

NNPDF COLLABORATIVE STUDY – KAREN QUANDT, RN, MN

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. Today I closed the survey titled, “Neurodegenerative Disease in Family Members of Patients with Niemann-Pick Type C”.

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 Institutes of Health.

Dr. Sidransky and Joie are interested in investigating the possible association of Parkinson’s disease, ALS (Lou Gehrig Disease) and Alzheimer’s disease (diagnosed before the age of 65), present in the families who also have a relative with NP-C.

Thank you,
Karen Quandt, RN,MN