

Familial syndromes associated with testicular and paratesticular neoplasms: a comprehensive review

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REVIEW AND PERSPECTIVES

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Abstract

A syndromic association between a subset of testicular/paratesticular neoplasms is well established. Such examples include Carney complex and large cell calcifying Sertoli cell tumor, Peutz-Jeghers syndrome and intratubular large cell hyalinizing Sertoli cell neoplasia, and VHL syndrome and clear cell papillary cystadenoma of the epididymis. However, recent studies proposed potential novel links between some testicular and paratesticular neoplasms with certain tumor syndromes. While more studies are still needed to solidify these associations, recent research suggests that a subset of Leydig cell tumors may arise in patients with hereditary leiomyomatosis and renal cell carcinoma syndrome or that some seminomas may occur in Lynch syndrome patients. Additionally, an association between testicular sex cord stromal tumors and paratesticular sarcomas with Familial adenomatous polyposis syndrome and DICER1 syndrome, respectively, has been proposed as well. This review provides a comprehensive overview of the intricate relationship between familial syndromes and associated testicular and paratesticular tumors, shedding light on their clinicopathological and molecular characteristics.

Keywords: Association; Familial syndromes; Paratestis; Testis; Tumors.

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