

Gestational Trophoblastic Disease

Gestational trophoblastic disease (GTD) is a major disease category in diagnostic gynecological pathology. Among common clinical specimens, morphologic assessment of hydatidiform moles continues to suffer from significant diagnostic inaccuracy and inter- and intra-observer variability, even among experienced pathologists. However, distinction of hydatidiform moles from non-molar specimens and precise sub-classification of hydatidiform moles are critical for patient care, as the risk of post-molar gestational trophoblastic neoplasms varies among subtypes of hydatidiform moles and so does the duration of clinical follow-up.

During the past decade, p57 immunohistochemistry and PCR-based DNA genotyping have emerged as powerful diagnostic measures to precisely classify hydatidiform moles. The modern diagnostic approach to molar gestations now requires integration of these ancillary techniques into an algorithm aiming to provide a refined diagnosis, accurate risk assessment of post-molar gestational trophoblastic neoplasms, and to guide appropriate clinical management.

At Yale Pathology, experienced gynecological pathologists are pioneers in the clinical validation and applications of molecular genotyping diagnosis of GTD. Our expert consultants offer diagnostic services to pathologists throughout the United States and abroad. As a leading institution, Yale Gynecologic Pathology has recently optimized the diagnostic algorithm with integration of histology, molecular genotyping and immunohistochemistry in the precise classification of molar gestations and gestational trophoblastic tumors. Yale expert consultants are authoritative writers in the field of GTD and are expert editorial member/chapter contributors to the 4th and 5th editions of the WHO blue book on classification of gestational trophoblastic disease.

- **Precise diagnosis of GTD is essential for patients**
- **In the last 10 years, p57 immunohistochemistry and PCR-based DNA genotyping have emerged as powerful diagnostic measures to precisely classify hydatidiform moles**
- **Yale Gynecologic Pathology has world-renowned experts who have pioneered integrating molecular technology into routine diagnoses of GTD**

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Our Services:

Histological Evaluation

The first step in the diagnostic work-up of GTD is morphologic evaluation on H&E-stained sections. Based on the microscopic features, the cases are triaged for further ancillary studies using published algorithm. Correlative morphological review is crucial for correct interpretation of ancillary studies, particularly molecular genotyping in the diagnosis of molar gestations.

Immunohistochemistry

P57 immunostain is often used in the diagnostic work-up of hydatidiform moles, particularly in the confirmation of complete moles. A variety of other immunohistochemical stains – hCG, hPL, GATA3, p63, inhibin, etc. – may be necessary for diagnostic classification of gestational trophoblastic tumors.

Molecular Genotyping Diagnosis

DNA genotyping has become the gold standard in the diagnosis and subclassification of hydatidiform moles with superb diagnostic sensitivity and specificity. Genotyping does not require fresh tissue and can be performed on formalin fixed paraffin-embedded tissue samples.

Genotyping may also play a crucial role in the diagnosis of gestational trophoblastic tumors and their risk scoring for clinical patient management.

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