

# ***COLLOQUIA IN CELLULAR SIGNALLING***

Venue: Medical University Vienna, Center for Physiology and Pharmacology,  
Institute of Pharmacology, Waehringerstrasse 13a, 1090 Vienna, "**Leseraum**".

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**Friday**      **25.10.2013 11:00 s.t.**      **Geoffrey Pitt** (host: A. Koschak)

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***"New roles for L-type Ca<sub>v</sub>1.2 Ca<sup>2+</sup> channels in development and non-excitable cells"***

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Abstract:

The identification of a gain-of-function mutation in *CACNA1C* as the cause of Timothy Syndrome (TS), a rare disorder characterized by cardiac arrhythmias and syndactyly, highlighted unexpected roles for the L-type voltage-gated Ca<sup>2+</sup> channel Ca<sub>v</sub>1.2 in non-excitable cells. How abnormal Ca<sup>2+</sup> influx through Ca<sub>v</sub>1.2 underlies the multiple phenotypes such as the accompanying syndactyly or craniofacial abnormalities in the majority of affected individuals is not readily explained by established Ca<sub>v</sub>1.2 roles nor known pattern of Ca<sub>v</sub>1.2 expression. We have uncovered new tissues in which Ca<sub>v</sub>1.2 is expressed during development and have defined new roles for Ca<sub>v</sub>1.2 during development.