Researchers define role of CEP290 in maintaining ciliary function

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A new study in the September 6 issue of the *Journal of Cell Biology* helps define the role of an important ciliary protein, CEP290. The results could be applied toward targeted gene therapy in cilia-related diseases.

Mutations in human CEP290 cause cilia-related disorders that range in severity from isolated blindness to perinatal death. CEP290 mutations are known to cause Meckel syndrome, Joubert syndrome, and NPHP—the most common syndromic form of cystic kidney disease in childhood—among others.

Although the exact role of CEP290 has been unclear, a team of researchers from the University of Massachusetts Medical School and Yale University now demonstrate that CEP290 is an integral component of the ciliary "gate" that bridges the transition zone between the cilia and cytoplasm. The protein plays an important role in maintaining the structural integrity of this gate, and thus has a crucial role in maintaining ciliary function.

More information: Craige, B., et al. 2010. *J. Cell Biol.* doi:10.1083/jcb.201006105
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