

# Understanding a molecular motor responsible for human development

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Another mystery of the human body has been solved by scientists who have identified how a molecular motor essential for human development works. They have also pinpointed why mutations in genes linked to this motor can lead to a range of human diseases.

Researchers at the University of Bristol have defined the composition of the human version of a [molecular motor](#), called 'cytoplasmic dynein-2', that is essential for normal [human development](#). Dynein 2 directs molecules into cilia as well as controlling their movement along cilia.

Cilia are slender protrusions that act as antennae on nearly all human cells. They are important in sensing signals that direct cell function.

Dysfunctional cilia are known to underlie a number of often chronically disabling and sometimes life-threatening genetic conditions. They affect multiple systems, causing blindness, deafness, chronic respiratory infections, [kidney disease](#), heart disease, infertility, obesity and diabetes.

These human diseases are collectively known as ciliopathies, many of which, including Jeune Syndrome, are linked to childhood development.

In the UK, one in every 100,000 babies is born with Jeune Syndrome - a [rare genetic disorder](#) that affects the way a child's cartilage and bones develop.

The new research, funded by the Medical Research Council and

published today [09 September] in the *Journal of Cell Science*, has explained exactly how the human cytoplasmic dynein-2 motor works for the first time.

This new knowledge could help with diagnosis and, in the long-term, scientists hope that they might be able to alter the function of the defective motor for therapeutic benefit.

Professor David Stephens, from the School of Biochemistry at the University of Bristol, led the research.

He said: "The discovery of new components of the motor gives us a great opportunity to work towards understanding how defects in dynein-2 lead to disease."

Building on work done in simple model organisms such as green algae, researchers have also shown that two genes associated with Jeune Syndrome (WDR34 and WDR60) are essential parts of the [human](#) form of this motor.

Both genes encode proteins required to form a functional dynein-2 motor, explaining why a mutation in either one leads to a ciliopathy.

The work also identified a novel component of dynein-2 (TCTEX1D2), providing another candidate gene that could be mutated in cases of Jeune Syndrome.

**More information:** 'Subunit composition of the human cytoplasmic dynein-2 complex' by David Asante, Nicola L. Stevenson and David J. Stephens in the *Journal of Cell Science*.

Provided by University of Bristol

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