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**Conjunctival lesion in a child**

**Case**

A 5-year-old healthy Asian boy was noted by his mother to have a spot hanging from his upper eyelid. On examination, there was a vascular, nonpigmented papillomatous mass in the superior fornix measuring at least 30 mm diameter. Initial suspion for giant conjunctival papilloma was raised. At surgery 1 week later, the mass was found larger at 50 mm diameter and rhabdomyosarcoma (RMS) was suspected, give the rapid growth. The mass was completely removed.

**Pathology**

Microscopic examination revealed a papillomatous mass with tumor in the substantia propria. The tumor consisted of malignant cells demonstrating strap configuration, rhabdomyoblasts, and mitoses. Immunoreactivity for striated muscle markers desmin and myogenein were positive. Portions of the tumor showed intense eosinophilia. FOX01 fusion protein was negative.

**Diagnosis**

Conjunctival RMS, embryonal type (botryoid pattern), FOX01 negative, low risk

**Discussion**

**Rhabdomyosarcoma**

There are 3 histopathologic subgroups of RMS including pleomorphic (rarely in orbit, most often in adults), embryonal (most common in orbit), and alveolar (most malignant). Differentiation of the various types shows only 70% concordance between laboratories.

**Embryona**l

* 50%-70% orbital RMS
* bipolar cells
* tapered cyto processes
* interlacing fascicles
* cross striations
* botryoid is variant with papillary configuration
* does not show FOX01 fusion protein abnormality

**Alveolar**

* 20%-30% orbital RMS
* poorly differentiated cells
* loosely arranged
* septa
* any focus of alveolar morphology is sufficient to classify the tumor as alveolar RMS

**RMS syndromes** – There are several cancer predisposition syndromes that can manifest RMS

* + - * Li Fraumeni familial cancer - p53 mutation
      * Neurofibromatosis
      * Noonan syndrome
      * Beckwith-Wiedermann syndrome
      * Costello syndrome

**Genetics** – Important genetic mutations relate to prognosis

Alveolar

* + - t(2;13)(q35-37;q14) most common – leads to PAX3-FOX01 fusion protein - poor prognosis
    - t(1;13)(p36;q14) less common – leads to PAX7-FOX01 fusion protein - better prognosis

Embryonal do not show these mutations. These mutations strongly predict prognosis [Williamson et al] and are performed in all cases. If negative, this implies embryonal and very low risk for metastasis.

**References**

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