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CASE REPORT

Navigating Ambiguous Genitalia: A Case of Dysgerminoma in an Afghan Boy

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Introduction: Ambiguous genitalia (AG) present significant challenges in both diagnosis and management. Often associated with disorders of sexual development (DSD), AG can lead to complex medical scenarios, including an increased risk of malignant transformation. This case report underscores the importance of recognizing the clinical implications of AG and the necessity for careful monitoring.

Case Presentation: We present the case of a 22-year-old male who presented with abdominal pain and distension. Upon physical examination, he exhibited characteristics consistent with ambiguous genitalia, including a small penis and an absent scrotal sac. The diagnostic imaging, particularly computed tomography (CT) scans, revealed a large solid mass in the left adnexa. This mass displayed lobulated outlines and speckled calcifications, which raised concerns for a potentially malignant process. Given the findings, the patient underwent surgical intervention, which included a total abdominal hysterectomy, bilateral salpingo-oophorectomy, and omentectomy to ensure complete removal of the tumor and surrounding affected tissues. Histopathological analysis of the excised tissue confirmed the diagnosis of dysgerminoma, a type of germ cell tumor known for its malignant potential.

Conclusion: This case highlights the critical need for vigilant surveillance for malignancy in patients with ambiguous genitalia. The inherent risk factors associated with AG necessitate a proactive approach to patient management, including regular imaging and timely surgical interventions. By adopting a comprehensive care strategy, healthcare providers can optimize patient outcomes and address the unique challenges posed by disorders of sexual development. Continuous monitoring and early intervention are paramount in mitigating the risks of tumor development in this vulnerable demographic.

Keywords: Ambiguous genitalia, dysgerminoma, disorders of sexual development, germ cell tumors, multidisciplinary approach

Introduction

Ambiguous genitalia (AG) refers to atypical development of external genitalia that makes it difficult to classify an individual as male or female at birth.[1] It may present as hypospadias, micropenis, or labial fusion, requiring early evaluation for gender assignment and exclusion of life-threatening conditions.[2] AG often stems from hormonal imbalances during fetal development,[3] with environmental exposures, such as endocrine disruptors, also implicated.[4] Incidence of AG varies from 1 in 4500 to 1 in 5500 for strict definitions, rising to 1 in 300 with broader criteria.[5] AG presents diagnostic challenges,[6] and can cause significant ethical and psychosocial distress.[7] These challenges often revolve around gender identity concerns, as individuals may struggle with their assigned gender, leading to potential gender dysphoria.[8] The stigma associated with AG and DSD can exacerbate feelings of isolation and anxiety, impacting both the individuals and their families.[9] The condition is also associated with elevated malignancy risk, particularly germ cell tumors, with incidences between 15% and 60%.[10–12] Modern diagnostic tools for evaluating AG include targeted genetic tests[13] and advanced imaging techniques.[14]

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Misdiagnosis remains common; one study found 47.2% of genetically female individuals were assigned male at birth.[11] Delayed diagnosis can lead to severe psychological and social consequences, as well as medical complications, such as,[15] missed opportunities for early intervention, and increased risk of complications such as malignant transformation.[16]

Effective management requires a multidisciplinary approach, including history-taking, physical examination, imaging, and karyotyping to determine etiology.[17] A major gap remains in understanding genetic causes and long-term psychosocial impacts, complicating diagnosis and treatment decisions.[18] This report discusses a unique case of a young adult presenting with abdominal pain, ambiguous genitalia, and a malignant germ cell tumor, underscoring the importance of vigilant surveillance.

Case History/Examination

A 22-year-old male presented with a significant medical history of abdominal pain and distension, previously uninvestigated for any underlying conditions. The physical examination revealed ambiguous genitalia, characterized by a small penis, absence of a scrotal sac, and a vagina-like structure (Figure 1). These findings raised concerns for a potential disorder of sexual development, warranting further evaluation. Upon abdominal palpation, a mass was detected in the lower abdominal cavity, prompting imaging studies for further assessment.

Case Presentation

Abdominal and pelvic computed tomography (CT) scans were conducted, revealing a large solid mass lesion with heterogeneous contrast enhancement and lobulated outlines in the mid and lower abdomen as well as the pelvis (Figure 2). The mass, likely arising from the left adnexa, measured approximately 20 cm x 19 cm x 12 cm in craniocaudal, transverse, and anteroposterior dimensions, with multiple speckled calcifications. The radiological findings

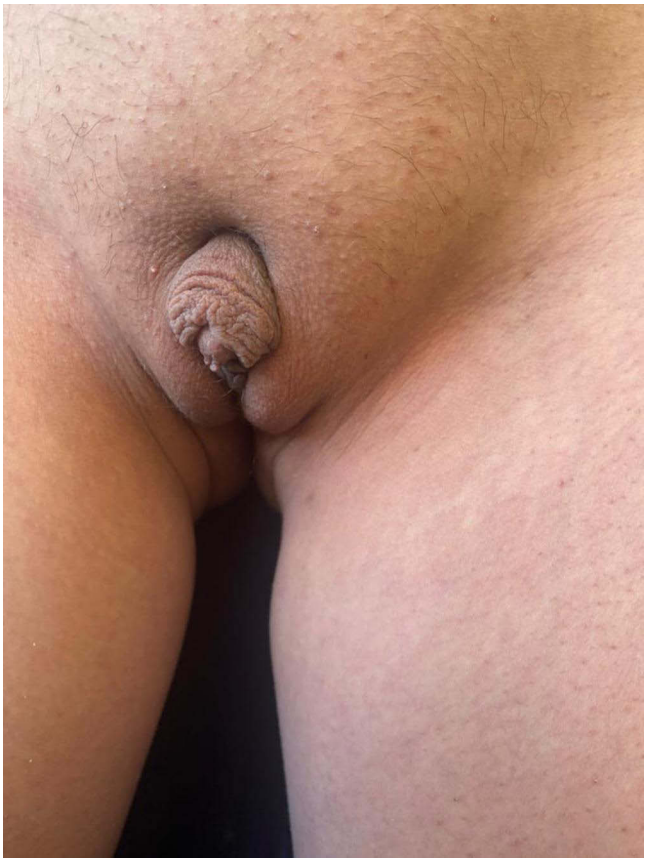

Figure 1 External genitalia of the patient demonstrating ambiguity, characterized by micropenis, prominent labia majora, an enlarged phallus, and absence of the scrotum.

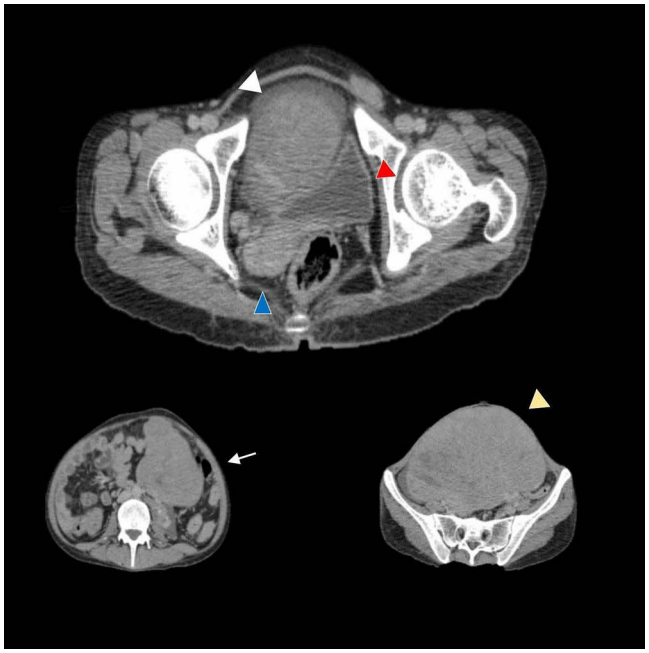

Figure 2 CT scan images depicting the uterus and a left adnexal mass. The white arrowhead indicates the left adnexal mass, the red arrowhead marks the urinary bladder, the blue arrowhead denotes the uterus, the yellow arrowhead highlights different levels of the mass, and the white arrow points to the mass adjacent to the right adnexa.

suggested a neoplastic lesion, with germ cell tumor and teratoma as differential diagnoses. Both adnexae were not separately identifiable, complicating the surgical approach. The mass was closely adjacent to the uterus, urinary bladder, bowel loops, and anterior abdominal wall, necessitating meticulous surgical planning to avoid bowel injury. Proximity to the iliac vessels, aorta, and inferior vena cava was noted; however, no encasement of the vessels was observed, which favored surgical intervention. Mild free fluid was also detected in the abdominopelvic cavities, a common finding in malignancy or significant pathology. Imaging showed multiple enlarged lymph nodes in the left paraaortic region, with one measuring approximately 28 mm x 22 mm, raising concerns for possible metastatic involvement. Serological evaluations indicated elevated levels of LDH, AFP, and β-hCG. A psychologist provided the patient with necessary advice regarding his condition. The patient was informed about his diagnosis and the surgical procedure, ensuring he understood all aspects. Written informed consent was obtained for the publication of images, emphasizing the patient's rights to privacy and confidentiality throughout the process. The patient underwent a total abdominal hysterectomy, bilateral salpingo-oophorectomy, and omentectomy. The surgical procedure successfully removed the uterus, cervix, bilateral adnexa, and omentum, with no peritoneal deposits observed during the operation (Figure 3A and B). Following surgical excision, the tumor checklist based on the College of American Pathologists (CAP) guidelines confirmed the tumor's location in the left ovary, with a gross size of 20×18 × 11 cm. Ovarian surface involvement was noted, while no gross lymph node metastasis was observed. Pathologic staging, including the FIGO classification, was reported as pT1c2NxMx, corresponding to FIGO Stage IC2.

Histopathological analysis of the complex mass in the left ovary revealed an infiltrative neoplasm with extensive necrosis, arranged in nests and cords. Neoplastic cells varied in size from intermediate to large, exhibiting moderate cytoplasm, hyperchromatic nuclei, and visible to inconspicuous nucleoli, with intersecting fibrous bands containing inflammatory cells, suggesting dysgerminoma. Immunohistochemical staining for OCT3/4 and CD117 was positive in the neoplastic cells, confirming the diagnosis of dysgerminoma (Figure 4A–C). The tumor was confined to the left ovary, with the cervix showing only chronic inflammation and nabothian cysts. The endometrium displayed atrophic changes, indicating no active pathology. The myometrium revealed benign endometrial glands surrounded by stromal cells, indicating adenomyosis. The right ovary and fallopian tube exhibited benign cystic follicles and para-tubal cysts. Sections from the omentum indicated chronic inflammation and reactive hyperplasia of mesothelial cells, with no evidence of tumor involvement.

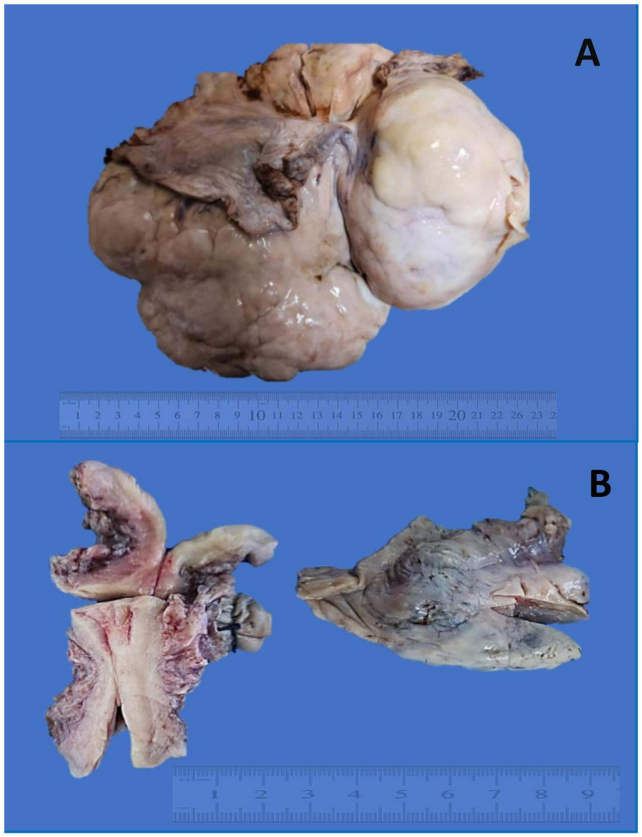

Figure 3 (**A**) Gross image of the left adnexal mass. (**B**) Sectioned uterus indicated by the white arrow, with the right ovary and fallopian tube highlighted by the white arrowhead.

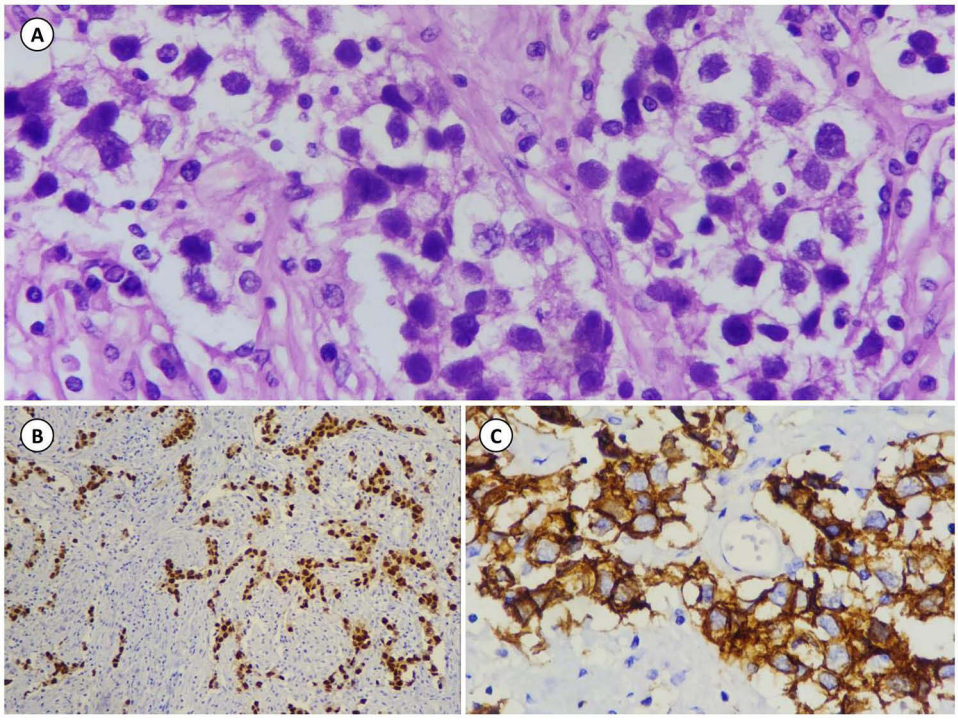

Figure 4 (**A**) H&E-stained image illustrating nests of large polygonal cells with eosinophilic cytoplasm and well-defined cell membranes, separated by fibrous septa containing lymphocytes and histiocytes. (**B**) Nuclear positivity of the neoplastic cells as shown by OCT3/4 staining. (**C**) Membranous positivity of the neoplastic cells demonstrated by CD117 staining.

The surgical intervention was successful, with the total abdominal hysterectomy, bilateral salpingo-oophorectomy, and omentectomy performed without immediate complications. Histopathological analysis confirmed the diagnosis of dysgerminoma localized to the left ovary, allowing for timely treatment and improved prognosis. Postoperatively, the patient was monitored for complications such as infection and bleeding, remaining stable and discharged after one night in the hospital. Approximately three weeks post-surgery, the patient began the bleomycin, etoposide, and platinum (BEP) chemotherapy regimen. During the course of chemotherapy, the patient was educated about potential side effects, including nausea, fatigue, hair loss, and the risk of infections due to myelosuppression. He was advised on supportive measures to mitigate these effects, such as antiemetic medications for nausea and maintaining good hygiene to reduce infection risk.

Discussion

Living with a rare condition like AG poses significant challenges and societal stigma.[19] Individuals with AG or disorders of sex development (DSD) face an increased risk of malignancy, primarily due to gonadal dysgenesis or undifferentiated gonadal tissue. These factors can lead to germ cell tumors (GCTs), such as gonadoblastoma and dysgerminoma, especially in those with Y-chromosome material.[20] GCTs are a diverse group of neoplasms that typically arise from germ cells in the gonads but can also occur in extragonadal sites. They are classified into seminomatous and non-seminomatous tumors, with the latter including teratomas, embryonal carcinoma, yolk sac tumors, and choriocarcinoma.[21] The etiology of GCTs remains unclear, but genetic predisposition, environmental factors, and DSD may play a role.[22] AG is usually identified at birth and can cause distress for families. The development of male and female sex organs originates from the same embryonic tissue, with male hormones guiding organ formation. A deficiency of these hormones in genetically male fetuses can result in ambiguous genitalia, while exposure in genetically female fetuses can have similar effects.[19] In many developing countries, children with this condition are often hidden due to fears of shame and associated stress.[23] Atypical gonadal development in DSD increases the risk of neoplasms, with GCTs being the most common, although rare ovarian malignancies have also been reported,[24] as seen in our case of dysgerminoma. Accurate diagnosis of AG requires evaluating external appearance, internal anatomy, genetic makeup, hormonal profiles,[25] and advanced imaging techniques like pelvic MRI and ultrasonography.[26] Timely surgical intervention may be necessary based on diagnosis and patient needs, with some advocating for deferring surgery until the patient is older and can participate in decision-making.[27] For male gender assignment, administering 10–25 mg of testosterone enanthate or cypionate intramuscularly once a month for three months is recommended to enhance penile response to androgens before surgery.[25]

In this case, a 22-year-old male, raised as a boy, was admitted due to severe abdominal pain. Clinical examination and imaging, including a CT scan, led to the diagnosis of AG, characterized by atypical external genitalia and variations in chromosomal and gonadal sex. Histopathological examination of the excised specimen revealed an infiltrative neoplasm with extensive necrosis in the left ovary, highlighting the increased risk of gonadal tumors in AG patients. This risk is often linked to undescended testes or dysgenetic gonads, which can foster malignant transformation.

The cervix examination showed chronic inflammation and nabothian cysts, typically benign and resulting from cervical gland obstruction. In the myometrium, findings included benign endometrial glands surrounded by stromal cells, indicating adenomyosis, which can cause dysmenorrhea and pelvic pain, possibly explaining the patient's abdominal discomfort. The right ovary and fallopian tube exhibited benign cystic follicles and para-tubal cysts, common incidental findings that do not indicate tumor involvement. This case underscores the importance of vigilant surveillance for malignancy in AG patients, particularly concerning the ovaries and testes. The heightened risk of tumor development necessitates a multidisciplinary approach, including regular imaging and potential surgical intervention. Clinicians should maintain a high index of suspicion for neoplastic changes in this population to ensure timely diagnosis and treatment. The incidental discovery of a large intra-abdominal mass and subsequent diagnosis of dysgerminoma highlight the critical need for timely assessment in DSDs. This case contributes to clinical understanding by illustrating how delayed diagnosis can conceal life-threatening malignancies. A comprehensive approach—incorporating advanced imaging, histopathology, immunohistochemistry, and multidisciplinary care—enabled accurate diagnosis and appropriate management. Furthermore, integrated psychosocial support and patient-centered communication are essential for addressing the psychological impact of DSD diagnosis and treatment. Ultimately, this case reinforces the significance of early recognition and multidisciplinary intervention in managing ambiguous genitalia with malignant potential.

Consent

The patient provided written informed consent for the publication of case details, including the accompanying images. Furthermore, the study received approval from the Ethical Committee of Khatam Al-Nabieen University (AF, knu.edu.af. rec 19-14-11, 2024).

Disclosure

The authors declare no conflicts of interest in this work.

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