

Note: The Cystinosis Research Network is providing the following information as a service to the community. CRN is not associated in any way with Mitomed Diagnostic Laboratory. Individuals wishing to obtain any of these testing services should first consult with their doctor as to the necessity and relevance to their particular situation. In addition, individuals bear the responsibility of determining if testing is covered by their individual insurance. CRN does not have resources available to provide any financial assistance in this regard.

Mitomed Diagnostic Laboratory at University of California, Irvine is now offering genetic testing for CTNS, the gene that causes cystinosis. The lab examines the gene for mutations, which are deleterious changes that affect how the gene works. Genetic testing has a detection rate of 85-90 percent for people with clinical symptoms. Genetic testing may be helpful in establishing carrier status in unaffected individuals. If possible, it is best to first test the person who has cystinosis. Once the mutations that run in a family are known, it makes carrier testing for other family members easier and more accurate. If the familial mutations are not known, then a negative test result can significantly decrease a person's carrier risk from that in the general population. Genetic testing may also be helpful in prenatal diagnosis and preimplantation genetics (PGD). Prenatal diagnosis can be arranged by our laboratory through GeneDx.

Please contact Mitomed Diagnostic Laboratory at 949-824-1886 if you have questions about genetic testing for cystinosis. You can also find more information, including requisitions and sample requirements, at our website: <http://mitomed.bio.uci.edu>.