



## National Human Genome Research Institute (NHGRI)

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#### Pendrin Knockout Mouse

NHGRI invention number:  
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#### **Key Words**

Pendred Syndrome, Pendrin,  
PDS Gene, COPD, Asthma,  
Knockout Mice

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#### **Summary**

Mutations in the human Pendred syndrome (PDS) gene cause the deafness and goiter disorder Pendred syndrome (PS). PDS encodes pendrin - a sodium-independent anion transporter that has been shown to play an important role in the regulation of blood pressure by the kidneys. In addition, pendrin has been implicated in the pathogenesis of asthma and chronic obstructive pulmonary disease (COPD). NHGRI investigators generated a pendrin knockout mouse that is deaf and shows vestibular dysfunction. Thus, the mouse can serve as a model for auditory dysfunction and deafness in PS, as well as for asthma and COPD.

#### **Potential Commercial Applications**

This mouse could be used to screen for and/or test candidate therapeutics targeting either the PDS gene or the pendrin protein. The tested compounds could be useful for treating deafness, goiter, and hypertension, as well as lung diseases characterized by inflammation, such as asthma and COPD. Either *in vivo* experiments or *in vitro* studies with isolated cells could be carried out.

#### **Related Article**

Everett et al., *Targeted Disruption of Mouse Pds Provides Insight About the Inner-Ear Defects Encountered in Pendred Syndrome*, 10 Human Mol. Genetics 153 (2001).  
<http://hmq.oxfordjournals.org/content/10/2/153.full.pdf>