

## **New Cystinosis Study from McGill University Montreal Children's Hospital (CTNS Nonsense Mutation Screen - CyNoMuS)**

The purpose of this message is to invite cystinosis patients to join our upcoming study concerning a specific type of genetic alteration called a “Nonsense Mutation”. Although cystinosis is caused by many different disruptions of the CTNS gene, we are particularly interested in “nonsense mutations” which trick the cell into stopping production of Cystinosin protein. Several pharmaceutical companies are working hard to develop medications related to a well-known antibiotic (gentamicin) that permits the cell to disregard nonsense mutations.

Nonsense Mutations are estimated to account for about 10-15% of cases in other genetic diseases, but the prevalence in cystinosis is unknown and there is some evidence that CTNS Nonsense Mutations are clustered in certain regions. Looking forward, cystinosis patients may wish to know whether or not they carry a nonsense mutation. We would like to survey the cystinosis community and characterize the prevalence and distribution of Nonsense Mutations among cystinosis patients in North America and Europe. This would simply involve (prepaid) mailing a sample of saliva to our research group at McGill University Children's Hospital in Montreal. We would analyze the sequence of your CTNS gene and let you know for future reference whether or not you carry a nonsense mutation.

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