The Challenges and Anxieties of Pregnancy in Cystinuria

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Abstract

Cystinuria is an inherited disease in which the reabsorption of cystine and dibasic amino acids from the renal tubules are disrupted, leading to cystine supersaturation in the urine and nephrolithiasis (1). Disease management in pregnant women is challenging due to concerns around radiation exposure related to imaging (2), the contraindication of thiol-based pharmacological treatment (3), the contraindication of some surgical procedures (4), and the risks of repeat surgical procedures (4). In this study we examine the experience of a cohort of previously pregnant cystinuric women with the aim of identifying areas of care which could be improved.

Aims

In this study, we aimed to identify key areas of patient's experiences during pregnancy which they found challenging or distressing. Specific areas of the patient's experience which we wished to evaluate included the availability of pre-pregnancy advice and support, problems which patients encountered during their treatment, and any additional areas of concern patients had about their care.

Methodology

From a prospective database of patients referred to our dedicated cystinuria clinic, we identified 37 women who were known to have had children. No exclusion criteria were applied. With the aid of patients from our cohort, we designed a provisional questionnaire which sought to evaluate the challenges encountered by this patient group during pregnancy. We met with one patient from our cohort at our clinic to discuss her experience of cystinuria and any areas of concern she had regarding her treatment. We used her input to revise our questions before a final draft was agreed upon and sent out to our patient group. The questionnaire was modified into a Google form, with drop down boxes for easy electronic completion. Answers to the questionnaire were recorded and tabulated into a spreadsheet for analysis. The genotype of each patient was also obtained from hospital records. Local governance approval was obtained.

Results

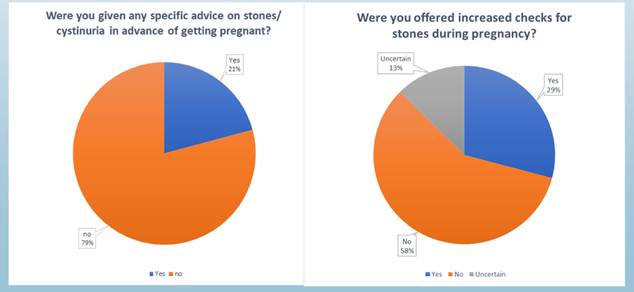
22 patients responded. Mean age 39 (range 22-59). Median age of diagnosis was 22 (range 9-40). Median number of children was 2 (range 1-4) and age of children was 4 months to 22 years.

74% were or had previously taken alkalinisation; 50% were or had taken thiol binding medication. The majority had been pregnant prior to referral to our service and are therefore likely to have been managed in general stone clinics.

Patient Support and Advice

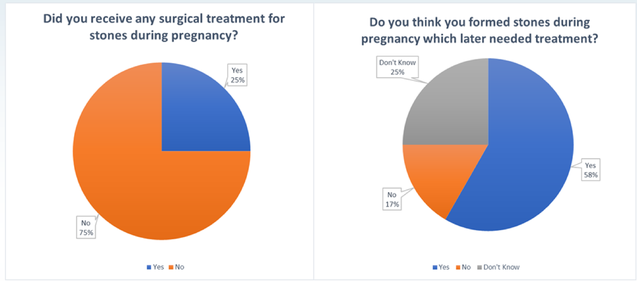
We found that many patients were under-supported, and anxiety among our patients was found to be common. 37% of patients reporting feelings of anxiety prior to their pregnancies due to cystinuria. Only 21% were given specific advice or reassurance from a healthcare professional before becoming pregnant. Despite increased risk of stone formation, only 42% of our cohort were offered increased checks for stone monitoring during their pregnancy.

Liaison between urology and obstetric teams also seemed to be lacking in many cases, as only 29% of patients felt that their midwives had an appreciation of their condition.



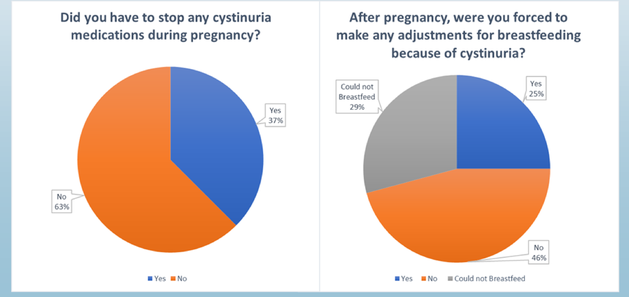
Symptoms and Stone Formation

Half of respondents reported suffering from pain due to stones during pregnancy. 58% felt they formed stones during pregnancy that subsequently needed treating. 32% passed stones during their pregnancy and 25% reported needing surgery during pregnancy.



Issues with Medication

37% of patients were forced to stop taking medication which had been prescribed to control their urinary cystine levels after becoming pregnant. Medication also negatively impacted the ability of patients to breastfeed. 29% of patients were unable to breastfeed their children at all, and a further 25% reported having to either discontinue medications in the post-natal period in order to be able to breastfeed or stop breastfeeding early.



Overall, whilst 64% do not think cystinuria is a barrier to pregnancy, 23% do still think it is. The majority (91%) report being concerned their children could have cystinuria and 77% have actually sought advice for them whilst 50% have actually been tested.

Of those with genetic testing, 11 had SLC3A1 mutation; 8 SLC 7A9 and 3 had both. There was no clinical differences experienced in pregnancy between the groups.

Conclusions

Cystinuria causes significant anxiety to patients considering pregnancy as well as challenges during and afterwards due to cessation of medication, frequent stone episodes and extra surgery. Patients should be counselled by urologists to ease the anxiety and offered extra ultrasound checks and easy access back to urological services if stone episodes occur. Liaising directly with the obstetric teams will help to improve their knowledge as well as patient experience. With appropriate support women should not see cystinuria as a barrier to childbirth. In future work, it would be interesting to explore the concerns for women yet to have children.

References and Contact

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