Nonsense suppression drug restores function in a mouse model of aniridia

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Congenital aniridia is a progressive disease that is associated with improper development of eye structures as well as abnormalities in the brain and pancreas. A variety of nonsense mutations in the *PAX6* gene are linked with aniridia; however, despite understanding the genetic basis of the disease, few treatment and prevention strategies are available.

In this issue of the *Journal of Clinical Investigation*, Cheryl Gregory-Evans and colleagues at the University of British Columbia evaluated a small molecule nonsense suppression strategy for relief of aniridia-associated defects in a mouse model of the disease. The authors developed a formulation of the nonsense suppression drug ataluren that could be given topically to postnatal aniridia mice. Administration of their ataluren-based formulation inhibited disease progression, reversed eye deformations, and restored eye function in aniridia mice.

In an accompanying commentary, José-Alain Sahel and Katia Marazova of the Institut de la Vision suggest that ataluren administration should be further explored as a therapeutic option for treatment of congenital eye defects associated with nonsense mutations.

More information: Postnatal manipulation of Pax6 dosage reverses congenital tissue malformation defects, *J Clin Invest*. DOI: 10.1172/JCI70462
Toward postnatal reversal of ocular congenital malformations, *J Clin Invest*. 2014;124(1):81–84. DOI: 10.1172/JCI73560