Repository Spotlight – A whole-genome sequence study identifies genetic risk factors for Neuromyelitis Optica

ACP Repository samples include DNA samples from people with MS and other demyelinating diseases, including neuromyelitis optica (NMO). As discussed in our March 2018 and October 2018 newsletters, these samples have helped researchers identify the genes associated with MS. NMO is a rare condition that affects the optic nerve and spinal cord, once considered to be a clinical subclass of MS but is now known to be a distinct disease. NMO is similar to MS in that both are autoimmune diseases where the myelin sheath around nerves is damaged. The difference lies in the target of the immune attack. Researchers from Biogen, the Broad institute, Massachusetts General Hospital, Harvard Medical School and the University of Texas Southwestern (UTSW) recently published groundbreaking work on the genetics of NMO using samples from the ACP Repository and from UTSW. Results from this collaboration reveal two specific genes associated with NMO, which are not linked to MS. Furthermore, these data show NMO is more genetically similar to systemic lupus erythematosus (SLE) than MS. Research on the genetics of autoimmune diseases is important from the perspective of understanding the pathophysiology of disease as well as identifying targets for new drugs. The identification of common autoimmune mechanisms can result in the discovery of drugs that may be useful in different disorders. This is just one of more than one hundred studies using the ACP Repository as a resource to advance and accelerate research into demyelinating diseases.