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Prof. Dr. Pedro Terradellas Brugada, co-promotor

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PhD in Medical Sciences
2014-2015

INVITATION to the Public defence of

Véronique BISSAY

To obtain the academic degree of '**DOCTOR IN MEDICAL SCIENCES**'

Clinical spectrum of SCN4A-associated skeletal muscle channelopathies**Thursday 18 June 2015**

Auditorium **Brouwer**, **17:00**

Faculty of Medicine and Pharmacy, Laarbeeklaan 103, 1090 Brussel

How to reach the campus Jette:

<http://www.vub.ac.be/english/infoabout/campuses>



Vrije Universiteit Brussel

Summary of the dissertation

SCN4A-associated non-dystrophic myotonias are rare diseases. Only in the last few years, prospective studies evaluated the sensitivity of symptoms and signs of myotonia in large cohorts of patients, leading to the conclusion that the phenotypes are frequently much more complex than previously thought. This clinical heterogeneity can also be applied to our patient cohort. Some of the observed phenotypes and related genotypes differ from what is commonly described in literature. Our findings contribute to visualize the genotypic and phenotypic characteristics and distribution of muscular sodium channelopathies in the region of Brussels.

Further, the diagnosis of these channelopathies remains challenging. We highlighted some pitfalls in the diagnosis, based on different observations in our patient population.

Finally, we observed a previously unreported overlap between Brugada syndrome and SCN4A-associated muscular channelopathies. The possible association between these two rare diseases, is supported by several publications, demonstrating the expression of skeletal muscle voltage-gated sodium channels in the cardiac muscle. The presence of a SCN4A variant is probably not solely responsible for an arrhythmic event, but would rather act as a modifier gene in Brugada syndrome.

Curriculum Vitae

Véronique Bissay was born on the 7th of January 1969 in Willebroek. In 1993, she obtained her medical degree at the University of Brussels, VUB, graduated with high distinction. She started the training in neurology (Prof Guy Ebinger) with special interest for multiple sclerosis, neuromuscular disorders and clinical neurophysiology. She obtained 2 additional diplomas in France, "Diplôme Inter Universitaire (DIU) de Neurophysiologie Clinique" in 2007 and "DIU des Neuropathies Périphériques" in 2011. At present, she works at the Universitair Ziekenhuis Brussel as clinical neurophysiologist and runs, as a neurologist, a private practice in Asse on part-time base.