North American Proceedings in Gynecology & Obstetrics

Special Issue
CAOG Conference Abstracts
Winter 2022

Atypical presentations of DICER1 syndrome in adults with juvenile granulosa cell tumor

Callie J. Hamai, BS [1] , Samantha Salmon, MD [2] , Arwa Mohammad, MD [3] , Jean R. Goodman, MD [4] , Mark Hunter, MD [5]

1. University of Missouri-Columbia School of Medicine, Columbia, MO, USA
2. Department of Internal Medicine, University of Missouri, Columbia, MO, USA
3. Department of Obstetrics and Gynecology, Mayo Clinic, Rochester, MN, USA
4. Department of Obstetrics, Gynecology and Women's Health, Columbia, MO, USA
5. Department of Obstetrics Gynecology and Women's Health, University of Missouri, Ellis Fischel Cancer Center, Columbia, MO, USA

Abstract

PURPOSE: Autosomal dominant DICER1 syndrome is diagnosed by heterozygous DICER1 loss of function mutation resulting in abnormal microRNA regulation and classically presents with pleuropulmonary blastomas in conjunction with a variety of tumors: cystic nephromas, Sertoli-Leydig cell tumors, multinodular goiters, and papillary thyroid carcinomas. Herein we describe two uncommon presentations of DICER1 syndrome in adults with juvenile granulosa cell tumors (JGCT), rarely seen with this syndrome.

METHODS: Two patients were identified who presented with JGCTs and were later diagnosed with DICER1 syndrome. The patients were retrospectively reviewed and their clinical cases reported.

RESULTS: Two female patients, one 20 years-old and one 25 years-old, were found to have complex abdominal masses on imaging following clinical evaluation of associated symptoms. Both were diagnosed with JGCTs by final pathology following exploratory laparotomy with mass removal and staging. Following the pathologic diagnosis, the patients underwent genetic testing and were found to be positive for heterozygous germline DICER1 pathologic variants. The first patient was positive for the DICER1 c.556delT variant and all three of her male children were subsequently found to carry the same mutation. The second patient was positive for the DICER1 c.2988-20_2988del variant. She had a history of malignant follicular thyroid carcinoma diagnosed at age 11. Both family histories were explored and showed the suggestion of undiagnosed DICER1 syndrome in multiple relatives.

CONCLUSIONS: The presentation of two cases of atypical DICER1 syndrome presenting with JGCTs suggests the association of JGCTs and DICER1 syndrome may be underrecognized and underreported.