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STORMORKEN SYNDROME DISEASE STIM1 PROTEIN STUDIED BY HIGH RESOLUTION NMR

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STIM1 serves as calcium sensor protein in the endoplasmic reticulum of the cell which extends into the cytosol and oligomerises upon calcium store depletion. The cytosolic part of STIM1 consists of one long and two short coiled coil domains directly involved in homo-oligomerization leading to spatial elongation of the STIM1 protein and activation of the Orai calcium channel. The Stormorken syndrome associated with a single point mutation (R304W) within this region of STIM1 results in permanent activation of Orai channel. Using high resolution solution state NMR we have found a helix elongation within the short coiled coil domain of the mutant close to the mutation position with respect to the wild type STIM1. These findings corroborate the increased propensity of this domain to form homomers destabilizing the resting state of STIM1, which leads to the increased channel activation.

Biography

Petr Rathner has completed his Double Degree Mgr/MSc in 2013 from Johannes Kepler University of Linz (Austria) and University of South Bohemia (Czech Republic). Currently, he is a PhD student in the group of Professor Norbert Müller Nanocell research (JKU Linz, Austria) focusing on biomolecular nuclear magnetic resonance spectroscopy.

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