www.jmscr.igmpublication.org
Impact Factor: 1.1147
IGM Publication Journal Of Medical Science And Clinical Research ISSN (e): 2347-176X
An Official Publication Of IGM Publication

Pattern of Congenital Anomalies in Neonatal Unit in Gharian Teaching Hospital

Ali. M .Ghareba, Samia. A. Abugella, Hanan. M. Alhadi

Department of Pediatrics, Gharian Teaching Hospital Gharian/ Libya
Department of pediatrics, Faculty of Medicine Al-jabal algarbi University

ABSTRACT

Congenital Anomaly is an anomaly that affects a body part or physiologic function and is present at birth.The exact antenatal prevalence of congenital anomalies in Libya is unknown. Early antenatal diagnosis of congenital anomalies is crucial for early counseling, intervention and possible fetal therapy. The objective of this study was to evaluate the pattern of congenital anomalies and malformations for all cases were admitted in neonatal care unit in our hospital. This hospital based prospective descriptive study highlights the point pattern of congenital anomalies in one year. The number of congenital anomalies were 73 cases more in males than Female (M: F = 1.5: 1%). The pattern of congenital anomalies included CVS (36.3%), GIT (16.5%), genitourinary (14.3%), musculoskeletal (11%), CNS (6.6%), respiratory (4.4%), etc. In CVS the most common anomalies were ventricular septal defect, atrial septal defect and patent ducts. In GIT group, commonly imperforated anus followed by esophageal Artesia. In musculoskeletal group, telipes was most common malformation followed by spinabifida. In CNS, group hydrocephalus was the most common malformation followed by anencephaly and meningomyeleceole. Frequency of congenital anomalies was more in vaginal delivery as compared to cesarean born babies (2.7 % vs. 1.58%). in still born as compared to live born babies (5.5 % vs. 1.5 %) Present study stress upon the importance to carrying out Congenital Anomalies through clinical examination of neonate at birth.

Keywords: Congenital, Abnormalities, Anomalies, NeonateCounseling,Malformation

Introduction:

A congenital anomaly (C A) is an abnormality of structure, functions or body metabolism that is present at birth (even if not diagnosed until later in life) and results in physical or mental disability, or is fatal. (1)

Congenital anomalies contribute a significant proportion of infant morbidity and mortality, as well as fetal mortality. Individual development begins with fertilization and extends into postnatal life at least until processes of growth give way to those of maintenance. Its progress and direction are determined by genetic and environmental factors acting singly and in combination. Developmental anomalies include all anatomical, physiological, or biochemical deviations arising during development, especially during organogenesis (including placentogenesis), irrespective of cause. This discussion is confined to events occurring in intrauterine life. (2)

A congenital anomaly is considered to be multifactorial (or polygenic) in origin when there is a combined influence of (a number of) genes and environmental factors that interfere with normal embryologic development. Multifactorial inheritance is considered when there appears to be a genetic component but there is no clear Mendelian pattern of inheritance. Multifactorial inheritance is the underlying etiology of most of the common congenital anomalies.(3)

Each year, eight million children are born worldwide with congenital anomalies, of which 3.3 million die before the age of five; 3.2 million of the survivors may be mentally and/or physically disabled. [4] The prevalence of birth defects is comparable all over the world; about 3% in the United States, [5] 2.5% in India, [6] and 2% to 3% in the United Kingdom. [7] The most prevalent conditions include congenital heart defects, [8] orofacial clefts, Down syndrome, [9] and neural tube defects.

Causes of Congenital Anomalies

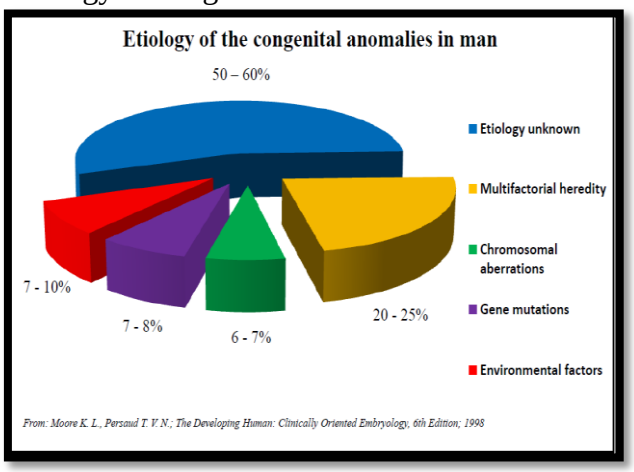
In spite of the frequency of congenital anomalies, the underlying causes for most remain obscure. It has been estimated that around 15%-25% are due to recognized genetic conditions (chromosome and single gene causes), 8%-12% are due to environmental factors (maternal-related conditions, drug or chemical exposures) and 20%-25% are due to multifactorial inheritance. The majority, 40%-60% of congenital anomalies, have unexplained causes.[10,11] Examples of infectious agents that can be transmitted to the fetus and have an adverse effect include rubella, cytomegalovirus, varicella and toxoplasma. A number of drugs have clearly been shown to be teratogenic.

A teratogen is a factor that has an adverse effect on an embryo or a fetus between fertilization and birth. The teratogenic risks associated with most maternal environmental exposures are not well-established. [12]

The global epidemic of thalidomide-induced limb defects seen in the 1960s resulted in today's practice of monitoring for congenital anomalies worldwide. Other examples of teratogenic agents include folic acid antagonists, anticonvulsants (Dilantin, Tegretol), coumarin derivatives and retinoids (Accutane). The most commonly used teratogenic agent is alcohol.

Recent research has reported increased risks for structural birth defects and chromosomal abnormalities with air pollution and proximity to hazardous waste sites, respectively. [13]

Maternal age is a risk factor for congenital anomalies, specifically chromosome problems, Maternal health conditions that contribute to increased risks for congenital anomalies include obesity, epilepsy controlled with anticonvulsant medications, and insulin-dependent diabetes, maternal thyroid disease, even when treated, as increasing the risk for congenital anomaly-affected pregnancies. [14, 15]Currently we register any diagnosis of CA from ICD-X Q00-Q99 group[16]. Figure 1. Etiology of congenital anomalies.

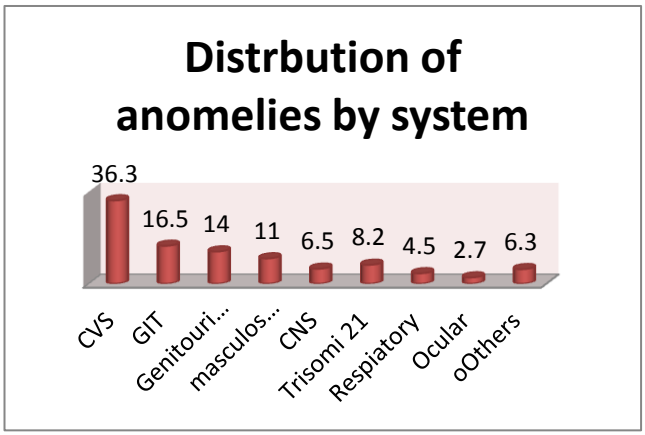


Material and Methods

This prospective study was undertaken in Gharian Teaching Hospital, Gharian, Libya. Our hospital provides medical care to about 5000 delivered women in every year who are Libyan and other nationality, originating and living in Aljabal algarbi area. All babies born in the department of Obstetrics and Gynaecology and admitted to Neonatal Unit in our hospital were included in the study and the Congenital malformations discovered at birth or before discharge from the neonatal nursery in infants born between Jan 2010 to Dec 2010 were studied. The data presented here include 1360 Births, Both mother and the baby were examined as a unit within 24 hours of birth and were further followed upto 72 hours. A detailed history was taken including all familial and

gestational factors and a physical examination of baby was done. The diagnosis of congenital malformations was based on clinical evaluation and all were logged. The clinical, radiological and laboratory data were entered in the medical records. Chromosomal studies were carried out in infants suspected to have a recognizable chromosome syndrome.

Congenital anomalies were coded according to the International Statistical Classification of Diseases and Related Health Problems, Chapter XVII (Q rubric) 'Congenital Malformations, Deformations and Chromosomal Abnormalities, 10th revision' (ICD-10) (World Health Organization, 1993).



Results

During this one year study, there were 4850 deliveries in our Hospital Out of 1360 newborns admitted to Nursery, 73 had one or other congenital anomaly accounting to an incidence of 5.4%. Out of these, 50 had single congenital anomaly and rest 23 had multiple malformations. Thus, there were total of 103 anomalies amongst 73 newborn babies.

The congenital anomalies were seen more (1.5 %) in neonates born to advanced maternal age (> 35 years). With the increasing parity frequency of congenital anomalies also increased, it accounted for 3.12 % anomalies when the neonates were born to mother having 2 or more siblings. There was higher frequency of congenital anomalies in males as compared to female babies (1.5% vs. 1 %). and also more in full term as compared to preterm (2.21% vs. 1.82%). Frequency of congenital anomalies was more in vaginal delivery as compared to cesarean born babies (2.7 % vs. 1.58%). Congenital anomalies were more common in stillborn babies as compared to live born babies.Consanguinity was noted in 3 parents out of 73 deliveries who had malformed babies. Exposure to drugs was not included because no data about it . Family history of congenital anomaly was unknown. None of mothers who delivered congenitally malformed babies gave history of exposure to radiation, smoking or alcohol during the pregnancy. Figure 2. Distribution of anomalies by systems.

Discussion

In the present study, the overall incidence of congenital anomalies was 1.5%, which was almost comparable with other studies (17-19). The incidence varied from 1.2% to 1.81% in these studies. With regard to pattern of congenital anomalies in the study, the most common system involved was CVS (36.3%) followed by GIT (16.5%), genitourinary (14%), musculoskeletal system (11%), CNS (6.6%),respiratory (4.4%) etc. This was comparable to studies conducted by other workers (17-19). Some studies however recorded higher incidence of CNS malformations followed by GIT and musculoskeletal system (20,21). It was observed in present study the congenital anomalies were more common in babies born mother aged 35 years and above. Similar observations were recorded in other studies also (4, 20,21). The congenital anomalies were seen more frequently in mothers who had parity of two and above which in our study was comparable to observations made by various authors (4, 7,8). Consanguinity as a factor, significantly increased rate of congenital malformations as reported by other authors too (13,14), but despite the high prevalence of consanguineous marriages in Saudi Arabia, the overall incidence of congenital anomalies was not higher than in other parts of the world as reported .(22)

Higher incidence of congenital anomalies in still born babies (5.5 %) compared to live born babies (1.5 %) was reported in our study which was comparable with study by Boyed PA (7) who recorded incidence of congenital

malformation in still born babies to be 13% as compared to 3.7% in live born babies.

Malformations of Cardiovascular system and Gastrointestinal system were more frequent than malformations of other systems.

There were 33 newborn with congenital heart disease (36.3%). The most common anomalies were ventricular septal defect, atrial septal defect and patent ductus arteriosis respectively. One-third of the cases had complex cardiac anomalies.
The incidence of GIT anomalies was (16.5%), most common anomalies were imperforated anus followed by esophageal Artesia.

Genitourinary malformation represented (14%) mainly congenital Hydrocele.
The incidence of congenital malformations of the musculoskeletal system in one of the studies conducted at Jammu was reported to be 13 % (16) while in our study 11% , it was revealed that the CNS anomalies include meningomyelocoele, anencephaly, hydrocephalus etc were less common in this study compared to others (17). Trisomy-21 represented 8.2 % of all congenital disorders, 4.5% respiratory anomalies and 2.7 % ocular. Four stillbirths out of 73 had congenital anomalies. the overall incidence of congenital anomalies we found was not higher than in other parts of the world.
The present study helps to know the pattern of congenital malformations prevalent in this part of the country. Observations made in this study also help us to know the possible correlation of various factors as to the cause of congenital malformations. Most of the observations are comparable with the similar studies undertaken in other country. However, some of the observations differ which is expected given the different nature of various studies like hospital versus community based, difference in geographical and environmental factors, difference in time period for follow up, criteria for classification used etc.

Recommendation:-

1.Patients should be informed that prenatal ultrasound at 18 to 20 weeks can detect major structural anomalies in approximately 60% of such cases.



Anencephaly

2. When a fetal structural anomaly is suspected or identified, a referral to a tertiary ultrasound unit should be made as soon as possible to optimize therapeutic options.



Omphalocel

3. Parental imaging should be considered in specific cases, depending on the fetal anomaly identified (e.g., potential dominant inheritance).



Bone anomaly

4. Parental blood testing and invasive prenatal testing may also be required to clarify the diagnosis for a fetus with isolated or multiple structural anomalies.(23)



Meningomyelocel

5. Maternal consumption of folic acid-containing prenatal multivitamins is associated with decreased risk for several congenital anomalies (24).



Ampegious Genetelia

6.Avoid potentially teratogenic industrial pollutants because of significant associations between the textile industry and anencephaly, and between the manufacture of engines and turbines and microcephaly(25).



Amelia



Umbalioal Cord Cyst

REFERENCES:

1. (March of Dimes Resource Center. *Birth Defects*. 1998. (Available www.modimes.org).
2. (Charles R. Green, M.B., Department of Pathology, University of Melbourne, Parkville, N. 2, Victoria, Australia.)
3. (March of Dimes Resource Center. *Birth Defects*. 1998. (Available www.modimes.org)
4. (March of Dimes Resource Center. Birth Defects 1998. Available from: www.modimes.org .)
5. Canfield MA, Honein MA, Yuskiv N, Xing J, Mai CT, Collins JS, Devine O, Petrini J, Ramadhani TA, Hobbs CA, Kirby RS. National estimates and race/ethnic-specific variation of selected birth defects in the United States, 1999-2001. Birth defects Res A Clin Mol Teratol 2006 Nov;76(11):747-756.
6. Patel ZM, Adhia RA. Birth defects surveillance study, year 2005. Indian J Pediatr 2005 Jun;72(6):489-491.
7. Boyd PA, Armstrong B, Dolk H, Botting B, Pattenden S, Abramsky L, Rankin J, Vrijheid M, Wellesley D. Congenital anomaly surveillance in England-ascertainment deficiencies in the national system. BMJ 2005 Jan 1;330(7481):27-31.
8. Lin AE, Herring AH, Amstutz KS, Westgate MN, Lacro RV, Al-Jufan M, Ryan L, Holmes LB. Cardiovascular Malformations: changes in prevalence and birth status, 1972-1990. Am J Med Genet 1999 May 21;84(2):102-110.
9. Centers for Disease Control and Prevention (CDC). Improved national prevalence estimates for 18 selected major birth defects-United States, 1999-2001. MMWR Morb Mortal Wkly Rep 2006 Jan 6;54(51):1301-1305.)
10. Stevenson RE. The Genetic Basis of Human Anomalies. In: Stevenson RE, Hall JG, Goodman RM (Eds.), *Human Malformations and Related Anomalies*. Vol. 1. New York: Oxford University Press, 1993: 115.
11. Nelson K, Holmes LB. Malformations due to presumed spontaneous mutations in newborn infants. *N Eng J Med* 1984; 320: 19-23.
12. (O'Rahilly R, Muller F. Teratology. In: *Human Embryology and Teratology*, 2nd Edition. Toronto: Wiley-Liss Publications, 1996: 110.)
13. Vrijheid M et al. Chromosomal congenital anomalies and residence near hazardous waste landfill sites. *Lancet* 2002; 359: 320-2)
14. Wolfberg AJ, Nagey DA. Thyroid disease during pregnancy and subsequent congenital anomalies, Abstract #274. Society for Maternal-Fetal Medicine Annual Meeting, New Orleans, 2002.)
15. Ritz B, Yu F, Fruin S, Chapa G, Shaw GM, Harris JA. Ambient air pollution and risk of birth defects in Southern California. *Am J Epidemiol* 2002; 155: 17-25.
16. Jones, K.L. Smith's Recognizable Patterns Of Human Malformation Sixth edition2005.
17. Gupta R K, Singh A, Gupta R. Pattern of Congenital Anomalies In Newborn at Birth: A Hospital Based Prospective Study. Scientific paper presented during the Proceedings at 42nd national conference of Indian academy of Pediatrics (Pedicon) at KolKatta, 2005.
18. Swain S. Agarwal A, Bhatia BD. Congenital malformations at birth. *Ind Pediatr* 1994; 31(10):1187-91.
19. Verma M. Chhatwal J. Singh D. Congenital malformations. A prospective study of

10,000 cases. *Ind J Pediat*.1991; 58(2): 245-52.

20. Datta V, Chaturvedi P. Congenital Malformations in Rural Maharastra. *Ind Pediatr* 2000:37; 988-1001.

21. Khatemi F, Mamorri GA. Survey of congenital major malformations in 10,000 newborns. *Iranian J Pediatrics* 2005:15; 315-20.

22. Hassib Narchi; Naji Kulaylat, Congenital Malformations:Are They More Prevalent In Populations With A High Incidence Of Consanguineous Marriages, Annals Of Saudi Medicine, Vol 17, No 2, 199.

23. Alain Gagnon, MD, Vancouver BC, Evaluation of Prenatally Diagnosed Structural Congenital Anomalies, No. 234, September 2009.

24. Y. Ingrid Goh, HBSc,1,2 Enkelejd Bollano, MD, Prenatal Multivitamin Supplementation and Rates of Congenital Anomalies AUGUST JOGC AOÛT 2006.

25. Eduardo E. Castilla,1'2 Hebe Campala, Economic Activity and Congenital Anomalies: Environmental Health Perspectives * Volume 108, Number 3, March 2000.