

# Ciliopathies lie behind many human diseases

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In recent years, cilia, microscopic, tentacle-like extensions from biological cells, have risen from relative obscurity and are now considered important to the understanding of many human afflictions. In a December *BioScience* article, George B. Witman, of the University of Massachusetts Medical School, and Jason M. Brown, of Salem State University, describe recent discoveries involving cilia-related diseases (called "ciliopathies") and highlight "model" species that could be useful for systematic study of ciliopathies.

Cilia perform a broad range of functions, including a starring role in cell signalling. Motile ones wiggle and so move fluids within the body, including [cerebrospinal fluid](#) in the brain. In humans, [cilia](#) are found on almost every cell in the body. Because of this, ciliopathies often make themselves known as syndromes with widely varying effects on a number of tissue types. For instance, the ciliopathy Jeune asphyxiating thoracic dystrophy involves the development of abnormally short ribs, accompanied by short limbs and, occasionally, the development of extra digits.

In primary ciliary dyskinesia, [motile cilia](#) are dysfunctional and fail to beat. This can lead to bronchitis resulting from the failure to clear mucus from the sufferer's airways. Male patients with primary ciliary dyskinesia are infertile because of impaired motility of the sperm's flagellum (flagella and cilia are structurally similar).

The article's authors point to a number of other human diseases in which cilia may play a role; for example, some cancers and [neurological](#)

[diseases](#) may be related to ciliopathies. Because of the limitations placed on research involving humans, the authors propose the use of model species ranging from the green alga *Chlamydomonas* to the house mouse to further study the role of cilia. They write, "We can anticipate that new and improved techniques will open new avenues for gaining further insight into these immensely important and ever more fascinating cell organelles."

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