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Correlation of prenatal ultrasound with fetal autopsy in central nervous system malformations: a neuropathological study

Nithyanandham V2, Umamaheswari Gurusamy2, Lalitha Natarajan1
INSTITUTION: 1. Fetal Medicine, PSG Institute of Medical Sciences and Research, Coimbatore, India.
2. Pathology, PSG Institute of Medical Sciences and Research, Coimbatore, TN, India.

Introduction

CNS malformations are among the most common congenital defects, with a prevalence of 5 to 10 per 1000 births. While radiological imaging has advanced significantly, a structured fetal autopsy remains critical to recognizing exact phenotypes, identifying syndromic associations, and providing a quality check for clinical audits. The study aimed to determine the spectrum of these malformations and correlate autopsy findings with antenatal imaging

Materials and methods

This was a retrospective analysis of 325 fetuses received for autopsy examination at our institution between January 2018-March 2025. 77 cases with CNS malformations/anomalies were identified. Antenatal ultrasound findings were reviewed and correlated with detailed neuropathological examination at autopsy. Cases were categorized as isolated CNS malformations, complex CNS defects, CNS malformations associated with other major system anomalies, and syndromic associations. CNS anomalies were further classified according to their anatomical distribution

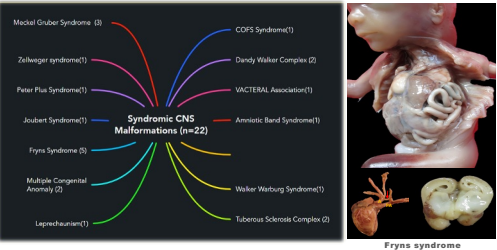
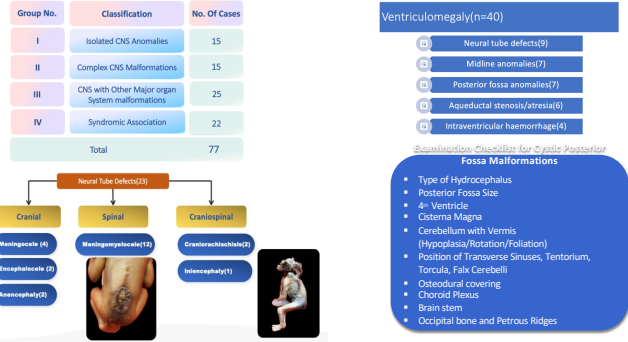
Results:

Spectrum of CNS Malformations

The study categorized 77 cases into several overlapping anatomical distributions:

- Ventricular System Abnormalities (50%):** The most frequent finding, primarily ventriculomegaly and aqueduct stenosis.
- Neural Tube Defects (30%):** Included myelomeningocele (the most common NTD), anencephaly, and rare cases like iniencephaly.
- Midline Anomalies (29%):** Agenesis of the corpus callosum was the primary finding (n=18).
- Posterior Fossa Anomalies (23%):** Included global cerebellar hypoplasia and Dandy-Walker malformation

Systemic and Syndromic Associations: Only 15 cases were isolated CNS anomalies; the rest involved other organ systems (25 cases) or specific syndromes (22 cases), such as **Fryns Syndrome**, **Meckel-Gruber Syndrome**, and **Walker-Warburg Syndrome**.



the most frequent extra-cranial organ system involvements were: **Musculo-skeletal system (32%), Cardiovascular System (CVS) (24%), Multiple Organ System Malformations (20%), Cranio-facial Dysmorphism (12%) and Gastrointestinal Tract (GIT) (8%)**: The specific cardiovascular findings identified across various cases and syndromes include **Septal Defects, Hypoplastic Heart Conditions, Outflow Tract and Great Vessel Anomalies, cardiac rhabdomyomas and anomalies of the venous system**

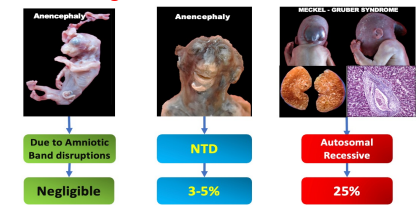
The Cardinal Finding: Autopsy vs. Ultrasound

The study highlights the superior diagnostic yield of autopsies over antenatal Ultrasound (USG):
37% Concordance: Near-complete agreement between USG and autopsy.

57% Major Additional Findings: Autopsy identified significant diagnostic information missed by USG, particularly for complex syndromes and internal malformations.

6% Discordance: Cases where USG and autopsy findings were entirely incomparable.

Counseling on risk of recurrence



Conclusion:

Perinatal autopsy remains the **"gold standard"** for diagnosing CNS malformations. It provides the critical **"final diagnosis"** by establishing an exact etiopathogenesis, which is essential for accurate genetic counselling regarding recurrence risks in future pregnancies.

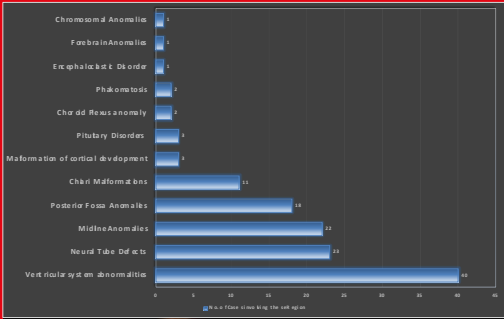


Figure 1. Anatomic categorization of fetal CNS anomalies



Figure 2. Ventriculomegaly



Figure 3. Posterior fossa malformations