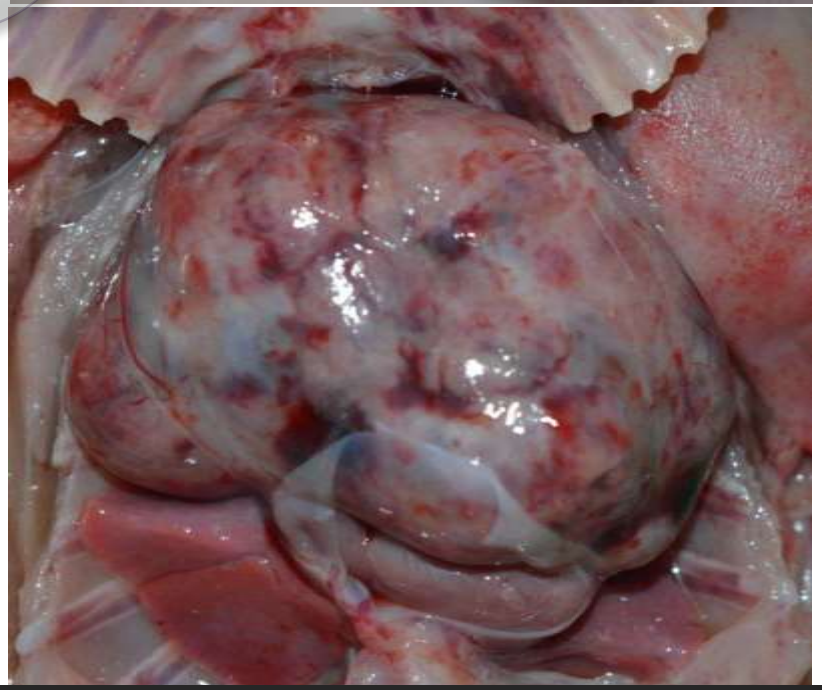
Severe hydrops fetalis due to large congenital tumor with unusual localization: Report of two cases



DAROUICH S, BELLAMINE H, MKAOUAR L, AYACHI A, MOURALI M. **CONGENITAL LARGE CUTANEOUS HEMANGIOMA WITH ARTERIOVENOUS AND ARTERIOARTERIAL MALFORMATIONS: A NOVEL ASSOCIATION**. FETAL AND PEDIATRIC PATHOLOGY 2019;38(1):85-90

DAROUICH S, TAAMALLAH N, BELLAMINE H. **FETAL MEDIASTINAL TERATOMA: MISINTERPRETATION AS CONGENITAL CYSTIC LESIONS OF THE LUNG ON PRENATAL DIAGNOSIS**. ABSTRACT. ECP 2019, NICE.

FETAL AND PEDIATRIC PATHOLOGY
2019, VOL. 38, NO. 1, 85–90
https://doi.org/10.1080/15513815.2018.1547337

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Congenital Large Cutaneous Hemangioma with Arteriovenous and Arterioarterial Malformations: A Novel Association

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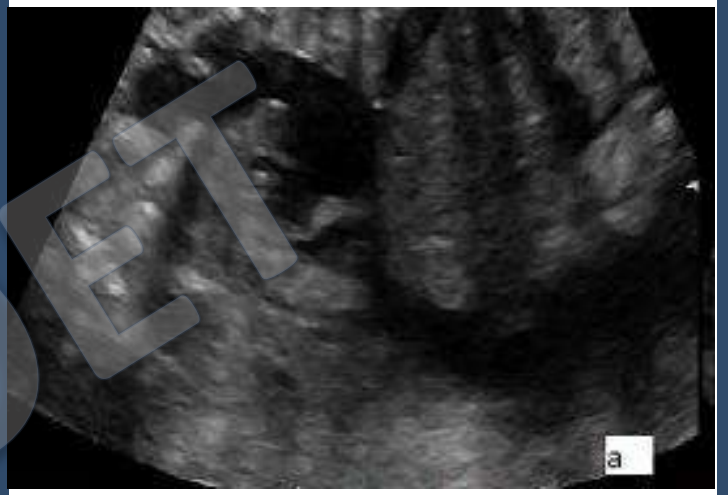
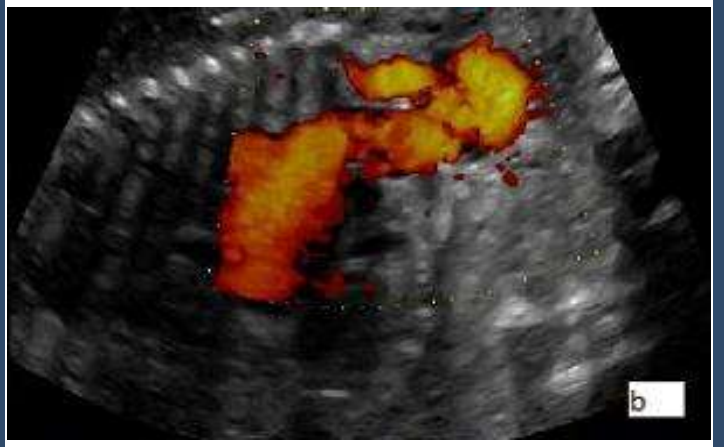
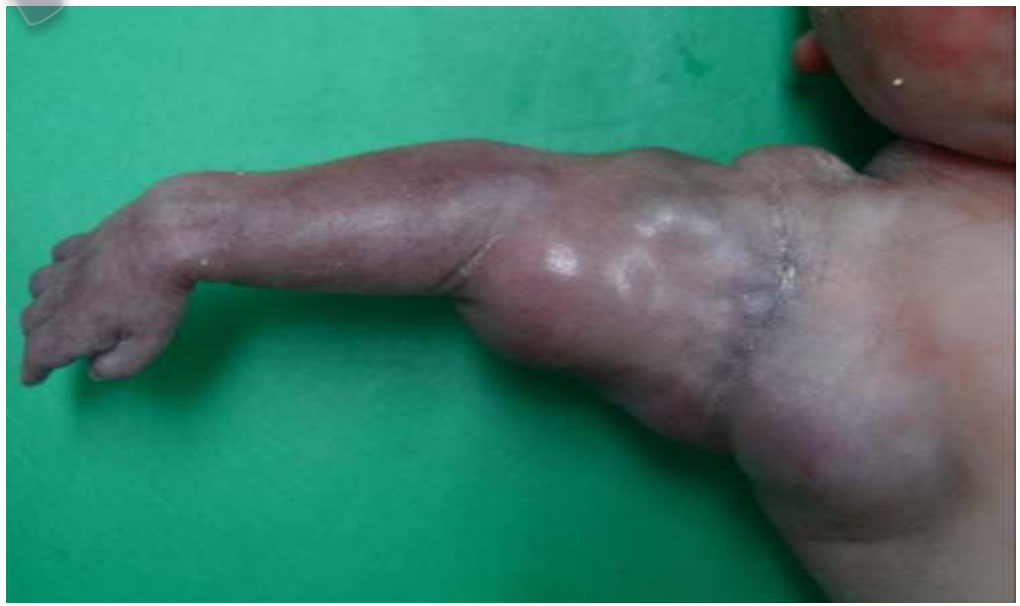
- **A 23-year-old woman, G1P1, A positive**
- **32 WG : hydrops fetalis, placentomegaly, elevated middle cerebral artery-peak systolic velocity with normal amniotic fluid volume**
- **On admission, anamnios (SRM)**
- **Ultrasound & Doppler imaging: dilatation was seen in the brachiocephalic trunk and right carotid and subclavian arteries, rich low velocity arterial flow within a large cutaneous mass located on the right upper limb suggesting hemangioma and a peak systolic velocity of middle cerebral artery >1.5 MoM**
- **Cesarean delivery : male neonate weighing 1700g, profoundly depressed at birth with Apgar scores of 1 at 1min and 4 at 5min. Unfortunately, postnatal respiratory care failed and the neonate died 30min after birth. Hematology workups and karyotype were not performed.**



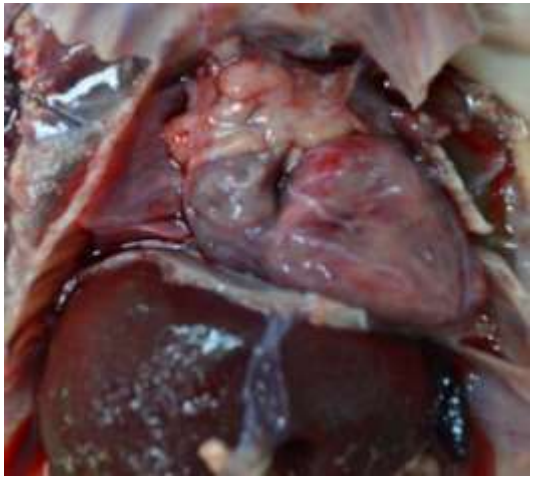


Autopsy

- Macroscopic examination showed a hydropic male fetus with body weight of 1616.8g, crown-heel length 35.5cm, crown-rump length 24cm and foot length 5cm, which were consistent with gestational age of 27-28 weeks.
- The hemangioma was confirmed as a 5x5x3.5cm raised reddish-violet and ulcerated mass with well-defined borders involving the posterior side of the right upper arm with marked perilesional edema.
- The tumor was associated with an axillary prominent mass on the same side.
- Diffuse cutaneous petechiae were observed (more evident over the abdomen).

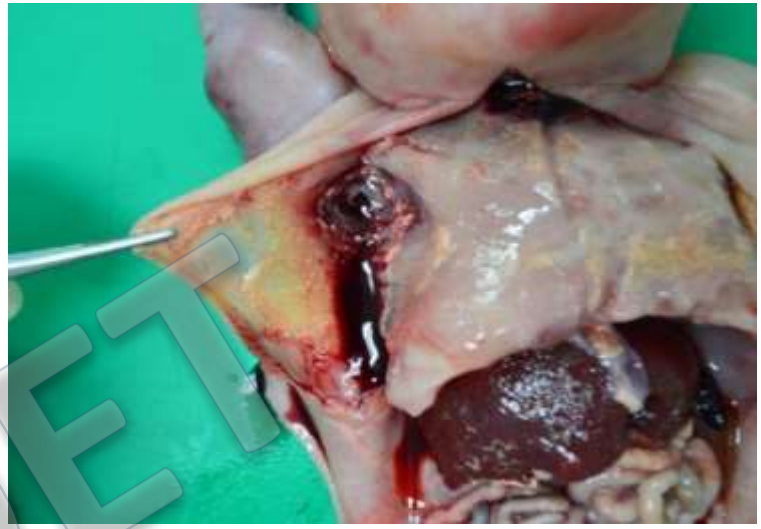


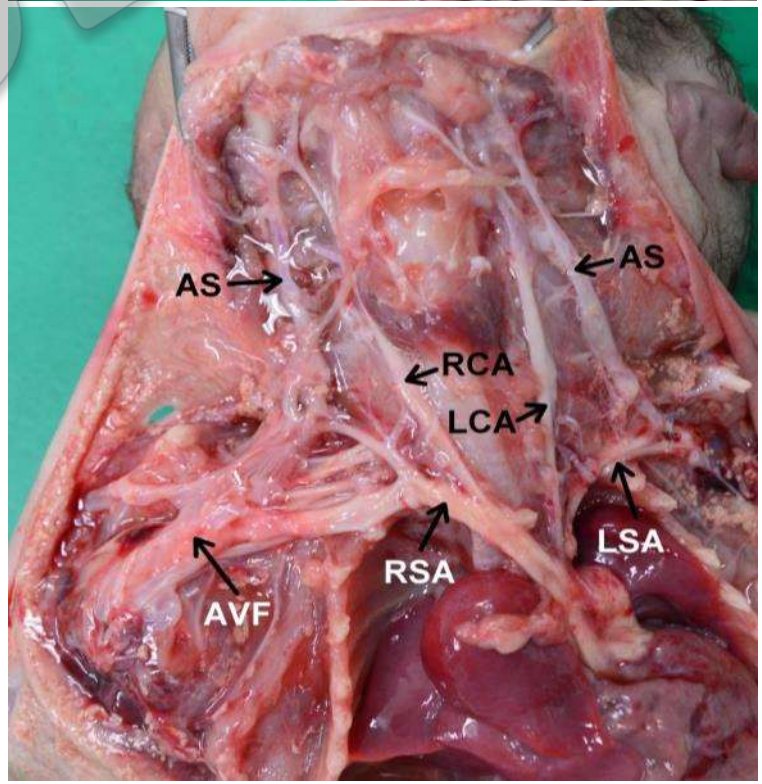
- Massive effusions of all serous cavities
- Thymic hypoplasia (1.35g; normal ranges 2.25–2.5)
- Lung hypoplasia (5.5g/4.25g; normal ranges 10.2–10.6/9.2–9.6)
- Myocardial hypertrophy without structural abnormalities (13.8g; normal ranges 5.5–6)
- There were no visceral petechiae.

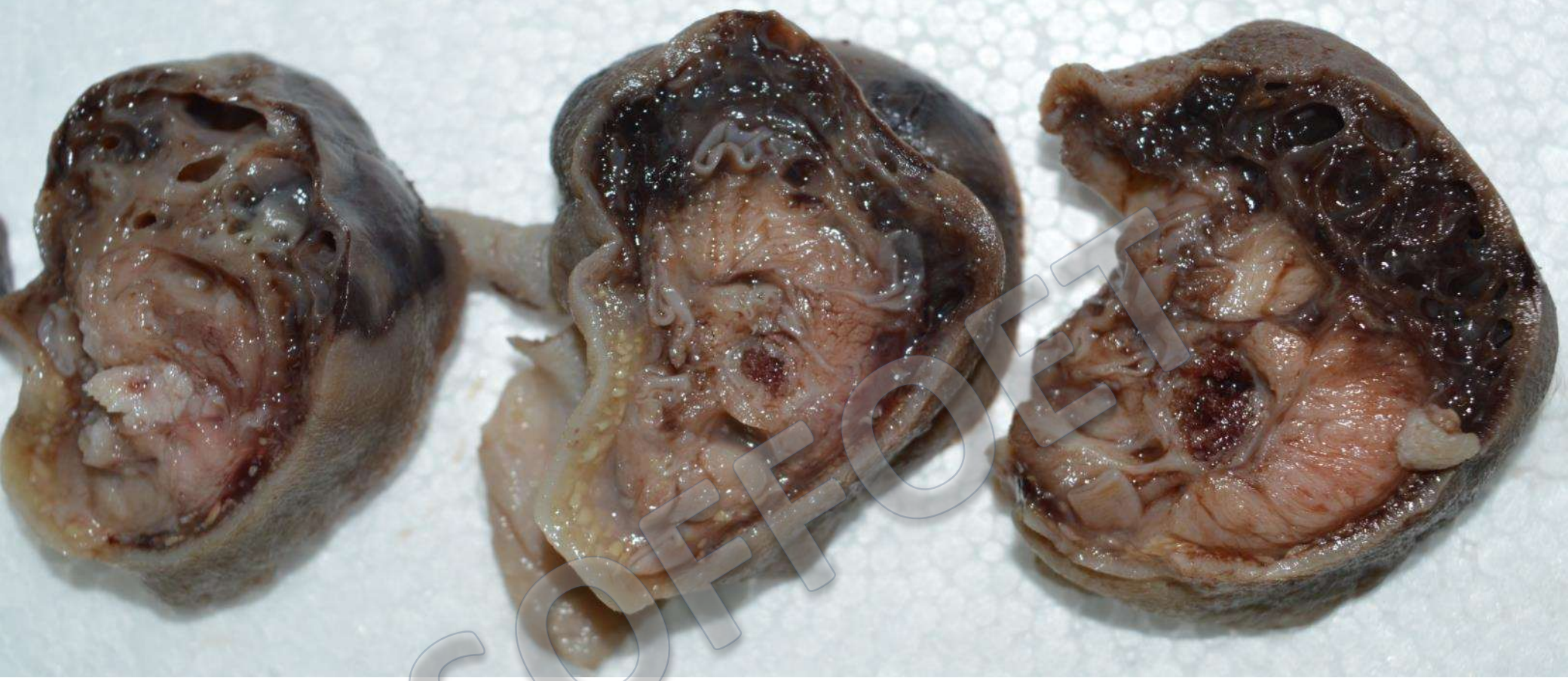


- The right axillary mass : large arteriovenous fistula of aneurysmal aspect, which was predominately fed by one vessel arising from the axillary artery, and emptied into a larger vessel tributary of the axillary vein.

- Other abnormal vascular shunts : bilateral anastomosis between the common carotid artery and the subclavian artery.

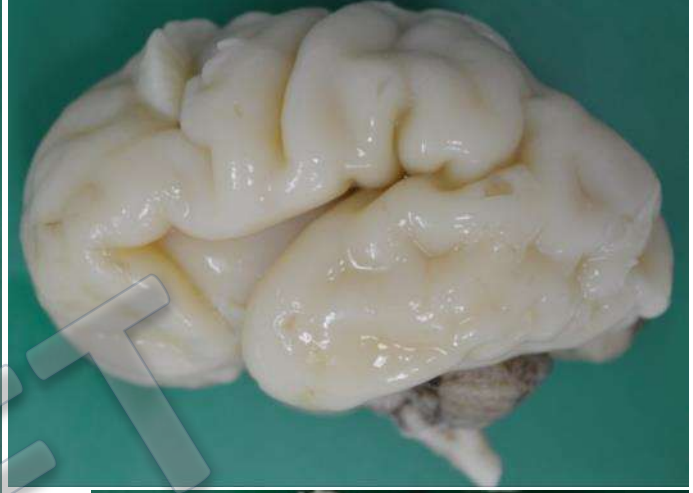


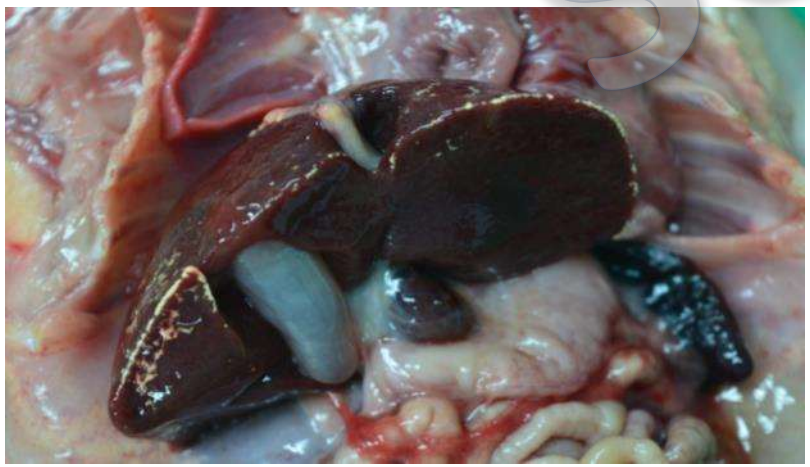


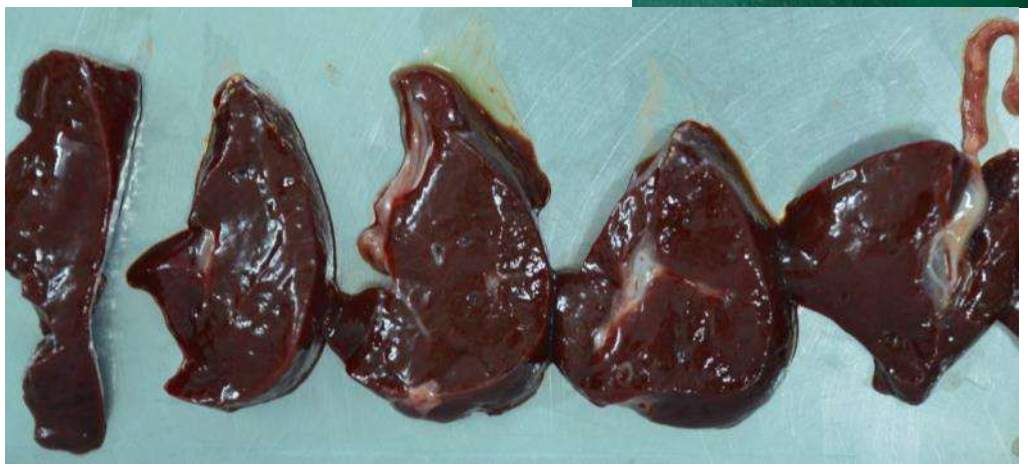


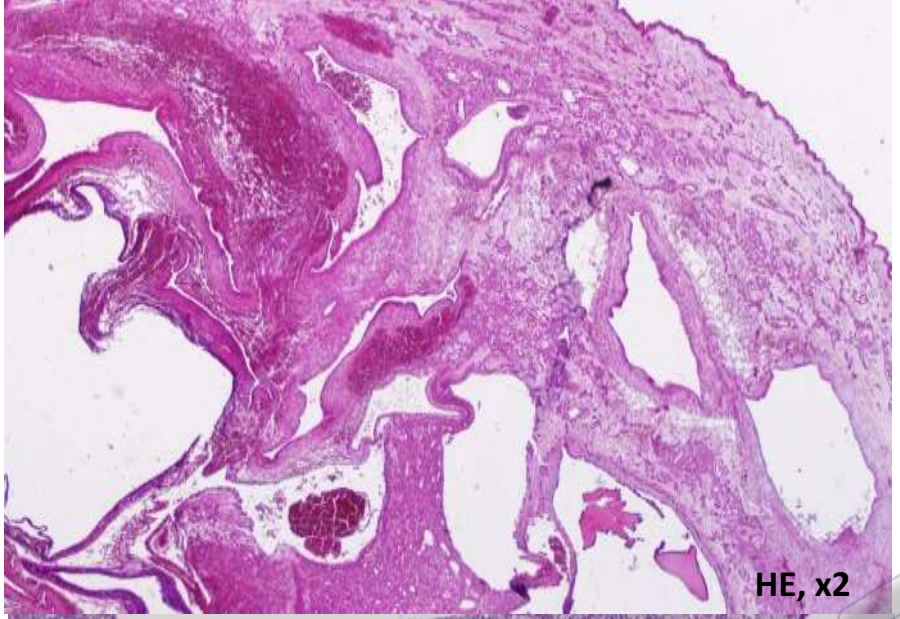
- Cut sections : tumor extension into dermal and subcutaneous layers with characteristic spongious architecture without involving the muscle.

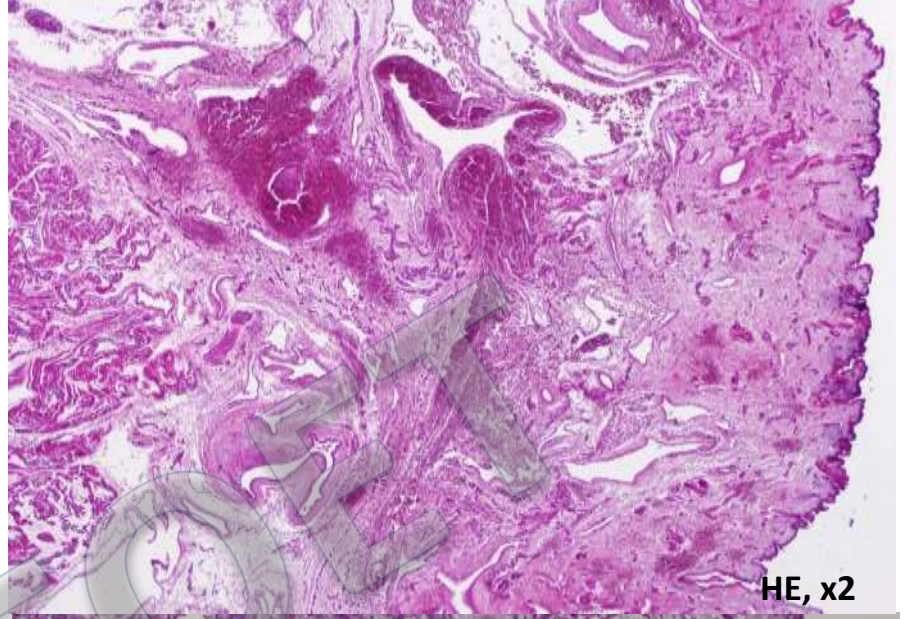
- Brain growth (144g; RR 185-200) and maturation were consistent with gestational age of **29-30 WG**, without structural abnormalities.
- No hemangioma was found in the liver or brain.

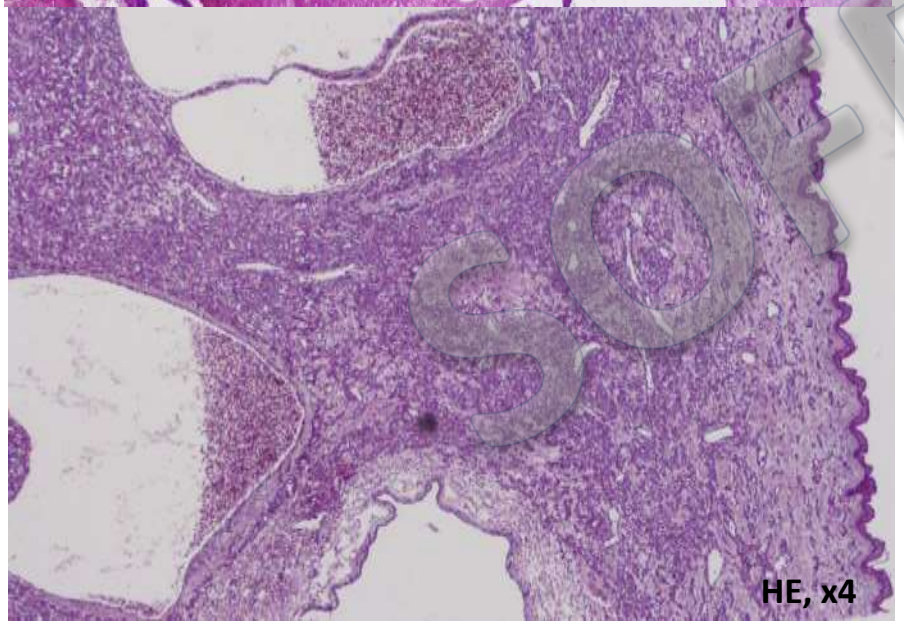


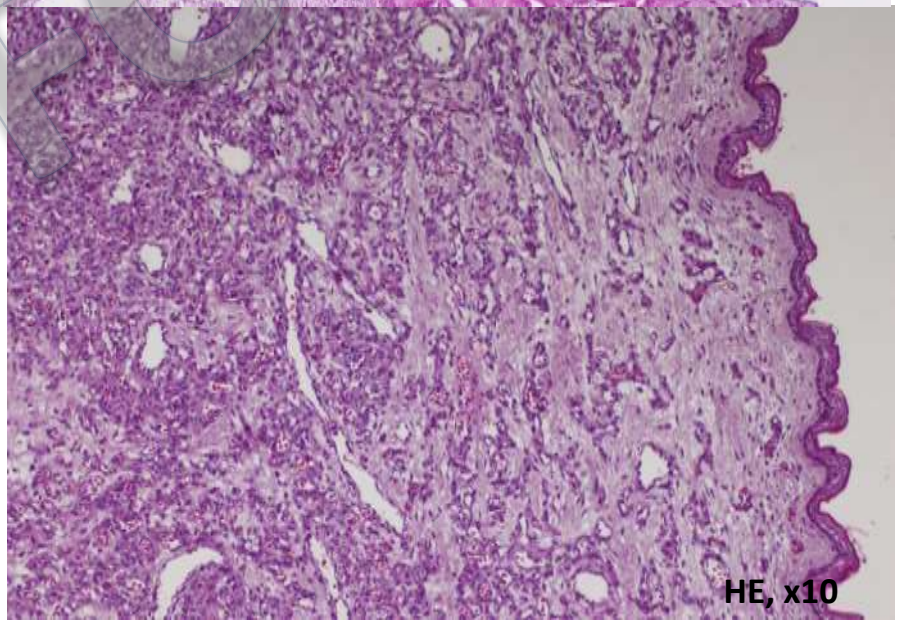








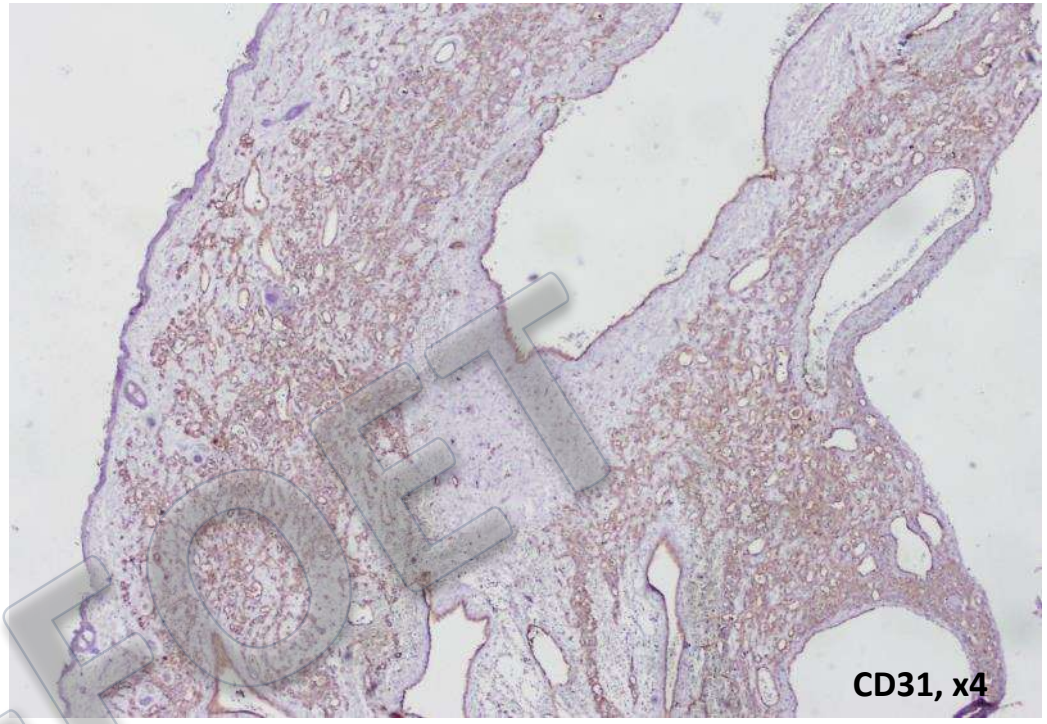




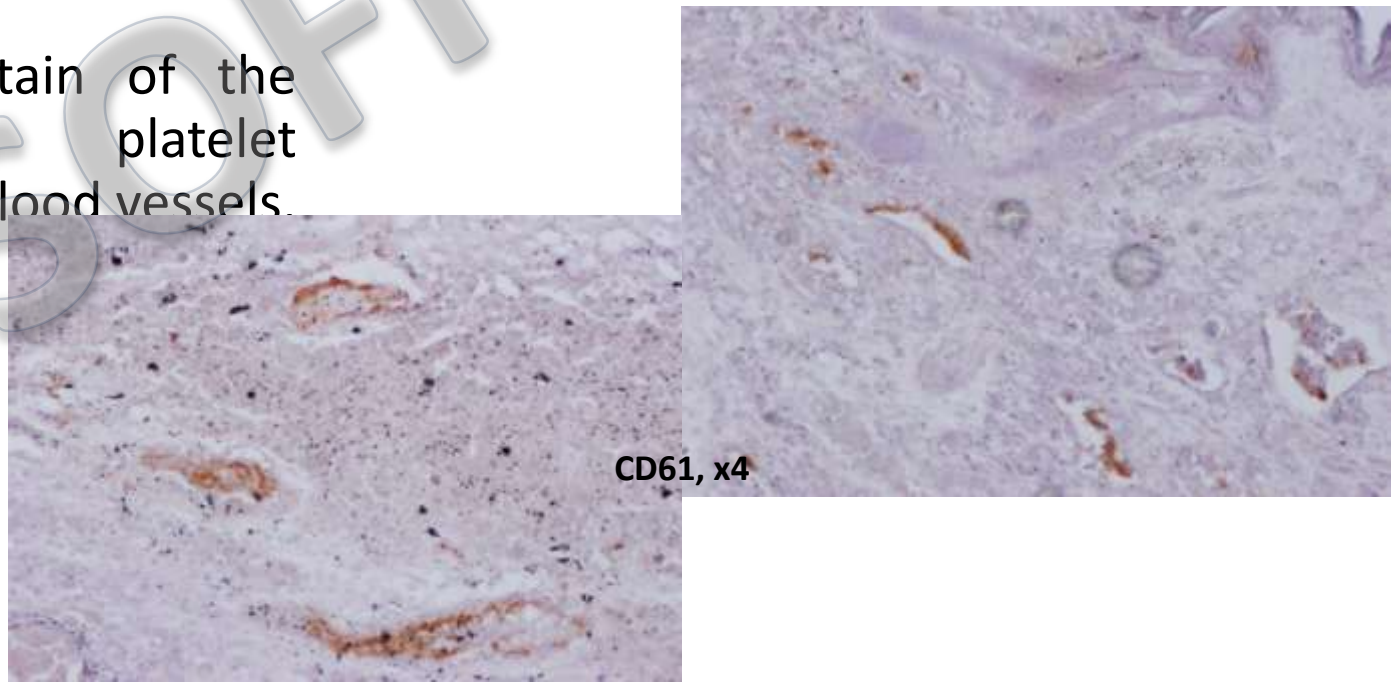
- **Histology: mixed capillary-cavernous hemangioma with many small capillaries lined by a thin single layer of endothelial cells supported by a connective tissue stroma and large, thin-walled vessels or sinusoids lined by a single layer of endothelium and separated by thin septa of connective tissue. The underlying skeletal muscle was not invaded by the lesion, but showed dilated vessels.**

IHC

- The dermal and subcutaneous vascular channels were lined by GLUT1-negative and mature CD31-positive endothelial cells.
- There was no Ki67 expression in the endothelial nuclei.



- The CD61 immunostain of the lesion showed platelet microthrombi in the blood vessels.



Additional findings

- Histological examination of liver sections showed hematopoietic islets with no evidence of other sites of extramedullary hematopoiesis. A faint Perls' staining was only seen in liver sections excluding an hemolytic process.
- There was no evidence of placental inflammation or membranes hemosiderosis. Perls' stain was performed on sections of fetal membranes that appeared negative.
- There was no evidence of erythroblastosis in placental and visceral vessels.

- Bone marrow showed increased number of megakaryocytes.

- The placenta was enlarged with a pale and friable appearance (weight 586.25g; normal ranges 270–290). Histology showed hydropic swelling of the villi and increased Hofbauer cells without specific lesions.

Discussion

- The aggressive clinical presentation of this vascular lesion : life-threatening complications (congestive heart failure, hydrops fetalis, coagulopathy).
- Vascular malformations increase the risk of hemodynamic complications.
- Elevated middle cerebral artery peak systolic velocity : arterioarterial shunts, hypertrophic cardiomyopathy, massive ascites
- Bilateral arterial carotid–subclavian anastomosis : these shunts seem to develop in response to the tremendous blood steal phenomena to ensure a sufficient blood supply to the brain.

  Defects in angiogenesis/vasculogenesis ? Atypical aortic arch branching variants ?
- The underlying pathophysiology is not fully understood. Research in the field of genetics of vascular anomalies has expanded over the last decade, leading to the detection of several causative somatic++ and germline mutations (proangiogenic function).
- The intriguing association between large congenital cutaneous hemangioma and vascular malformations as well as the symmetric intrauterine growth restriction raise the possibility of a common genetic mechanism.

PS-16-008

Foetal mediastinal teratoma: misinterpretation as congenital cystic lesions of the lung on prenatal diagnosis

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Background & Objectives: Teratomas are among the most common congenital tumours and frequently reported to be associated with foetal hydrops. The thoracic teratomas are commonly misinterpreted on prenatal ultrasound, being confused with congenital adenomatoid malformation of the lungs, diaphragmatic hernia or bronchopulmonary sequestration. The aim of this case report is to document this challenge.

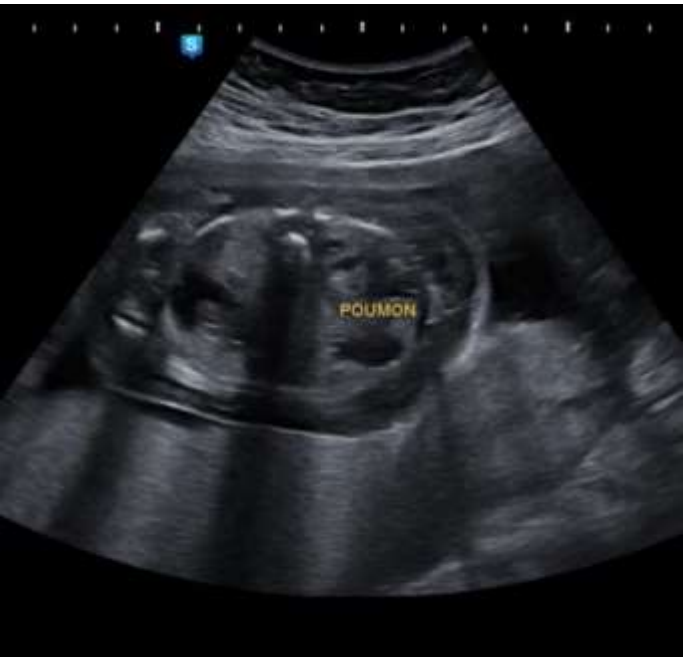
Methods: In the present case, we correlated prenatal diagnosis with autopsy findings of mediastinal teratoma complicated by severe hydrops foetalis.

Results: The ultrasound revealed severe hydrops foetalis, placentomegaly and multiple pulmonary cysts that were suggestive of cystic adenomatoid malformation of lung. Autopsy of the 24-weeks gestation stillborn male foetus showed a 54.3g, 6.1 x 4.4 x 2.8-cm, large and well encapsulated polycystic mediastinal teratoma with thymic, cardiac and pulmonary hypoplasia, and moderate post-hemorrhagic hydrocephalus. Histological study confirmed that the tumeral mass was consistent with immature teratoma and revealed placental diffuse chorangiomatosis and chorangiosis.

Conclusion: This case emphasizes that congenital mediastinal teratoma should be considered in the differential diagnosis of non-immune hydrops, particularly if a thoracic cystic lesion is detected on ultrasonography.

- A 31-year-old woman, G2P2, one full-term healthy male child, A positive, TORCH screening was negative
- Referred for investigation of hydrops fetalis
- The first morphology ultrasound perfomed at 17 weeks gestation didn't show anomalies
- Repeated fetal ultrasound, carried out at 23 weeks gestation, detected 1) placentomegaly, 2) severe hydrops fetalis with skin edema, ascites and pleural effusions, 3) heart displaced to the left hemithorax by a large mixed cystic and solid mass that was interpreted as cystic adenomatoid malformation of the lungs, 4) some hyperechoic spots or linear plaque with acoustic shadows
- Fetal karyotyping showed a 46, XY formula
- Pregnancy was terminated seven days later (24 WG) after parental consent as the outcome was predicted to be very poor

virchows arch (2019) 475(suppl 1): 1. https://doi.org/10.1007/S00428-019-02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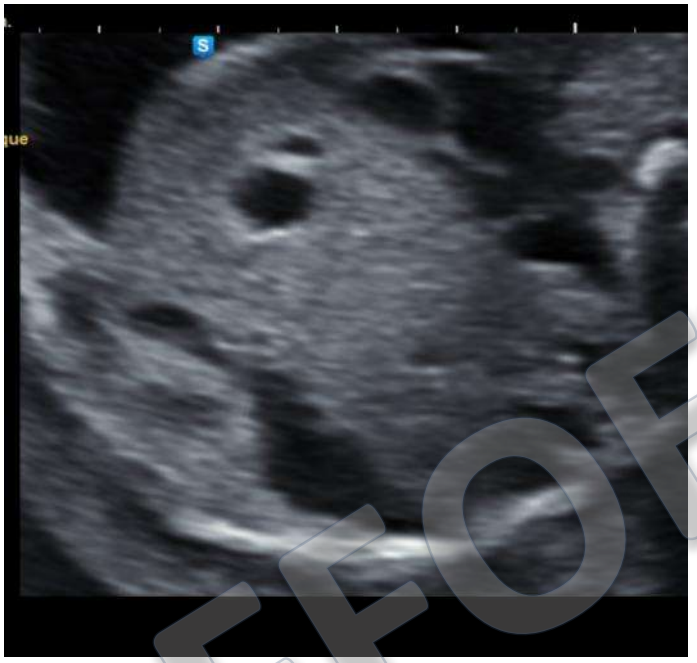

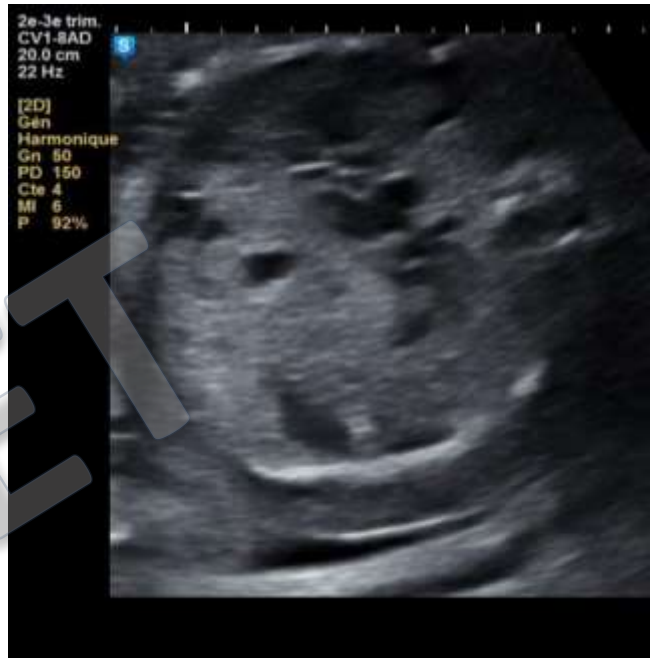
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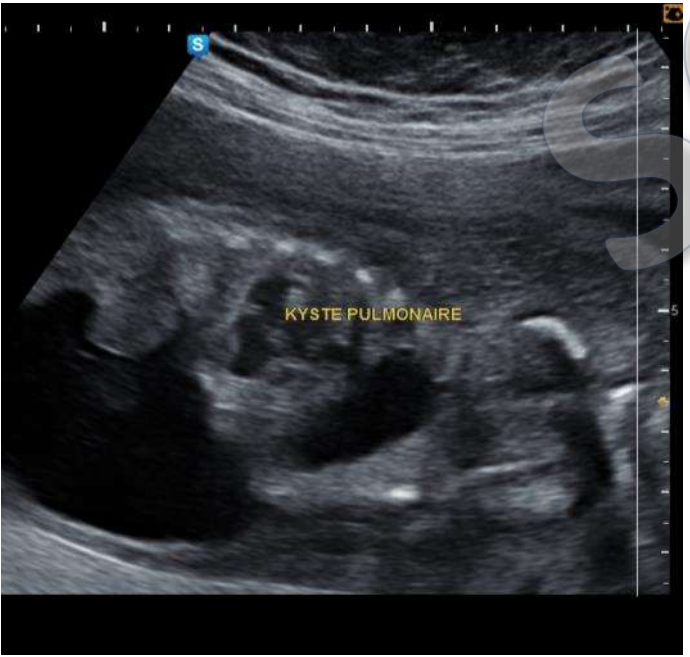
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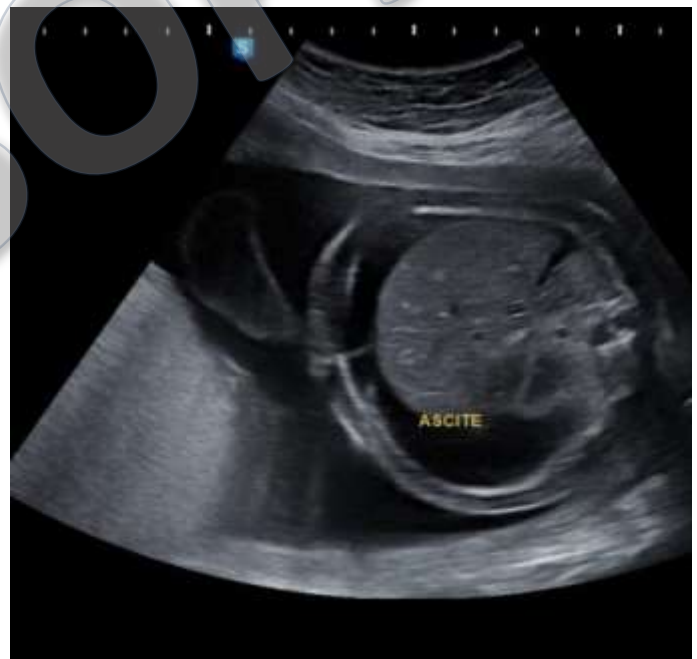
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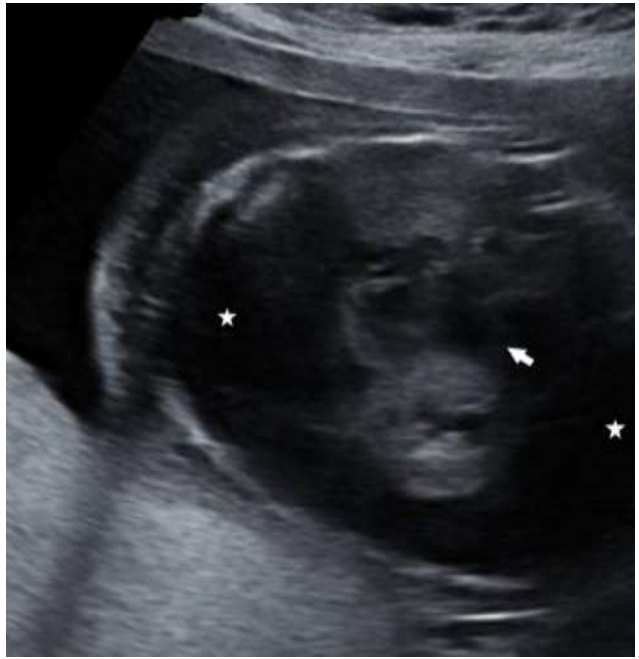
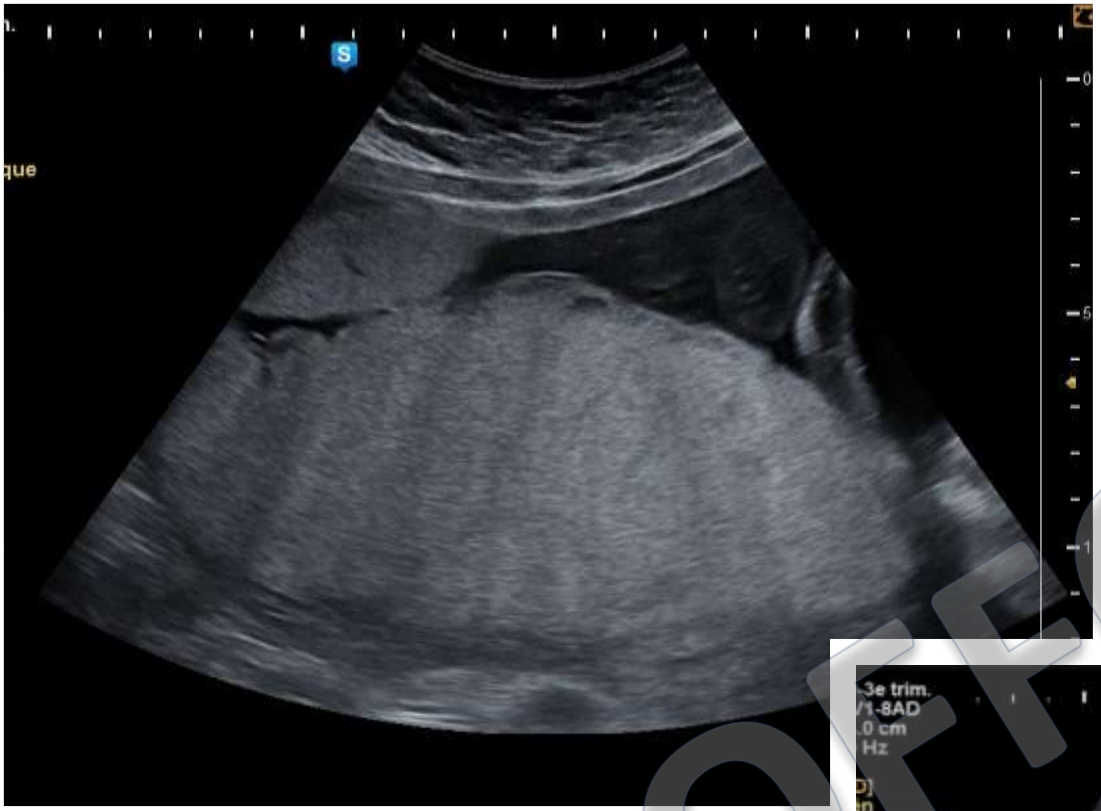
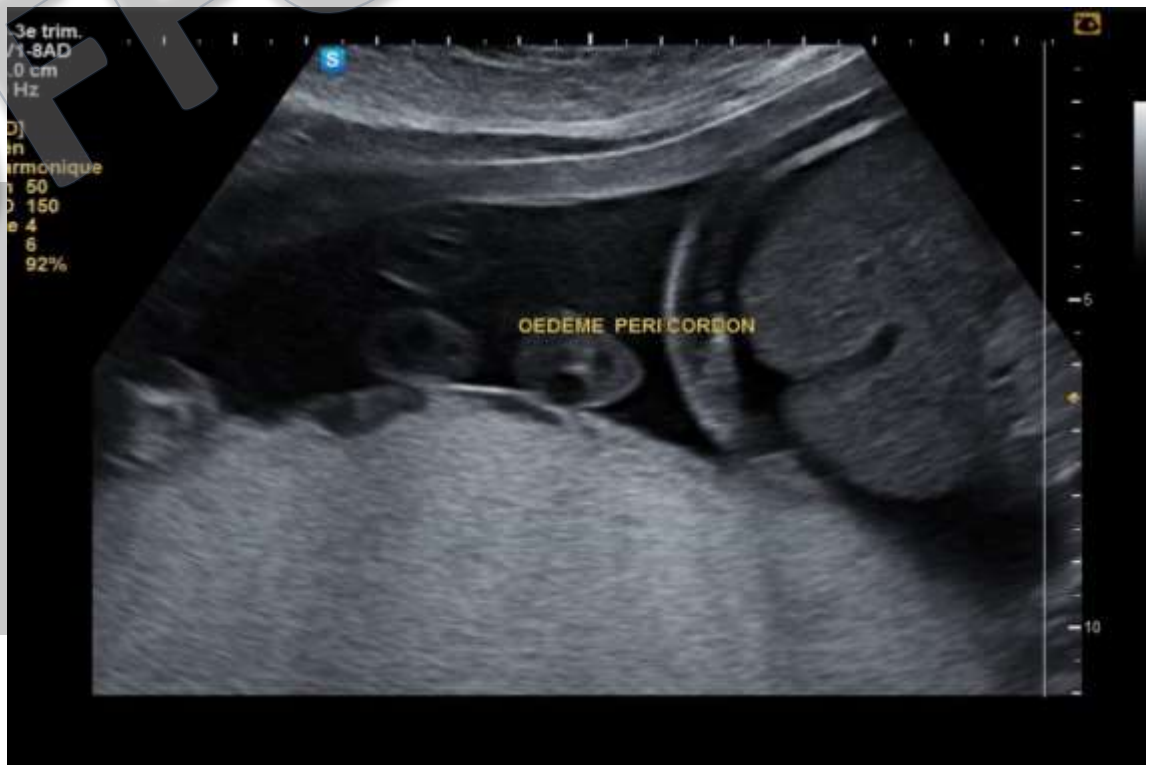


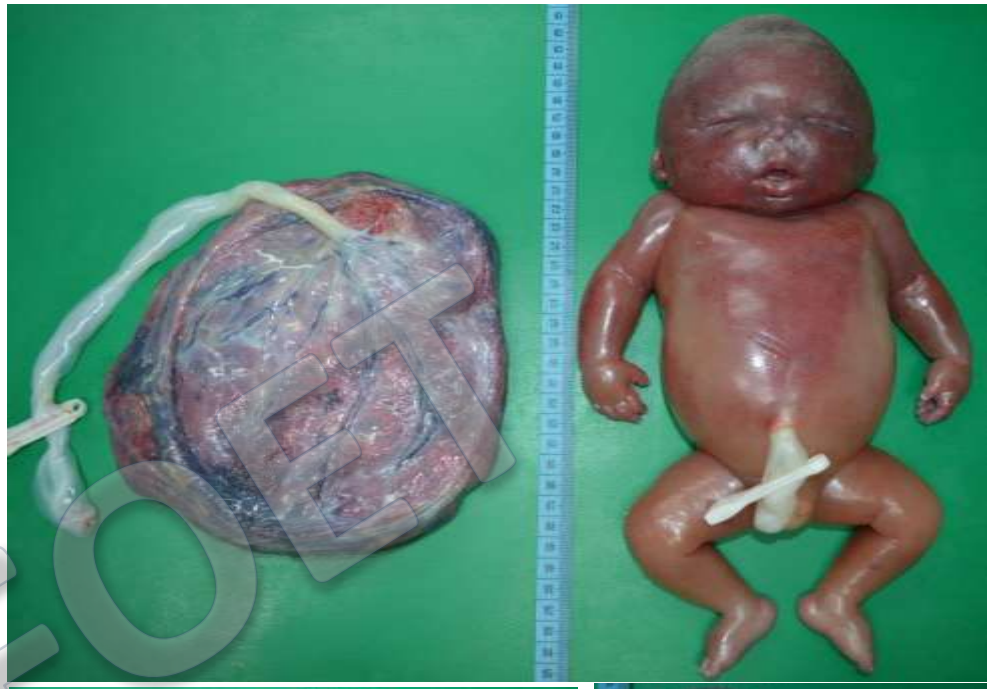


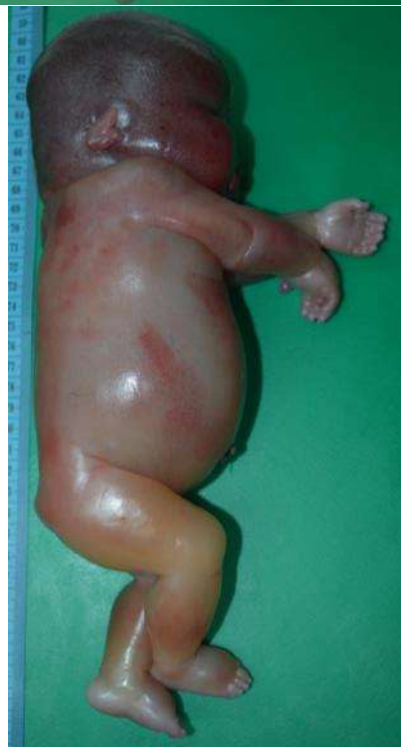


Autopsy

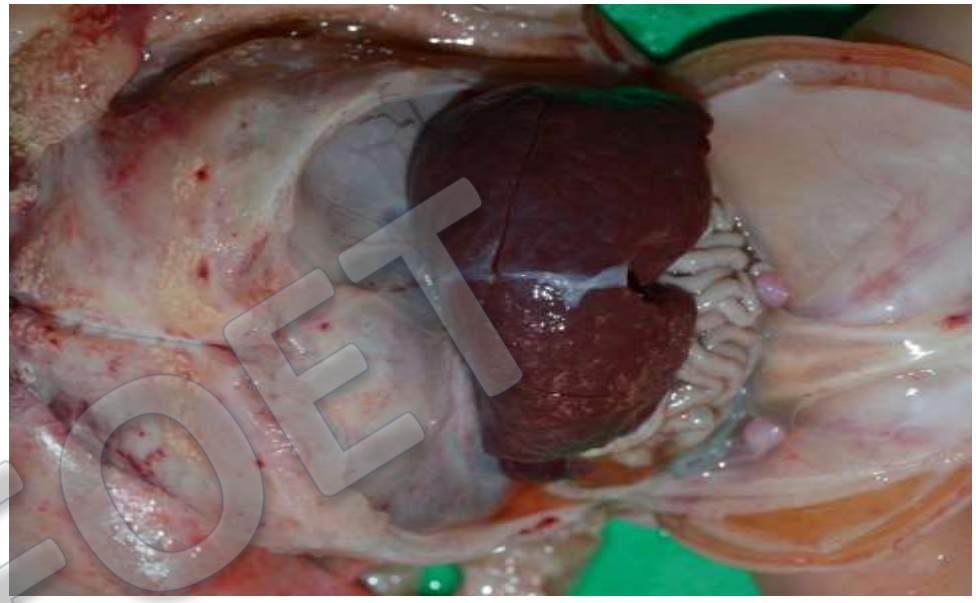
- Hydropic fetus weighed 1323.5g (expected ranges 540-630) with a crown to heel length of 30.5 cm which was within normal ranges for gestational age.

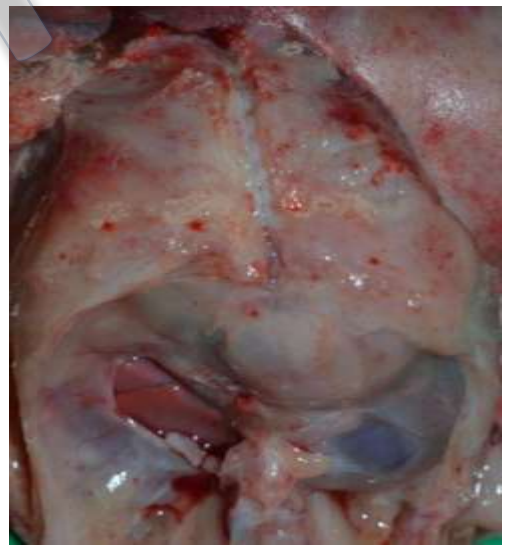
- Placentomegaly (weight 555g ; expected ranges 205-220). The placenta was dark-red despite the hydrops.

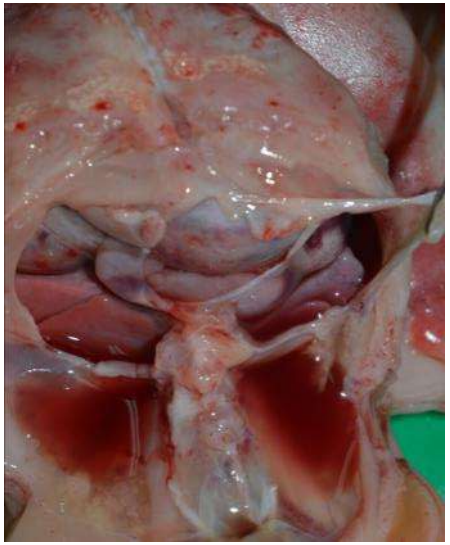


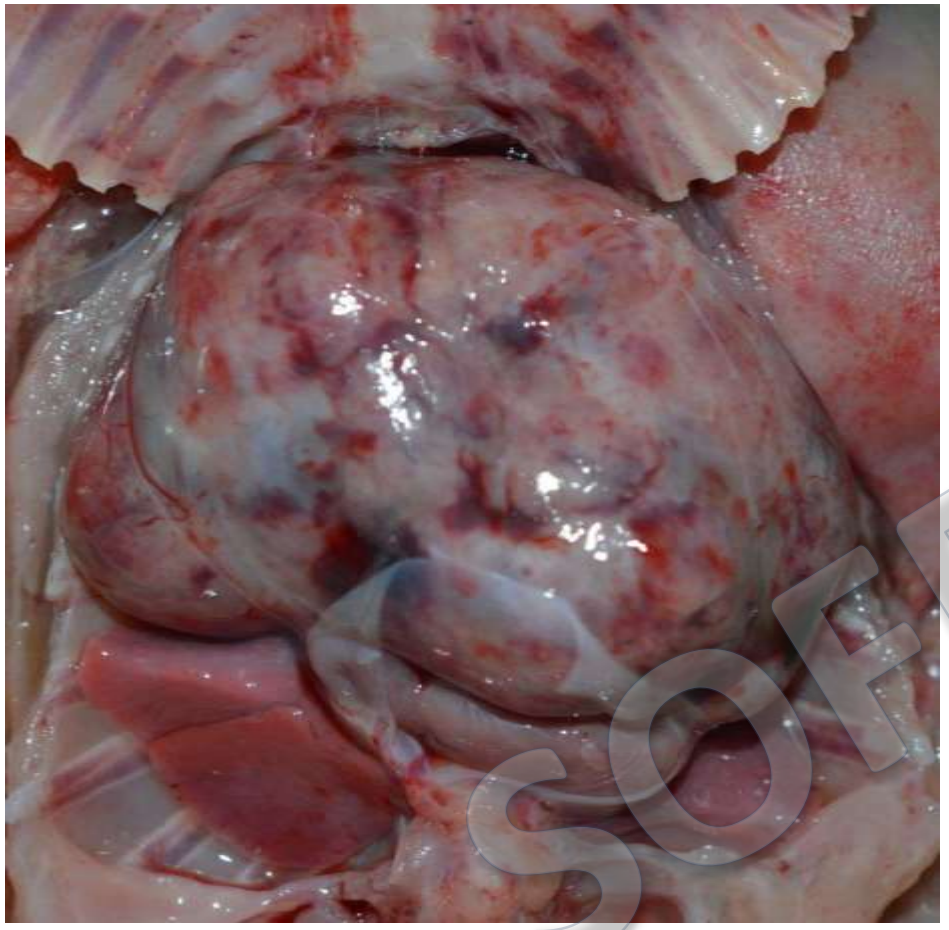


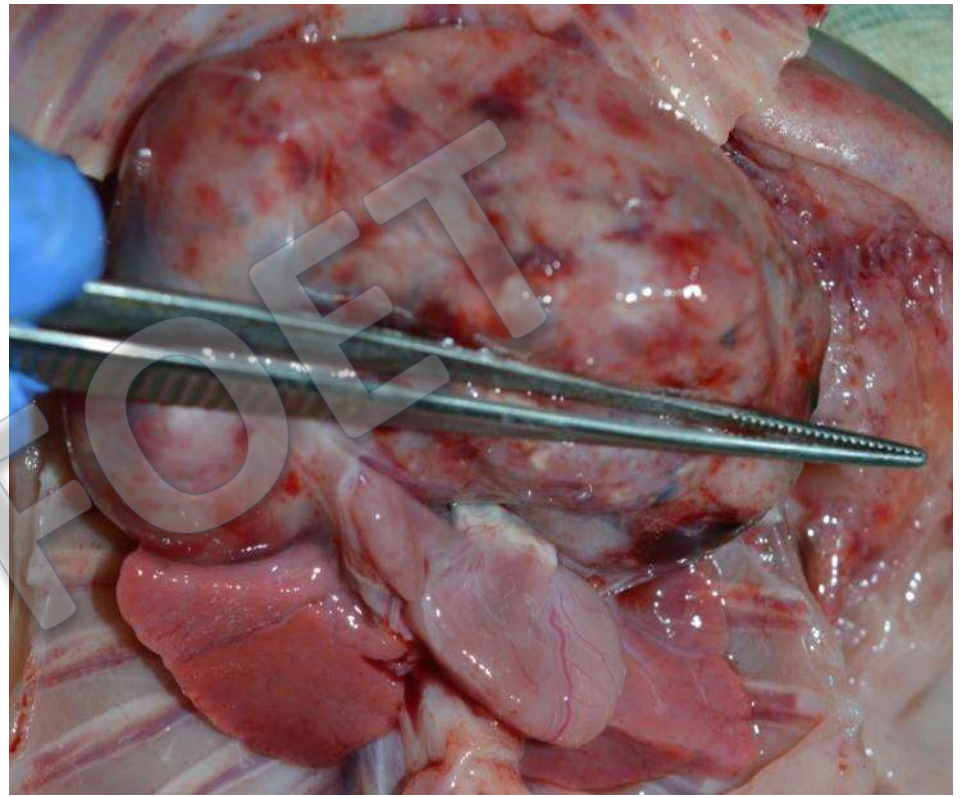
- Massive ascites and pleural effusions.







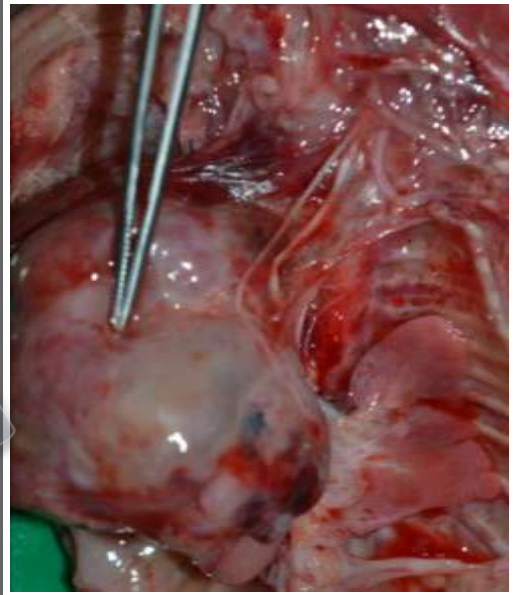


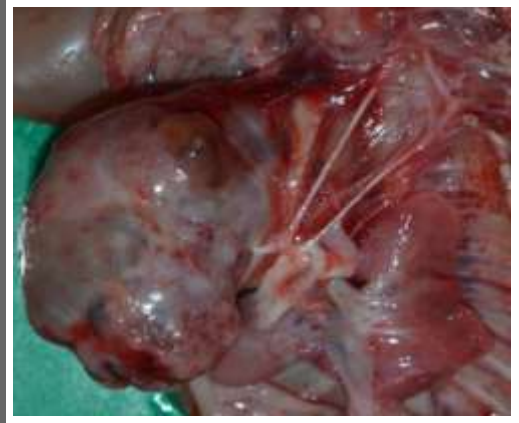


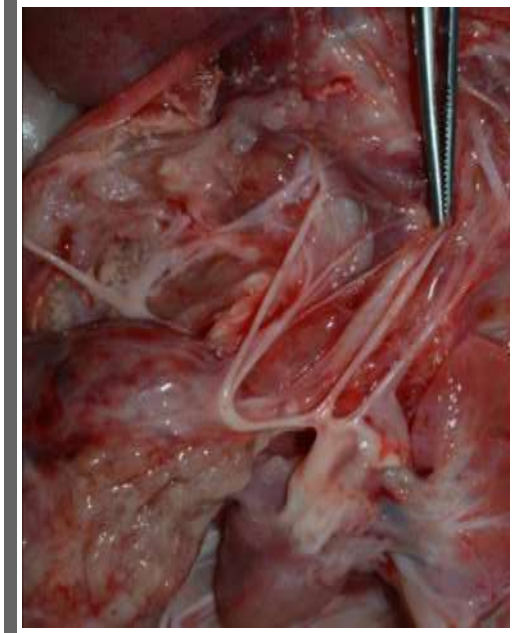
- Well-encapsulated lobulated mass, measuring 6.1x4.4x2.8cm and weighting 54.3g, occupying the thoracic cavity, displacing and compressing the heart, great blood vessels, airways and lungs.
- The tumoral mass appeared to arise in the anterior mediastinum and did not directly involve the thymus or invade the pericardium, heart, lungs or blood vessels
- The tumor capsule was fimly attached to the pericardium

- The tumor incorporated the great vessels, brachiocephalic artery, left common carotid and subclavian arteries and superior vena cava.

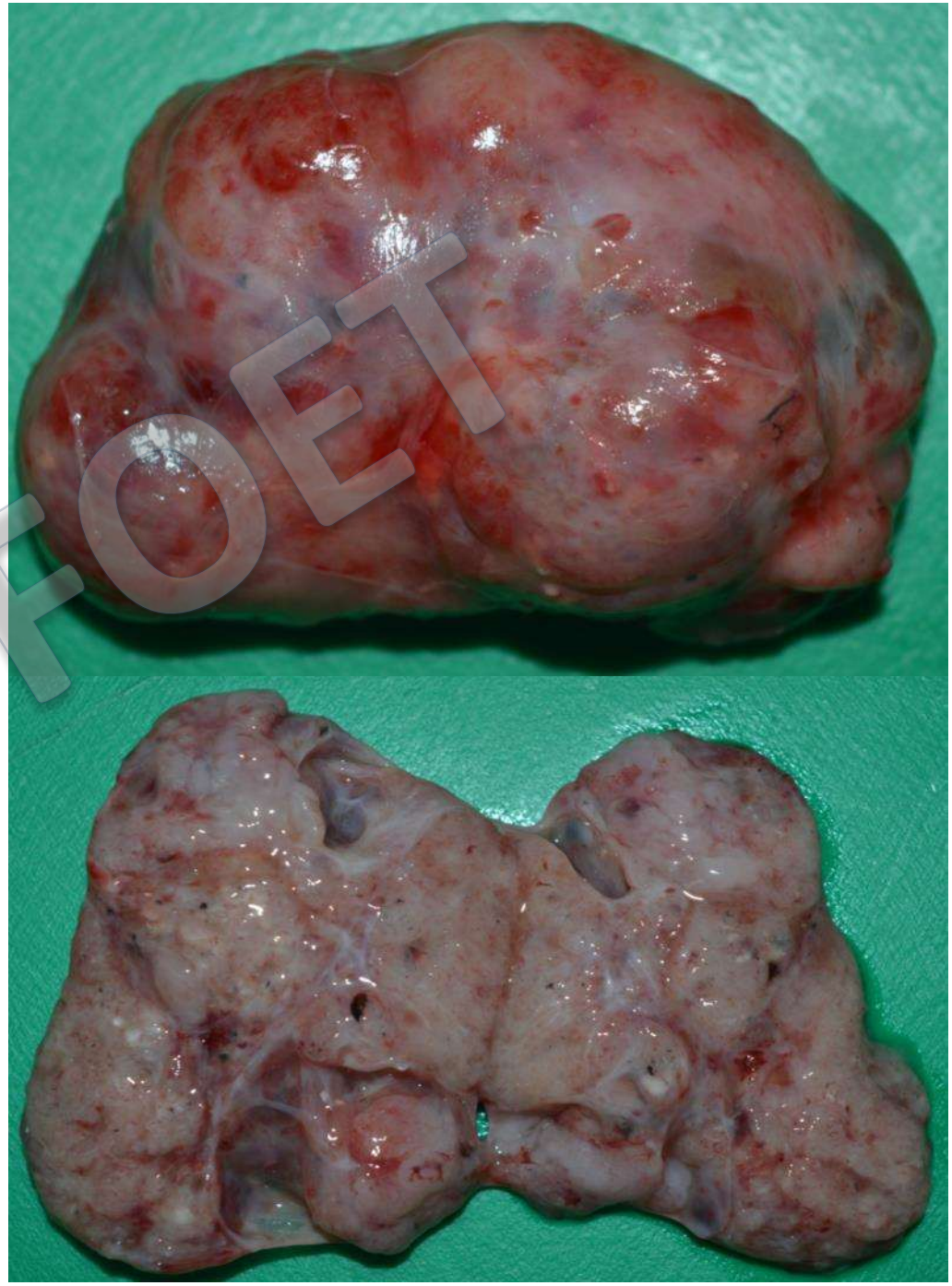
- The ectopic cervical thymus was very hypoplastic .



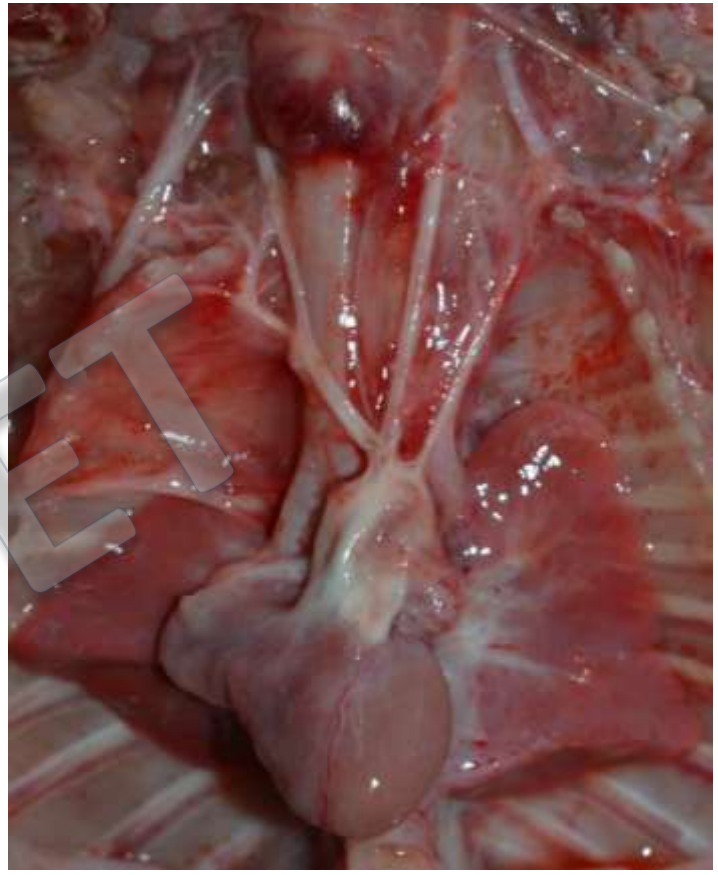


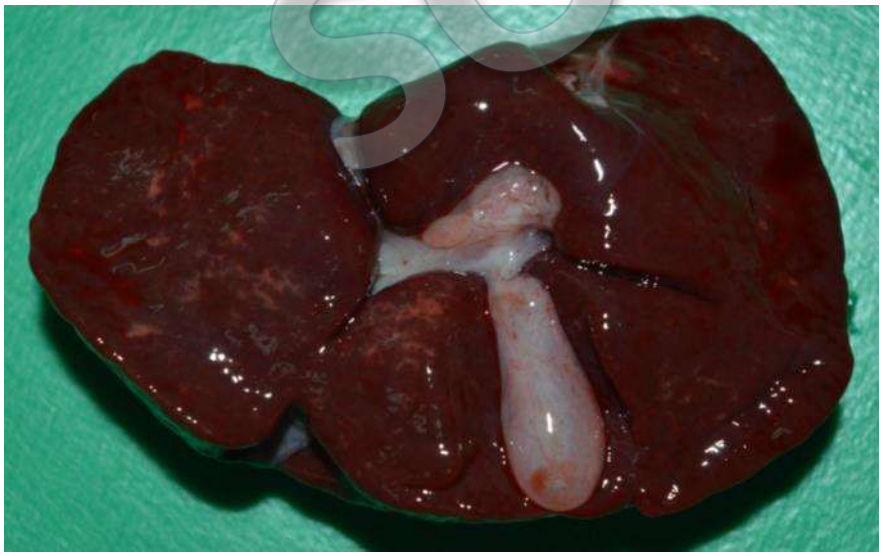


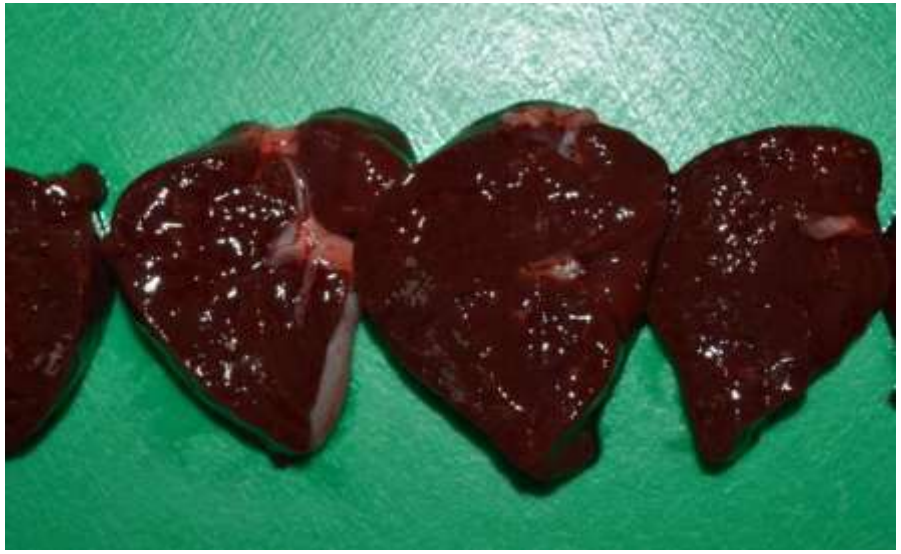
- The cut surface showed both solid and cystic components and calcifications.

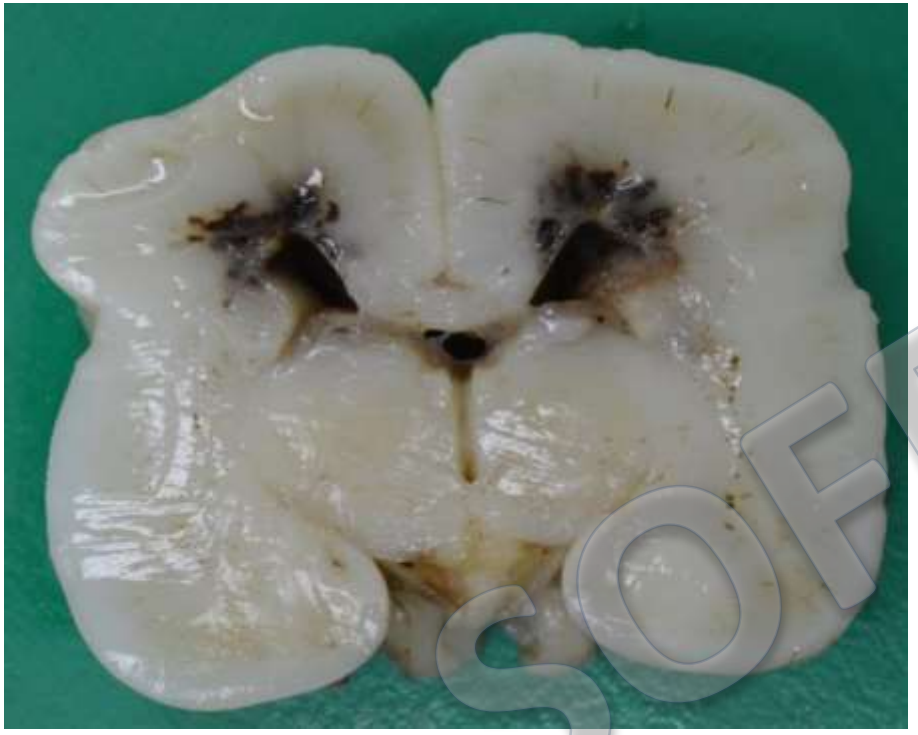


- The heart weight was below the lower limit of normal (2.35g ; expected ranges 3.5-4g).
- The combined lung weight was 6.35g (expected ranges 14.7-17g) with a lung weight to body weight ratio of 0.0047 (reference ratio ≥ 0.015).
- Liver was very enlarged (weight 31.9g; expected ranges 21-22.5).





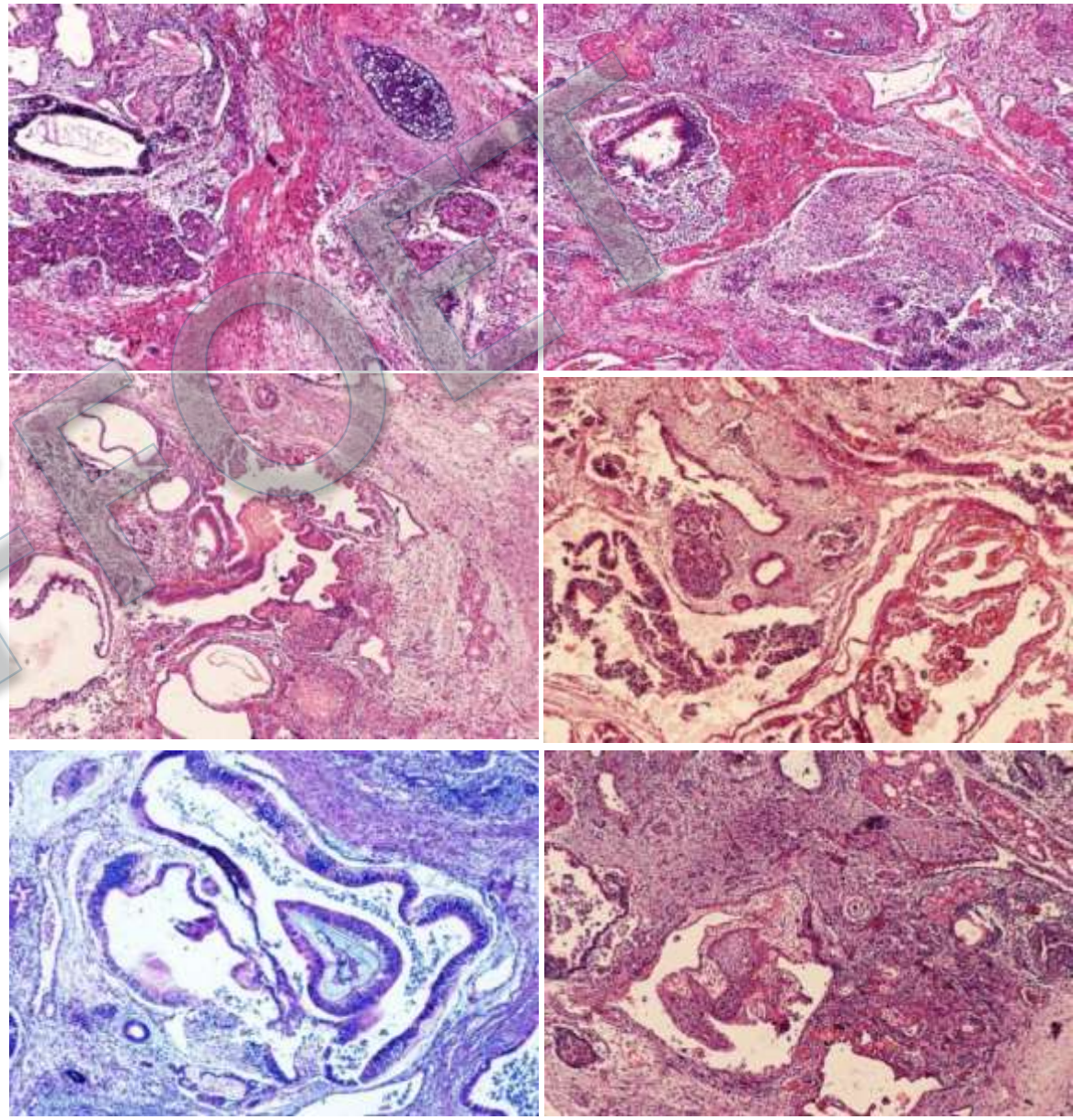




- Brain: massive and diffuse periventricular hemorrhage, diffuse hyperemia involving the gray and white matter, moderate bi-ventricular dilatation.

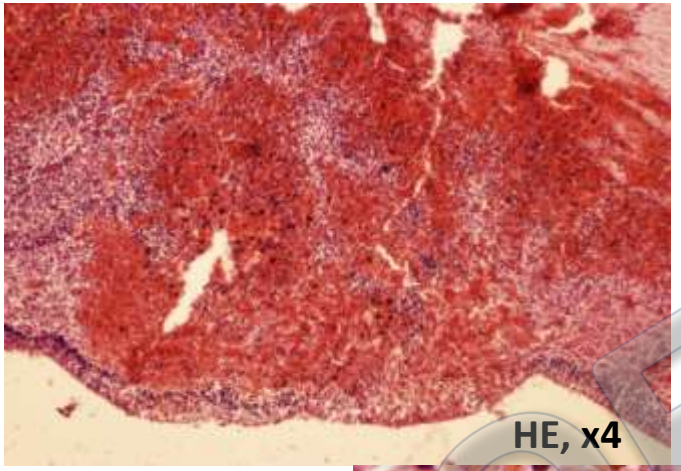
HISTOLOGY

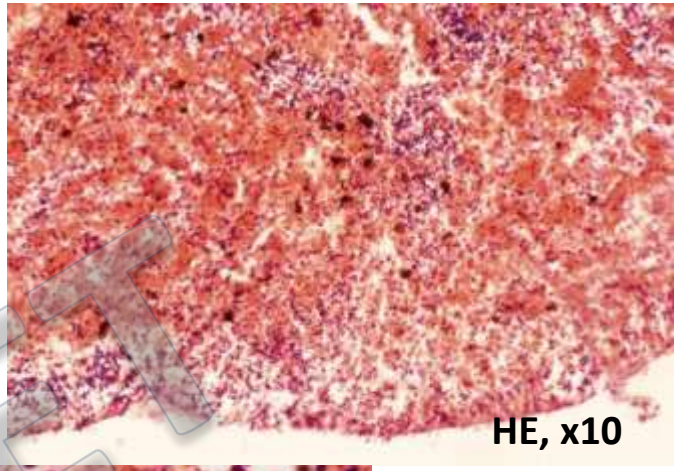
Tumor was composed of mature and immature tissues derived from the three embryonic germinal layers.

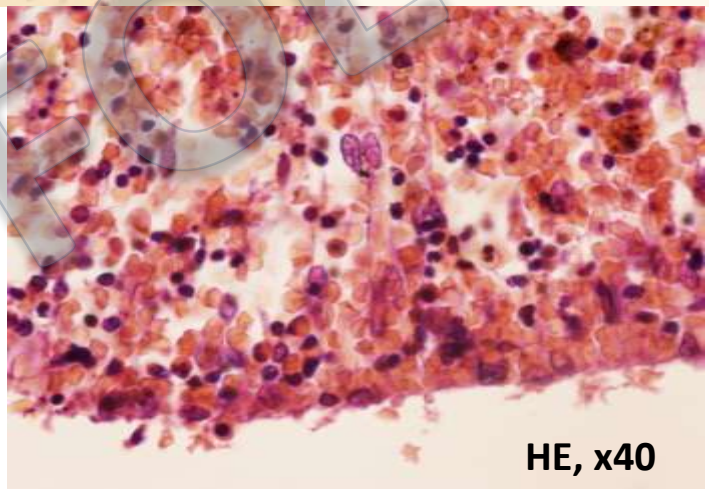


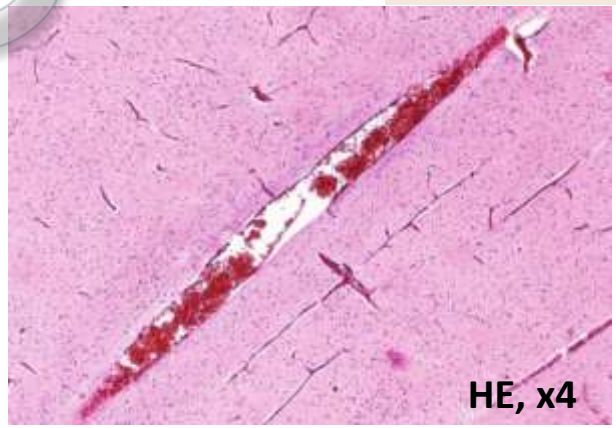
HE, x4

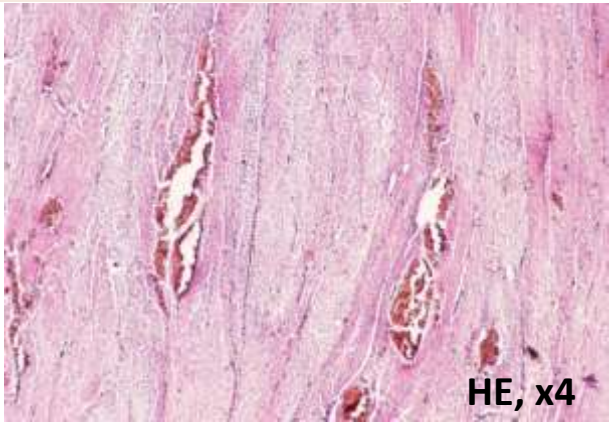
- Massive germinal matrix hemorrhage with extension into the lateral ventricles.
- Significant capillary congestion in the gray and white matter.



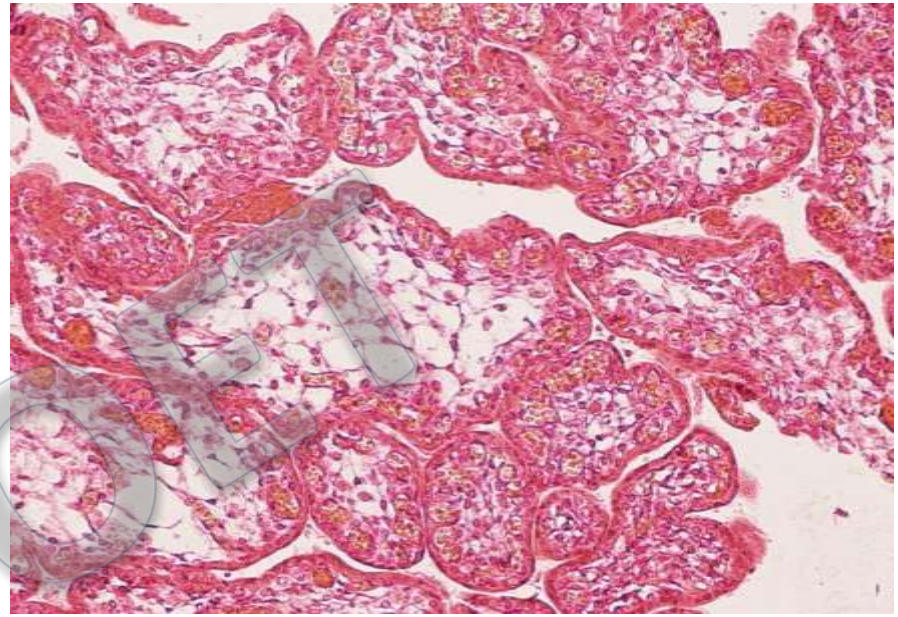


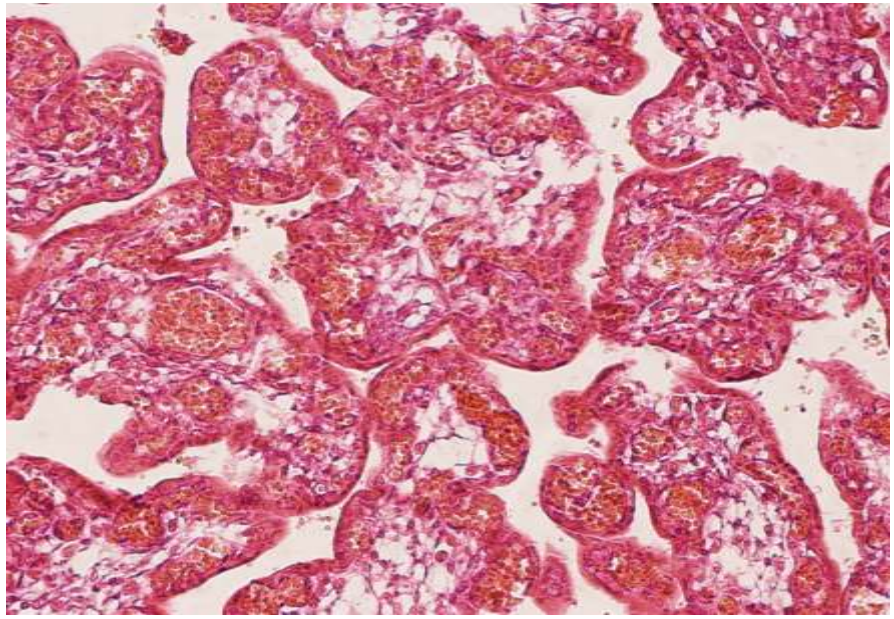






- Villous edema along with diffuse multifocal chorangiomatosis and chorangiosis without ischemic changes or fetal erythroblastosis.





Discussion

- **Prenatal diagnosis of the mediastinal teratoma**

Bronchogenic cysts, congenital cystic adenomatoid malformation of the lung, diaphragmatic hernia or bronchopulmonary sequestration, especially in the case of a large mass occupying the whole thoracic cavity, similarly to this case

➢cystic and solid components, calcification spots, acoustic shadows

Rapid and severe development of hydrops fetalis

- **Pathogenic mechanisms involved in the mediastinal teratoma associated fetoplacental complications**

➢The direct compression at the heart and great blood vessels, causing increase in venous pressure and decrease in the cardiac output (severe fetoplacental anasarca)

➢The compression effect of the teratoma at the heart and lungs (severe hypoplasia of these vital organs). Pulmonary hypoplasia may be as well explained by the compression effect of the massive ascites and pleural effusions

➢Decreased cardiac output (hypoxic-ischemic brain damage)

➢ Fetal hemodynamic changes (i.e., decreased cardiac output and obstruction of the venous return) may be the underlying mechanisms of the chorangiomatosis

- **Prognosis** : severe hypoplasia of the heart and lungs (limiting factor of survival); the outcome depends on the stage of development of mediastinal vital organs at the time of diagnosis

➢The extent of tumor and tumor effects suggest reasons for poor outcomes despite advancements in in-utero tumor resections. It is important to evaluate by ultrasonography not only lungs and heart development, but also brain compromise.

Conclusion

- These cases emphasizes that congenital **hemangioma** and **mediastinal teratoma** should be considered in the **differential diagnosis of non-immune hydrops**, particularly if a vascular anomaly or a thoracic cystic lesion is detected on Doppler ultrasound, respectively.

- Although hese tumors are benign, they may cause perinatal death because of their significant size and hemodynamic complications.

- The **full autopsy is strongly recommended** following fetal demise as it gives **significant additional informations**, particularly on **vascular malformations** (hemangioma) and **cerebral ischemic changes** (teratoma) which are poorly investigated on prenatal imaging.