



## This week in techniques

Summary	Licensing status	Publication and contact information
Mice carrying a mutation in <i>phosphodiesterase</i> 6C ( <i>Pde6c</i> ) could serve as a model of human achromatopsia, a congenital retinal dystrophy characterized by low visual acuity, photophobia, involuntary eye movements and lack of color discrimination. Analysis of 24 achromatopsia patients showed that all of them carried mutations in <i>PDE6C</i> , which were not found in healthy controls. Mice bearing a <i>Pde6c</i> -inactivating mutation exhibited achromatopsia-like symptoms. Next steps could include using this model to study the role of PDE6C mutations in achromatopsia and to identify possible therapeutic approaches.  SciBX 2(43); doi:10.1038/scibx.2009.1616	Patent and licensing status unavailable	Chang, B. et al. Proc. Natl. Acad. Sci. USA; published online Oct. 26, 2009; doi:10.1073/pnas.0907720106 Contact: Bernd Wissinger, University Clinics Tubingen, Tubingen, Germany e-mail: wissinger@uni-tuebingen.de
	Mice carrying a mutation in <i>phosphodiesterase</i> 6C ( <i>Pde6c</i> ) could serve as a model of human achromatopsia, a congenital retinal dystrophy characterized by low visual acuity, photophobia, involuntary eye movements and lack of color discrimination. Analysis of 24 achromatopsia patients showed that all of them carried mutations in <i>PDE6C</i> , which were not found in healthy controls. Mice bearing a <i>Pde6c</i> -inactivating mutation exhibited achromatopsia-like symptoms. Next steps could include using this model to study the role of PDE6C mutations in achromatopsia and to identify possible therapeutic approaches.	Mice carrying a mutation in <i>phosphodiesterase</i> 6C ( <i>Pde6c</i> ) could serve as a model of human achromatopsia, a congenital retinal dystrophy characterized by low visual acuity, photophobia, involuntary eye movements and lack of color discrimination. Analysis of 24 achromatopsia patients showed that all of them carried mutations in <i>PDE6C</i> , which were not found in healthy controls. Mice bearing a <i>Pde6c</i> -inactivating mutation exhibited achromatopsia-like symptoms. Next steps could include using this model to study the role of PDE6C mutations in achromatopsia and to identify possible therapeutic approaches.  SciBX 2(43); doi:10.1038/scibx.2009.1616