

# Niemann-Pick Disease Type C Family Survey Updates

KAREN QUANDT, RN, MN writes .....

## Neurodegenerative Disease in Family Members of Patients with Niemann-Pick Type C

### Final Progress Report – Lay Summary

Thank you to all the families that participated in my recent research survey. Thanks also go to the National Niemann-Pick Disease Foundation, The Niemann-Pick Disease Group of the United Kingdom and to the Ara Parseghian Medical Research Foundation, all of whom assisted with this survey. Here is a summary of the results:

57 families answered the survey

29 families had a family member with another neurodegenerative disease- 50.9%

28 families did not have a family member with another neurodegenerative disease – 49.1%

Summary of other neurodegenerative diseases that the families have in their health history:

17 cases of Alzheimer disease- in 14 different families

8 cases of Amyotrophic lateral sclerosis-(Lou Gehrig's disease) - In 6 different families

8 cases of Parkinson disease

4 cases of Multiple Sclerosis - in 3 different families

2 cases of Huntington Disease- in the same family

1 case of Multiple System Atrophy

Several families had 2 different neurodegenerative diseases in their family health history.

I discussed the findings of my recent Niemann-Pick type C (NP-C) family health history survey of neurodegenerative diseases with Dr. Ellen Sidransky and Joie Davis, nurse practitioner, both of whom work at the National Institute of Health. As you know, my family history survey was done anonymously and I am unable to re-contact participants directly. Dr. Sidransky and Joie are interested in investigating the possible association of Parkinson disease, ALS (Lou Gehrig Disease) and Alzheimer disease (diagnosed before the age of 65), present in the families who also have a relative with NP-C. Specifically, the individuals of interest would be relatives of a patient with a known NP-C mutation and our aim would be to see if the relative with a neurodegenerative disease also carries a NP-C mutation. The study would involve a cheek brush test of the relative. If your family has a member with Alzheimer disease, Parkinson disease or Lou Gehrig's disease and meets the above mentioned criteria, and may be interested in this study or have additional questions, please contact Joie Davis at [jdavis@mail.nih.gov](mailto:jdavis@mail.nih.gov), or phone 301-435-6691.

Thank you,  
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