IBMPFD

Motor Neuron Genotype-Phenotype Correlation Study

Title	Motor neuron genotype-phenotype correlation
Short name	IBMPFD
Principal investigator	Michael Benatar, MD, PhD
Site-PI	-
Summary	IBMPFD is a rare disorder in which affected individuals may have muscle weakness, Paget's disease of bone, and/or dementia. Muscle weakness in this disorder has typically been attributed to a disease of muscle known as inclusion body myopathy (IBM). The only identified genetic cause of IBMPFD is mutation of the VCP (valosin-containing protein) gene, although mutations in other genes are believed to cause IBMPFD in families without VCP mutations. We have recently found that mutations in VCP may also cause familial ALS and that ALS sometimes occurs in families with IBMPFD. Based on these observations, the goal of this study is to further explore the extent to which muscle weakness in people with IBMPFD might be due to motor neuron degeneration.
Eligibility Criteria	Individual affected with muscle weakness, Paget's disease or dementia in a family known to have IBMPFD, irrespective of whether family is known to harbor a mutation in the VCP gene
Funding Agency	ALS Association
Collaborators	Paul Taylor, MD, PhD (St. Jude Children's Research Hospital) Bryan Traynor, MD (National Institutes of Health) Conrad Chris Weihl, MD, PhD (Washington University)
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Link(s)	http://www.alsa.org/news/archive/2011-new-research-grants.html http://www.alsa.org/news/archive/vcp-mutations-as-a-cause-of.html http://alsn.mda.org/news/vcp-gene-implicated-familial-als-ibm
Presentations/ Publications	Exome sequencing reveals VCP mutations as a cause of familial ALS. Neuron 2010 68(5):857-864