Case Report

Rare association of xeroderma pigmentosum with ovarian Sertoli–Leydig cell tumor: A case report and literature review

Anwesa Pal[1], Manoranjan Mahapatra[2], Anirban Talukder[1], Bhagyalaxmi Nayak[3], Mamita Nayak[4], Janmejaya Mohapatra[5], Ashok Kumar Padhy[2], Nihar Ranjan Rout[1]

From [1]MCh Resident, [2]Associate Professor, [3]Professor, Ex Head, [5]Professor and Head, Department of Gynae-Oncology, [4]Assistant Professor, Department of Oncopathology, Acharya Harihar Post Graduate Institute of Cancer, Cuttack, Odisha, India

ABSTRACT

Xeroderma pigmentosum (XP) is a rare autosomal recessive disorder caused by deoxyribonucleic acid repair defects, predisposing patients to early cutaneous malignancies and, less frequently, internal tumors. We report the case of a 19-year-old female with XP who presented with abdominal pain, distension, and secondary amenorrhea for 3 years. Imaging revealed a large solid-cystic ovarian mass with elevated CA-125. She underwent fertility-sparing staging laparotomy, and histopathology confirmed a well-differentiated Sertoli–Leydig cell tumor of the ovary (Federation of Gynecology and Obstetrics Stage IA). No adjuvant treatment was required. She remains disease-free after 18 months, with restoration of normal menstruation. This rare association emphasizes close surveillance in XP patients for early detection of internal malignancies.

Key words: Deoxyribonucleic acid repair, Ovary, Sertoli–Leydig cell tumor, Xeroderma pigmentosum

Xeroderma pigmentosum (XP) is a rare autosomal recessive genodermatosis caused by nucleotide excision repair pathway defects, leading to genomic instability and increased cancer risk [1,2]. Patients typically develop early-onset skin cancers and may experience neurological degeneration [1-3]. Improved survival has led to recognition of early internal malignancies, including central nervous system, hematologic, and visceral cancers [3,4]. Ovarian tumors remain extremely rare in XP, particularly sex cord-stromal tumors such as Sertoli–Leydig cell tumors (SLCTs), which constitute <1% of ovarian tumors [5,6]. These tumors often present in young women with abdominal symptoms or hormonal manifestations [7]. Only a few SLCT cases in XP patients have been documented [8,9].

This case highlights the clinical features, surgical management, and follow-up of a young XP patient with SLCT.

CASE REPORT

This is a case of a 19-year-old girl with a known history of XP since infancy presented with progressively increasing abdominal swelling and dull aching lower abdominal pain for the past 2 months (Fig. 1a). These symptoms were associated with constipation and urinary frequency for 2 weeks, likely due to pressure effects from the mass. She also reported secondary amenorrhea for 3 years, raising concern for possible endocrine dysfunction. Her history was significant for excision of a right eye limbal squamous cell carcinoma at 11 years of age, consistent with the malignant predisposition seen in XP. The patient was born of a non-consanguineous marriage and diagnosed with xeroderma pigmentosum in infancy based on classical cutaneous photosensitivity, freckling, and progressive pigmentary changes. There was no family history of similar illness or malignancy. She had normal developmental milestones and no neurological deficits. Menarche occurred at 13 years of age, with initially regular cycles before the onset of secondary amenorrhea 3 years prior to presentation.

On examination, a firm, non-tender abdominopelvic mass was palpable, corresponding clinically to approximately a 20-week gravid uterus size.

Ultrasonography revealed a large solid-cystic mass arising from the left adnexa (Fig. 1b). Further characterization with magnetic resonance imaging (MRI) of the pelvis confirmed a 17 × 16 × 15.6 cm well-defined mixed solid-cystic lesion with internal septations (Fig. 1c). Positron emission tomography-computed tomography scan demonstrated an fluorodeoxyglucose-avid pelvic mass without evidence of nodal disease

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Correspondence to: Anwesa Pal, MCh Resident, Department of Gynae-Oncology, Acharya Harihar Post Graduate Institute of Cancer, Cuttack, Odisha - 753007, India. E-mail: mckanwesa23@gmail.com

or distant metastasis, suggesting localized pathology. Serum tumor marker CA-125 was significantly elevated at 329 U/mL (Fig. 1d).

She underwent staging laparotomy on 17 February 2024, which revealed a 15 × 9 cm left ovarian mass with torsion and dense adhesions. Considering her young age and fertility concerns, a fertility-sparing left salpingo-oophorectomy was performed. Gross examination showed a predominantly solid tumor with tan-yellow areas and focal necrosis. Frozen section was suggestive of a sex cord-stromal tumor (Fig. 2a).

Final histopathological evaluation confirmed a well-differentiated SLCT, characterized by Sertoli cells arranged in tubules and Leydig cells in clusters. Immunohistochemistry demonstrated strong positivity for inhibin-B, calretinin, and CD99, supporting the diagnosis (Fig. 2). There was no evidence of peritoneal or nodal spread.

She was staged as the International Federation of Gynecology and Obstetrics (FIGO) IA and managed with surveillance without adjuvant chemotherapy. Her menstrual cycles resumed 2 months post-operatively, and at 18-month follow-up, she remains asymptomatic with no clinical or radiological recurrence (Fig. 2).

DISCUSSION

SLCTs are uncommon ovarian neoplasms representing <1% of all ovarian tumors and are typically diagnosed in young women during their reproductive age [6,7]. Their etiology is not fully understood; however, a genetic basis is suggested because approximately 10–20% of SLCTs are associated with DICER1 mutations, especially in familial tumor predisposition syndromes [6]. Whether defective DNA repair mechanisms in XP interact synergistically with tumor-predisposing genetic mutations, such as DICER1, to accelerate ovarian tumorigenesis remains to be elucidated. Clinically, SLCTs produce androgens in one-third of cases due to the presence of functional Leydig cells, resulting in virilizing symptoms such as hirsutism, deep voice, clitoromegaly, or oligomenorrhea [7]. Our patient uniquely demonstrated only amenorrhea without virilization, highlighting that endocrine manifestations are variable and the absence of virilization does not exclude SLCT.

XP is characterized by defective nucleotide excision repair leading to unchecked accumulation of DNA mutations after ultraviolet (UV) and oxidative exposure [1-4]. Although cutaneous malignancies predominate, internal cancers may arise due to systemic

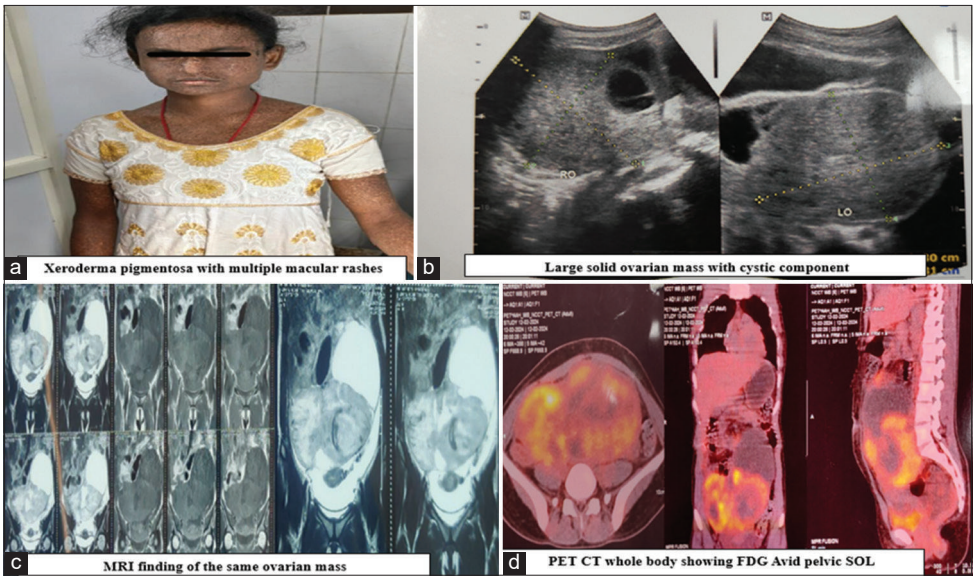

Figure 1: Clinical and radiological presentation. (a): Phenotypic appearance of xeroderma pigmentosum; (b): Ultrasound of pelvic mass; (c): Magnetic resonance imaging pelvis showing solid-cystic mass; (d): Positron emission tomography-computed tomography showing fluorodeoxyglucose-avid lesion

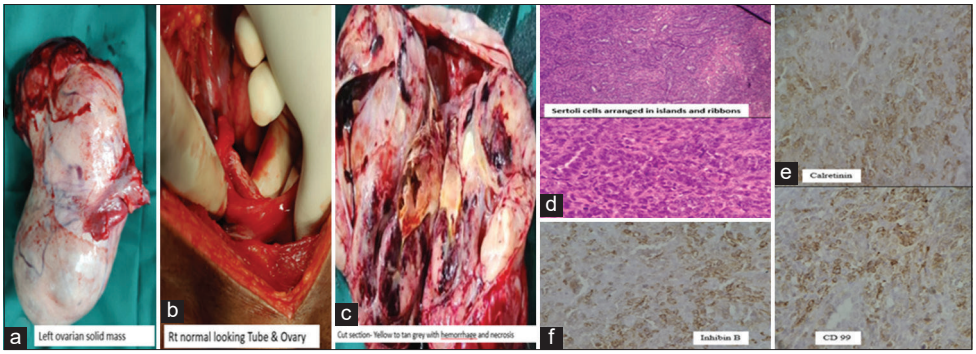

Figure 2: Gross, microscopic, and immunohistochemical findings. (a): Large solid left ovarian mass; (b): Normal right adnexa; (c): Cut section demonstrating necrosis; (d-f): Histology and Immunohistochemistry positive for inhibin, calretinin, and CD99

Table 1: Comparison of the previous cases with the present one

Feature	Bdioui *et al*., 2021 [8]	Yurchenko *et al*., 2023 [9]	Present case
Age	27 years	Teenage/young adult	19 years
Differentiation	Poorly differentiated	Variable	Well differentiated
Treatment	Surgery+Chemo	Surgery	Surgery only
Outcome	Good	Good	Disease-free at 18 months

Table 2: Grades and stages of tumor

Tumor grade	Standard management	Prognosis
Well-differentiated	Surgery only	Excellent (>90% survival)
Intermediate/poorly differentiated	Surgery+ Chemotherapy	Intermediate
Advanced stage	Multimodal	Variable

DNA repair impairment. Several mechanisms may contribute: (a) Persistent oxidative stress → increased double-strand breaks and somatic mutations; (b) Hormonal cycling in ovaries → repeated proliferation and DNA replication errors; (c) Lack of repair of endogenous estrogen-induced DNA adducts; and (d) Chromosomal instability in rapidly dividing gonadal stromal cells. Thus, XP creates a molecular environment highly conducive to neoplastic transformation even in organs shielded from UV radiation. This case supports growing evidence that XP is an independent risk factor for early-onset gonadal tumors, particularly SLCTs [8,9]. Only two previously reported patients closely resemble this clinical scenario and their differences are mentioned in Table 1.

SLCT diagnosis can be challenging as symptoms mimic common gynecologic conditions, imaging features overlap with germ-cell and epithelial tumors, and tumor markers like CA-125 may be elevated despite stromal origin. Thus, histopathology remains the gold standard. Inhibin-B, calretinin, and CD99 are highly sensitive markers confirming sex-cord stromal differentiation [6]. Pre-operative differential diagnosis included germ cell tumor (dysgerminoma), epithelial ovarian carcinoma, granulosa cell tumor, teratoma, and ovarian lymphoma. These differentials justify the decision for intraoperative frozen section and staging.

Treatment of SLCT depends on grade and stage (Table 2). In XP, clinicians must consider the risk of toxicity and hypersensitivity associated with chemotherapy and radiation, respectively. Thus, avoiding unnecessary DNA-damaging therapies is crucial. Our patient was rightly managed with surveillance only.

Due to lifelong cancer predisposition in XP, structured follow-up is recommended through clinical review and abdominal/pelvic examination, serum CA-125 monitoring (though non-specific), MRI/ultrasound preferred over computed tomography to reduce radiation exposure, and long-term monitoring beyond the standard 5-year window. Our patient has remained disease-free for 18 months both clinically as well as by imaging and regained normal menstrual cycles, a strong indicator of a favorable outcome.

What Makes This Case Unique is its earlier onset as XP patients present SLCT in the 2nd decade, not the 3rd–4th decade; organ-confined disease as FIGO IA in most reported cases; good prognosis as surgical cure is possible in well-differentiated tumors; and fertility preservation is feasible as it is crucial in young XP individuals with shortened life expectancy due to malignancy burden. This case adds valuable evidence by demonstrating: XP can predispose to ovarian tumors at a very young age, SLCTs in XP may retain low-grade biology, fertility-preserving surgery is effective and safe, need for heightened vigilance for internal malignancies in XP, importance of multidisciplinary management (Dermatology + Oncology + Genetics); and growing documentation will help establish guidelines for surveillance and risk-stratification in XP patients.

CONCLUSION

This rare case underscores the need for vigilant surveillance for internal malignancies in XP patients. Early diagnosis and fertility-preserving surgery offer promising outcomes in well-differentiated SLCT.

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