This week in therapeutics

Indication	Target/marker/pathway	Summary	Licensing status	Publication and contact information
Ophthalmic disease				
Blindness	<i>Nicotinamide nucleotide adenylyltransferase 1 (NMNAT1)</i>	<p>Studies with human tissues identified multiple <i>NMNAT1</i> mutations that could help diagnose Leber's congenital amaurosis (LCA). Previous <i>Drosophila</i> studies have shown that <i>Nmnat1</i> deficiency leads to degeneration of photoreceptor cells. In four independent studies, genomics analyses identified associations between multiple <i>NMNAT1</i> SNPs and LCA in patients who did not harbor mutations in known LCA-associated genes. In fibroblasts and red blood cells from the patients with LCA, functional analyses showed that the <i>NMNAT1</i> SNPs restricted the protein's enzymatic activity. Ongoing work includes further investigations of the effects of the SNPs on NMNAT1 function and the role of the mutant proteins in LCA.</p> <p>SciBX 5(31); doi:10.1038/scibx.2012.821 Published online Aug. 9, 2012</p>	<p>Patent and licensing status unavailable for findings in first and second studies</p> <p>Findings in third study unpatented and available for partnering</p> <p>For findings in fourth study, patent application filed in China by BGI; available for licensing and partnering</p>	<p>Koenekoop, R.K. <i>et al. Nat. Genet.</i>; published online July 29, 2012; doi:10.1038/ng.2356 Contact: Rui Chen, Baylor College of Medicine, Houston, Texas e-mail: ruichen@bcm.edu</p> <p>Perrault, I. <i>et al. Nat. Genet.</i>; published online July 29, 2012; doi:10.1038/ng.2357 Contact: Josseline Kaplan, Institut National de la Santé et de la Recherche Médicale (INSERM), Paris, France e-mail: josseline.kaplan@inserm.fr Contact: Jean-Michel Rozet, same affiliation as above e-mail: jean-michel.rozet@inserm.fr</p> <p>Falk, M.J. <i>et al. Nat. Genet.</i>; published online July 29, 2012; doi:10.1038/ng.2361 Contact: Eric A. Pierce, Massachusetts Eye and Ear Infirmary, Boston, Mass. e-mail: eric_pierce@meei.harvard.edu</p> <p>Chiang, P.-W. <i>et al. Nat. Genet.</i>; published online July 29, 2012; doi:10.1038/ng.2370 Contact: Ming Qi, Zhejiang University School of Medicine, Hangzhou, China e-mail: ming_qi@urmc.rochester.edu</p>