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The “Compliance Gap” in Surgical Decision-Making for Bardet–Biedl Syndrome with Vaginal Reatresia and Intellectual Disability: A Case for Hysterectomy

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Case Report

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Abstract

Background: Bardet–Biedl syndrome (BBS) is a rare autosomal recessive ciliopathy characterized by rod-cone dystrophy, obesity, polydactyly, renal dysfunction, and genital anomalies[1]. In female patients, vaginal atresia often leads to hematometrocolpos and cyclic abdominal pain after menarche[2]. The optimal surgical management for BBS patients with concomitant vaginal atresia and intellectual disability remains poorly defined.

Case presentation: We report a 13-year-old female with BBS10-related Bardet–Biedl syndrome and intellectual disability, who presented with recurrent hematometrocolpos from complete vaginal reatresia. Infant vaginoplasty for hydrometrocolpos failed due to discontinuation of postoperative dilation.At age 13, recurrent obstruction led her parents to choose hysterectomy, weighing her cognitive status, disease trajectory, and family support. The laparoscopic procedure was uneventful.

Conclusions: For adolescent BBS patients with vaginal atresia and intellectual disability, hysterectomy may be a more appropriate and humane surgical option than vaginoplasty. Clinical decision-making for this unique population should extend beyond anatomic reconstruction to address the compliance gap—the fundamental mismatch between the demanding lifelong postoperative care required for vaginoplasty and the patient's limited capacity to comply—which is collectively shaped by cognitive function, disease trajectory, and family support systems. These factors, often overlooked, critically determine postoperative outcomes and quality of life.

Background

Bardet–Biedl syndrome (BBS) is a rare autosomal recessive ciliopathy, with an estimated incidence of 1:160,000 in Northern European populations and 1:13,500 in certain Arab populations [3]. The disease is characterized by a constellation of primary features, including rod-cone dystrophy, obesity, postaxial polydactyly, renal abnormalities, genital anomalies, and cognitive impairment[1–5].

Genital anomalies are a well-recognized component of the BBS phenotype, with an incidence of 59%–98%[7]. In females, these abnormalities include vaginal atresia, transverse vaginal septum, uterine hypoplasia, uterine duplication, and hydrometrocolpos. Hydrometrocolpos stems from congenital vaginal atresia—the most common genital tract anomaly in BBS. It may present prenatally or neonatally as fluid accumulation from fetal cervical and endometrial gland secretions under maternal estrogen stimulation

[8–15], or after puberty as hematocolpos, manifesting with cyclic abdominal pain and a pelvic mass [2].

Vaginoplasty is currently the standard procedure for isolated vaginal atresia, aiming to restore anatomy and preserve fertilit[6]. However, when vaginal atresia is a local manifestation of BBS, a multisystemic disorder, management is substantially more complex. Given that cognitive impairment affects approximately 50% of BBS patients—with severity ranging from mild to moderate[5, 7]—the therapeutic

paradigm fundamentally shifts: the patient confronts not only anatomical obstruction but also lifelong self-care limitations and progressive multisystem decline.

Literature on the management of adolescent BBS patients with vaginal atresia is scarce and limited to case reports. Most reported cases underwent vaginoplasty, but postoperative complications—including sepsis, renal failure, and restenosis—were exceedingly common[2, 9, 17]. In other instances, hysterectomy was performed; however, the rationale for this choice and its impact on long-term quality of life were not thoroughly addressed[18, 19].

Notably, the "compliance gap" has never been independently incorporated into surgical decision-making in the existing literature. For an adolescent female with intellectual disability, whether the clinical benefits of uterine preservation and vaginal reconstruction outweigh the associated physical suffering and caregiving burden remains unclear. Using a case of a 13-year-old female BBS patient with vaginal atresia and intellectual disability, combined with a literature review, this study aims to explore whether hysterectomy offers greater clinical rationality and humanistic value than vaginoplasty in patients with a compliance gap.

Case presentation

A 13-year-old premenarcheal female (BMI 31.1 kg/m²) presented with 3-month cyclic lower abdominal pain. Pubertal development was Tanner II; menarche had not occurred.

Prenatal ultrasound at 27 weeks revealed a 6 cm retrovesicular cystic mass. Postnatally, she was found to have complete vaginal atresia, postaxial polydactyly, and ureteral dilatation. At 3 months of age, laparotomy confirmed hydrometrocolpos, and vaginoplasty with drainage was performed. Postoperatively, a vesicovaginal fistula developed, necessitating removal of the vaginal drain. At age 3, night blindness, hyperphagia, and progressive obesity emerged. At age 4, genetic testing confirmed BBS10 compound heterozygosity (paternal c.1514_1520del; maternal c.980G > T p.Gly327Val), and intellectual disability was documented.

At current presentation, she exhibited attention deficit, reduced verbal fluency, and emotional lability. Examination confirmed vaginal reatresia(Fig. 1); MRI and ultrasound revealed hematometrocolpos and mild pelvicalyceal dilatation. Renal function was mildly impaired (creatinine 69 μmol/L; uric acid 372 μmol/L).

After counseling, the parents chose hysterectomy over repeat vaginoplasty, citing severe intellectual disability, progressive BBS, and exhausted caregiving. Laparoscopic hysterectomy was uneventful; pathology confirmed hematometrocolpos(Fig. 2). She was discharged on day 2 and recovered well at 1-year follow-up.

Discussion

This case illustrates a distinct therapeutic challenge: managing a 13-year-old female with BBS, vaginal atresia, and intellectual disability. In contrast to prior reports favoring vaginoplasty[2, 17], we ultimately chose hysterectomy. We contend that, in this particular subgroup, the "compliance gap"—a critical determinant of therapeutic goals—should inform surgical decision-making. For this patient, hysterectomy appears a judiciously weighted and reasonable option. The ensuing discussion examines this choice across three domains: progressive disease trajectory, adherence capacity, and caregiver burden.

I. Progressive nature of BBS and intellectual disability: clinical benefit of fertility preservation approaches zero

BBS is a progressive degenerative disorder. With advancing age, patients experience worsening visual impairment, progressive obesity, and deteriorating renal function, resulting in a continuous decline in overall quality of life[6, 15, 20]. In individuals with significant intellectual impairment, the ability to self-manage these progressive conditions is severely limited, rendering them more vulnerable to the cumulative burden of disease progression and **diminishing their capacity to benefit** from routine surveillance and therapeutic interventions.

Under these circumstances, uterine preservation for fertility—a traditional surgical objective—holds little clinical relevance in this patient population. Three reasons support this view:

First, although a broad spectrum of genital anomalies has been documented in females with BBS—partly accounting for their diminished fertility[3]—the patient’s intellectual disability precludes independent parenting. Childbearing in this context would be neither fair to the patient nor ethically defensible for any potential offspring.

Second, the genetic risk is substantial. BBS follows autosomal recessive inheritance, with a 50% recurrence risk if the partner is a carrier, and 100% if the partner is also affected. In a patient unable to comprehend genetic risks or provide informed consent, actively preserving fertility raises serious ethical concerns.

Third, pregnancy itself carries high risks. BBS patients are predisposed to obesity, hypertension, and renal insufficiency, conditions that significantly increase the risk of severe pregnancy complications such as preeclampsia and renal failure, rendering maternal safety difficult to guarantee[21,22].

Therefore, in this patient, the "theoretical fertility potential" afforded by uterine retention represents an illusory benefit, with a weight far outweighed by the avoidance of pregnancy-related risks and the considerations of the surgical procedure itself.

II. Long-term management following vaginoplasty: a disproportionate burden in intellectually disabled patients

The success of vaginoplasty depends not only on surgical technique but also on long-term postoperative maintenance. The neovagina requires regular, sustained stenting to prevent contracture and reatresia, along with ongoing dilation to maintain patency[16].

For patients with intellectual disability, such long-term adherence is virtually unattainable. They are unable to comprehend the purpose of stenting, cooperate with dilation, or tolerate the associated discomfort—often leading to spontaneous removal of devices or refusal of care, culminating in exceptionally high rates of reatresia. In this patient, vaginal drainage performed at 3 months of age failed to address the obstruction after pubertal hormonal activation, illustrating that palliative drainage cannot substitute for definitive surgical correction.

Previous reports indicate that even when vaginoplasty is anatomically successful, improper stent placement or inadequate maintenance can lead to recurrent genital tract obstruction, retrograde menstruation, and genital tract abscesses, necessitating reoperation[2, 16]—which, in patients with preexisting renal impairment, may progress to renal failure and even death[17]. Our patient presented with mild renal insufficiency preoperatively, underscoring the importance of heightened vigilance in such cases.

Caregiver burden as a surgical criterion

Recent literature has increasingly recognized the profound impact of BBS on caregivers. Forsythe et al., in a large multi-country study of 242 caregivers, found that hyperphagia had a significant negative impact on caregiver sleep, mood, work, leisure, and relationships. Over half of caregivers reported that caring for a child with BBS resulted in a reduction in work hours (20%), temporary leave from work (19%), or permanent retirement (15%)[23].

This case extends these considerations to surgical decision-making. Thirteen years of caring for a severely intellectually impaired child had already imposed an immense and unrelenting burden on the family. The failed infant procedure signaled that reconstruction was unsuitable; repeat surgery would demand intensive postoperative care, while risking further complications. In contrast, hysterectomy definitively resolved hematometrocolpos, cyclic pain, and infection risk, eliminated menstruation, and substantially eased the caregiver burden, thereby improving quality of life. The decision was jointly reached after a comprehensive assessment of these cumulative burdens.

Implications for clinical practice

This case argues against considering "fertility preservation" an immutable gold standard in BBS patients with vaginal atresia and intellectual disability. Therapeutic decision-making should move beyond a purely anatomical reconstruction paradigm to integrate cognitive status, disease trajectory, social support networks, and familial burden—thus operationalizing the principles of patient-centered, individualized medicine.

Limitations

Study limitations include the single-case design and the relatively short follow-up duration. Ongoing evaluation of long-term quality of life and caregiver outcomes is required. Larger case series and multicenter studies are needed to establish evidence-based guidelines for this distinct patient population.

Conclusions

This case highlights a critical therapeutic dilemma in the management of BBS-related genital anomalies: the "compliance gap" between demanding postoperative requirements of reconstructive surgery and limited capacity for adherence in patients with severe intellectual disability. We propose that surgical decision-making in BBS should be stratified by neurological status, with hysterectomy considered a legitimate option in cognitively impaired patients in whom reconstructive approaches have failed—or, given the compliance gap, are predictably destined to fail. By incorporating caregiver burden and therapeutic care capacity into the decision-making process, clinicians can help families make informed choices that align with the overarching goals of minimizing morbidity and optimizing quality of life.

Abbreviations

BBS
Bardet–Biedl syndrome
MKKS
McKusick–Kaufman syndrome

Declarations

Ethics approval and consent to participate
This case report was reviewed by the Academic Development Committee, which determined that formal Institutional Review Board (IRB) approval was not required as the report describes a single anonymized patient case. The study was conducted in accordance with the principles of the Declaration of Helsinki. Written informed consent for publication was obtained from the patient’s parent.

Consent for publication
Written informed consent was obtained from the patient’s parent for publication of this case report, including all clinical data. They understand that the information will be freely available online and accessible to the public, and that once published, the work cannot ordinarily be removed from the public record. A completed consent form is archived by the corresponding author and is available upon request by the journal.

Availability of data and materials
Data sharing is not applicable to this article as no datasets were generated or analyzed during the

current study.

Competing interests
The authors declare no competing interests.

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Authors' contributions
SSZ conducted the literature search, collected the data, drafted the initial manuscript, and revised the manuscript. HMJ managed the patient and reviewed the manuscript. All authors read and approved the final manuscript.

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Figures



Figure 1

The patient presented with obesity, Tanner stage II breast development, a surgical scar on the right palm from previous excision of postaxial polydactyly, and vaginal atresia

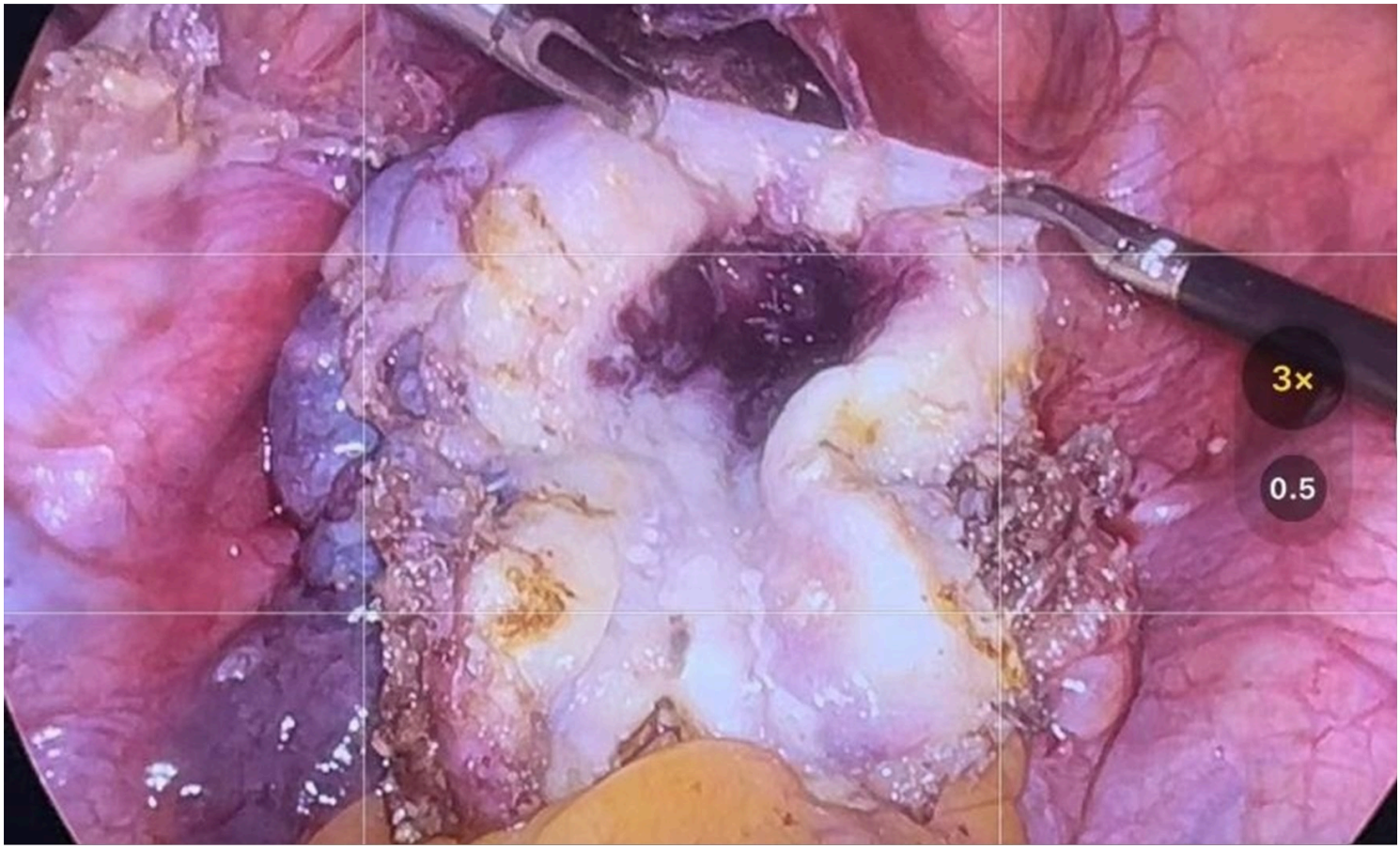


Figure 2

Surgically resected hypoplastic uterus with hematometra.