Familial isolated congenital penile torsion

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A R T I C L E   I N F O
Article history:
Received 29 December 2012
Received in revised form 6 April 2013
Accepted 8 April 2013

Key words:
Congenital penile torsion
Hypospadias
Genetics

A B S T R A C T
Congenital penile torsion is a three-dimensional aberration in penile development producing abnormal helical structures of the corpora. We present a case of isolated congenital penile torsion in brothers. This is the first indication of a potential hereditary component in the development of isolated congenital penile torsion in three decades.

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Congenital penile torsion (CPT) is a three-dimensional abnormality of rotation of the corporal bodies producing an aberrant helical structure [1]. The first case of CPT was described with hypospadias in 1857 [2].

The severity of the deformity is based on the degree of glanular angulation into mild (<45°), moderate (45–90°) or severe (>90°) [3]. The structural deformity is almost exclusively counterclockwise; in a series of 370 only two of the deformities were clockwise [4,5]. CPT can occur in isolation or in combination with hypospadias or chordee; 34 cases have been reported illustrating this association [4].

A limiting factor in diagnosis is glans accessibility, therefore the condition may remain unrecognized until circumcision or the foreskin becomes retractile. There has been a paucity of data to define the incidence of isolated congenital torsion, but it was historically considered a rare disease [2,6–8]. In a prospective study of 274 neonates, mild torsion was described in 1.5% and severe isolated torsion in 0.7% [9]. However, a recent study exploring the incidence in 370 neonates that were exclusively circumcised found an incidence of isolated neonatal penile torsion of 30%, although the overwhelming majority only had a mild deformity (87% with an angle of 11–20°). No neonates were observed to have a degree of deviation of greater than 60° of the glans [5]. The natural history of this group has not been defined.

The indication for interventions are cosmetic and functional [10,11]. It is generally regarded that deformities of greater than 60° should be considered for corrective surgery in the absence of associated hypospadias or chordee [4]. The functional sequelae of isolated CPT in adulthood are not well described. However in a study of more than 11,000 men presenting to infertility/sexual dysfunction clinics in Egypt, 12% had penile torsion. The degree of torsion was only mild in 80% of cases, but 5% had torsion of more than 60° [12].

1. Case report

A four-year-old boy presented with isolated CPT of 90° counterclockwise rotation. This was surgically corrected with penile degloving, rotational correction and redundant skin was resected. Three months following this he was observed to have a successful correction with no residual rotation.

Following this his six-year old brother presented with difficulty directing his urinary stream. He was initially treated with steroid cream because of adhesive foreskin which was not retractile. Following this the foreskin could be retracted revealing an isolated torsion of 30°. He was brought forward for corrective surgery and circumcision for severe phimosis. The phallus was degloved to
2. Discussion

The only report that acknowledges a potential hereditary component in the etiology of CPT was published three decades ago. In this paper Paxson et al. observed congenital torsion in five newborns, three of whom had a paternal history of penile torsion [7]. By contrast, the etiology and molecular genetics of hypospadias has been described much more comprehensively. The overall risk of a brother of an affected infant also having hypospadias is 9% [13,14]. There is 18% concordance amongst same-sex twins in both mild and severe hypospadias [15].

It has been recognized that familial clustering may be at least partly due to shared environmental exposures [16]. Nevertheless, several candidate genes have been identified for further population-based evaluation including SRD5A2, HSD17B3 and the AR genes [15]. DGKK has been implicated as a major risk gene in a population based European study with demonstrated expression on preputial tissue [17].

Several androgen receptor antagonists have been implicated in animal rat models including dichlorodiphenyltrichloroethane (DDT) metabolites. These have been correlated to reproductive abnormalities including hypospadias at exposure levels similar to some human populations [18]. The culminating multifactorial model for hypospadias states that when genetic susceptibility and antiandrogen environmental exposure surpass a threshold the malformation results.

The etiology of isolated CPT is poorly understood [10,19]. There is a cacophony of models for the abnormal development in CPT, which is mirrored by a variety of surgical approaches to achieve correction. CPT has been at least partially attributed to false attachment of the ectoderm (skin and darts fascia) [2,20]. Based on this, penile degloving and mobilization of the tunica has been proposed as a corrective measure [11]. This technique is successful in correcting the great majority of cases, but not universally [4,19]. A dorsal darts rotation technique may provide more scope to treat severe or refractory cases and can be integrated into coexisting hypospadias or chordee repairs [3].

Asymmetric development of the corpora cavernosa and subsequent corporal disproportion may be the another contributing factor in cases of CPT [19]. Zhou et al. consistently observed a hypoplastic left corpus cavernosum, and often a fibrous band attaching the left corpus to the pubic peristome. To this end, release of the fibrous bands and suturing of the lateral aspect of the tunica albuginea to the pubic peristome was employed with success [19].

A more invasive approach in severe cases involves excision of angled elipses from the corporal tissue and plication, the so-called modified Nesbit procedure [21]. Though effective, it is associated with neurovascular injury, hemorrhage and penile shortening.

Bhat et al. have suggested that CPT may be related to eccentric fusion of the ectodermal/endordermal folds as in Fig. 1 [4]. This lateral fusion leads to aberrant mesodermal proliferation and unilateral attachment of fascial layers of the penis and spongiosum. Circumstantial evidence for this is the fact that deviation of the median raphe correlates with deviation of the meatus in almost all cases.

All the surgical approaches to CPT have had results that have not been replicated in the literature, no studies have reported long-term outcomes, and there is no consensus on the most effective approach to repair [10]. Furthermore, the fundamental cause of the anatomical deformity remains elusive. Bauer and Kogan have hypothesized that the mechanism causing the malformed Buck’s fascia is the fetal heel in the groin [10]. This does not explain the preponderance to counter-clockwise torsion.

Conflict of interests
None.

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Fig. 1. Embryo-pathology of the congenital penile torsion. (A) The development of urethral folds bridging the urethral groove up to but not including the glans during the 10th week of development. (B) At the end of the 3rd month the fusion of endoderm and ectoderm has occurred at the midline raphe. (C) and (D) illustrate eccentric development of the urethral folds, their fusion off midline results in CPT. (For interpretation of the references to color in this figure legend, the reader is referred to the web version of this article.)