PRESS RELEASE

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GENETICISTS HAVE ADVANCED IN UNDERSTANDING THE CAUSES OF HUMAN GROWTH DISORDERS

The surface of almost every human cell is covered by tiny hair-like structures - primary cilia. Their dysfunction leads to many disorders, including impaired growth. Pavel Krejčí's team from the Institute of Animal Physiology and Genetics CAS and Masaryk University of Brno, in collaboration with their colleagues from the University of California in Los Angeles has taken a crucial step towards understanding the origins of serious growth disorders. This paves the way to finding possible treatment.

"We have described new mutations in the GRK2 gene that lead to primary cilia dysfunction and ultimately, to fatal growth impairment," says Pavel Krejčí, describing the results of his work. Mutations in GRK2 kinase cause Jeune syndrome, a genetic bone growth disorder that is currently untreatable. The importance of this research is emphasised by the fact that Pavel Krejčí's work was featured on the cover of the November issue of the prestigious scientific journal *EMBO Molecular Medicine*.

> *"Understanding these mechanisms is essential for the development of treatment methods as well as for uncovering the genetic burden in families at risk."*

For many years, Pavel Krejčí's team has been focusing on the study of genetic syndromes caused by dysfunctional cilia (so-called ciliopathy). Ciliopathies are serious, often fatal diseases, with a very limited range of treatment options.

Primary cilia have originally been involved in cell motility (flagella, pili), but throughout evolution they have specialised in detecting and transducing extracellular cues, similarly to the antenna on a radio. Mutations in genes involved in the formation of cilia or regulation of their specific signalling functions lead to disruptions in intercellular communication (faulty antenna). Ciliopathies comprise a diverse group of disorders affecting a variety of organs, including bone growth.

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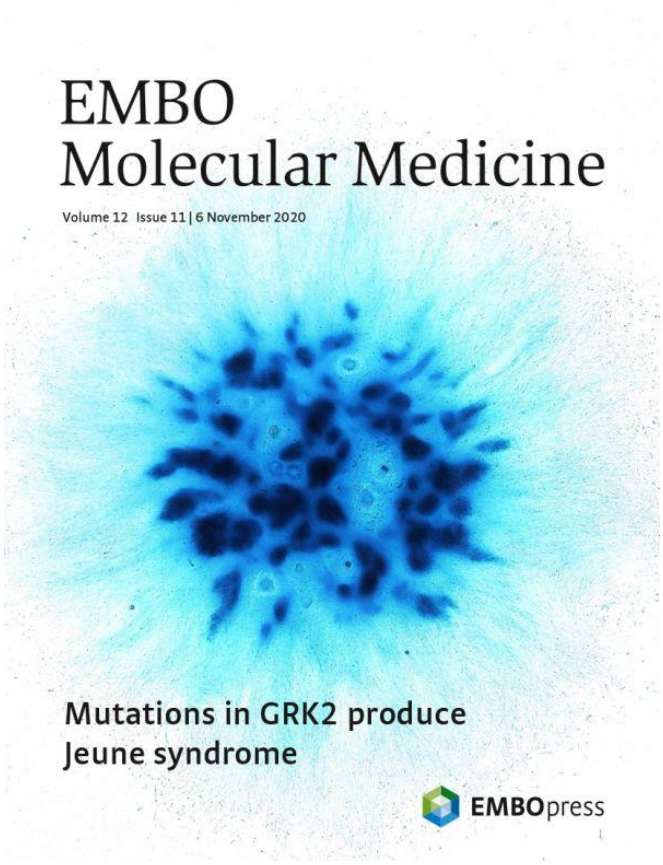
"Studying the pathology of ciliopathies helps to clarify the basic processes of skeletal development and growth. Understanding these mechanisms is essential for the development of treatment methods as well as for uncovering the genetic burden in families at risk," explains Pavel Krejčí.

Jeune syndrome is a genetic bone growth disorder that is characterised by a narrow chest, short ribs, shortened long bones and the occasional presence of polydactyly (extra fingers). The disease occurs in 1 in 100,000-130,000 people and is often fatal, or the patients live only a few years with severe respiratory distress. Jeune syndrome is currently untreatable.



The cover page of the prestigious EMBO Molecular Medicine journal with an image from Pavel Krejčí's publication.

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Link to the publication: https://www.embopress.org/journal/17574684