Original Research Article

Unusual Congenital Deformities detected in Perinatal Autopsy- A Series of 5 Cases

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Abstract

Autopsy literally means to see for one self - is the dissection and examination of a dead body to determine the cause of death and establish evolution of a disease. There are 3 main types of autopsies-(1) Medico-Legal/Forensic/coroner's autopsy, (2) Clinical /pathological autopsy, (3) Anatomical or academic autopsy. In this study we performed pathological autopsy of perinatal death cases and presenting a series of 5 cases with rare (unusual) congenital deformities along with review of literature.

Key words: Autopsy, perinatal death, congenital deformities.

Introduction

"To the living we owe respect but to the dead we owe the truth" - Voltaire.

"We must turn to nature itself, to the observations of the body in heath and disease, to learn the truth." -Hippocrates. An autopsy is a medical procedure that consists of a thorough examination of a body to determine the cause of death and to evaluate any disease or injury that may be present. An autopsy is an important audit tool that often discloses new information about the cause of death[1]. There are 3 main types of autopsies[2],- 1) Medico-Legal Autopsies - These are performed as prescribed by applicable law in case of violent, suspicious or sudden deaths; 2. Pathological autopsies - Are performed to diagnose a particular disease or for research purposes; 3. Anatomical/Academic autopsies - Are performed by students of anatomy for study purposeonly.

In this study we performed pathological autopsy of perinatal death cases.

Perinatal death is a common clinical problem and the family seeks and deserves answers regarding the cause of death. The clinical utility of the perinatal autopsy are as following[3]

1. Confirm the clinical diagnosis (concordant),
2. Change the clinical diagnosis (discordant).
3. Add (significant unexpected finding noted).

In this study we present a series of 5 cases of perinatal autopsies with rare congenital malformations.

Materials & Methods

Ethical Consideration

The study was approved by the Institutional Ethics Committee.

This study includes the perinatal deathwhich is defined as follows (Most widely accepted definition for international comparison)[4,5].

1. >= 500 gm. birth weight at 22 or more weeks of gestation and
2. Death between birth and at the end of the neonatal period.

All these cases were referred from the Department of obstetrics and Gynecology to our department with written consent obtained from the parents in each case for autopsy.

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- In each case autopsy was done according to the standard protocol in the following order -

1. Anthropometry.
2. External examination.
3. Internal examination.
4. Examination of the placenta and umbilical cord.
5. Tissue taken for histopathological study.

Case-1

This is a case of elderly primi (maternal age 37 years) presented with intrauterine fetaldeath (IUFD).The body was received and perinatal autopsy was done according to the protocol described.

On gross examination, no structures of the face developed except a midline aperture with single round blue color eyeball likestructure. On opening of the cranial cavity, it was found that failure of the forebrain to separate into two halves was there. On histopathological examination of the round blue colored structure, the components of eye ball wereconfirmed. The final diagnosis was Holoprosencephaly.

Case-2

In this case 32 years female 2nd gravidapresented with lower abdominal pain and tenderness. AbdominalUSG confirmed there was intrauterine fetal death.

On gross examination of the body, there were short forearm, syndactyly and polydactyly in the foot & imperforate anus. On dissection, there was absent kidney. On X-Ray of the fetus, it showed absence of the radius bone. The final diagnosis was done as Ellis Van Creveld syndrome.

Case-3

34 years female, primigravida presented with IUFD. On gross examination, there was anencephaly, spine bifida and a large intracranial mass replacing the intracranial contents. On histological examination of the intracranial mass, it was found to be Mature Cystic Teratoma containing mature elements derived from all the three germ layers.

The final diagnosis was made as Neural Tube Defect with Congenital Intracranial Teratoma.

Case - 4

32 years female term pregnancy presented to the emergency with labour pain. The baby died soon after birth because of failure to

breath. On autopsy examination there was an abnormal opening in the diaphragm on left side through which part of liver, spleen and portion of intestine were herniatedin to chest cavity with collapse of lung and secondary dextrocardia.
The final diagnosis was Congenital Diaphragmatic Hernia with Secondary Dextrocardia.

Case- 5
A 30 years female of term pregnancy having difficulty in expulsion. On cesareansection it was found to be a case of dichorionic diamniotic twin pregnancy, one with normally developedfetus and placenta and another placenta with multiple grapes like vesicles which was confirmed to bepartial mole on histopathological examination. The final diagnosis was made as term twin pregnancy with hydatidiform mole.

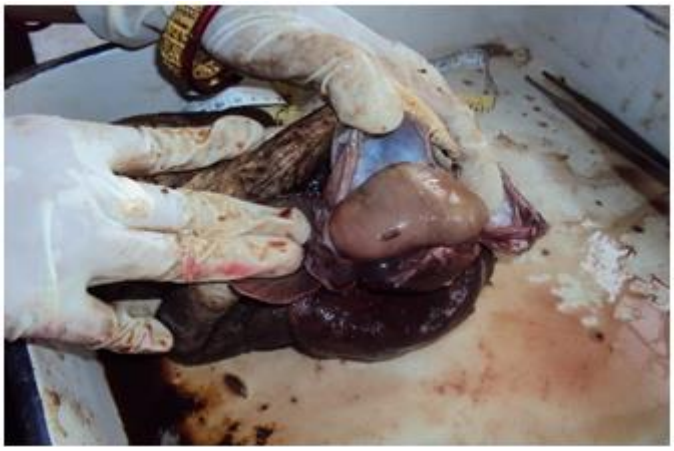
Discussion

Case-1: (Holoprosencephaly)
Holoprosencephaly is a cephalic disorder in which the prosencephalon (the forebrain of theembryo) fails to develop into two hemispheres. Normally the forebrain is formed and the face begins to develop in 5th and 6th weeks of gestational period. Hox gene which guide placement of embryonicstructures, fail to activate along the midline of the head allowing the structures that are normally paired on the left and right to merge. The condition can be mild or severe. Holoprosencephaly consists of spectrum of defects or malformations of the brain and face. According to the National Institute of Neurological Disorders and Stroke (NINDS), "In most cases of holoprosencephaly, the malformation isso severe that babies die before birth."[6,7].
In our case the baby was died before birth and the fetus presented with most severe facial defecti.e., Cyclopia (i.e. development of single eye), and the nose is in the form of a proboscis (tubular appendage). The condition is also referred to as Synophthalmia or cyclocephaly and is very rare.

Case 1(Holoprosencephaly)

Figure 1: A: cyclopia

B .Proboscis



C. Failure of forebrain to separate

Case - 2: (Ellis Van Creveld Syndrome)
It is also known as Chondroectodermal Dysplasia. This is an inherited disorder of bone growth results in very short stature and narrow chest with short ribs and syndactyly /polydactyly, congenital heart defect and finger nail dysplasia[8]. In our case the baby had very small forearms (absent radius bone on X-Ray), syndactyly and polydactyly in left foot, finger nail dysplasia, flattened pinna, ASD (Atrial Septal Defect) & absent kidney.

Case 2(Ellis Van Creveld Syndrome)

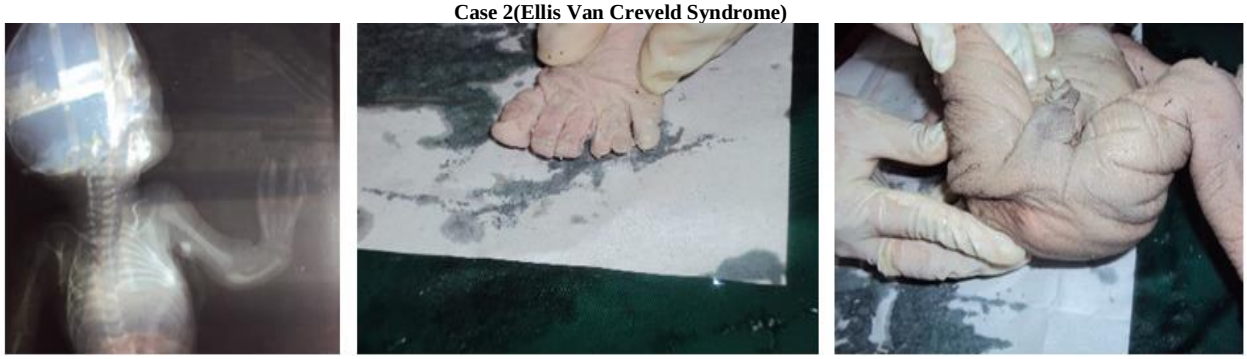


Fig 2 A: Absent radius bone **B. Syndactyly and polydactyly** **C .Imperforate anus**

Case -3:(Congenital Intracranial Teratoma)

Intracranial teratoma are rare, according for 0.5 - 2.0% of intracranial tumors[9]. They are typically benign. They develop from embryonic cells which become "misinvolved" during formation of primitive streak in the 3rd. week of life. Some of these cells become "misenfolded" as intracranial rests of tissue.Sometime the intracranial mass is massive and replaces the intracranial contents[10]. Our case was of massive mass replacing the intracranial contents and on histology it had mature elements derived fromall the three germ layers. The case was also associated with spina-bifida.

Case 3 (Congenital intracranial teratoma)

Figure 3

Case-4: Congenital Diaphragmic Hernia (CDH)

Diaphragmatic hernia is a birth defect in which there is an abnormal opening in the diaphragm most commonly on the left side. The incidence of CDH ranges from approximately 0.8- 5/10,000 births[11-14]. Inspire of advances made in the medical and surgical management of CDH the mortality and morbidity remain high[15,16,17]. In our case the defect was on left side through which part of liver, spleen, stomach and portion of intestine were present in the chest cavity with collapse of lung and secondary dextrocardia. The baby died soon after birth because of failure to breath.

Case 4(Congenital diaphragmatic hernia)

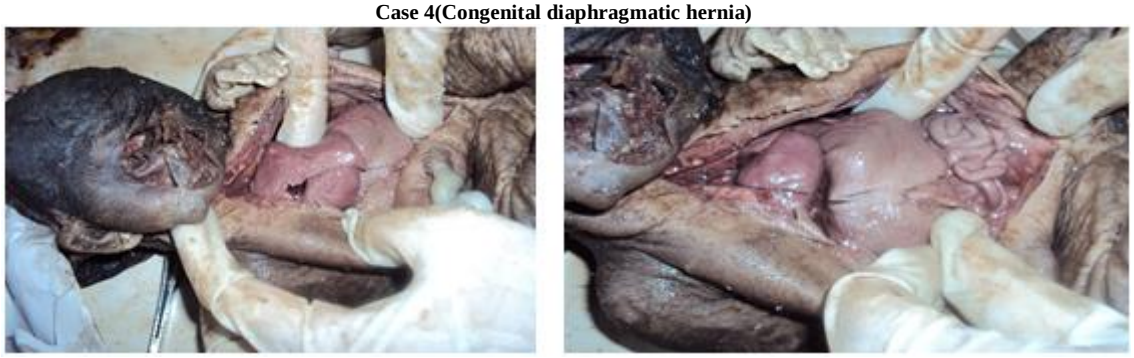


Fig 4: Dextrocardia and herniation of viscera

Case-5: (Term Twin Pregnancy with Hydatidiform Mole)

The coexistence of hydatidiform mole with a normal fetus is rare. The incidence ranges from 1 in 20,000to 1 in 100,000 pregnancies[18]. The probability of carrying one fetus up to term with associated molar pregnancy is low because of development of complications like pre-eclampsia and vaginal bleeding, thyrotoxicosis and increased risk of persistent trophoblastic disease[19].

In our case on caesariansection, it was found to be a case of dichorionic diamniotic twin pregnancy with one normal fetus and placenta and another placenta with multiple grapes like vesicles which was confirmed to be of partial mole on histopathological examination.

Case 5: Term twin pregnancy with hydatidiform mole)

Figure 5

Conclusion

In our case series we have reported unusual congenital abnormalities in perinatal autopsy study. The clinical utility of perinatal autopsy are as follows -

1. Confirm the diagnosis (clinical and autopsy diagnoses concordant).
2. Change the diagnosis (clinical and autopsy diagnoses discordant).
3. Add (significant unexpected findings noted on the autopsy although the clinical diagnosis was not altered to conclude carefully performed perinatal autopsy is the single most useful step in identifying the cause of perinatal death[20].

According to Bove KE et al, 1997 autopsy in perinatal deaths provides an etiology and summary of contributing factors in most cases and allow improved management in futurepregnancies[21].

Conflicts of interest

None.

Reference

1. Gordijin SJ, Erwich JJ, Khong TY. Value of the perinatal autopsy; critique pediatr Dev, Patho, 2002, 54800-488.
2. Strasser, Russell S. (2008) Autopsies in Ayn Embar Seddon, Allan D Pass (eds) Forensic Science. Salem Press PP 95 ISBN 978-1587 654237
3. Saller D.N. et al : JAMA, the journal of the American Medical Association 1995, Vol. 273, No. pp. 663-665,ISSN 0098- 7484.
4. Mosby's Medical Dictionary, 8thediction 2009, Elsevier
5. Definition of perinatal death for internationalcomparison. Wikipedia.
6. "Holoprosencephaly Information Page | National Institute of Neurological Disorders and Stroke". www.ninds.nih.gov.
7. Totori-Donati P, Rossi A, Biancheri R (2005). "Brain
8. Ruiz-Perez, V. L. et al. Mutations in a new gene in Ellis-van Creveld syndrome and Weyers acrodental dysostosis. Nat. Genet. 24, 283–286
9. Sandow BA, Dory CE, Aguiar MA, and Ahuhamad AZ: Bestcases from the AFIP, congenital intracranial teratoma, Radiographic 2004; 24 1165-1170.
10. Nanda Anil, Luis Schut, Leslie N. Suttan. Congenital forms of intracranial teratoma, Surg Neurol 1982;17:383-38
11. Colvin J, Bower C, Dickinson JE, Sokol J. Outcomes of congenital diaphragmatic hernia: a population-based study in Western Australia. *Pediatrics.* 2005;**116** (3):e356–363. doi: 10.1542/peds.2004-2845.
12. Gallot D, Boda C, Ughetto S, Perthus I, Robert-Gnansia E, Francannet C, Laurichesse-Delmas H, Jani J, Coste K, Deprest J, et al. Prenatal detection and outcome of congenital diaphragmatic hernia: a French registry-based study. *Ultrasound Obstet Gynecol.* 2007; **29**(3):276–283. doi: 10.1002/uog.3863.
13. Yang W, Carmichael SL, Harris JA, Shaw GM. Epidemiologic characteristics of congenital diaphragmatic hernia among 2.5 million California births, 1989–1997. *Birth Defects Res A Clin Mol Teratol.* 2006;**76**(3):170–174. doi: 10.1002/bdra.20230.
14. McGivern MR, Best KE, Rankin J, Wellesley D, Greenlees R, Addor MC, Arriola L, de Walle H, Barisic I, Beres J, et al. Epidemiology of congenital diaphragmatic hernia in Europe: a register-based study. *Arch Dis Child Fetal Neonatal Ed.* 2015;**100**(2):F137–144. doi: 10.1136/archdischild-2014-306174.
15. Brownlee EM, Howatson AG, Davis CF, Sabharwal AJ. The hidden mortality of congenital diaphragmatic hernia: a 20-year review. *J Pediatr Surg.* 2009;**44**(2):317–320. doi: 10.1016/j.jpedsurg.2008.10.076.
16. Mah VK, Zamakhshary M, Mah DY, Cameron B, Bass J, Bohn D, Scott L, Himidan S, Walker M, Kim PC. Absolute vs relative improvements in congenital diaphragmatic hernia survival: what happened to "hidden mortality" *J Pediatr Surg.* 2009;**44**(5):877–882. doi: 10.1016/j.jpedsurg.2009.01.046.
17. Stege G, Fenton A, Jaffray B. Nihilism in the 1990s: the true mortality of congenital diaphragmatic hernia. *Pediatrics.* 2003;**112**(3 Pt 1):532–535. doi: 10.1542/peds.112.3.532.
18. Malhotra N, Deka D, TakkarD,Kochar S, Goel S,SharmaMC:Hydatiform mole withcoexistinglive fetus in dichorionic twin gestation. Eur J ObstetGynecolReprod Biol 2001, 94:301-303.
19. Matsui H, Sekiya S, Hando T, Wake N, Tomoda Y: Hydatidiform mole coexistent with a twin live fetus: a national collaborative study Malhotra N, Deka D, Takkar D, Kochar S, Goel S, Sharma MC: Hydatiform mole with in Japan. Hum Reprod 2000, 15:608-611.
20. Saller D.N. et al: JAMA, the journal of the American Medical Associtiaon 1995, Vol. 273, No. 8, pp. 663-665,ISSN O098-7484.
21. American College of Obstetricians and GynecologistsDiagnosis and management of fetal death. ACOG no. 176.Washington DC American College of Obstetricians andGynecologists, 1993.
22. Bove KE and the Autopsy committee of the college ofAmerican pathologists. Practicce guidelines for peri natal autopsy Arch Pathol Lab Med 1997; 121: 368-76.