서울醫大學術誌 제22권 제 2 호
The Seoul Journal of Medicine
Vol. 22, No, 2, June 1981

Ectopia Cordis

—Report of 2 autopsy cases—

Je G. Chi, Sun Hyung Lee, Sung Hoe Park and Dong Hee Lee*

Department of Pathology, College of Medicine, Seoul National University
and
*Department of Obstetrics & Gynecology Cheil Hospital**

Ectopia cordis is a rare form of congenital anomaly in which the heart is completely or incompletely outside the mediastinum. Abbott (1936) defined this entity as "a displacement so that the heart passes out of the thorax and comes to lie either upon the outer surface of the body or in the abdominal cavity."

Recently an experience of ectopia cordis, so called thoraco-abdominal type, and an additional autopsy case among the hospital records prompted this report.

REPORT OF CASES

Case 1(A80-17): A male premature infant, 37 weeks of gestation, was admitted to Cheil Hospital on July 2, 1980, immediately after delivery, because of ectopia cordis and eventration of abdominal viscera. After a few minutes, he expired.

The pregnancy was complicated by polyhydramnios of more than 1600ml. His mother had a history of ingesting herb medicine for extended time during her second month of pregnancy.

At autopsy, the body was rather small weighing 1250gm. Crown to rump and crown to heel lengths were 27.5cm and 45.5cm, respectively. Left ear was extremely hypoplastic, 0.5 ×0.7cm in maximum extents, with atresia of the external auditory canal. The right ear lobe was prominent. The ear measured 3.0×1.5cm. The anterior fontanel measured 4.0cm×3.0cm without bulging out. In the midline, extending from the lower end of sternum to a point caudad to and including the umbilicus, there was a large defect of anterior abdominal wall, measuring 4.7cm×2.0cm. The lower portion of the sternum was defective. Rectus abdominis muscles were laterally displaced and made borders of the defect. It was rather hypoplastic and was covered by thin skin which was continuous to parietal peritoneum. Large herniation of viscera was present through the defect, including whole heart, whole liver, whole stomach, small intestine and most part of large bowel, a portion of esophagus, a whole spleen embedded in omental sac, and a whole pancreas.

There was an umbilical stump which was 5cm in length and 0.6cm in diameter, containing three vessels. Scrotum was not redundant and rather flat, without testes inside. Sacral dimple was present with hairs. Extremities had no anomalies and appeared normal. Anus was perforated. The pleural cavity contained some amount of clear serous fluid as well as extremely hypoplastic lungs which weighed 4.3gm in toto. The cut surface of the lung was pale gray. The right lung showed incomplete lobation. Hilar portions were inconspicuous and trachea and main bronchi were unremarkable. The heart protruded from the abdominal defect which measured 2.0cm in diameter. The heart was

pendulous, globular, and weighed 3.5gm. It was attached to the body only by the large vessels. The right ventricle was larger and prominent than the left. Opening the heart the papillary muscles were moderately hypertrophied. There was no septal defect. The aorta measured 3.5 cm in circumference and was rather hypoplastic and somewhat elongated. In the proximal portion of the right ventricle, a small pulmonary artery, 1.1cm in diameter, was given off. There was patent foramen ovale, measured 0.5cm in diameter. Ductus arteriosus was prominent and elongated, measuring 0.8cm in length. All major vessels were normal. Venous drainage and outflow tracts were normal. The spleen was 1.5gm in weight. The organ was round and embedded in omental sac. Outer surface was rather smooth. Thin muscular diaphragm had a round ventral defect, 2cm in diameter and was hypoplastic. It extended to the level of the 12th rib on each side. Through this defect was the heart protuding outside the body wall. The stomach was totally extra-abdominal as well as a portion of esophagus. Small intestine was 79cm in length. The color was greenish dark purple. The lumen was filled with green colored meconium. Large intestine was 32cm in length and had a stout thick wall. Greenish ragged pseudomembranous tissue was attached to serosal surface of the intestine. The liver was rectangular in shape, and weighed 45gm. Outer surface was smooth. Greenish ragged material was attached to the outer surface. The nature was same as mentioned above. Ductus venosus drained to the liver and made a landmark dividing it to the left and right lobes. A gelatinous tissue arising from the umbilical cord was attached to the left margin of the liver surface. It measured 4cm in length without containing vessels. Gall bladder was totally absent, being replaced by green grayish fibrous tissue which had the same nature as the tissue attached elsewhere. The right and left kidneys weighed 2.0gm and 2.2gm, respectively. They were normally located. Each organ was firm, lobulated and dark purplish, and the capsule stripped with ease. The cortex measured up to 2mm in thickness, and the corticomedullary junction was distinct. The adrenal relationships to the kidneys were very unusual. The right adrenal gland was lobated at the renal pelvis parallel to the long axis of the kidney.The left adrenal gland was located at the upper most part of the kidney. Cut surface showed reddish brown medulla and pale yellowish cortex.

Case 2(A76-8): This was a deadborn fetus to a young mother who was exposed to acute carbon monoxide intoxication during her first trimester. She was in unconscious state at least for several hours before she regained the consciousness.

Grossly malformed body weighed 2.5kg. There were complex anomalies involving body wall and extremities. The head was of normal size, but had two large bony defects of the skull, including a large one, 4×4cm involving left posterior parietal region. A thin layer of dura was covering the brain on this region. The other defect, 8×6cm was in the area of anterior fontanel. Left upper extremity was absent. And cleft lips and palates were present. All the intrathoracic and abdominal viscera including liver were found outside the body cavity. There was no membrane covering eviscerated organs. There was no need of any incision to expose the organs. A total absence of the left upper extremity was noted. There was bilateral talipes equinovarus.

The neck organs were unremarkable. Both lungs were hypoplastic and showed no fissure in the left lung. The heart and lungs together weighed 15gm. The heart was not covered by the pericardium, but was located upside down in superior mediastinal region. The was no

sternum formed. The ventricle was above the level of the atria, and major vessels from the heart pursued their normal courses. Except for patent foramen ovale there was no intrinsic cardiac malformation. Upper small intestine and stomach were unremarkable except for mucosal congestion of the stomach. The liver and pancreas were normal in size and shape. The left kidney showed a moderate hydronephrosis and proximal hydroureter. The genitalia were ambiguous. There was a small phallus, uterus and vagina left to the midline urinary bladder.

The brain was slightly asymmetric in terms of size of each hemisphere. The gyral pattern was well formed, although the lobe determination could not be easily made. Serial coronal sections showed ventricular hypoplasia in the right together with massive ependymal nodule along the ependymal surface. The left hemisphere showed partially preserved anterior horn and occipital horn. There were multiple grayish white mottlings of the brain substance. The corpus callosum was absent in the left. The cerebellum also showed deformities in the distribution of dentate nucleus. A large nodule measuring up to 1.5cm in diameter was also seen along the ventricular surface. The cerebellar hemispheres showed abnormal folial pattern mimicking whorls of the hair in the head. The brainstem showed abnormal shape of the midbrain together with aqueductal stenosis. Microscopically abnormal mixture of neurons, glial cells and other cellular components were confirmed.

REVIEW AND DISCUSSION

It was cited that the first one of ectopia cordis recorded in the literature was the Stensen's case in 1671, which was complicated with tetralogy of Fallot(Byron 1948; Logan et al 1965).

Over two hundreds such cases were reported to date. However, there are no convincing data of its incidence, pathogenesis and etiology. Charateristically presented in one case was the association with maternal ingestion of quinine in early pregnancy. However, it is still ambiguous on its exact etiology(Crittenden, 1959).

Since the first attempt of anatomical classification by Weese in 1818, it is conventional to classify ectopia cordis as cervical, thoracic, abdominal, and thoracoabdominal types. (Blatt and Zeldes, 1942; Byron, 1948; Logan 1965). This classification of the anomaly does not relate to its pathogenesis and is nothing more than a structural descriptive interpretation. It has been emphasized that the vascular connections to heart should be considered in classifyng the anomaly(Willis, 1978).

In the thoracic form, vascular connections must pass through a midline defect in the sternum. It is distinguished by abdominal heart where vascular connections pass through a diaphragmatic defect. The thoracoabdominal variety is not, therefore, a mere combined form of anomaly described above and rather a specific complex anomalies, the embryological mechanism of which is different from each other. (Cantrell et al., 1958; Crittenden et al., 1959)

The embryological mechanism, however, remains problematic, not only because of its rarity, but also because of different embryological interpretations. According to Patten(1960), thoracic ectopia could be present when the ventral body walls catch the heart outside their place of convergence rather than inside as occurred normally.

Another contention by Taussig(1947) was that ectopia cordis represents an arrest in the development of the heart, of such a nature that it fails to complete its descent into the thorax.

Additionally, the conclusion of Cantrell et al. (1958) appears to be a scientifically based compromise. They have designated the thoracoabd-

ominal type as a specific syndrome, which embraces a midline supraumbilical abdominal wall defect, in the diaphragmatic pericardium, and congenital intracardiac defects. The variable morphological presentation of this anomaly may be explained by the participation of both transverse septum, a specific mesoderm, and ventral migration of the paired primordial structures; defects of the former may cause diaphragmatic, pericardial and intracardiac anomalies, and failure of the latter, including sternal anlagen and abdominal myotomes results in sternal fissure and midline abdominal wall defect with diastasis recti.

Table 1 is a summary of the cases reported in the literature, and the most frequent type of anomaly is the thoracic tupe.

One of our cases is a full-blown form of Cantrell's pentalogy except an absence of intracardiac anomalies. This finding is noteworthy, because only rarely was the heart found to be normal (Byron, 1948; Cantrell et al., 1958). Left superior vena cava, aortic and pulmonary stenosis, transposition of aorta, cor triloculare or biloculare, atrioventricularis communis, atrial septal defect, tetralogy, and tricuspid atresia have been described. Among them the most frequent type of cardiac anomalies was interventricular septal defect (Blatt and Zeldes, 1942; Millhouse and Joos, 1959; Cantrell et al., 1958).

Associated malformations in ectopia cordis are harelip, cleft palate, anencephalus, and hydrocephalus (Blatt and Zeldes, 1942; Millhouse and Joos, 1959). It is reported in the literature that multiple congenital malformations coexist with ectopia cordis as in our second case, which consisted of agenesis of gallbladder, atresia of left external auditory canal with hypoplastic ear lobe and markedly hypoplastic lungs.

Table 1. Distribution of sites of defect in Ectopia Cordis

	Blatt & Zeldes (1942)	Byron (1948)	Major (1953)	Kanagas-untheram (1962)
Cervical	2	4	4	3
Thoracic	28	81	81	30
Abdominal	18	38	38	4
Thoracoabdominal	4	10	15	6
Unclassified	—	9	9	5

Despite sporadic successes of surgical correction (Major 1953), the overall life expectancy of ectopia cordis is limited, due to various unfavorable factors such as insufficient thoracic space, occasional large high liver and short inferior vena cana, other significant malformations (Major, 1953; Cantrell et al., 1958; Crittenden, 1959; Logan et al., 1965).

Summary

Two autopsy cases of ectopia cordis are described. The first case was a 37 weeks premature who died soon after birth. The mother took unknown kind of herb medicine for an extended period of time during her second month of pregnancy. This famale baby showed a typical thoraco-abdominal type of ectopia cordis. Although there was no intracardiac anomalies she showed atresia of left auditory canal, pulmonary hypoplasia, agenesis of gall bladder, abnormally shaped liver, spleen and urinary bladder together with meconium peritonitis.

The second case was a deadborn fetus to a mother who was exposed to acute carbon monoxide poisoning during her first trimester. The degree and duration of coma were not identified, although she was in unconscious state at least for several hours. This baby showed severe external anomalies that were characterized by almost total absence of thoracic and abdominal wall, along with amelia of left upper extremity. He also had skull bone defect, lung hypoplasia and severe central nervous system malformations.

=國文抄錄=

이소심(異所心)의 2부검증례

지제근 · 이선형 · 박성희 · 이동희
서울대학교 의과대학 병리학교실 및 제일병원산부인과

이소심(異所心)은 심장이 심낭에 의하여 완전히 싸이지 않은 채 흉강내의 정상위치에서 벗어나 체외 혹은 체내의 기타 장소에 위치하는 희유한 선천성기형이다.

저자들은 서울대학교 의과대학 병리학교실에서 경험한 2례의 이소심 부검예를 기술하였다. 본 예들은 모두 사산 내지 신생아기에 사망한 예로서 산모가 임신기간중 한약(제 1 례)을 복용하였거나 연탄가스중독(제 2 례)에 걸렸던 기왕력을 가졌음이 특징이었다.

제 1 례는 흉복부형으로 심장내 기형은 없었으나 좌측귀의 외이도의 폐착, Omphalocele, 폐 저형성, 간장 및 비장의 형태이상 및 대변성 복막염등이 합병되어 있었으며, 제 2 예는 좌상지결손, 두개골 부분결손, 폐 저형성 및 심한 중추신경계 기형을 동반하였다.

REFERENCES

Abbott, M.E.: *Atlas of congenital cardiac disease. New York, American Heart Association, 1936(quoted).*

Blatt, M.L. and Zeldes, M.: *Ectopia cordis, report of a case and review of the literature. Am. J. Dis. Child.*, **63**:*515, 1942.*

Byron, F.: *Ectopia cordis, report of a case with attempted operative correction. J. Thorac. Surg.*, **17**:*717, 1948.*

Cantrell, J.R., Haller, J.A. and Ravitch, M.M.: *A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium, and heart. Surg. Obs. and Gyn.*, **167**:*602, 1958.*

Crittenden, I.H., Adams, F.H. and Mulder, D.G.: *A syndrome featuring defects of the heart, sternum, diaphragm, and anterior abdominal wall. Circulation*, **20**:*396, 1959.*

Kanagasuntheram, R. and Verzin, J.A.: *Ectopia cordis in man, Thorax*, **17**:*159, 1962(quoted).*

Logan, W.D., Crispin, R.H., Patterson, J.H. and Abbott, O.A.: *Ectopia cordis: Report of a case and discussion of surgical management. Surgery*,**57**:*898, 1965.*

Major, J.W.: *Thoracoabdominal ectopia cordis, report of a case successfully treated by surgery. J. Thorac. Surg.*, **26**:*39, 1953.*

Millhouse, R.F. and Joos, H.A.: *Extrathoracic ectopia cordis: Report of cases and review of literature. Am. Heart J.*, **57**:*470, 1959.*

Patten, B.M.: *The development of the heart, S. E. Gould (ed.) "Pathology of the heart." Charles C. Thomas Publisher, Springfield, Ill., 1960, p.24*

Taussig, H.B.: *Congenital malformations of the heart, New York, 1947, The Commonwealth fund.*

Todd, R.B.: *Abnormal condition of heart, in the cyclo paedia of anatomy and physiology. London, Longman et al., 1836~1939, Vol. 2, p.387 (quoted).*

Willis, H.J.: *The heart, arteries and veins. McGraw-Hill book company, A Blackiston publication, 4 th ed., 1978.*

LEGENDS FOR FIGURES

Fig. 1. Exterior view of the body (Case 1), showing a complete extrusion of the heart(H)outside the body, together with other eviscerated organs including stomach(S).

Fig. 2. Whole body roentgenogram taken under the same position of the body in Fig. 1. No skeletal anomalies were detected except for a small chest cage.

Fig. 3. Anterior view of the chest organs in case 1. In comparison with the heart(H), two lungs are small and hypoplastic.

Fig. 4. Top of the cranial vault, showing a round bony defect (arrows) in parietal bone. Case 1.

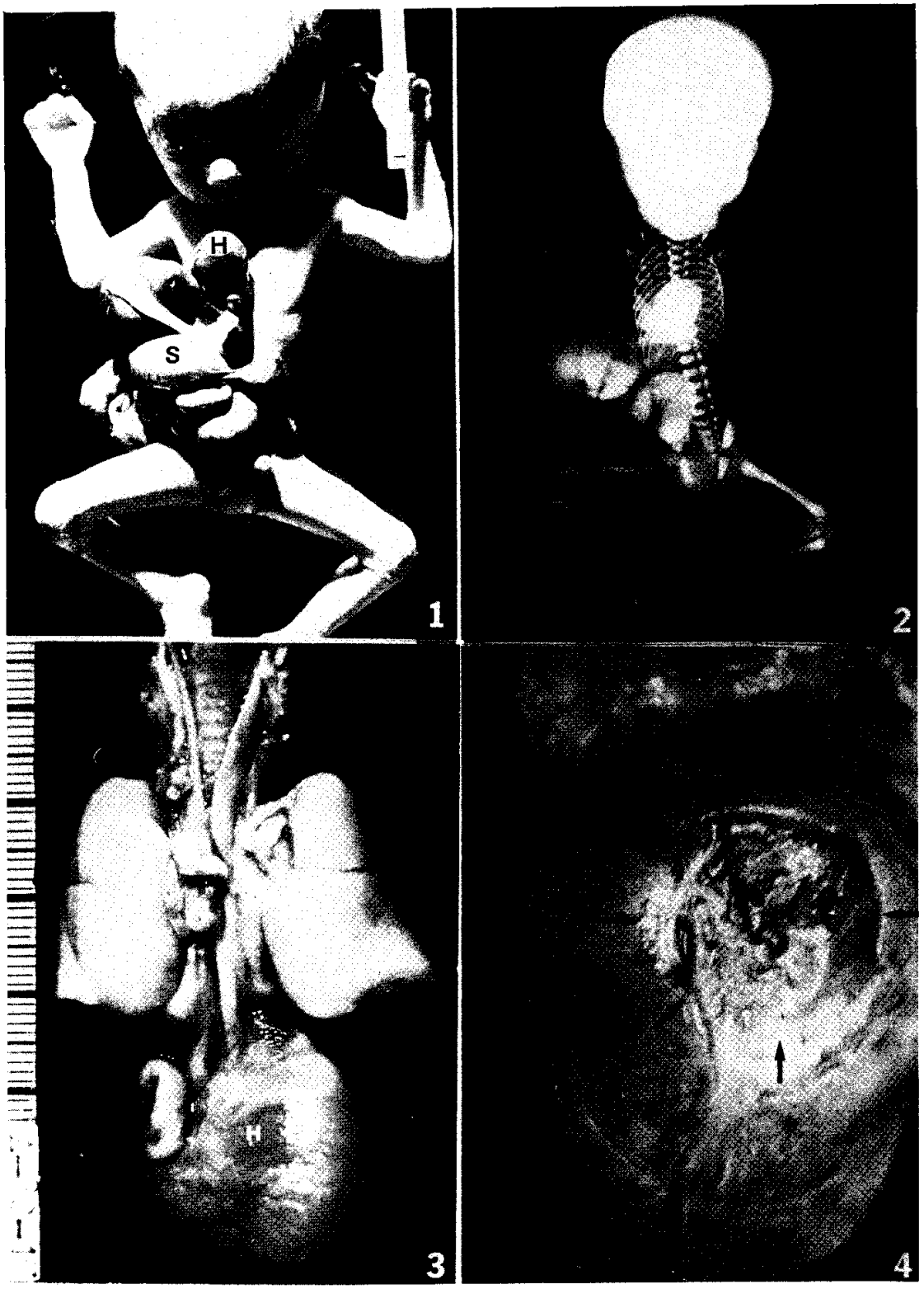
Fig. 5. Ventrolateral view of the body (Case 2). No incision was made before taking this picture. The heart is outside the body wall which is poorly formed. There was no anterior body wall in the chest and abdomen. L: liver.

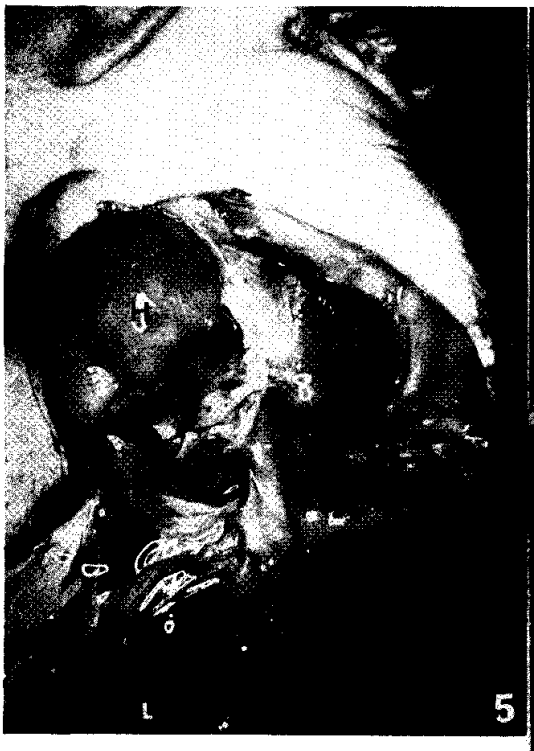
Fig. 6. Transverse section of the cerebellum at the level of mid-pons, showing distorted and malformed fourth ventricle and asymmetry of the dentate nuclei(D) along with hypoplasia of the cerebellar hemisphere.

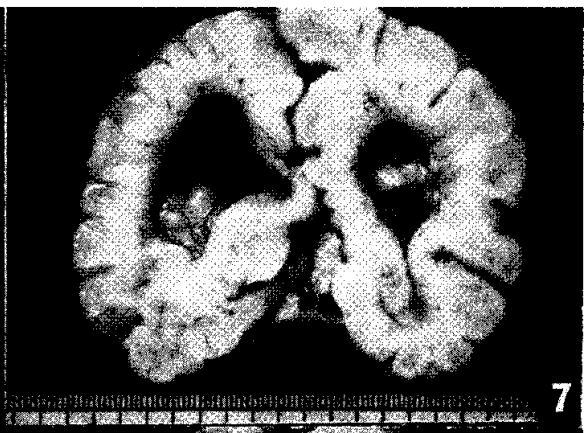
Fig. 7. Coronal section of the brain shows multiple heterotopic gray matter nodules along the ventricular wall in both lateral ventricles.

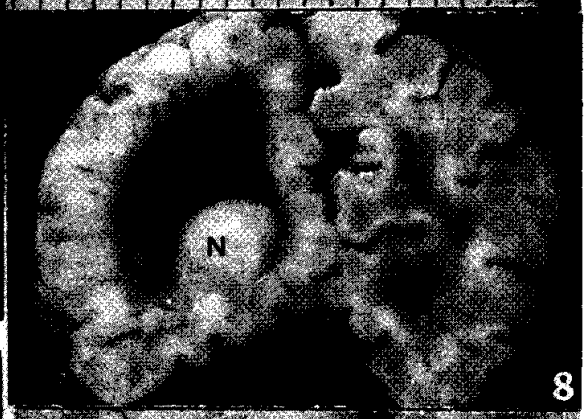
Fig. 8. Asymmetrical posterior horns of the lateral ventricles and peculiar round mass (N) in the left lateral ventricle are shown.

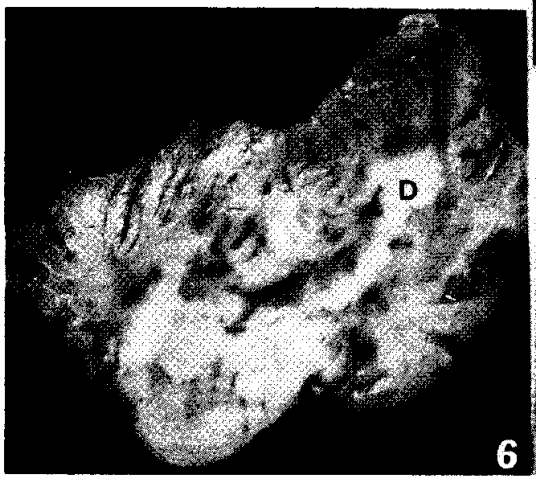
Fig. 9. The corpus callosum(CC) of the right hemisphere is not formed, and instead Probst bundle(P) can be seen. Ventricular cavity in the right hemisphere is not formed anteriorly. Irregular margin of the caudate nucleus(C) in the left and abnormal corpus striatum can be seen.

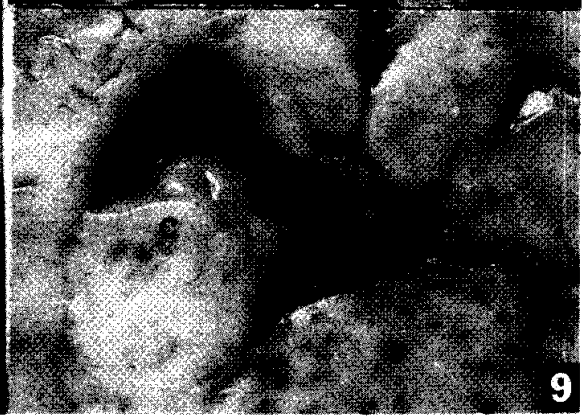

H
S
1
2
H
3
4


L
5


7


N
8


D
6


9