



SPOTLIGHT ON RESEARCH: Opening New Doors to Understanding Multiple Sclerosis

Researchers at the University of California, San Francisco (UCSF) have identified the gene for a disease that may hold clues in understanding the severely debilitating auto immune disorder, Multiple Sclerosis (MS). The disease, Autosomal Dominant Leukodystrophy (ADLD) is thought to be extremely rare. Like MS, which affects nearly 1 in 1,000 Americans, ADLD affects the myelin, which protects the nerve fibers of the Central Nervous System (CNS). MS results primarily from the body's own immune system attacking the myelin sheath, in contrast, ADLD is not an autoimmune disorder.

Dr. Ying-Hui Fu in the Department of Neurology, led her team in localizing the gene to chromosome 5 and positionally cloning the disease gene (lamin B1) showing the disease to result from a duplication and one extra copy of the lamin B1 gene in patients. Lamin B1 is an important protein in the nuclear envelope and is important for many biological functions including nucleocytoplasmic transport and transcriptional regulation.

The same protein-Lamin B1 that is produced in excess in ADLD patients is also one of the proteins that are found in the plaques found in the brains of patients with MS. While auto antibodies to Lamin B1 have not been identified in MS, previous studies have shown that Lamin B1 is also a protein against which auto antibodies are formed in other auto immune disorders. The researchers are further studying these intriguing links to MS by making a mouse which also has an extra copy of this gene. They hope that this mouse model may shed further light not only on ADLD, but also on the more complicated mechanisms that underlie MS.