1,2].

Incidence of congenital diseases in different in different races. Congenital abnormalities occur norandomly according to Vectral theory [3]. The gene studies help in confirming the incidence of congenital heart diseases[4]. Due to unavailabily data in our population about incidence of various congenital anomalies, a 5 years surey was conducted on pediatric population, in order to find out incidence of various anomalies. The antenatal checkup and ultrasonography can also help us detecting the anomalous child, which can be confirmed by using several tests during intrauterine life.

METHODOLOGY:

Population understudy included less than 8 years children including neonates, 3380 in number. Multiple life threatening and mild congenital disorders were studied in this study. Research was conducted at Mayo Hospital Lahore from 2010 to 2015. The past history and maternal history was collected from the hospital where patient was born. To further confirm disease, investigations were performed, family history about the disease was taken. Ultrasonography, electrocardiography were also performed besides gene analysis. Patients were stratified in groups containing 1000 in each group.

American software to plot data into tables, graphs etc. was used. It was rechecked using SPSS version 11.5 which helped in categorization of data quickly.

RESULTS:

The understudy topic helps in collecting incidence of congenital diseases in children less than 8 years old. Study was conducted over a period of 4 years. Results obtained from study have been presented in the form of tables. 902 patients were found to have congenital abnormality. Males were more commonly suffering from it than females. The male to female incidence was 539 and 362 each, respectively. Amino acids metabolism abnormality was found to be most common, making 7.8% of total followed by congenital heart diseases which were 6%. All the data collected was compared with hospital data in order to avoid any error, and bring precise results in front.

Table 1: Incidence of Chromosomal abnormalities.

Disorder	Male	Female	Case	Percentage
Mental retardation	3	3	6	0.2
Fragile X syndrome	1	-	1	0.02
Down	21	14	35	1.0
Wolf Hirschhorn	1	-	1	0.2
Total	26	17	44	1.3

Table 2: incidence of inborn error of metabolism.

Amino acid metabolism disorder	Male	Female	Cases	Percentage
Hypercalcemia	41	20	61	2
Hypocalcemia	38	23	61	2
Cholesterolemia	24	3	27	0.9
Galactosemia	2	-	2	0.06
MPS	8	4	12	0.6
Tyrosinemia	1	-	1	0.02
PKU	4	3	7	0.2
Favism	14	10	24	0.7
Hypouricemia	10	7	17	0.5
Acidosis	7	2	9	0.2
Total	149	72	221	7.18

Table 3: central nervous system disorders

Disorder	Male	Female	Cases	Percentage
Meningomyelocele	8	9	17	0.5
Anencephaly	4	3	7	0.2
Omphalocele	13	5	18	0.5
Spina bifida	4	-	4	0.1
Hydrocephaly	13	8	21	0.6
Microcephaly	20	17	37	1.1
Cerebral palsy	8	8	16	0.5
Epilepsy	5	4	9	0.26
Total	75	54	129	3.76

Table: 4 reproductive organs disorders

Disorders	Male	Female	Cases	Percentage
Testicular abnormalities	17	-	17	0.5
Hypospadias	47	-	47	1.4
Cryptorchidism	21	-	21	0.7
Epispadia	2	-	2	.05
Bladder exrophy	1	-	1	.02
Renal disorder	4	4	8	.01
Turner	6	5	11	0.5
Total	98	9	107	3.3

Table 5: sensory disorders

Disorders	Males	Females	Cases	Percentage
Hereditary Deafness and Blindness	13 4	11 5	24 9	0.7 0.3
Total	17	16	33	1.0

Table:6 thorax, abdomen and respiratory tract abnormalities

Disorders	Males	Females	Cases	Percentages
Esophageal atresia	16	9	25	.75
Laryngomalacia	5	30	8	.23
Hirschsprungs	17	7	34	1.0
Gastroschisis	3	2	5	.14
Anal atresia	6	2	8	.23
Total	47	23	70	2.35

Table 7: hematological disorders

Disorder	Male	Female	Cases	Percentage
Thalassemia major	8	6	14	0.4
Thalassemia minor	4	2	6	0.2
Hemophilia	6	-	6	0.2
Von wilibrand disease	1	-	1	0.02

Table 8: skeletal, muscular and neuromuscular

Disorder	Male	Female	cases	percentage
marfan	1	-	1	.02
osteogenesis	2	-	2	.05
imperfecta	1	-	1	.02
achondroplasia	-	-	-	.02
spondyloepiphyseal	4	2	6	.05
dysplasia	1	-	1	.02
polydactylty	1	-	1	.02
ectrodactyly	3	1	4	.05
Meckel syndrome	1	1	2	.02
pierre robin syndrome	6	-	6	.02
amyotropic lateral sclerosis				
duschene muscular dystrophy				
total	20	5	25	0.5

Table 9: miscellaneous

Disorders	Males	Females	Cases	Percentage
CHD	90	112	202	6
Wardenburg syndrome	1		1	.02
Tuber sclerosis	2		2	.05
Vater association	1	1	2	.02
Total	94	113	207	6.12

DISCUSSION:

High prevalence of cousin marriages in Asian population is the major cause of high incidence of congenital anomalies [5,6]. Generation to generation transmission of such snomalies can be reduced by non- consanguineous marriages [7].

According to understudy title, results have clearly shown that the incidence of congenital anomalies is more in male population as compared to female population [8]. Besides that the incidence of diseases are also different. Most commonly occurring disease is amino acids metabolism disorder, followed by congenital heart diseases, other diseases according to their order of occurrence have been mentioned as follows; chromosomal disorders, inborn errors of metabolism, nervous system diseases, abdominal and thoracic anomalies, urinary tract anomalies and miscellaneous disorders, respectively.

Current study has been conducted over a large Pakistani population visiting a tertiary care hospital over a period of 5 years. Purpose behind this study was to find out the incidence of diseases among our population, as the incidence varies from population to population. Less research data was found in our population, that's y need was felt to conduct a survey to find the rate.

CONCLUSION:

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.

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