ACARDIAC ANCEPS FETUS – A CASE REPORT

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ABSTRACT

An interesting rare case of lethal fetopathy is described. This case is based on morphological and histological confirmation. Fetus acardius is a parasite for its vascular circulation from the donor twin. The twin reversed arterial perfusion syndrome is an extremely rare manifestation of feto-fetal transfusion in twin pregnancy where the affected twin receives retrograde vascular supply from the healthy twin. Acardiac twin syndrome is a rare complication affecting monozygotic twins where one twin fails to have normal development.

In this report we present an acardiac anceps fetus seen at term gestation and delivered by caesarean section, the other twin is a healthy, well formed baby. At the time of caesarean section, the live baby was delivered first and as the placenta was extracted, the malformed fetus was attached to the chorionic plate of the same placenta. A thin cord with one vascular channel was the only communication between placenta and malformed fetus which was later delivered and collected for the study.

KEY WORDS: Monozygotictwin; Fetal monster; Twin-Twin transfusion syndrome

1.INTRODUCTION

Acardiac twin is a lethal anomaly occurring in 1 in 35000 live births[1]. The pathological basis of acardiac twin is still not known but most proposed theory is the presence of placental vascular arterio-arterial anastomosis between both umbilical arteries of foetuses[2]. The normal umbilical blood flow is reversed in the acardiac twin and blood flows from umbilical artery of normal twin (pump twin) to acardiac twin. This is called TRAP sequence(Twin Reverse Arterial Perfusion). This is the most extreme manifestation of twin-twin transfusion syndrome[3].

Monochorionic placentation occurs in 2/3 of monozygous twins and approximately 0.3% of all spontaneous conceptions. Vascular anastomoses within the placenta allow inter twin transfusion to occur, which in most cases is normal event[4,16]. Imbalance in this flow may lead to clinical sequelae with acute, chronic or inter twin transfusion. TRAP is the most bizarre of inter twin transfusion. The abnormal circulation results in early tissue hypoxia leading to secondary atrophy of heart and other major organs[5,6]

The TRAP sequence leads mainly to four categories of phenotypes:acardius anceps,acardius acephalus,acardius acormus and acardius amorphous[17]. We present this unusual case of acardiac anceps twin with absent heart, upper extremities, thorax and maldeveloped abdomen and lower extremities.

2.CASE REPORT

A 26 year old primigravida, with term pregnancy, married life of 2 years was an unbooked case from a rural area of Andhra Pradesh,India, with poor socio-economic background.
She presented with history of draining liquor and non progression of labour and was taken up for emergency caesarean section after routine blood investigations including HbSAg and HIV.. On caesarean section, first a healthy male baby was delivered, umbilical cord was cut and baby was handed over to the paediatrician. The baby had apgar score of 9 and weighed 2.1kg .While trying to deliver the placenta, the surgeon could find a round mass with small stumps of lower limbs attached to the same placenta with a very thin umbilical cord. This malformed fetus was delivered and placenta was extracted. The weight of the malformed fetus is 900gm ,collected from the operation theater and later studied in the anatomy dissection hall at Osmania medical college.

3. GROSS EXAMINATION

We could identify the cranial, caudal ends and dorsal, ventral surfaces for the malformed fetus.

4. INSPECTORY FINDINGS

A rounded globular brownish mass covered by skin and hair weighing about 900gm.
The caudal end was identified with small stumps of lower limbs with feet attached to it in flexed position .The author tried to inspect the genitalia between the lower limbs and a small elevation was seen resembling primitive phallus without scrotal sac.There was no anal orifice.
At the cranial end, a rounded swelling with two primitive eyes, nasal pit and oral cavity on the anterior aspect were observed. There was scanty hair on the head.
SURFACES: Dorsal surface was convex and hair was seen in the caudal aspect.
Ventral surface has cut umbilical cord and poorly identifiable structures.

5. PALPATION

The fetal mass was soft and cystic, yielding with pressure.
At the cephalic end and on dorsal surface, bone like consistency was felt. At the caudal end of fetus inspectory findings were confirmed. The cephalic globular end of the fetal mass was extended and recognisable facial features like eyes, nasal pit and oral cavity were found.
Examination of external genitalia: A small phallus was found without scrotal sac.

6. AUTOPSY OF THE SPECIMEN

One incision was given on ventral surface extending from below the primitive face to the primitive genitalia.Thick flaps measuring 2cm thickness were opened and and on retraction,only intestinal loops were found, no other recognisable viscera like liver , kidney, pancreas or pelvic organs were found.
A second incision was given on the dorsal surface and did not find any recognisable vertebral column. The incision was extended to the cephalic end of the fetal mass and no recognisable brain structures were found.
Lateral incisions were extended and,cystic cavities were seen and no thoracic cage, heart or lungs were found.

7. INVESTIGATION

To confirm the structures and contents of this malformed fetus, the author

performed some investigations. These include, the radiological and histopathological examination.

8. RADIOLOGICAL EXAMINATION

The fetus was sent to the diagnostic center for x-ray (skiagram) where academic research is active. There is a separate x-ray facility for study of dead and abnormal foetuses. The trained radiologist opinion was taken and the findings are as follows:

9. FINDINGS

1. Fetal skull vault formed with minimal overriding.
2. Entire fetal spine could not be visualised
3. Facial bones partly formed
4. Both upper arm bones faintly visualised
5. Pelvic girdle formed
6. Both femurs formed
7. Leg bones tibia and fibula are absent on one side
8. Both feet formed
9. Shoulder girdle partly formed
10. Rib cage not visualised

The following samples were collected and sent to histological examination. Skin, umbilical cord, intestine and tissue from cranial cavity.

10. HISTOPATHOLOGY REPORT

A) Section from skin: shows epidermis and dermis. Epidermis lined by 2-3 layers of stratified squamous epithelial cells with basal cell layer and keratinization. Stratum granulosum and stratum spinosum not seen. Pigmentation noted in basal cell layer. Dermis shows loose areolar tissue with rudimentary hair follicles along with the hair shaft. Focal aggregates of small round (? Mesenchymal cells) around the hair the hair follicles and also loosely floating. No erector muscle noted. Congested vessels noted, nerve bundles not seen.

B) Umbilical cord :- Sections from umbilical cords shows 3 vessels, two of them are round with thick muscular layer and presence of RBC's other appears oval with thin muscular layer and few RBC's, adventitial layer is not appreciated in all 3 vessels. These vessels are surrounded by mesenchymal tissue with peripheral lining of flattened small round cells with dark round (? Nuclei) at places giving hobnail pattern suggestive of amnion. Adjacent to large arteries 2 long thin walled congested capillaries are seen.

C) Intestinal segment: - Section taken from the loops of alimentary canal shows partial villous processes lined by flattened to cuboidal to columnar cells interspersed (? Goblet cell), mucosal glands noted at places. Below this there is a diffuse mononuclear cell aggregates, one focus shows aggregates of these round cells (? Peyers patches). There is well developed muscular layer, loose areolar tissue is seen below this, lumen contains slough tissue.

D) Nervous tissue: - Section studied show brain tissue predominantly neural tissue with fibrillary background. There are few foci of well organized cortical matter, this is enclosed with in a linear membranous 2-3 spindle cell layer (? meninges) rich in congested vessels. Below this there are places of calcified areas noted with blotchy and linear streaks. One focus shows papillary and tubular process lined by flattened to cuboidal to columnar epithelium admixed with primitive neural tissue (? Choroid plexus).There is diffuse calcification throughout the tissue, no neuronal or glial tissue appreciated.

DISCUSSION

Acardic fetus is a rare anomaly. The pathogenesis of acardia is not fully understood. However, monochorial placentation with substantial arterio-arterial anastomosis between the two fetal circulations and twin reverse arterial perfusion are consistently present[7]. Other organs may have existed and subsequently undergone atrophy in early pregnancy due to hypoxia[8].

Coulam *et al*. reported cardiac activity in a monochorionic gestational sac containing 2 embryos between 5 and 7 weeks, one twin lost cardiac activity at 7 weeks[9,10]. A diagnosis of acardiac twin was made at 10 weeks when growth of the lower extremities was observed and no development above the thorax was noted.The reversal of blood flow through the umbilical cord of acardiac twin was noted by 14 weeks.

Lattanzi *et al*. reported that the severity of the syndrome depends upon the type of the anastomosis arising between the vascular networks of the two foetuses. There are arterio-arterial and veno-venous anastomoses[11]. They found placental characteristics are major contributor to adverse outcome of monochorionic twins. Anastomoses in TRAP sequence were more likely to be of deep rather than superficial type Masuzaki et al. showed that chromosomal microdeletion or gene mutation can affect normal embryogenesis[12]. The acardiac twin shows variable grades of developmental disruption and, consequently, no two cases are similar.Since monozygotic twins are formed from single zygote, scientists theorize that an error occurs early in cell division in only one of the two groups of cells formed during the process.

Monozygotic twinning are more common in IVF pregnancies, hence increased risk for TRAP sequence is also associated with IVF. Two cases of acardia have been associated with maternal epilepsy and use of anti convulsants primidone and oxcarbazepin. Normally the cephalic pole is most severely affected, being most distal to retrograde perfusion[13].

Depending on the state of disruption, acardiac anomalies are divided into four categories: acardius anceps,acardius acephalus, acardius acormus and acardiusamorphus. Acardius anseps is the most developed form, characterized by a partially developed head, brain tissues, and facial structures. The body and extremities are also developed. The acardus acephalus is the most common type of acardiac twin. These twins do not develop a head but may have an underdeveloped skull base. They have legs but donot have arms. They generally found to lack chest and upper abdominal organs on autopsy. In acardius acormus, there is only development of the isolated head without a body trunk and is rarest of all types[13,14]. Acardius amorphous is presented by a shapeless and disorganised mass containing skin, bone, muscle, cartilage, fat and blood vessels but no recognizable human organs . However, not all cases will fall into one of the above categories.

A single artery in the umbilical cord is seen in 50% of cases . The prenatal diagnosis of an acardiac fetus can be difficult. Diagnostic criteria by ultrasound usually include: absent cardiac motion, poorly defined head and trunk, increase in soft tissue mass and existence of reverse blood flow through the umbilical cord. In some cases, the diagnosis of acardia was established at autopsy.[13]

Mohanty *et al*. reported an unusual case of complete absence of upper extremities and thoracic organs[14].They followed the patient with serial ultrasounds,and in the last ultrasound at 38 weeks only the amorphous mass was seen while the pump twin was normal.They speculated that

cessation of blood flow and hypoxia cause growth arrest and absorption of the hypoplastic limbs[15,16].

Histopathologic report has shown that in the brain tissue there was diffuse calcification throughout the tissue, no neuronal or glial tissue appreciated.The intestinal tissue shows well developed muscular layer, loose areolar tissue is seen below this, lumen contains slough tissue.The umbilical cord shows two arteries and one vein.The skin tissue shows epidermis and dermis.The Stratum granulosum and stratum spinosum are not seen.No erector muscles and nerve bundles seen in the dermis.

Acardiac fetuses,due to variation in vascular anastomosis at placental site, have various phenotypes, but in our literature review this type of acardiac fetus has not been reported previously. Our case illustrates clearly how various phenotypes may occur as a result of atrophy of structures and organs which attempted to form early on in acardiac twins, and how the phenotype may change as gestational age progresses supported by radiological and histopathological evidence.

CONCLUSION

Normally,in acardia cephalic pole is most affected being most distal to retrograde perfusion.However in the present case, there was no development of brain,thorax and upper extremities and partially developed abdomen and lower limbs which has been confirmed by radiological and histological investigations.

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Fig1.1.1 ACARDIAC ANCEPS FETUS

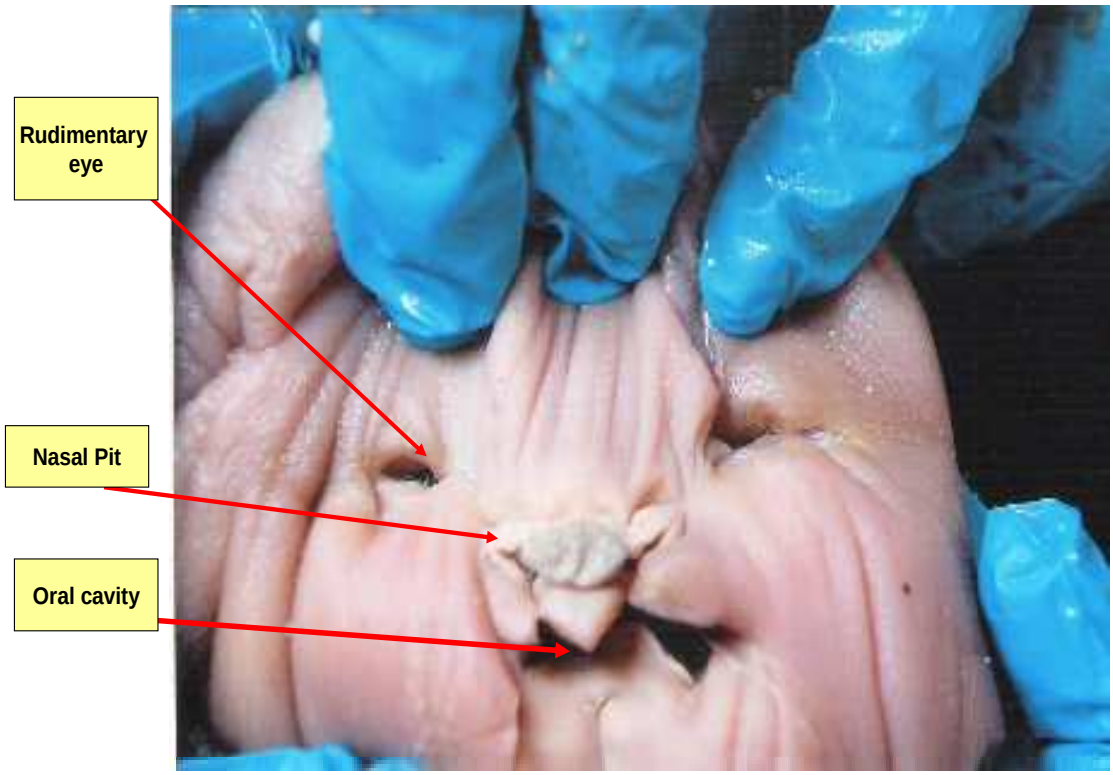

Fig1.1.2 ACARDIAC ANCEPS FETUS

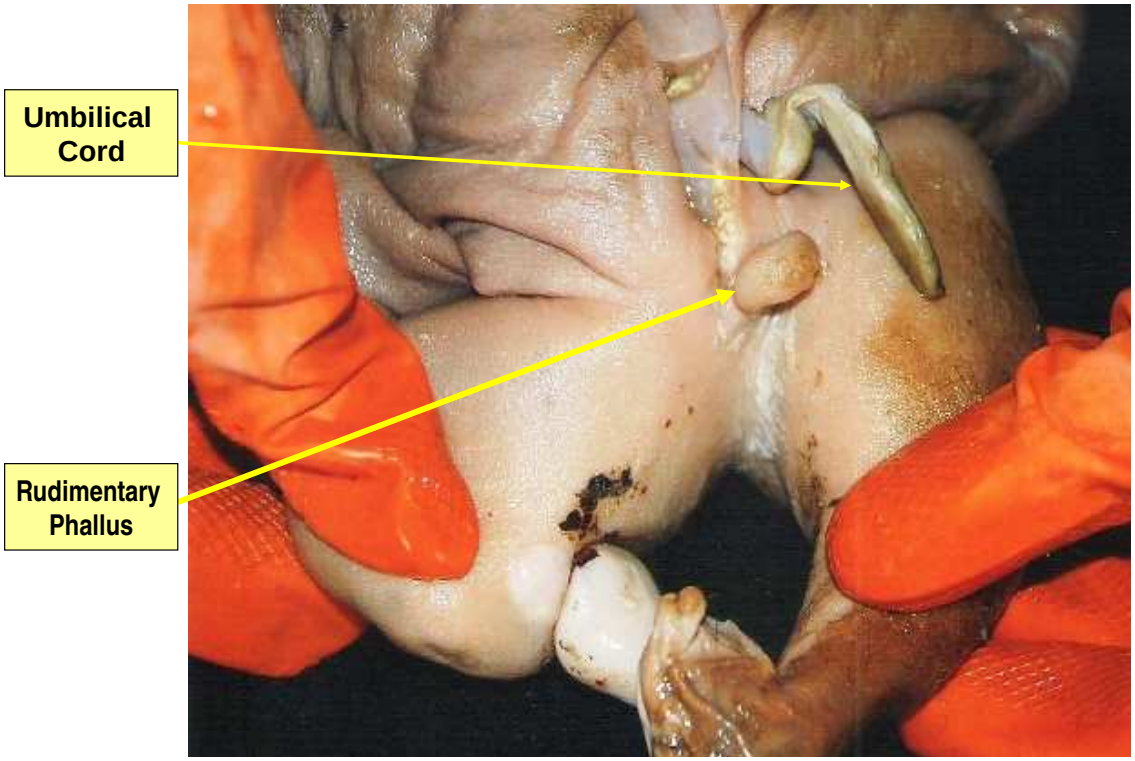

Fig. 1.1.3 ACARDIAC ANCEPS FETUS

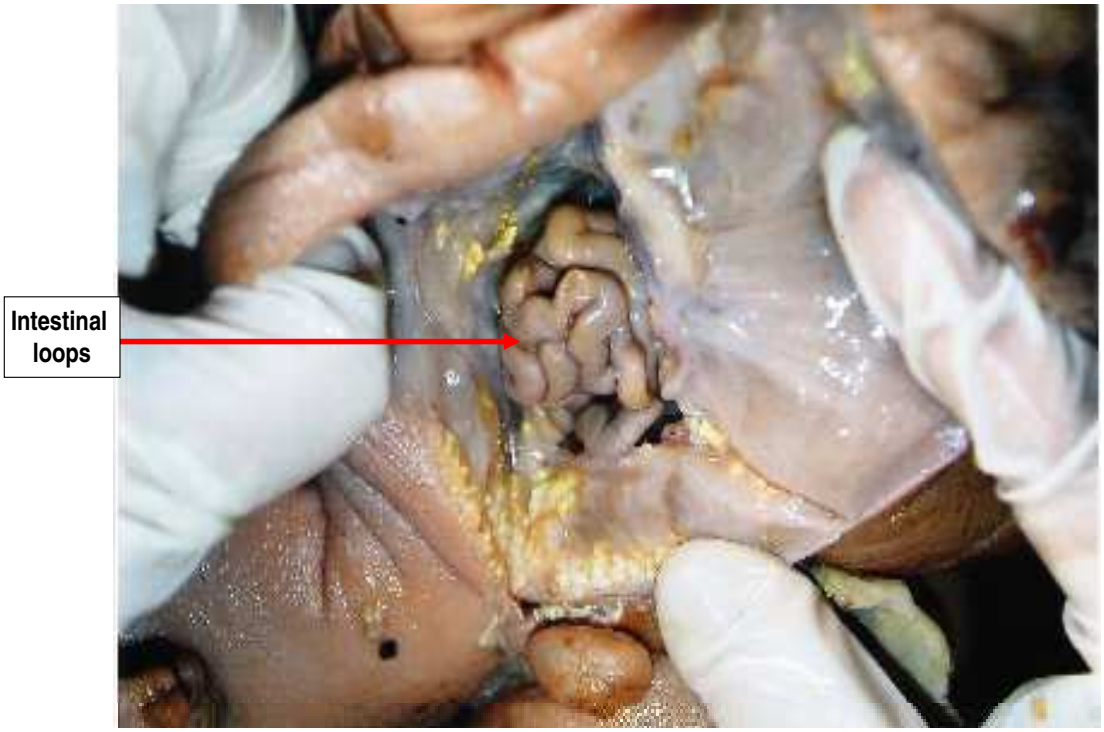

Fig. 2.1.4 ACARDIAC ANCEPS FETUS

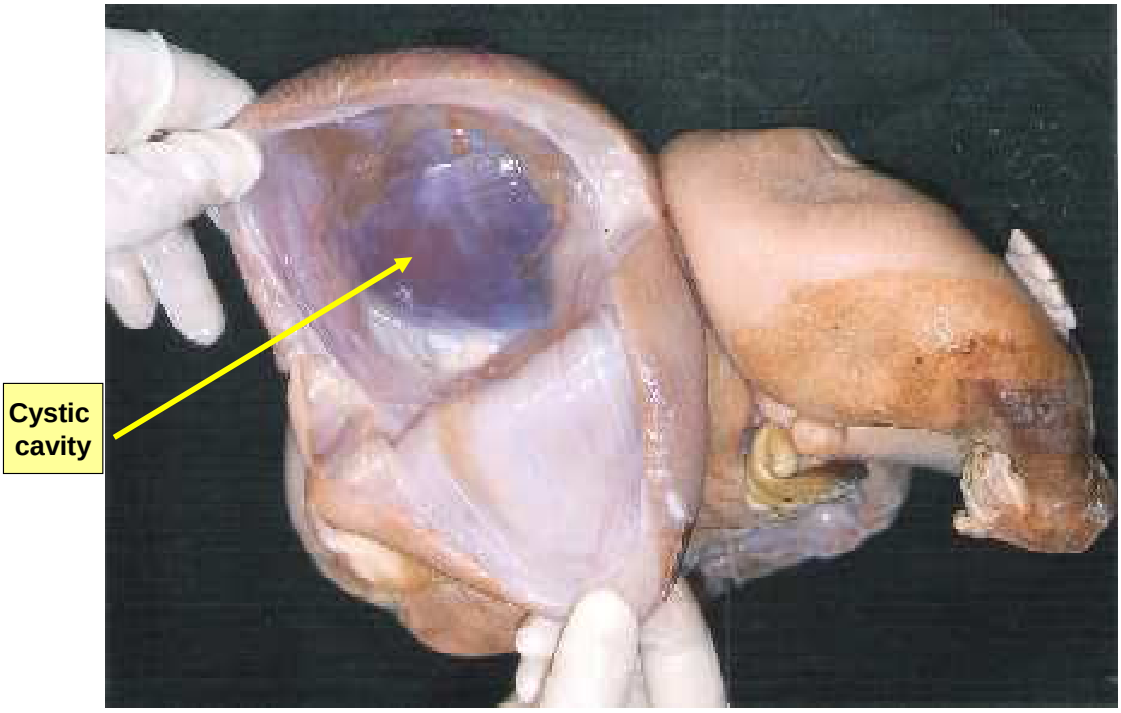

Fig. 3.1.5 ACARDIAC ANCEPS FETUS

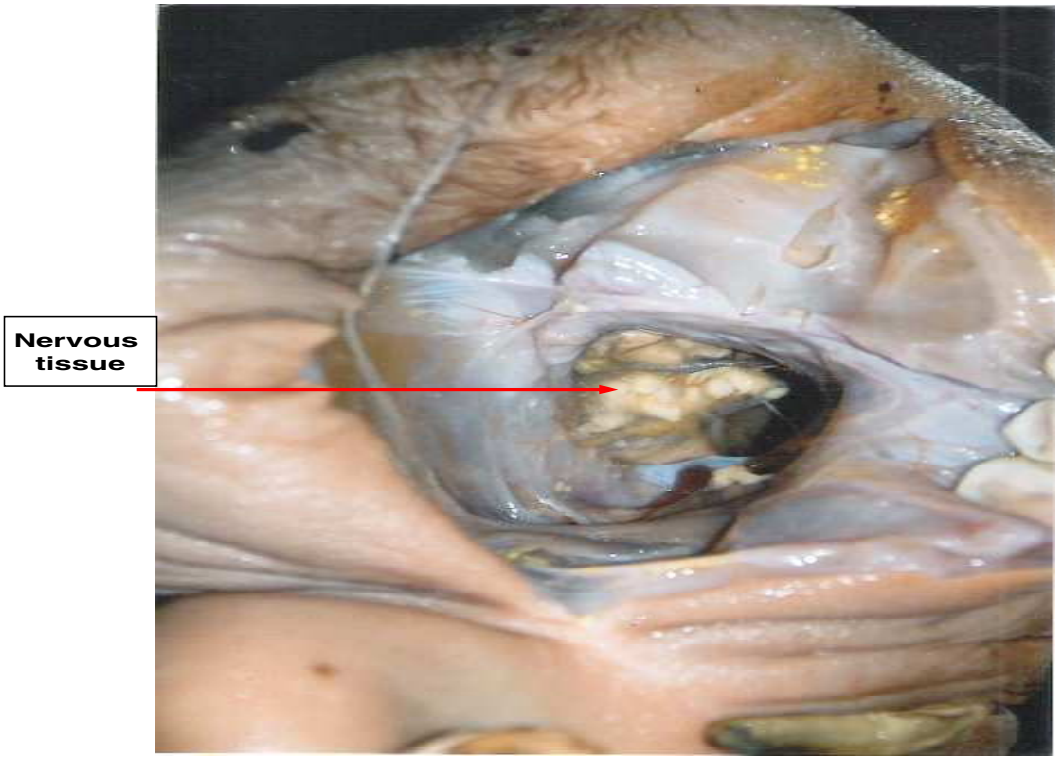

Fig. 4.1.6 ACARDIAC ANCEPS FETUS

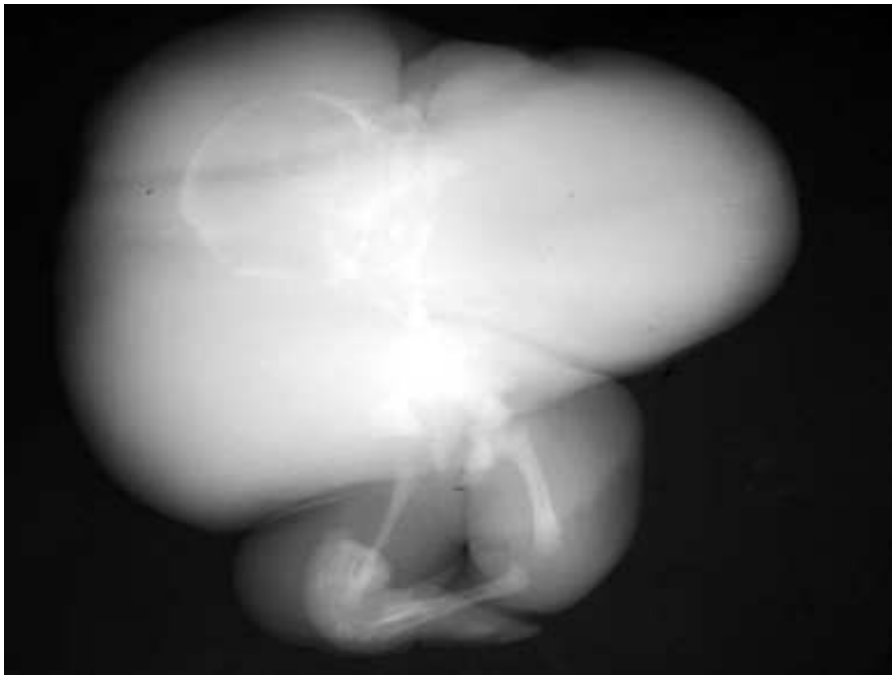

Fig. 5.1.7 Acardiac Anceps Fetus

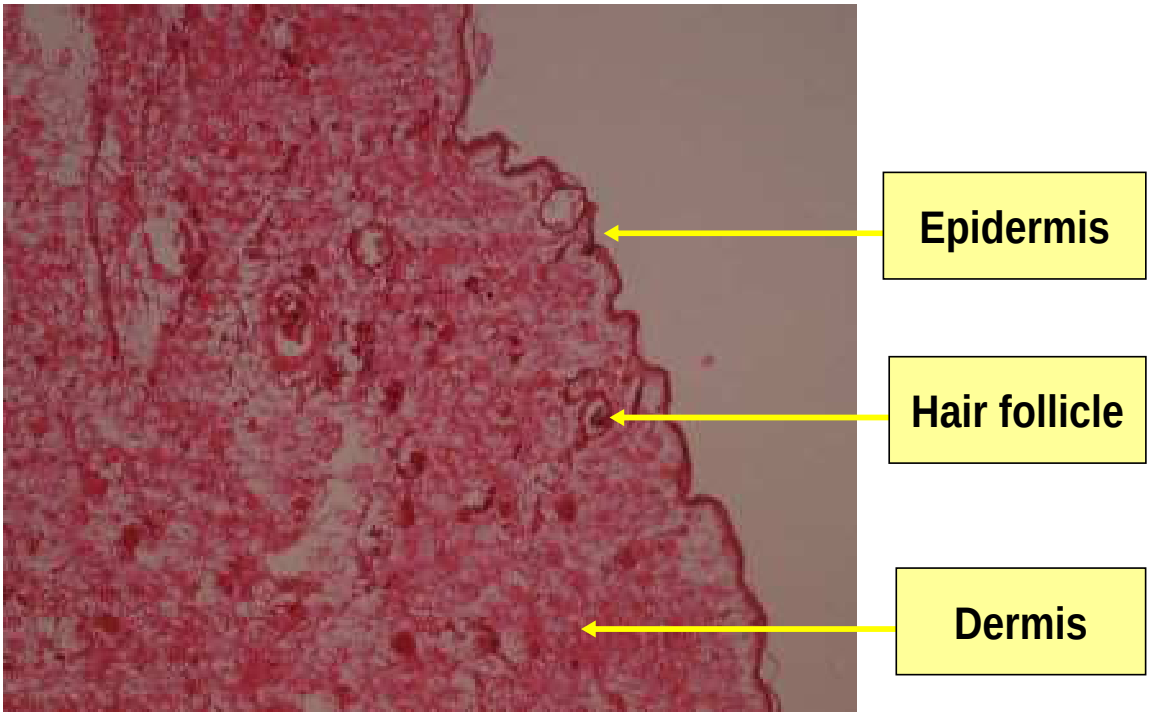

Fig. 1.1.8 Skin section

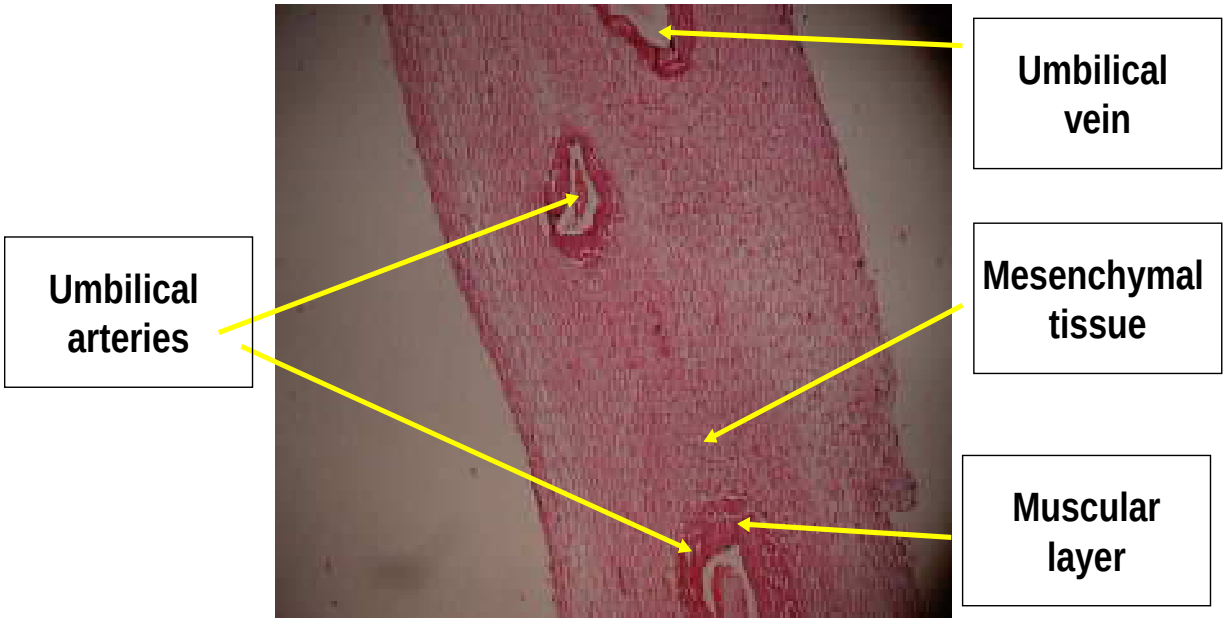

Fig. 1.1.9 Umbilical Cord

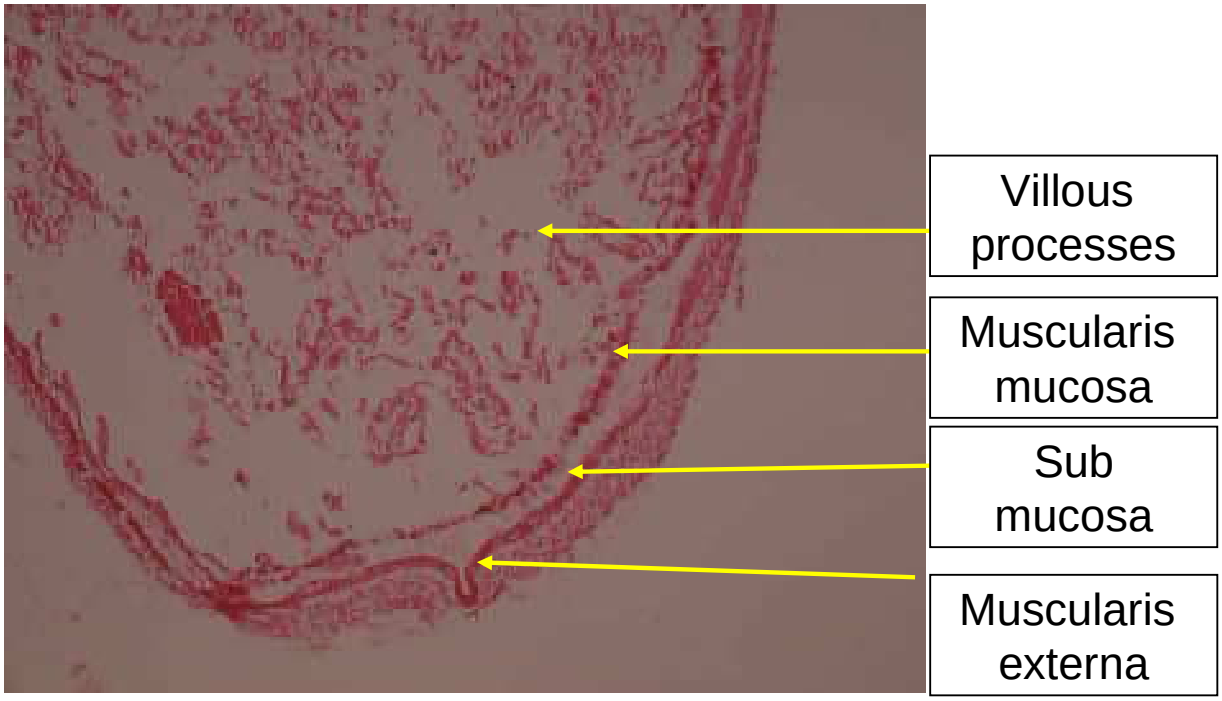

Fig. 1.1.10 G.I. Tract