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Giant ovarian cystadenoma in association with Cowden syndrome

Inês Lemos Vargas Damásio, Valeriano Alberto Pais Horta Leite, Rita Joana Alves Martins dos Santos

Instituto Português de Oncologia de Lisboa Francisco Gentil, Lisboa, Portugal

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PTEN hamartoma tumour syndromes (PHTS) are a spectrum of hamartomatous overgrowth syndromes caused by germline mutations in the phosphatase and tensin homologue (*PTEN*) gene. Cowden syndrome (CS) is considered the prototype of PTHS. Diagnostic criteria for CS were first established by Salem and Steck [1] in 1983 and later revised in 1996 by an international consortium of researchers [2]. Since then, various modifications to this consensus have been proposed, and in 2013 Pilarski et al. [3] presented revised and evidence-based clinical criteria covering the spectrum of PTEN-related clinical disorders. However, due to the wide spectrum of clinical manifestations, the clinical diagnosis of PTHS remains challenging in some cases.

A 20-year-old woman reported a 1.5-month history of rapid and progressive increase of abdominal volume. She had a previous history of total thyroidectomy due to a multinodular colloid goitre at age 7 years. Pelvic magnetic resonance imaging (MRI) showed a large cystic tumour filling the abdominopelvic cavity with an apparent origin in the left ovary, measuring 38.5 × 31 × 18 cm. The patient underwent left anexectomy and cystectomy of the right ovary. The histological analysis revealed a 38 cm mucinous cystadenoma of the left ovary and a 3 cm serous cystadenoma of the right ovary.

The patient had no other clinical features that suggested a hereditary cancer syndrome, namely mucocutaneous lesions or macrocephaly. Family history was assessed: her father and 14-year-old brother had macrocephaly. Her father also had a history of intestinal polyps, multinodular goitre, and nephrectomy due to an arteriovenous fistula. Genetic testing was performed, and a pathogenic missense c.144C>A, p.(Asn48Lys), germline mutation in exon 2 of the *PTEN* gene was identified. The last medical visit occurred at age 23 years, and the patient referred regular menses and had no complaints. Pelvic ultrasound revealed normal-sized right ovary with multiple follicles. The patient is under active surveillance due to the risk of development of other tumours, namely breast and colon cancer.

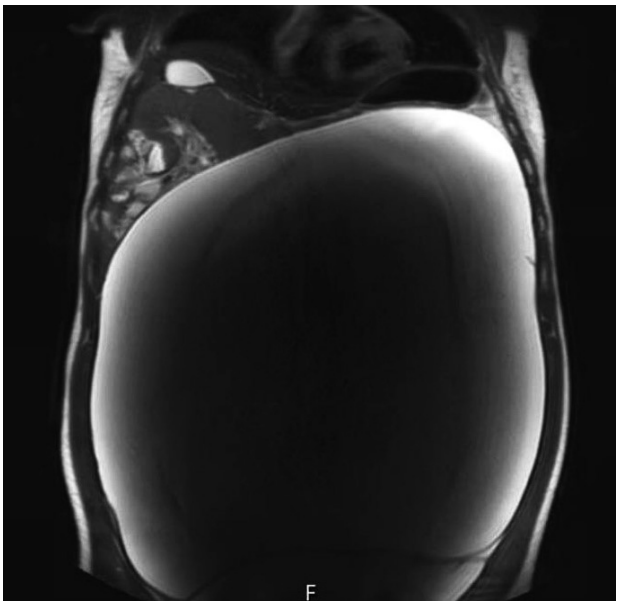

Figure 1. *Giant ovarian cystadenoma*

Benign ovarian tumours have been rarely described in PHTS, and they are not included in the latest clinical criteria revision of Pilarski et al. [2]. To the authors' knowledge, this is the first case of an association between a pathogenic mutation of the PTEN gene and a very large benign ovarian tumour in a patient who lacks the more common features of Cowden syndrome.

Inês Lemos Vargas Damásio, Largo Manuel Emídio da Silva nº 9, 4º direito, 1500-415 Lisboa, Portugal, tel: 000351963358265; e-mail: ines.damasio@hotmail.com

This report emphasizes the phenotypic variability that characterizes CS and the importance of assessing family history in young patients with a history of benign ovarian tumours.

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Conflict of interest

The authors declare that they have no conflicts of interest concerning this article.

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References

1. Salem O, Steck W. Cowden's disease (multiple hamartoma and neoplasia syndrome). J Am Acad Dermatol. 1983; 8(5): 686–696, doi: 10.1016/s0190-9622(83)70081-2, indexed in Pubmed: 6863628.
2. Nelen MR, Padberg GW, Peeters EA, et al. Localization of the gene for Cowden disease to chromosome 10q22-23. Nat Genet. 1996; 13(1): 114–116, doi: 10.1038/ng0596-114, indexed in Pubmed: 8673088.
3. Pilarski R, Burt R, Kohlman W, et al. Cowden syndrome and the PTEN hamartoma tumor syndrome: systematic review and revised diagnostic criteria. J Natl Cancer Inst. 2013; 105(21): 1607–1616, doi: 10.1093/jnci/djt277, indexed in Pubmed: 24136893.