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Letter to the Editor

Letter to Editor - Utility of genetic work-up for 46, XY patients with severe hypospadias

It was really difficult for us to understand the context of this article, which we believe is a combination of clinical observations of hypospadias and scientific data from genetic studies [1]. The authors have included a very small number of hypospadias patients with heterogeneous anatomy, and simply classify them as "standard" and "severe" hypospadias. As per the published literature, the classification of these cases as standard, or cases with a small glans size, etc., lacks a conclusive scientific/anatomical basis. It is sobering to see that the term "standard hypospadias" is not present in human anatomical and surgical texts. The authors could have utilized the normal anatomical characteristics of penis for a better segregation of these cases. Moreover, in this study, the clinical findings of the patients were termed as "anatomical characteristics". However, the authors seem to show a departure from the normal postnatal anatomical characteristics of penis, e.g., septum glandis and frenulum. The septum glandis is a well-documented fibrous median partition of the glans penis and the frenulum is its epithelial extension to the foreskin [2–4]. It is unfortunate to see that many surgeons (including the senior author) have shown a consistent denial in accepting these normal

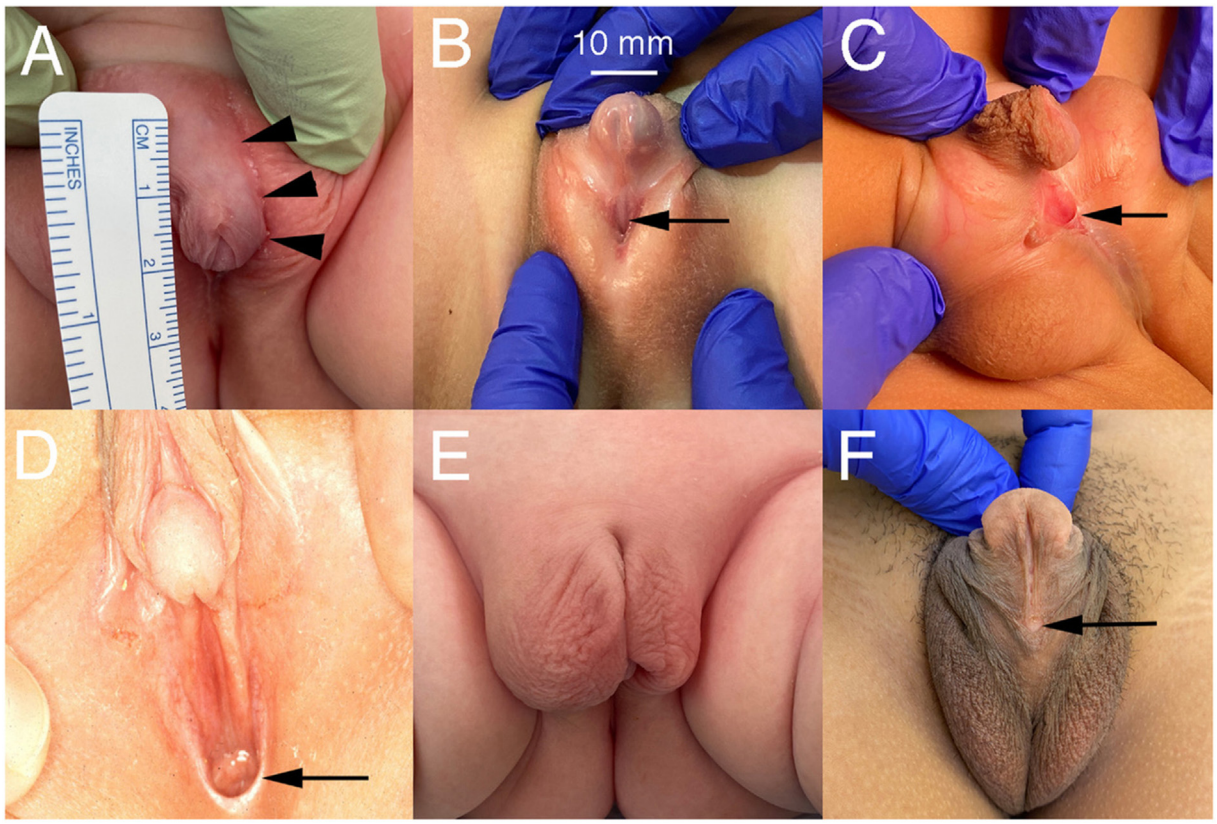

Fig. 1 The visible structure in Fig.1-D is a blind ending vaginal remnant, not a prostatic utricle. The prostatic utricle is an embryonic remnant that cannot be observed on clinical examination.

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postnatal anatomical characteristics published hundreds of years ago [5].

The study is supplemented with different types of surgical repair techniques performed on hypospadias patients. However, we found no correlation between the genomic test results and the patients' surgical procedures and/or outcomes. It is also controversial and not clear how the authors included patients with testicular descent and excluded undescended testicles associated with hypospadias. Among those 14 patients, there are patients with orchiopexy (patient 1), orchiectomy (patient 4,8), hernia repair (patient 7), intraabdominal testis (patient 14). Obviously, there is little to no significant data on the results which can be used in genetic counselling of the patients classified as severe hypospadias with descended testes. It will be really interesting to see the reference no 46 (neither visible in the reference list nor properly cited in the text), highlighting that patients with severe hypospadias are at risk for testicular ascent or acquired cryptorchidism.

The genital malformation shown in Fig. 1-D (which cannot be seen in high resolution) was described as "perineal ectopic meatus with visible prostatic utricle". As we could see, this image shows the so-called "pseudovaginal, perineoscrotal" type of urethral opening in the external genitalia and clinical evidence of a blind ending vagina. This type of clinical finding is a typical presentation of the external genitalia in patients with 5-alpha reductase enzyme deficiency. According to the current embryological definitions, prostatic utricle is named for a müllerian duct remnant at a higher level in the posterior urethra between the two ejaculatory ducts, denoting the fusion of the caudal end of the müllerian duct, but retaining its proximal portion.

The authors' conclusion that patients with severe hypospadias with descended testes can benefit from molecular genetic testing derives mainly from a limited number of observations, making this work more of a theoretical than a scientific manuscript.

Conflict of interest

None.

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