

Orgasmic Anejaculation Due to Prostatic and Right Seminal Vesicle Agenesis in a Patient with Prune-Belly Syndrome: A Rare Case Report and Literature Review

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Abstract

Case Report

Background: Prune-Belly syndrome (PBS) is a rare congenital disorder defined by the triad of abdominal wall muscular deficiency, severe urinary tract anomalies, and bilateral cryptorchidism. Genital anomalies affecting the prostate and seminal vesicles are less well studied in adulthood, particularly regarding their impact on ejaculatory function and fertility. **Case presentation:** We report the case of a 20-year-old single male student, followed since childhood for complete PBS bilateral vesico-ureteral reimplantation on a psoas-hitch bladder for bilateral megaureter and bilateral orchidopexy at age 6, simple right nephrectomy for pyonephrosis in 2021, an annually exchanged left double-J stent, and clean intermittent self-catheterization. He presented with orgasmic anejaculation since the onset of sexual activity. Clinical examination was unremarkable (BMI 25.2 kg/m², normally positioned urethral meatus, scrotal testes of normal size and firm consistency, palpable vasa deferentia without abnormality, no varicocele or palpable mass). Transrectal ultrasound failed to identify any prostatic tissue. Pelvic MRI confirmed complete prostatic agenesis together with agenesis of the right seminal vesicle, with probable ectopic termination of the right vas deferens into the left seminal vesicle. Hormonal work-up (FSH 12.8 IU/L, LH 7.1 IU/L, testosterone 11.76 IU/L) showed no major abnormality. **Discussion:** The combination of prostatic and seminal vesicle agenesis with PBS is an exceptional, likely under-reported, finding that highlights the embryological relationship between urogenital sinus derivatives and Wolffian duct derivatives in this condition. The resulting anejaculation reflects an anatomical rather than a neurogenic or psychogenic mechanism, which has direct implications for fertility management, favoring surgical sperm retrieval over penile vibratory stimulation or electroejaculation. **Conclusion:** This case highlights the value of systematically assessing sexual function and internal genital anatomy (transrectal ultrasound, pelvic MRI) in adult patients with PBS, and the need for early fertility-preservation counselling in this population.

Keywords: Prune-Belly syndrome; prostatic agenesis; seminal vesicle agenesis; anejaculation; male infertility; Wolffian duct malformation.

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INTRODUCTION

Prune-Belly syndrome (PBS), also known as Eagle-Barrett syndrome, is a rare congenital disorder, estimated to occur in approximately 1 in 30,000 to 50,000 live births, and affecting boys in the vast majority of cases [3]. It is classically defined by a triad combining deficiency of the anterior abdominal wall musculature, dilating urinary tract anomalies (mega bladder, megaureters, vesicoureteral reflux), and bilateral

cryptorchidism [4]. In reality, PBS is a multisystem disease with highly variable clinical expression and severity, ranging from in-utero lethal forms to phenotypes compatible with a near-normal life [4].

Pathophysiologically, two main theories are classically opposed: a functional lower urinary tract obstruction in utero (secondary to prostatic hypoplasia altering the configuration of the prostatic urethra during voiding), leading to secondary bladder distension and

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abdominal wall atrophy; and a primary disorder of genitourinary mesenchymal development occurring between the 5th and 10th weeks of gestation, which better accounts for the associated anomalies of the upper genital tract and testes [3,4]. Genetically, most cases remain sporadic, although variants in genes involved in embryonic genitourinary myogenesis (including PIEZO1) have recently been reported, opening the way to more structured genetic counselling [3].

Bilateral cryptorchidism, present in nearly all cases, is the best-documented genital anomaly of PBS. In contrast, anomalies of the prostate and seminal vesicles are far less frequently described, particularly in adulthood, and their impact on ejaculatory function remains poorly characterized. A survey-based study of adults with PBS found that half of male respondents reported anejaculation or retrograde ejaculation, without systematic anatomical correlation [4]. Furthermore, complete prostatic agenesis and seminal vesicle agenesis are, in isolation, exceptional anomalies, more commonly associated with the spectrum of mesonephric (Wolffian) duct malformations, which classically also includes congenital bilateral absence of the vas deferens, Zinner syndrome, or a single seminal vesicle associated with ipsilateral renal agenesis [8-11].

We report here the case of a young patient with complete PBS presenting with orgasmic anejaculation related to prostatic agenesis and right seminal vesicle agenesis, with a probable ectopic termination of the contralateral vas deferens. To our knowledge, this precise combination has not previously been reported in such detail in the literature, which justifies the interest of

this observation both from a pathophysiological standpoint and for the practical management of fertility in these patients as they reach adulthood.

CASE PRESENTATION

The patient is a 20-year-old single male student, with no relevant family history, followed since early childhood for Prune-Belly syndrome. His surgical history includes bilateral vesico-ureteral reimplantation on a psoas-hitch bladder for bilateral megaureter combined with bilateral orchidopexy performed at age 6, followed by initiation of a clean intermittent self-catheterization programme. A simple right nephrectomy was performed in 2021 for pyonephrosis. A left double-J ureteral stent is maintained in place, with regular annual exchanges.

The patient presented with orgasmic anejaculation that is, the occurrence of orgasm without any perceived antegrade or retrograde emission of semen present since the onset of his sexual activity. There was no new voiding complaint and no associated pelvic pain.

On clinical examination, the patient was in good general condition, with a body mass index of 25.2 kg/m². The urethral meatus was normally positioned and of normal caliber. Both testes were scrotal, of normal size and consistency (firm, non-atrophic) (Figure 1). Palpation of both vasa deferentia revealed no macroscopic abnormality (palpable vasa, no nodule or induration). There was no varicocele and no palpable pelvic or scrotal mass.

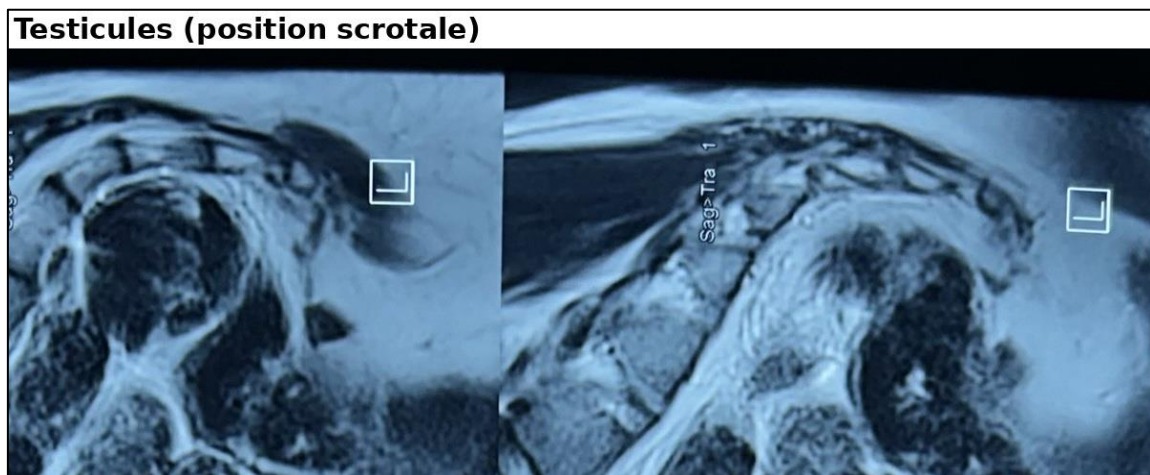


Figure 1: Sagittal T2-weighted MRI centered on the scrotum : testes in normal position, with homogeneous signal, no focal lesion or atrophy, consistent with the clinical examination. (Image anonymized; cropped to the region of interest.)

Transrectal ultrasound failed to identify any distinguishable prostatic tissue, raising the suspicion of prostatic agenesis. A complementary pelvic MRI confirmed complete prostatic agenesis, together with agenesis of the right seminal vesicle (Figure 2). It also

demonstrated a probable ectopic termination of the right vas deferens into the left seminal vesicle, suggesting a defect in the midline convergence of the right-sided mesonephric derivatives.

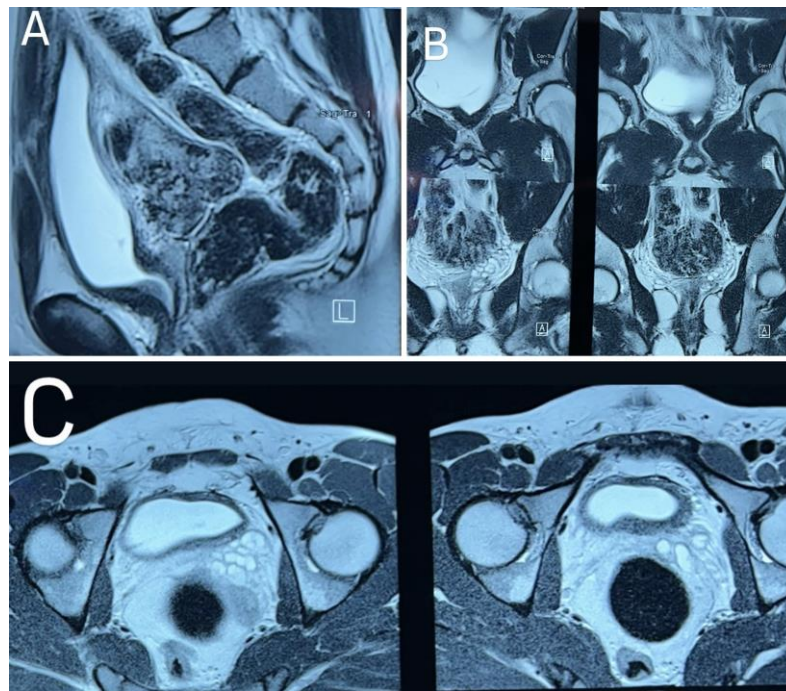


Figure 2: Pelvic MRI (T2-weighted sequences). A : mid-sagittal view showing no identifiable prostatic structure in the retrovesical position, consistent with prostatic agenesis. B : coronal view confirming the absence of a recognizable prostate gland and right seminal vesicle. C : axial view at the pelvic level showing asymmetry of the seminal remnants, with no identifiable right seminal vesicle. (Images anonymized; cropped to the region of interest.)

Hormonal work-up showed a plasma FSH of 12.8 IU/L, a plasma LH of 7.1 IU/L, and a testosterone level of 11.76 IU/L, with no pattern suggestive of peripheral or central hypogonadism at the time of assessment. Semen analysis and a search for spermatozoa in post-orgasmic urine were proposed as complementary work-up, to exclude an unrecognized retrograde component and to guide the fertility-preservation strategy.

DISCUSSION

Spectrum of genital anomalies in Prune-Belly syndrome

Bilateral cryptorchidism, found in nearly all boys with PBS, is classically accompanied by prostatic hypoplasia of variable degree, itself implicated in the obstructive pathophysiological theories of the syndrome [3,4]. Complete prostatic agenesis, however, is a far rarer occurrence, reported only sporadically in the PBS literature, most often incidentally on imaging performed in adult patients presenting with sexual or fertility complaints. Likewise, reports of PBS associated with scrotal, dental, or gastrointestinal anomalies illustrate the wide phenotypic variability of this multisystem disease [5,6,7].

Embryological basis of combined prostatic and seminal vesicle agenesis

The prostate arises from the urogenital sinus, an endodermal structure, under the influence of testicular androgens, whereas the seminal vesicles and vasa deferentia develop from the mesonephric (Wolffian)

duct, a mesodermal structure. Although these two embryological lineages lie in close proximity within the pelvis and both depend on an adequate androgenic environment, they remain histogenetically distinct. The coexistence, in our patient, of prostatic agenesis with unilateral seminal vesicle agenesis and contralateral ectopic termination of the vas deferens argues in favour of an early, global disturbance of caudal urogenital development, rather than an isolated defect confined to a single lineage. This hypothesis aligns with the theory of a primary genitourinary mesenchymal disorder proposed in PBS pathophysiology, which could jointly affect sinus-derived and Wolffian-derived structures [3,4].

Isolated anomalies of Wolffian duct derivatives are, on the other hand, well described outside the context of PBS: unilateral or bilateral vasal agenesis, single seminal vesicle, seminal vesicle cyst associated with ipsilateral renal agenesis (Zinner syndrome), or ectopic fusion of the contralateral seminal pathways, as reported in several recent observations [8,9,10,11]. These malformations share with our case an ectopic or aberrant course of the seminal tract, reflecting a common defect in the migration or regression of the mesonephric duct.

Anejaculation: from presumed functional cause to anatomical proof

In patients with PBS, ejaculatory disorders have long been presumptively attributed to prostatic hypoplasia and bladder-neck incompetence, favouring mainly retrograde ejaculation [4]. Our case illustrates that orgasmic anejaculation may also correspond to a

genuine anatomical absence of the accessory glands normally responsible for most of the ejaculate volume (prostatic and seminal vesicle secretions), rather than to a simple bladder-neck functional disorder. This distinction is essential, as it directs diagnostic and therapeutic management along different paths.

From a diagnostic standpoint, a careful clinical examination (assessment for varicocele, palpation of the vasa, evaluation of testicular position), hormonal work-up, search for spermatozoa in post-orgasmic urine, and dedicated pelvic imaging (transrectal ultrasound followed by MRI when in doubt) are recommended whenever obstruction or agenesis of the distal genital tract is suspected, particularly when an associated renal anomaly is already known [11].

Implications for fertility preservation

The treatment options classically proposed for anejaculation penile vibratory stimulation and electroejaculation have proven effective mainly in neurogenic (spinal cord injury) or psychogenic anejaculation, situations in which the accessory genital apparatus is anatomically present but its reflex or voluntary activation is impaired [12,13,16]. In these indications, electroejaculation allows retrieval of spermatozoa usable for intrauterine insemination or intracytoplasmic sperm injection (ICSI), with meaningful cumulative pregnancy rates reported across several series [13,15]. In contrast, in the setting of prostatic and seminal vesicle agenesis, as in our patient, the absence of the glandular structures that normally produce the ejaculate makes the efficacy of these stimulation techniques highly unlikely, since no functional seminal reservoir exists to be mobilized.

In this context, surgical sperm retrieval directly from the testis or epididymis (TESE/PESA), followed by ICSI, represents the most realistic option for a paternity project, provided that spermatogenesis is preserved which is suggested in our patient by testes of normal volume and consistency and an unremarkable hormonal work-up [14]. Psychosexual counselling is also recommended, as anejaculation can have a substantial impact on the sexual life and self-image of a young adult.

Overall follow-up of the adult patient with Prune-Belly syndrome

Beyond the fertility question, this case is a reminder of the need for prolonged urological and nephrological surveillance in adult patients with PBS, given the risk of progression to chronic kidney disease and the complications inherent to long-term intermittent catheterization [1,2]. A multidisciplinary approach combining urology, nephrology, andrology and, where relevant, reproductive medicine, appears best suited to this population, whose life expectancy and quality of life have improved considerably over recent decades.

CONCLUSION

The combination of prostatic and right seminal vesicle agenesis with Prune-Belly syndrome, resulting in orgasmic anejaculation, is a rare finding that broadens the spectrum of genital anomalies described in this condition. It highlights the value of systematic anatomical assessment by pelvic imaging (transrectal ultrasound, MRI) in any PBS patient reporting an ejaculatory disorder, in order to distinguish an anatomical from a purely functional cause, and to guide fertility-preservation counselling as early as young adulthood.

DECLARATIONS

Patient consent: Written informed consent was obtained from the patient for publication of this case report and associated imaging data, in accordance with the CARE (CAsE REport) guidelines. Data were anonymized.

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