Rare disease

Fowler's syndrome and pregnancy

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Summary

We present a favourable outcome of a pregnant woman with underlying Fowler's syndrome. This is a rare disorder affecting young women with painless urinary retention, and as there is no known cure, the main concern is to ensure adequate bladder emptying. Our patient went through self-catheterisation and suprapubic catheters after which she was finally successfully managed on a sacral nerve stimulator (SNS).

Upon getting pregnant the stimulator was switched off due to safety concerns, and from then on, bladder emptying was managed with a suprapubic catheter (which was regularly changed over the gestation). The main concern thereafter was recurrent hospital admissions with urinary tract infections and pelvic pain requiring parenteral antibiotics and analgesia.

She underwent an uncomplicated elective caesarean section at 36 weeks and delivered a healthy female infant. The SNS was successfully reactivated in the postnatal phase, and the suprapubic catheter was removed upon achieving adequate urethral output.

BACKGROUND

Fowler's syndrome is a relatively rare disorder with significant implications during pregnancy due to difficulty in maintaining adequate bladder emptying and the risk of recurrent urinary tract infections with their associated morbidity to the mother and fetus.

CASE PRESENTATION

We report the successful outcome of a pregnancy in a 32-year-old patient with Fowler's syndrome. Mrs P was diagnosed with the condition 10 years prior to the index pregnancy, following recurrent urinary tract infections associated with symptoms of voiding dysfunction and difficulty with defecation. Although she was initially managed with intermittent self-catheterisation and anti-cholinergic agents, followed by the long-term insertion of suprapubic catheters, the results were disappointing. She finally had placement of a sacral nerve stimulator (SNS) at the National Hospital for Neurology and Neurosurgery (NHNN) in London with a good clinical response.

Mrs P's first presentation to our Obstetric Department was at 7 weeks gestation with a urinary tract infection and an infected suprapubic catheter site. Her SNS had been deactivated at the NHNN due to concerns about the safety of the device during pregnancy. Mrs P responded well to medical management, and she was discharged on a low-dose-maintenance regimen of antibiotic to cover the remainder of her pregnancy.

Mrs P had multiple admissions under the care of the urology and obstetric teams during the second and third trimester with recurrent urinary tract infections associated with significant pelvic pain. These required parenteral antibiotic treatment and changes of her suprapubic catheter. The suprapubic pain was difficult to manage, with Mrs P requiring opioid analgesia to stabilise her symptoms, initially as an inpatient but subsequently in the community.

With the onset of the third trimester Mrs P's urinary symptoms worsened, resulting in more frequent and prolonged admissions. At 31 weeks gestation she was administered a course of betamethasone to promote fetal lung maturation, and an elective caesarean section was planned for 36 weeks. Serial ultrasound examinations demonstrated normality of fetal growth and liquor volume.

At her request her SNS was reactivated by the NHNN at 32 weeks gestation. However, Mrs P chose to leave it in its non-functional state until she had delivered despite the numerous admissions, courses of systemic antibiotics, and insertions of central venous access and doses of analgesics that this approach entailed.

OUTCOME AND FOLLOW-UP

At 36+2 weeks, Mrs P underwent an uncomplicated elective caesarean section. A healthy female infant weighing 3.15 kg was delivered through a low transverse (Cohen) incision (2 cm above the fistula tract from the indwelling suprapubic catheter) and uterine entry by a transverse incision (at junction of upper and lower segment) without bladder reflection. There was no evidence of extension or damage to the fistula tract, and clear urine was observed to be draining postoperatively.

During the postpartum period, the SNS implant was 'turned on' as planned and suprapubic catheter clamping was initiated. Her urethral output increased gradually to achieve bladder residual urine of <100–150 ml, after which the suprapubic catheter was removed. The remainder of her postnatal recovery was uncomplicated.

DISCUSSION

First described in 1985, Fowler's syndrome is the association of painless urinary retention occurring as an isolated disorder in a young woman associated with polycystic ovaries and abnormal urethral sphincter electromyogram

(EMG). The full aetiology of this condition remains to be explained, but it has been hypothesised that the disorder is due to a hormonally sensitive channelopathy, which results in a sustained involuntary contraction of the striated urethral sphincter. This in turn has an inhibitory effect on detrusor contractions as well as the desire to void.¹

The classical presentation is with a woman in her 20–30s with an intermittent inability to pass urine with a poor stream on voiding. This may happen spontaneously or may be noted following an operative procedure (gynaecological, urological or even ENT) or following childbirth.²

The gold standard investigation is a urethral sphincter EMG. The diagnostic findings compatible with Fowler's syndrome are complex repetitive discharges and decelerating bursts, which when transduced to the audible spectrum sound like the noise of a helicopter and a whale under water, respectively. Such abnormal activity has been suggested to prevent adequate relaxation of the sphincter during voiding, causing outflow obstruction of the bladder, an increased residual volume of urine and eventually failure of the detrusor muscle.³

As there is as yet no cure for this condition, the aim of treatment is to try to ensure bladder emptying. A small number of patients have a poor urinary stream, although some void almost normally. If the residual volume is low, no further intervention is necessary. Patients with larger residual volumes need more definitive treatment, which is carried out by means of either intermittent self-catheterisation or placement of a suprapubic catheter. A number of individuals are unable to self-catheterise as a consequence of extreme urethral sensitivity and spasm. Sacral neurostimulation is reserved for the more severe candidates, as this has shown to be the only effective therapy in women with urinary retention and voiding dysfunction secondary to urethral sphincter over activity.⁴

Successful placement of the SNS is by a two-stage process. The purpose of the first stage is to identify patients who develop improved voiding with sacral nerve stimulation. These patients go on to receive the SNS (Interstim Therapy), with a control 'unit' implanted in the subcutaneous fat of the buttock or anterior abdominal wall with electrodes located close to the sacral roots. Despite the confirmed efficacy, the procedure has significant complications, including lead migration, pain, infection and device failure.⁴

The inception of pregnancy creates several complications in an already complex clinical scenario. The foremost concern is to establish a method for ensuring that the bladder empties completely. As in Mrs P's case complications are common – specifically recurrent urinary tract infections, suprapubic pain and repeated hospital admissions for control of symptoms.

Sacral neurostimulation presents a significant dilemma in pregnancy, as there is no evidence regarding its safety profile. The advice from clinicians managing patients with SNS devices is to deactivate the stimulator.⁵ The leaflet provided by the company (Medtronic) openly states that 'the safety and effectiveness of this therapy has not been established for pregnancy, unborn fetus, or delivery.'⁶ It is notable that the experience of experts managing similar patients (those who accepted the risks and did not wish their SNS to be turned off) does not show any evidence of adverse effects

on the fetus or pregnancy (except for a sporadic finding of an isolated fetal limb defect in one case) (S Elneil, personal communication). Nevertheless, at present there are not enough patient data to reach a definitive conclusion regarding the safety profile of the SNS during pregnancy.

The implications in pregnancy for such an approach are compounded by the increased risk of urinary tract infections associated with invasive drainage of the maternal bladder. Meta-analyses of studies evaluating bacteriuria in pregnancy have concluded that there are confirmed associations with preterm delivery and low birth weight. In addition, there are increased risks of pre-eclampsia, anaemia, chorioamnionitis and postpartum endometritis. Fetal risks include fetal growth restriction, stillbirth, perinatal mortality, mental retardation and developmental delay.⁷

There is no evidence in literature to determine the ideal mode of delivery specifically for a case of Fowler's syndrome. There is some evidence that women with SNSs should be offered a caesarean section to avoid possible sacral lead damage or displacement during vaginal delivery.⁵ Most of the available evidence relates to impact of childbirth on the pelvic floor and the effect of caesarean delivery on prevalence of postpartum urinary incontinence.^{8–9} In general, a first delivery by caesarean section appears to reduce the short-term and long-term risk of urinary incontinence, but other factors (age, obstetric history, obesity, instrumental delivery) are also important.^{8–9}

With the available evidence to guide the ideal mode of delivery and no clear reference to impact of vaginal delivery on the pathophysiology of the disorder, we concluded that an elective caesarean section would be the safest course of action. This decision was made in conjunction with the urologists, keeping in mind the chronic and incurable nature of the underlying disease and to prevent any possible further damage to the bladder and urethral sphincter that may result from a vaginal delivery.

To our knowledge based on literature search, our case is the first of its kind to be published. We hope it provides optimism to those with a similar condition who wish to embark on a pregnancy and to the clinicians who look after them.

Learning points

- ▶ A pregnant patient with Fowler's syndrome requires multi-disciplinary care involving obstetricians and urologists for an optimal outcome.
- ▶ Recurrent urinary tract infection is to be anticipated and aggressively managed.
- ▶ Delivery by caesarean section is appropriate to prevent damage to the lower urinary tract (and SNS).
- ▶ Further research work is required on certain aspects of care (safety of sacral nerve stimulator implant in pregnancy, impact of mode of delivery on disease).

Competing interests None.

Patient consent Obtained.

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