

This week in techniques

| Approach | Summary | Licensing status | Publication and contact information |
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| Markers | | | |
| Risk assessment of hereditary sensory neuropathy associated with dementia and hearing loss | Genetic association studies identified DNA (cytosine-5)-methyltransferase 1 (DNMT1) mutations that could help predict risk of hereditary sensory neuropathy associated with dementia and hearing loss. In a genomewide linkage analysis and exome sequencing study, mutations that caused premature degradation of DNMT1 were identified in four affected families but were absent in healthy subjects. In cell culture, expression of the mutant protein disrupted protein folding, enzymatic activity and heterochromatin binding compared with expression of the wild-type protein. Next steps could include confirming the association in additional patients. | Findings unpatented; patenting or licensing being considered by the Mayo Clinic | Klein, C. <i>et al. Nat. Genet.</i> ; published online May 1, 2011; doi:10.1038/ng.830 Contact: Christopher J. Klein, Mayo Clinic, Rochester, Minn. e-mail: klein.christopher@mayo.edu |
| | <i>SciBX</i> 4(18); doi:10.1038/scibx.2011.529 Published online May 5, 2011 | | |