

Title: *DICER1*-Related Disorders *GeneReview* - Supplemental Material: Pathologic Findings, Part 2

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Pathologic Findings, Part 2

Ovarian sex cord-stromal tumors

- SLCT comprises uniform small tubules and trabecular profiles (Sertoli component) accompanied by solid collections of eosinophilic polygonal cells (Leydig cells).
- JGCT comprises solid and/or cystic nodules of polygonal cells with a pale mucoid background or granular mucoid microcysts.
- Gynadroblastoma is a composite neoplasm with features of SLCT and JGCT.

Cystic nephroma (CN). A circumscribed unilateral or rarely bilateral multiloculated cyst in an otherwise normal kidney is the gross appearance. Delicate septa divide the lesion into variably sized locules. The interstitium of the septa is composed of benign appearing spindle cells. The considerable morphologic overlap between CN and Type I PPB is thought to reflect their similar pathogenesis.

Thyroid gland neoplasia including multinodular goiter (MNG), adenomas, or differentiated thyroid cancer. A range of benign to malignant lesions of the thyroid is seen.

The most common benign lesion is multinodular or adenomatous hyperplasia, characterized by variably sized follicular nodules. There may be accompanying lymphocytic thyroiditis.

The most common thyroid malignancy is papillary thyroid carcinoma with either the classic papillary pattern or follicular variant pattern.

Criteria for fine needle aspiration biopsy of thyroid nodules should follow the Bethesda System for Reporting Thyroid Cytopathology (TBSRTC) [Baloch et al 2008] with management based on previously established guidelines from the American Thyroid Association (thyroidguidelines.net/revised/taskforce), or the American Association of Clinical Endocrinologists/European Thyroid Association/Associazione Medici Endocrinologi collaborative (www.aace.com/files/thyroid-guidelines.pdf).

Ciliary body medulloepithelioma (CBME). Basic histologic features are neuroblastic or embryonic-like neural tubules and Homer Wright rosettes accompanied by hyaluronic acid-rich stroma. A teratoid variant may have cartilage or immature skeletal muscle.

Botryoid-type embryonal rhabdomyosarcoma (ERMS). Similar to Type I PPB, botryoid ERMS show a subepithelial layer of embryonal rhabdomyoblasts beneath an

intact epithelium (cambium layer). *DICER1*-related ERMS have the typical botryoid features as well as small nodules of cartilage not seen in other non-uterine cervical ERMS.

Nasal chondromesenchymal hamartoma (NCMH). The most common finding is a polypoid mass with nodules of immature to mature cartilage surrounded in part by a spindle cell stroma without rhabdomyoblastic differentiation. Other patterns include aneurysmal bone cyst-like foci, immature mesenchyme with a mucoid background, and cysts lined by respiratory epithelium.

Pituitary blastoma. These ACTH-producing tumors typically have a mixed pattern of gland and/or rosette-like structures intermixed with small primitive-appearing cells with blastemal features and larger secretory cells.

Pineoblastoma. The constituent cell has a high nuclear to cytoplasmic ratio with an intense hyperchromatic, round to ovoid shaped nucleus. This neoplasm resembles medulloblastoma and other primitive neuroectodermal brain tumors, in which both Homer-Wright and Flexner-Wintersteiner rosettes can be seen [Dahiya & Perry 2010].

References

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