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Case Presentation

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Precision Treatment of Advanced Squamous Cell Carcinoma Arising from Mature Cystic Teratoma of the Ovary with Homologous Recombination Deficiency

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Abstract

Introduction and Importance: Squamous-cell carcinoma arising from Mature Cystic Teratoma of the ovary (MCTO-SCC) is rare. Due to unclear pathogenesis and limited therapeutic experience, treatment options for this rare histological type of ovarian tumor were constrained, with advanced-stage

patients exhibiting extremely poor prognosis. Although the efficacy of poly ADP-ribose polymerase (PARP) inhibitors has become increasingly prominent in homologous recombination repair deficiency (HRD)-positive or Breast Cancer susceptibility gene (BRCA)-mutated epithelial ovarian cancers, the clinical significance of HR (Homologous Recombination) deficiency status in guiding the diagnosis and treatment of MCTO-SCC remains not well-defined.

Case report: A 65-year-old female patient was diagnosed with stage IIIC MCTO-SCC of the right ovary. After undergoing primary debulking surgery (PDS), she received six courses of combined chemotherapy with paclitaxel and carboplatin. Based on her molecular profile revealing a BRCA2 germline mutation, the patient received maintenance therapy with Fluzoparib for up to 14 months. Compared to the reported median survival of only 8 months in stage III MCTO-SCC patients in the published literature, this case demonstrated significantly improved outcomes, achieving a progression-free survival (PFS) of 35 months (with no recurrence observed to date). This suggests the clinical value of PARP inhibitors in the treatment of MCTO-SCC with HR deficiency and BRCA gene mutation.

Conclusion: This study aims to provide a practical clinical paradigm for the precision treatment of MCTO-SCC with HR deficiency, establishing a novel HR deficiency status-guided therapeutic approach for this rare and highly aggressive ovarian malignancy with historically poor prognosis.

Keywords: Ovarian neoplasms; Malignant transformation of mature teratoma; Squamous cell carcinoma; Homologous recombination repair deficiency; PARP inhibitors

Abbreviations: MCTO-SCC: Squamous-Cell Carcinoma arising from Mature Cystic Teratoma of the ovary; PARP: Poly ADP-Ribose Polymerase; HRD: Homologous Recombination Repair Deficiency; HR: Homologous Recombination; BRCA: Breast Cancer Susceptibility Gene; PDS: Primary Debulking Surgery; PFS: Progression-Free Survival; SCC: Squamous Cell Carcinoma; HGSOC: High-Grade Serous Ovarian Cancer; CT: Computed Tomography; CK5/6: Cytokeratin 5and 6; P63/40/16: Tumor Protein 63/40/16; PAX-8: Paired box 8; NUT: Nuclear Protein in Testis; TP53: Tumor Protein 53; CDKN2A: Cyclin-Dependent Kinase Inhibitor 2A; TMB: Tumor Mutational Burden; CA125: Cancer Antigen 125; SCCA: Squamous Cell Carcinoma Antigen; CR: Complete Response; HRR: Homologous Recombination Repair; ATM: Ataxia-Telangiectasia Mutated gene; LCOCs: Less Common Ovarian Cancers; ARID1A: AT-Rich Interaction Domain 1A

Introduction

Mature Cystic Teratoma of the Ovary (MCTO) accounts for 10%-20% of ovarian neoplasms, with the vast majority being benign. The malignant transformation rate is only 0.17%-2%, and among these cases, 80% develop into Squamous Cell Carcinoma (SCC) [1]. Due to the rarity of MCTO-SCC and paucity of research, its pathogenesis remains unclear, resulting in insufficient clinical experience and restricted treatment options [2]. Currently, the conventional paradigm of surgical resection followed by platinum-based chemotherapy shows limited efficacy, with advanced-stage patients having a dismal prognosis—the 5-year survival rate for stage III-IV disease is below 30% [1,3-5] (Table 1).

Table 1: Prognostic data from published literatures on MCTO-SCC.

Author and Year	Type of Study Design	Number of cases	Method	5-year survival rate (stage III, stage IV)
LI C et al. (2019)	Retrospective Cohort Study	435	Kaplan-Meier	26.2%, 0%
CHIANG A J et al. (2017)		52		0%, 0%
CHEN R J et al. (2008)		188		20.6%, 0%
KIKKAWA F et al. (1997)		37		0%, 0%

In recent years, pivotal clinical trials (SOLO-1, PAOLA-1/ENGOT-OV25, and PRIMA/ENGOT-OV26) have demonstrated that PARP inhibitors (Olaparib, Niraparib) significantly improve outcomes in HRD-positive/BRCA-mutated High-Grade Serous Ovarian Cancer (HGSOC) patients [6–8]. Concurrently, emerging evidence suggests the potential value of HR deficiency status in guiding precision therapy for rare histological subtypes of ovarian cancer. For instance, Jessica D. St. Laurent et al. reported a BRCA1 germline-mutated stage IV carcinosarcoma patient who achieved PFS >3.5 years with PARP inhibitors maintenance therapy [9]. Huang X et al. [10] documented a case of advanced HRD-positive ovarian clear cell carcinoma, showing remarkable prognostic improvement under PARP inhibitors treatment. However, molecular subtype-driven individualized strategies for rare ovarian malignancies remain in the early exploratory phase, with an urgent need for robust evidence-based data to validate these approaches.

This study presents a case of stage IIIC MCTO-SCC carrying a BRCA2 somatic mutation that achieved sustained remission following PARP inhibitors maintenance therapy after Primary Debulking Surgery (PDS) and platinum-based chemotherapy. Through this illustrative case, we explore HR deficiency status-guided precision therapy strategies, offering novel insights for improving outcomes in this rare malignancy.

Case Presentation

A 65-year-old female patient was admitted to Sun Yat-sen Memorial Hospital of Sun Yat-sen University with a history of persistent abdominal distension lasting over 2 months and worsening edema in both lower limbs for 1 month on March 7, 2023. The patient had no known history of diabetes, hypertension, or other comorbidities, and no family history of similar conditions. Upon gynecological examination, a palpable large pelvic mass, abdominal distension, positive mobile dullness, and lower limb edema were observed. Laboratory analysis revealed an elevated Cancer Antigen 125(CA125) level of 147 U/mL. Abdominal Computed Tomography (CT) imaging revealed a 16.8×14.5×10.3 cm cystic and solid mass in bilateral adnexa, resulting in uterine compression, significant effusion in the abdomen and pelvis, compression of pelvic and abdominal structures, and bladder compression. These imaging findings suggest widespread tumor metastasis **(Figure 1).**

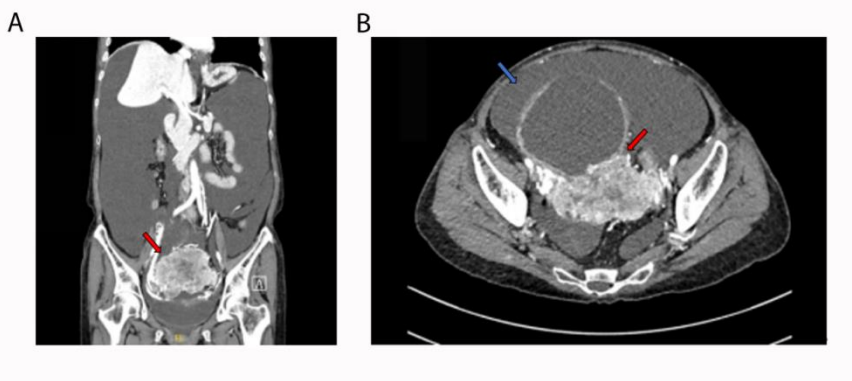

Figure 1: Computed tomography (CT) imaging before surgery. A) Coronal CT image. Cystic and solid mixed masses (indicated by red arrows) can be seen in the bilateral adnexa, with a size of approximately 16.8×14.5×10.3 cm. B) Axial CT images. A large amount of effusion can be seen in the abdomen and pelvis (indicated by the blue arrow).

The patient underwent surgery on March 14, 2023. During the operation, a huge mass was seen in the pelvis, which was explored as originating from the right ovary, with a size of about 15×10×12 cm. The base of the tumor is closely adhered to the sigmoid colon, accompanied by a large amount of pelvic effusion. The ovarian mass was bluntly separated from the sigmoid colon. Some of the tumors remained in the intestinal canal, which was hard and about 7×7×2 cm in size. Bilateral pelvic and abdominal para-aortic lymph nodes were not significantly enlarged. Abdominal hysterectomy, omentectomy, and sigmoid metastasis resection surgery were performed to achieve R0 resection.

Finally, the postoperative pathological diagnosis proved a mature cystic teratoma of the right ovary with moderately differentiated squamous cell carcinoma, tumor thrombus was positive in vessels, sigmoid colon metastatic carcinoma, and peri-intestinal lymph nodes metastatic carcinoma, conferring stage IIIC ovarian cancer (FIGO 2014) **(Figure 2).** Immunohistochemical examination showed CK5/6, P63, P40, P16, and Ki-67 were positive, while PAX-8 and NUT were negative, which confirmed that squamous cell carcinoma originated from teratoma malignant transformation. Further molecular profiling was performed through high-throughput gene sequencing on paraffin-embedded tumor tissue from the right ovarian mass and matched peripheral blood samples at the Molecular Diagnostics Center of Sun Yat-sen Memorial Hospital, utilizing targeted region probe capture technology and Illumina platform-based next-generation sequencing. The results revealed: a BRCA2 p.E2298* nonsense mutation (A nonsense mutation can disrupt gene function primarily by leading to the degradation of the mRNA transcript through nonsense-mediated mRNA decay, often resulting in a complete absence of the functional gene product **[11]**. Class I pathogenic variant **[12]**), concurrent TP53 p.E285K, and CDKN2A p.D108Y mutations, with an HRD score of 13 and a Tumor Mutational Burden (TMB) of 101.69 mutations/Mb.

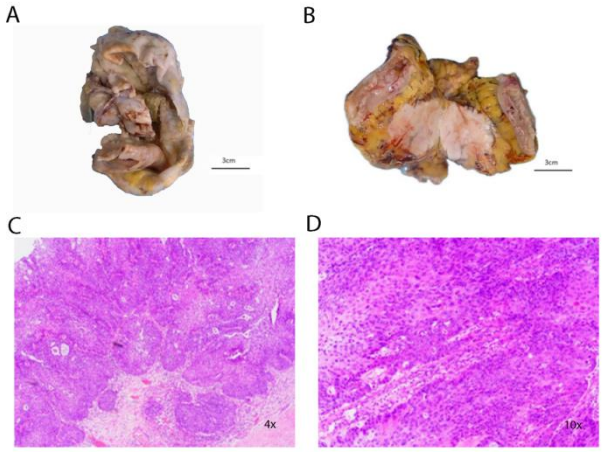

Figure 2: Histological and Gross Findings. A) The right ovarian tumor contains a poorly demarcated, grayish-white solid component. B) A metastatic lesion is identified in the sigmoid colon, with dimensions of 7×7×2 cm. C) HE staining of right ovarian tumor. The tumor is composed of moderately differentiated squamous cell carcinoma. D) HE staining of right ovarian tumor. Tumor cells are observed infiltrating adjacent stroma, with focal tumor thrombi present in lymphatic/vascular channels.

After surgery, the patient received carboplatin (AUC6) combined with paclitaxel (175 mg/m2) intravenous chemotherapy for a total of six courses. CA125 decreased from 147.0 U/mL preoperatively to 85.2 U/ml postoperatively and further decreased to 13.8 U/ml (within the normal reference range) after the first course of chemotherapy, Squamous Cell Carcinoma Antigen (SCCA): 3.22 ng/ml. After that, the tumor markers (CA125 and SCCA) of the patient stabilized in the normal range **(Figure 3)**, and no uncontrolled tumor signs were found. One month after completing chemotherapy (September 2023), the patient started PARP inhibitor (Fluzoparib, 150 mg bid) maintenance treatment, during which adverse reactions were mild, mainly manifested as venous thrombosis of lower limbs, which did not affect the medication. The patient discontinued the drug in November 2024, and the clinical efficacy evaluation Complete Response (CR). During follow-up, CA125 and SCCA levels remained stable within normal limits **(Figure 3).** A follow-up CT scan in February 2025 revealed no evidence of tumor recurrence **(Figure 4).**

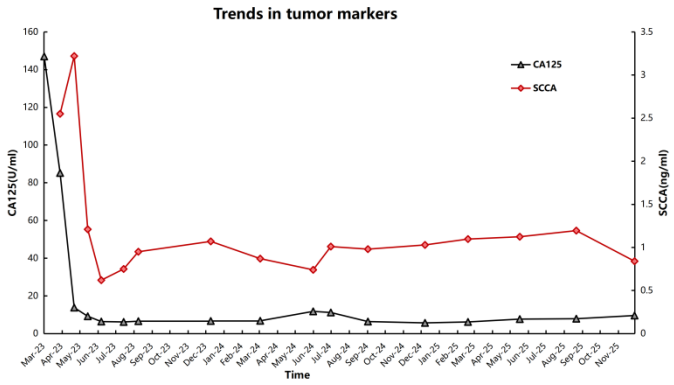

Figure 3: Trends in tumor markers (CA125 and SCCA). CA125 decreased from 147.0 U/mL preoperatively to 85.2 U/ml preoperatively and further decreased to 13.8 U/ml (within the normal reference range) after the first chemotherapy, SCCA: 3.22 ng/ml. After that, the tumor markers (CA125 and SCCA) of the patient stabilized in the normal range.

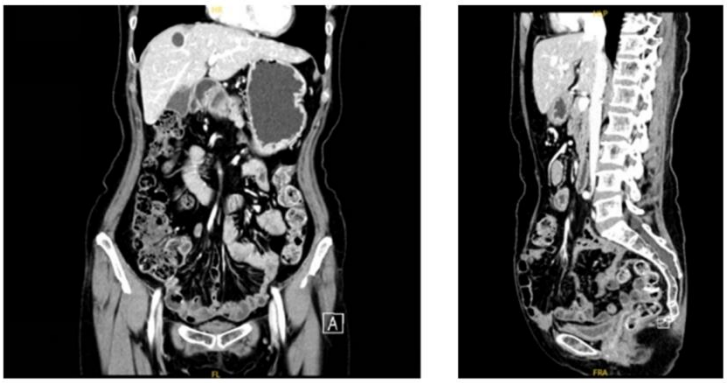

Figure 4: CT Imaging Performed in February 2025. No evidence of recurrence was observed.

Discussion

Squamous cell carcinoma is the most common pathological type of ovarian teratoma malignant, and it is also the main source of ovarian squamous cell carcinoma **[1,13].** Due to the rarity, unclear pathogenesis, innate platinum resistance, and high invasiveness of MCTO-SCC, there is a lack of treatment experience and poor prognosis for late-stage patients **[14,15].** Previous studies have shown that the 5-year survival rate of stage III patients with malignant teratoma squamous cell carcinoma is only 20.6%, and 0% of stage IV patients **[1].** At present, precision treatment strategies, represented by PARP inhibitors, are primarily focused on common ovarian epithelial tumors like high-grade serous carcinoma, while there are few related studies on rare pathological types of ovarian tumors, such as malignant germ cell tumors. The clinical significance of these approaches in MCTO-SCC was still unclear. Recent genomic analysis by Liang et al. **[16]** on six

MCTO-SCC cases revealed that 5/6 cases carried Homologous Recombination Repair (HRR) gene mutations (including BRCA1, ATM, etc.). The prevalence of HR deficiency was significantly higher than in epithelial ovarian cancer (approximately 50%) [17], suggesting that MCTO-SCC may exhibit a high frequency of HR deficiency. Moreover, HR deficiency might be a key molecular characteristic in a subset of MCTO-SCC cases. In this case, the patient's BRCA2 mutation further supports the literature findings that HR deficiency also exists in MCTO-SCC tumors. This highlights the importance of routinely testing HRR genes and performing HRD scoring in MCTO-SCC patients—even in the absence of family history—to identify potential candidates who may benefit from targeted therapies.

The patient in this case presented with an advanced-stage tumor and extensive metastases, carrying a somatic BRCA2 mutation (p.E2298*, class I variant) and an HRD score of 13. Our team designed a personalized treatment plan comprising surgery, platinum-based chemotherapy, and PARP inhibitor therapy guided by HR deficiency status. Following maintenance therapy with Fluzoparib, the patient achieved progression-free survival exceeding 27 months (with no recurrence to date), demonstrating significant prognostic improvement. This case highlights the critical role of HR deficiency status in guiding therapeutic decision-making for rare ovarian tumors and demonstrates that HRD-guided PARP inhibitor therapy may represent a promising therapeutic strategy for such malignancies. However, further evidence-based medical research is required to validate the generalizability of this therapeutic strategy. At present, the treatment of Less Common Ovarian Cancers (LCOCs) such as MCTO-SCC still faces multiple challenges. Firstly, the prevalence and biological characteristics of HR deficiency remain unclear. Secondly, novel targeted therapies for common mutations, such as TP53 and ARID1A, are still under development [18], and clinical experience is insufficient. In the future, it is urgent to carry out multi-center research and establish a molecular profiling database and registry system for rare ovarian cancers. This would provide high-quality data to support therapeutic decision-making for these malignancies, which currently have limited treatment options and suboptimal outcomes. Additionally, further research is required to explore targeted therapies and their underlying molecular mechanisms in LCOCs, ultimately advancing more precise and effective treatment strategies for MCTO-SCC and other rare ovarian neoplasms.

Conclusion

This case demonstrates that a HR deficiency status-guided combined modality approach — surgery followed by chemotherapy and PARP inhibitor maintenance therapy, can significantly improve outcomes in advanced MCTO-SCC patients. For patients carrying BRCA1/2 mutations and HRD positivity, this comprehensive treatment strategy holds important clinical value for broader application. Moving forward, additional high-quality studies are needed to further validate the efficacy of this approach. Concurrently, enhancing the molecular understanding of these exceptionally rare tumors, along with exploring targeted therapies and their underlying mechanisms, will be critical to advancing precision medicine in rare ovarian cancers. Such efforts may ultimately provide greater survival benefits for patients with MCTO-SCC and other rare ovarian malignancies.

Consent

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Authors Contribution

Bingzhong Zhang, Ruilin Lei and Guocai Xu conceived the idea for the study and were responsible for the supervision and revising the manuscript. Peijun Fang and Yun Long drafted the manuscript and constructed the figures. Xiaoliang Lv and Lin Lin carried out the information collection and follow-up of the patient. Yiou Zhang and Ruixin Li carried out the literature review. Wenyan Xu and Qin Chen carried out revised the manuscript. All authors read and approved the final manuscript.

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Conflict of Interest

The authors declare no conflict of interest in this study.

Ethics Approval

The study protocol was approved by the Scientific Research Ethics Committee of Sun Yat-sen Memorial Hospital, Sun Yat-sen University, with Ethics No.: SYSKY-2025-372-01.

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