Section: Anatomy

Original Research Article

GUT ANOMALIES IN ANENCEPHALY IN WESTERN INDIAN POPULATION

Swati Yadav[1], Prabhjot Kaur Chhabra[2], Mahindra Kumar Anand[3]

[1]Assistant Professor, Department of Anatomy, Santosh Medical College, Ghaziabad
[2]Associate Professor, Department of Anatomy, Mahatma Gandhi Medical College & Hospital, Jaipur.
[3]Professor & Head, Department of Anatomy Santosh Medical College, Ghaziabad

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Corresponding Author:
Dr. Prabhjot Kaur Chhabra,
Email: pkkjaipur@gmail.com

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Abstract

Background: Population of India is growing day by day. Thousands of children are born with birth defects. Congenital malformations contribute to an increase in the handicapped population of India resulting in concurrent increase in burden on the health care system of the community. Therefore, this study was undertaken to find the occurrence of gut malformations. **Materials and Methods:** 46 dead fetuses with anencephaly were included in the study. All the fetuses were preserved, dissected and observed for gut malformations. **Result:** Gut malformations were observed in 26% of anencephalic fetuses. In some of the fetuses, spleen was present in right hypochondrium, Megacolon, omphalocoele, cleft lip and palate were also observed. **Conclusion:** Congenital anomalies may be caused by genetic or environmental factors. Most anomalies are a result of complex interaction between genetic and environmental factors. Presence of anomalies in two or more organ systems indicate some common factors affecting development of multiple systems. Further genetic study and research is needed in order to prevent such malformations.

INTRODUCTION

Population of India is growing day by day. Thousands of children are born with birth defects. These congenital anomalies contribute to an increase in the handicapped population of India resulting in concurrent increase in burden on the health care system of the community. Limited financial resources for health can be better utilized by salvaging normal babies and preventing congenital malformations. Congenital malformations have emerged as a major cause of stillbirths and neonatal mortality. It is a common cause of morbidity and mortality not only in the newborn but also in childhood and beyond.[1] Due to renewed interest in anencephaly for organ transplant a detailed systemic internal study was carried out on anencephalic fetuses.

MATERIALS AND METHODS

This study was conducted in Pramukhswami Medical College and Shri Krishna hospital Karamsad, Municipal hospital Anand, J S Chauhan hospital, Baria, Shamalji Saravjanik hospital Godhara, Dr. P K T nursing home, Godhara and B J Medical College and associated civil hospital, Ahmedabad. Data was collected and manuscript was prepared at Santosh medical college, Ghaizabad.18075 consecutive deliveries were observed and fetuses were scanned for apparent central nervous system defects. 46 fetuses with apparent central nervous system anomalies were collected after obtaining written consent from parents .The gestational age of fetuses ranged from 18weeks to 40 weeks. This study was part of bigger project. Demographic data was collected and 46 fetuses were dissected for gastrointestinal tract anomalies.

Fixation and Dissection of specimens: All the specimens were fixed in buffered formalin. After 4 to 6 weeks of fixation with buffered formalin each specimen was dissected for central nervous system and other system malformations. A midline incision was given from suprasternal notch to symphysis pubis. Skin, fascia and muscles were reflected. Bilateral subcostal incisions were given to open abdominal cavity. Sternum was cut in the midline and thoracic cavity was opened. Thoracic viscera were observed for any anomaly and their position was noted. Diaphragm was studied for diaphragmatic hernia. Position and malformations if any, were noted for all the abdominal and pelvic viscera.

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RESULTS

46 anencephalic fetuses were collected and dissected for study. In the present study 12(26%) fetuses with anencephaly had associated systemic anomalies in the abdomino-thoracic region. Total 24 gut anomalies were detected [Table 1] namely, Megacolon (1A), Superior mesenteric artery lying behind the 3rd part of duodenum(1B) ,Spleen in right hypochondrium (1C), Omphalocele [Figure 2A], Intestines adherent to the undersurface of liver [Figure 2B], Enlarged liver occupying entire abdominal cavity [Figure 2C], Kidney seen in Thoracic Cavity(3A), Appendix in subhepatic position(3C), Diaphragmatic Hernia (left), Malrotation of gut, Enlarged liver and intestines in thoracic cavity, Presence of caecum and appendix on left side of abdominal cavity, Bilateral Diaphragmatic Hernia and Cleft lip and Cleft palate.

Table 1: Internal malformations in abdominal cavity found in anencephalic foetuses

S. No.	Gastrointestinal tract malformations	Anencephalic fetuses affected
1.	Megacolon	04
2.	Omphalocele	04
3.	Diaphragmatic Hernia (left)	02
4.	Enlarged liver occupying entire abdominal cavity	04
5.	Other gut malformations	10

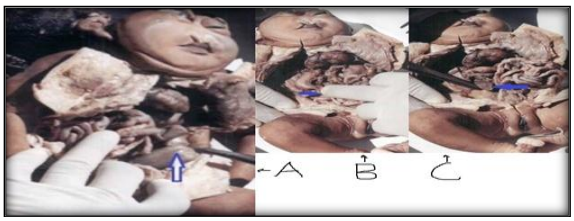

Figure 1: Anencephaly foetus with Megacolon (A), Superior mesenteric artery behind third part of duodenum (B) and spleen in right hypochondrium (C).

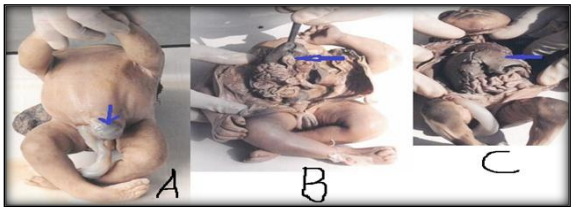

Figure 2: Anencephaly foetus with Omphalocoel(A),Intestine adherent to undersurface of liver (B) and Enlarged liver occupying entire abdominal cavity(C).

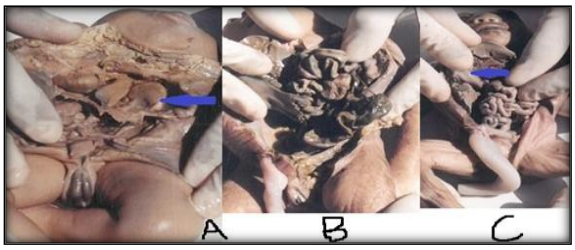

Figure 3: Kidney seen in Thoracic Cavity(A), Megacolon(B), Appendix in subhepatic position(C)

DISCUSSION

Although, the cause of most congenital anomalies is unknown but the complex interaction between genetic and environmental factors can result in malformations.

During the first two weeks of development, exposure to teratogenic agents usually kills the embryo rather than causing congenital anomalies. During period of organogenesis, teratogenic agents disrupt development and may cause major congenital anomalies. During fetal period teratogens can produce morphological and functional abnormalities, particularly of the brain and eye.[2,3]

In India estimated of 303,000 neonates die within the first 28 days of life every year from congenital anomalies. The impact of congenital anomalies is severe in middle and low-income countries.[4,5]

Present study finds 24 gut malformations in 46 anencephalic fetuses. Paduranga et al,[6] found 6 gut malformations in their study on 41 fetuses besides other systemic anomalies. A.M. Vare and P.C. Bansal,[7] also found malrotation of gut, abnormal position of appendix, enlarged liver, umbilical hernia and diaphragmatic hernia in their study on 41 anencephalic fetuses.

Presence of omphalocele seen in four anencephalic fetuses in the present study may be due to the insufficient formation or improper migration of mesoderm from the primitive streak. The enlarged liver present in the omphalocele sac in all four fetuses might have prevented the reduction of physiological hernia and hence led to the umbilical defect.

The common congenital gastrointestinal tract anomalies seen in babies not affected by congenital CNS malformations are cleft lip and cleft palate, esophageal atresia, tracheo-esophageal fistula, pyloric stenosis, ileal atresia, omphalocele, imperforate anus and diaphragmatic hernia.[8-11] This suggests that gut anomalies seen in anencephalic fetuses either are secondary to neural tube defects or share the same risk factors as neural tube defects.

The presence of such internal anomalies confirms the observations made by Jenne.E.Bell et al,[2] that in fetuses with localized external defects such as congenital CNS malformations, the internal defects were more extensive. M.L. Kulkarni, Mathew Kurian,[12] also found other system anomalies associated with anencephaly in fetuses. B.Vishnu Bhat and Lokesh Babu,[13] observed that postmortem examination of fetuses with external anomalies and of stillborn fetuses was useful in

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detecting internal malformations and this increased the total number of defects by 1½ times.

Tan et al,[14] recorded 9.4 % while David TJ,[15] recorded 84% associated anomalies. Gole et al,[16] in a study showed that nearly 80% of fetuses had associated malformations. Spina bifida was seen in 9 fetuses and cleft palate in 8 fetuses. Earlier studies have also reported cleft lip and palate to be more common in male anencephalic fetus.[5,14,17] Scott, J.M found that in one fetus the kidney was seen in thoracic cavity.[18] This is in alignment with our study. Another Study showed that Diaphragmatic hernia is one of the associated defect with anencephaly but not usually familial.[19] Anencephaly may also be associated with defect of internal organs like hypoplastic lungs, syndactyly, cyclopia, club foot, cleft palate, imperforate anus, renal defects, cardiac defects, large thymus, absence of thumb and radius, large thymus, and reduced size of adrenal gland.[20] Most of Neural Tube Defects are associated with omphalocele, diaphragmatic hernia & cleft lip. It was first described in 1981 by Czeizel & named "schisis association"(SA).[21]

CONCLUSION

The present study finds the prevalence of gut anomalies in anencephalic fetuses. Anomalies associated with apparent malformations are important for embryologists for analyzing inner defects. The study is of interest for academicians, researchers and is helpful for gynecologists and radiologists for accurate diagnosis and interpretation. Further research is needed at genetic level to find the cause of associated gut malformations in anencephalic foetuses. Related nutritional deficiencies need to be analyzed to prevent congenital anomalies at earliest stage.

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