



Berkeley
UNIVERSITY OF CALIFORNIA



UCSF Clinical & Translational
Science Institute

University of California San
Francisco IRB Approval
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Interested in taking part in a research study on fertility issues in males with nephropathic cystinosis?

About the Study: Dr. Minnie Sarwal (Nephrologist, UCSF), Dr. James Smith (Director of Male Reproductive Health, UCSF), and Dr. Polina Lishko (Human Reproductive Health Specialist, University of California Berkeley) are conducting a study to learn the cause of fertility issues in adult, male patients with nephropathic cystinosis who are treated with cysteamine. This study is currently jointly funded by Cystinosis Ireland (CI) and Cystinosis Research Network, USA (CRN). While the majority of men with cystinosis have not fathered children, there have been cases of some men becoming a father through assisted fertility. This has not been an easy journey and this study aims to find out more on the causes of male infertility in the hope it might help future treatment.

Primary Goals of the Study:

1. To better understand what causes male fertility issues in nephropathic cystinosis
 2. To design novel therapies to potentially prevent young boys from losing male reproductive function, and
 3. Patients with current infertility issues will be offered an option for sperm preservation for future use.
- Follow-up to be discussed with Dr. Smith after initial visit.

What the Study Involves:

- Participation in this study would involve one visit to UCSF. The visit would include the following: Clinical examination, a blood test, ultrasound (of the testes which is a painless procedure similar to ultrasound used for pregnant women) examination for testicular volume and architecture, semen analysis, sperm motility (this means how fast sperm swim which can indicate the health of the sperm), morphology, the concentration of white blood cells, the level of fructose in the semen (semen is an organic bodily fluid, which contain sperms. Higher the absolute fructose concentration in the semen, lower is the number of sperms), and pH (pH is a measure of how acidic/basic water is. The pH of semen plays a crucial role in maintaining quality of sperm ensuring fertilization). Testicular and epididymal (coiled tube behind each testis) biopsies, sperm will be obtained by the TeSE (a procedure to collect sperm directly from testes), well-established techniques, performed routinely by Dr. James Smith. We will also collect semen via masturbation.
- All interested participants will have an online consultation via zoom with Dr. Smith to discuss this process prior to the visit to UCSF. Please see below the contact info if you would like to participate.
- If you are travelling to UCSF, part payment will be provided to help defray the cost of the travel. All clinic, blood draw, imaging and procedure costs at UCSF will be covered. For biopsies and TeSE local anesthesia will be used. When the biopsy is taken, you will feel pressure or minor discomfort. Mild narcotic to relieve the pain will be given. You would need to take it easy and rest for a week or two to recover fully.
- You will receive a free consultation with Dr. Smith, counselling for your results, and the test results will be available to your care team.

Benefits of Participating: Participation in this study will help us to understand the causes of male infertility in cystinosis. Understanding the cause will provide novel insights into new surgical and therapeutic approaches to either prevent or reverse male infertility in cystinosis. You will get a detailed report of your clinical, biochemical, and semen examinations. You will not be involved in treatment at this phase, however we hope that the results from this research will lead to the development of future treatment trials. All male participants with cystinosis will be offered the option of sperm preservation.

Who May Participate: You are eligible to participate if you are greater than 18 years of age (no upper limit) with a diagnosis of nephropathic cystinosis.

If you would like to participate in this study or if you would like more information, please contact minnie.sarwal@ucsf.edu or the study coordinator, Jim Cimino at 415/514.0192 or email to jim.cimino@ucsf.edu