Lay Summary BRAIN UK Ref: 22/007 Alpha-synuclein pathology in metachromatic leukodystrophy Dr Daniel Erskine, Newcastle University

Metachromatic leukodystrophy (MLD) is a rare genetic disease that normally affects children and leads to damage to the brain and premature death. In contrast, Parkinson's disease (PD) is a relatively common disease that affects older people and is thought to be caused by a protein called alpha-synuclein building up in brain cells and killing them, but it is not known why this occurs. MLD is caused by a problem with a gene that has also been linked to PD, and a recent study suggested alpha-synuclein also builds up in the brains of children with MLD. We want to examine whether the alpha-synuclein built up in MLD brains is similar to that in PD. This is important as the cause of MLD is already known, so if alphasynuclein is the same as in PD, it would suggest that the changes to the brain that cause MLD may also cause PD. This is important as it would help us understand what causes PD and could identify new ways to treat both PD and MLD.

Abbreviations:

MLD - Metachromatic leukodystrophy

PD - Parkinson's disease