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Fetal endocrine organ abnormalities: prenatal ultrasound–autopsy correlation revealing occult endocrine and placental pathology

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Introduction

Congenital abnormalities of fetal endocrine organs are uncommon and remain poorly characterized in prenatal pathology. The pituitary, thyroid, and adrenal glands play pivotal roles in fetal growth, maturation, and homeostasis. Developmental disturbances affecting these organs may contribute to malformations, growth abnormalities, and adverse perinatal outcomes. Visualization of fetal endocrine structures during routine antenatal ultrasonography is challenging, resulting in potential underdiagnosis.

Aims & Objectives

To explore the occurrence of abnormalities in major fetal endocrine organs, with an emphasis on the associated placental pathology and the subsequent effects of these abnormalities on the development and function of other fetal organs.

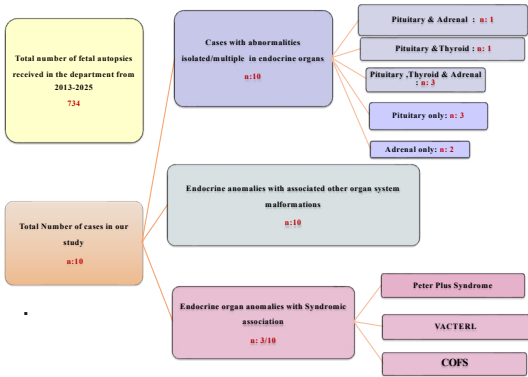
Objective:

- To identify the occurrence of abnormalities in major endocrine organs of fetus.
- To identify the associated placental pathology these abnormalities.
- To analyze the possible impact of abnormal endocrine organs in the other organs of the fetus.

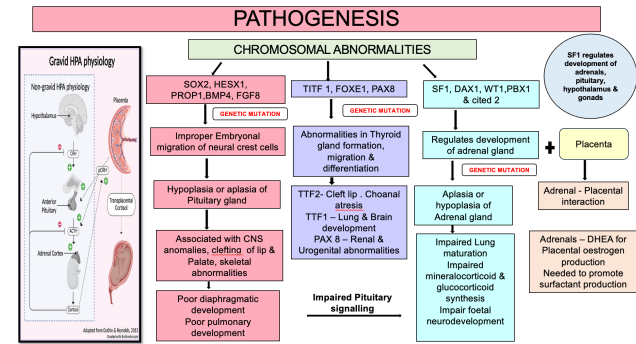
Materials And Methods

A retrospective descriptive study was performed over a 13-year period in the Department of Pathology at a tertiary care centre in South India. Clinical details, prenatal ultrasound findings, and detailed autopsy examinations were reviewed. Fetuses demonstrating abnormalities of endocrine organs were analyzed with respect to gestational age, associated congenital anomalies, and placental histopathology

Results:



Associated placental and cord abnormalities included Fetal vascular malperfusion, Villous oedema, Intervillous thrombohematoma, Hyper coiled cord, Single umbilical artery, Velamentous cord insertion, Retroplacental hematoma and Hypoxia induced changes



Discussion:

Fetal endocrine organ abnormalities are rare but clinically significant, occurring in only 10 of 734 fetal autopsies, yet they were consistently associated with complex multisystem congenital malformations.

None of the fetal endocrine organ abnormalities were detected on prenatal ultrasound, highlighting the limitations of antenatal imaging for these rare anomalies.

Pituitary abnormalities were the predominant endocrine anomaly, frequently occurring with Cleft lip & Palate, ocular and central nervous system malformations

Adrenal and thyroid abnormalities were frequently associated with multisystem congenital malformations, reflecting the integral role of these endocrine organs in foetal development

Placental pathology was consistently observed in association with fetal endocrine abnormalities, with features of fetal vascular malperfusion, villous oedema, cord abnormalities, and hypoxia-related changes, suggesting a close fetoplacental interaction in these developmental disorders.

Conclusion

Perinatal autopsy is indispensable for identifying occult foetal endocrine abnormalities not detected on prenatal ultrasound, defining their association with complex multisystem congenital anomalies and placental pathology, and providing crucial insights into foetal development, adverse perinatal outcomes, and counselling for future pregnancies

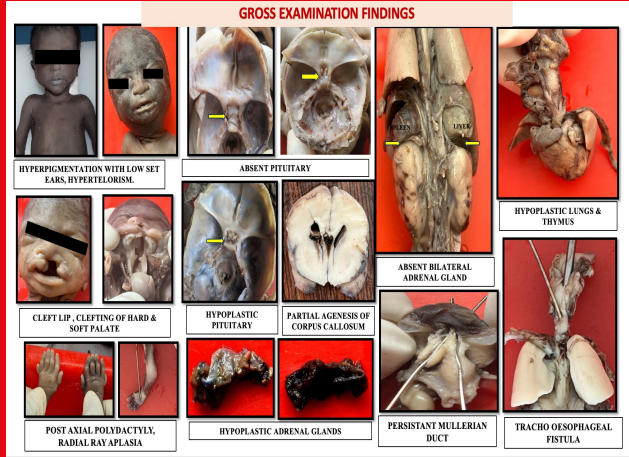


Figure 1. Gross examination findings at autopsy

FETAL GROWTH RESTRICTION	5
SKIN HYPERPIGMENTATION	2
HIRSUTISM	1
CLEFT LIP & PALATE	4
OCULAR ANOMALIES – MICROPTALMIA, ANOPHTHALMIA	2
FACIAL DYSMORPHISM	6
PULMONARY HYPOPLASIA	3
CARDIOVASCULAR ABNORMALITIES (ASD, VSD, DOUBLE OUTLET RIGHT VENTRICLE, PERSISTENT LEFT SVC, FLABBY HEART, BICUSPID AORTIC VALVE)	6
CENTRAL NERVOUS SYSTEM ANOMALIES (MICROCEPHALY, HYDRANENCEPHALY, PARTIAL AGENESIS OF CORPUS CALLOSUM, CEREBELLAR HYPOPLASIA, OCCIPITAL ENCEPHALOCOELE, HYPOPLASTIC CEREBELLUM)	6
THYMIC HYPOPLASIA	3
PERSISTENT UROGENITAL SINUS	1
POLYCYSTIC RENAL DYSPLASIA	1
SKELETAL ABNORMALITIES (RADIAL APLASIA, ABSENT THUMB, POST AXIAL POLYDACTYLY, B/L CTEV, FIXED FLEXION DEFORMITY)	4
PERSISTENT MULLERIAN DUCT SYNDROME	1

Figure 2. Associated abnormalities in other organ systems