ORIGINAL ARTICLE

FREQUENCIES OF CONGENITAL ANOMALIES AMONG NEWBORNS ADMITTED IN NURSERY OF AYUB TEACHING HOSPITAL ABBOTTABAD, PAKISTAN

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Background: Congenital anomalies play a significant role in perinatal and neonatal morbidity and mortality. The frequency of these congenital anomalies varies in different populations. Objective of this study was to find out the frequencies of congenital anomalies admitted in nursery of Ayub Teaching Hospital, Abbottabad. **Methods**: In this descriptive, cross-sectional study all patients admitted in NICU from October 2009 to January 2010 were included. The patients were examined for major and minor congenital anomalies. The observations were recorded in tabulated form. **Results**: A total of 2,360 patients were admitted in NICU during the study period. One hundred patients were noted to have congenital anomalies. The most frequent anomalies involved the central nervous system (31%). Meningomyelocele was the commonest defect (71%, 22 out of 31 cases of CNS defects), among these males were more (77%, 17 out of 22 of meningomyelocele cases) than females (14 out of 31). These were followed by patients born with congenital heart defects (16%). Patients with urogenital anomalies (6%) were all male except for one who had ambiguous genitalia. **Conclusions**: Cases of meningomyelocele were the commonest presenting congenital anomaly. More stress should be laid on the role of peri-conceptional vitamin supplementation like folic acid for the primary prevention of congenital defects.
Keywords: congenital anomaly, birth defect, encephalocele, anencephaly, Down's syndrome, trisomy

INTRODUCTION

As many infectious diseases have been controlled by use of vaccines and antibiotics, congenital anomalies are increasing playing a significant role in neonatal mortality and morbidity.[1,2] Treatment and rehabilitation of these morbid children is difficult and costly.[2,3] Finding the variation in the frequency of congenital anomalies may be helpful for us in planning healthcare measures for possible prevention of such anomalies.[4] Congenital anomalies are a major cause of perinatal and neonatal death, both in developed and developing countries. They are presumed to be more prevalent in populations with cousin marriages.[5]

Congenital anomalies are either single isolated defects or present as multiple anomalies in a single individual. A syndrome is defined as a pattern of multiple abnormalities that are related by pathophysiology and result from common, defined aetiology.[6] Major congenital anomalies occur in approximately 2–3% of births with a variable frequency in different populations ranging from 1.07% in Japan[7] to 4.3% in Taiwan.[8]

Dysmorphology is the study of abnormalities of human form and the mechanisms that cause these abnormalities. It is estimated that 1 in 40, or 2.5% of newborns, have a recognisable malformation or malformations at birth. In about half the cases a single malformation is found, while the other half display multiple malformations.

About 10% of paediatric hospital admissions have genetic conditions, 18% have congenital defects of unknown aetiology. Forty percent of surgical admissions are patients with congenital malformations. About 20–30% of infant deaths and 30–50% of deaths after the neonatal period are due to congenital abnormalities.[6]

Major congenital anomaly/malformation is defined as a structural abnormality present at birth which has a significant effect on function or social acceptability; examples: ventricular septal defect, cleft lip. Minor congenital anomaly/malformationis defined as a structural abnormality present at birth which has minimal effect on clinical function but may have a cosmetic impact, e.g., preauricular pit, developmental variant/variation: a cosmetically and functionally insignificant structural deviation from the usual, of prenatal origin, and usually familial, e.g., 5th finger clinodactyly.[9] The history and physical findings should lead to an initial impression and differential diagnosis. These will guide selection of preliminary tests, the content of initial counselling of the family and development of an immediate plan for management, which can be modified as new information is developed and synthesised. Initial impression should fit into one of three categories[9]:

- Single (isolated) malformation
- Multiple malformations, recognisable pattern (syndrome identification)
- Multiple malformations, pattern not recognized

Structural anomalies are considered overt when they are visible on inspection, otherwise they are considered "occult". Considering the elimination or control of some infectious diseases, congenital anomalies are increasingly playing a major role in the mortality and morbidity of children.[4]

Congenital anomalies are a major cause of admission and prolonged stay in nursery; they are also an important cause of early and late neonatal deaths. The causes for these anomalies are multifactorial.[10] Forty percent (40%) cases are due to unknown causes, other commonly known causes include[11]:

- Chromosomal defects
- drugs
- chemotherapy
- radiation exposure
- cousin marriages

RESULTS

A total of 2,360 patients were admitted in NICU during the 16 months of the study period. One hundred patients were noted to have congenital anomalies. Fifty-seven babies were male and 41 were female (n=100), there was one baby with ambiguous genitalia and in one patient sex could not be established (Table-1), this baby had multiple congenital anomalies too (Photograph-1, 2). The most frequent anomaly was spina bifida. The commonest defect was meningomyelocele (22%), with one case of occipital encephalocele (Table-2). These were followed by anomalies of the heart, cases of congenital heart disease constituted 16% of the total anomalies. Twelve (12%) presented with gastrointestinal anomalies of which imperforate anus was the most common (3%). Nine percent presented with musculoskeletal anomalies out of which talipes equino-varus was the most common (6%) (Table-3).

Most of the admitted patients (40%) were discharged after necessary investigations and counselling, 25% expired, 20% were referred to other hospitals for further management, mostly including cardiac cases or surgical cases. All expired patients were in the early neonatal or perinatal age. Of these, 15% patients left without medical advice. In 68% of patients complications like prematurity, low birth weight, respiratory distress, sepsis, intestinal obstruction and seizures etc. were present. Thirty-one percent patients presented with multiple anomalies for example patient with Down's syndrome also presented with dextrocardia or congenital heart defects, based upon the obvious defects such cases were included in only one group to avoid repetition. Cross-tabulation was used to correlate variables of gender, type of defects and outcome of the patients (Table-4).

Table-1: Frequency table for sex of the baby

Gender	Number of patients	Percentage
Male	57	57
Female	41	41
Not established	2	2
Total	100	100

Table-2: Major systems involved in congenital anomalies

Congenital Anomaly (involving)	Number	%
Central nervous system	31	31.0
Cardio vascular system	16	16.0
Urogenital system	6	6.0
Respiratory system	4	4.0
Gastro intestinal system	10	10.0
Muscoloskeletal system	9	9.0
Chromosomal defects	5	5.0
Dysmorphism	8	8.0
Others	11	11.0
Total	100	100.0

Table-3: Type of defect (n=100)

Type of Defect	Number	%	M	F	A
Central nervous system	**31**	**31**	**17**	**14**	
Dandy walker malformation	1	1.0	-	1	
Meningomyelocele	22	22.0	13	9	
Anencephaly	1	1.0	1	-	
Sacrococcygeal teratoma	1	1.0	1	-	
Hydrocephalus	4	4.0	1	3	
Encephalocele*	1	1.0	0	1	
Generalized brain atrophy	1	1.0	1	-	
Cardiovascular system	**16**	**16**	**10**	**6**	
Congenital heart disease	16	16.0	10	6	
Urogenital system	**6**	**6**	**5**	**-**	
Congenital hydronephrosis	1	1.0	1	-	
Infantile polycystic kidney disease	1	1.0	1	-	
Pujo /hydronephrosis	1	1.0	1	-	
Renal agenesis	1	1.0	1	-	
Ambiguous genitalia	1	1.0	-	-	1
Hypospedias/micropenis	1	1.0	1	-	
Respiratory system	**4**	**4**	**2**	**2**	
Tracheoesophageal fistula	4	4.0	2	2	
Gastrointestinal system	**10**	**10**	**7**	**3**	
Gut diverticulosis	1	1.0	-	1	
High anal atresia	1	1.0	-	1	
Imperforate anus	3	3.0	3	-	
Duodenal atresia	2	2.0	1	1	
Omphalocele	2	2.0	2	-	
Esophageal atresia	1	1.0	1	-	
Musculo skelatal system	**9**	**9**	**6**	**3**	
Craniosynostosis	2	2.0	2	-	
Talipese equino-varus	6	6.0	4	2	
Arthrogryoposis multiplex congenital	1	1.0	-	1	
Chromosomal	**5**	**5**	**3**	**2**	
Down syndrome	5	5.0	3	2	
Dysmorphism	**8**	**8.0**	**1**	**6**	**1**
Cleft lip	**2**	**2.0**	**1**	**1**	
Cleft palate	**1**	**1.0**	**1**	**-**	
Colloidon baby	**7**	**7.0**	**3**	**4**	
Metabolic disorder	**1**	**1.0**	**1**	**-**	
Total:			**57**	**41**	**2**

M=male, **F**=female, **A**=ambiguous/un-established sex, *Photograph 4,5

Table-4: Sex of the baby, outcome of the patient, congenital anomaly, and cross tabulation

Congenital Anomaly	Sex of the Baby	Outcome of the Patient				Total
		Dis.	Ref.	L	Ex.	
CNS	Male	9	2	2	4	17
	Female	7	2	4	1	14
	Total	16	4	6	5	31
CVS	Male	6	2		2	10
	Female	3	2		1	6
	Total	9	4		3	16
Urogenital	Male	1	2	1	1	5
	not established		1			1
	Total	1	3	1	1	6
Respiratory	Male		1		1	2
	Female		2			2
	Total		3		1	4
GIT	Male	2	3		2	7
	Female	2			1	3
	Total	4	3		3	10
Muscoloskeletal	Male	2			4	6
	Female			1	1	2
	Total	2		1	5	8
Chromosomal	Male	1	1		1	3
	Female	1	1			2
	Total	2	2		1	5
dysmorphism	Male				1	1
	Female	2		4		6
	Not established				1	1
	Total	2		4	2	8
others	Male	2	1	2	1	6
	Female	2		1	3	6
		4	1	3	4	12

Dis=Discharged, **Ref**=Referred, **L**=LAMA, **Ex**=Expired

Photograph-1, 2: Baby with multiple anomalies and absent external genitalia, anal orifice was normal

Photograph-3: Large thoracolumbar meningomyelocele with ruptured sac

Photograph-4: Occipital encephalocele

Photograph-5: Anencephaly

Photograph-6: Congenital hydrocephalus

Photograph-7: Colloidon baby

Photograph-7: Baby with features of Down's syndrome, also having neonatal hyperbilirubinemia

Photograph-8: Baby with features of Trisomy 18

Photograph-9: Arthrogryoposis multiplex congenital

Photograph-10: Club feet/Talipese equino-varus

DISCUSSION

Congenital anomalies if overt can be picked up easily at birth by trained paediatricians, anomalies like congenital defects of the heart are apparent in seven to ten days even if not apparent at or soon after birth. Sometimes patient are informed beforehand about the anomalies on antenatal ultrasounds, most common of these include hydrocephalus renal anomalies, heart defects, and anomalies of the lungs so that antenatal counselling can be done and necessary management plans can be laid out.

In a study discussing the pattern of congenital malformations in consanguineous versus non consanguineous marriages in Iran published in the Health Journal , genitourinary anomalies were the most common followed by musculoskeletal and then cardiovascular.[12] Studies from eastern Saudi Arabia showed the most affected systems were the CNS, Musculoskeletal and renal. Males were more common than females.[10,13]

A study conducted on congenital malformations among newborns in Kenya reported that most common anomalies involved the musculoskeletal system followed by anomalies of the CNS, among which hydrocephalus was most common followed by anencephaly and microcephaly and then chromosomal in which Down's syndrome was very common. Polydactyly was found to be single most common malformation. More males than females were observed but difference was not statistically significant.[14]

In a study conducted in North-western Iran showed that Central Nervous system anomalies were the most prevalent with anencephaly being the commonest followed by spina bifida and then hydrocephaly. These were followed by defects of the musculoskeletal system among which club feet or talipeses equino varus were most common followed by polydactyly. Digestive system defects were next in which imperforate anus was most common and then came urogenital anomalies like hypospedias followed by chromosomal defects like Down syndrome. The results of our study were similar to this study done in north-western Iran except that in Iran more females than males were observed to have congenital anomalies but difference was not statistically significant.[4]

In Southern Iran a study to find out the patterns and frequencies of congenital anomalies in newborns reported that genitourinary anomalies were most common followed by musculoskeletal and then cardiovascular. Theses were followed by CNS anomalies with spina bifida being the commonest. In gastrointestinal system imperforate anus was most common. It was followed by chromosomal anomalies

in which, Down's syndrome was most common. Gender was not mentioned in this study.[5]

CONCLUSION & RECOMMENDATIONS

Meningomyelocele was the most common defect noted. More stress needs to be laid on prescribing pre-conception vitamin supplementations. Extensive collaboration between the obstetrician and paediatricians is required for antenatal diagnosis of many congenital anomalies so that proper parental education and counselling may be done.

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