Researchers identify mechanism for Frank-Ter Haar syndrome

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An international team of investigators at Sanford-Burnham Medical Research Institute (Sanford-Burnham, formerly Burnham Institute for Medical Research), Nijmegen Centre for Molecular Life Sciences and other organizations have discovered that TKS4, a protein implicated in cancer metastasis, also plays a significant role in Frank-Ter Haar syndrome (FTHS), a rare fatal disorder. The research was published on February 12 in the *American Journal of Human Genetics*.

Children with FTHS suffer from skeletal, cardiovascular and eye abnormalities and usually die in infancy or early childhood. Hans von Bokhoven, Ph.D., at Nijmegen, with assistance from clinical colleagues in several different countries, studied 12 families who had been affected by FTHS. The team mapped the condition in 7 of these families to a mutated SH3PXD2B gene, which normally contains the code to create the TKS4 protein. Dr. von Bokhoven then began collaborating with Sara A. Courtneidge, Ph.D., at Sanford-Burnham, who had been studying the relationship between TKS4 and invadopodia, cellular protrusions that facilitate cancer cell migration and invasion. The Courtneidge group, together with Sanford-Burnham researchers José Luis Millán, Ph.D., and Pilar Ruiz-Lozano, Ph.D., and researchers at University of California, San Diego, determined that mice without TKS4 showed virtually identical traits to FTHS patients, confirming the results of the gene mapping. Interestingly, one FTHS family with a normal SH3PXD2B gene was nevertheless deficient in TKS4, indicating a consistent mechanism for the disease.

"This research illustrates the importance of collaboration in the scientific discovery process, and how the study of one disease, in this case cancer, can have a profound impact on the understanding on another." said Dr. Courtneidge, "In the future, we hope to use our mouse model to study the disease in more depth, as well as to determine whether other genes involved in invadopodia formation are also associated with FTHS."

Provided by Sanford-Burnham Medical Research Institute

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