This week in therapeutics

Indication	Target/marker/ pathway	Summary	Licensing status	Publication and contact information
Renal disease				
Renal disease	Centrosomal protein 290 kDa (CEP290)	<p>Mouse and cell culture studies suggest hedgehog pathway agonists could help treat Joubert syndrome, a genetic cerebellar disorder that causes renal failure and other symptoms. A mouse model of Joubert syndrome was generated from embryonic stem cells carrying a disruption in <i>Cep290</i>, the most common mutation in Joubert syndrome. The mice showed downregulation of the hedgehog pathway and a ciliopathy phenotype similar to the human syndrome, including retinal degeneration, cerebellar abnormalities and progressive polycystic kidney disease. In <i>ex vivo</i> cultures of collecting duct cells from the model and renal epithelial cells from patients with Joubert syndrome, a hedgehog pathway agonist improved spheroid formation and increased numbers of cilia compared with no treatment. Ongoing work includes identifying less toxic agonists by screening small compound libraries in cell-based assays.</p> <p>SciBX 7(28); doi:10.1038/scibx.2014.837 Published online July 24, 2014</p>	Unpatented; licensing status not applicable	<p>Hynes, A.M. <i>et al. Proc. Natl. Acad. Sci. USA</i>; published online June 19, 2014; doi:10.1073/pnas.1322373111</p> <p>Contact: John A. Sayer, Newcastle University, Newcastle upon Tyne, U.K. e-mail: john.sayer@ncl.ac.uk</p>