

VCFS Research Study

Drs. Kucherlapati and Finn of the Department of Genetics are conducting a VCFS research study at Children's Hospital, Massachusetts General Hospital, and Brigham and Women's Hospital in Boston, Massachusetts.

This study is looking at the relationship between changes in genes and the observed medical conditions seen in patients with VCFS/DiGeorge Syndrome, and their family members. We are looking for children and adults with VCFS, and their family members (parents, siblings) both with and without VCFS who are interested in participating in a research study.

Participation would involve a visit to the hospital, lasting about two hours. At the visit, patients with VCFS will have a physical examination, a review of their medical and family history, and a blood sample collection. Family members will also be asked to provide a small sample of blood. We will use this blood to isolate DNA and look for changes in genes that might be associated with different medical conditions in VCFS. We hope that the results of this study will help us understand VCFS and provide better care to these patients in the future.

For more information about this study, please contact the study coordinator, Erica Tworog-Dube, M.S., C.G.C., at 617-525-4490 or etworog-dube@partners.

Posting to websites, listservs, and meetings:

NSGC newsletter and listserv

VCFS family member listserv

Chromosome 22 Central website

VCFS Educational Foundation website

Genetics meetings – Annual basis

Harvard Partners Center for Genetics and Genomics website