SciBX
Science-Business eXchange

THE DISTILLERY

This week in therapeutics

Indication	Target/marker/pathway	Summary	Licensing status	Publication and contact information
Neurology				
Neurology	Adenosine monophosphate deaminase 2 (AMPD2)	*In vitro* and genetic studies identified a subset of patients with pontocerebellar hypoplasia (PCH) who could benefit from treatment with purine precursors. A genetic screen of 30 patients with early onset PCH and their families identified a subset in the cohort with mutations in *AMPD2*, an enzyme necessary for biosynthesis of the purine nucleotide guanine. In patient neural progenitor cells, a feedback loop regulating purine biosynthesis was disrupted and treatment with purine precursors rescued protein translation and cell viability. Next steps could include developing animal models of PCH based on mutations in *Ampd2*. *SciBX* 6(34); doi:10.1038/scibx.2013.932 Published online Sept. 5, 2013	Patent and licensing status unavailable	Akizu, N. *et al. Cell*; published online Aug. 1, 2013; doi:10.1016/j.cell.2013.07.005 **Contact:** Joseph G. Gleeson, University of California, San Diego, La Jolla, Calif. e-mail: jogleeson@ucsd.edu