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LETTER TO THE EDITOR

# The IVS8-2A>G (c.913-2A>G) mutation and the PAH deficiency populations of Central Europe

**Urh Groselj · Mojca Zerjav Tansek · Katarina Trebusak Podkrajsek · Tadej Battelino**

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The recent report by Sterl et al provides a very interesting account on the genetic characteristics of the Austrian PAH deficiency population, not just rounding the genetic data on the Central European populations but also positioning Austria as their very integral part, with evidently strong links to Eastern Europe (Sterl et al 2012). However, the IVS8-2A>G (c.913-2A>G) mutation—one of the presumably five novel mutations in the Austrian PAH deficiency population where it is found in 3/294 (1 %) of the alleles—has been previously reported in the Croat population (its frequency is not shown) (Karacic et al 2009). It is also found in the Slovene population where it represents 4/214 (2 %) of the alleles (Groselj et al 2012).

The simultaneous presence of the IVS8-2A>G (c.913-2A>G) mutation in the three populations is not surprising considering the close geographical proximity and also the common historical roots of the three populations in the Austro-Hungarian Empire, in what has been traditionally known as *Mitteleuropa*.

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U. Groselj · M. Z. Tansek (✉) · T. Battelino
Department of Pediatric Endocrinology, Diabetes and Metabolic Diseases, University Children's Hospital Ljubljana, Ljubljana, Slovenia
e-mail: mojca.zerjav-tansek@mf.uni-lj.si

U. Groselj
e-mail: urh.groselj@kclj.si

K. T. Podkrajsek
Center for Medical Genetics, University Children's Hospital Ljubljana, Ljubljana, Slovenia

T. Battelino
Department of Pediatrics, Faculty of Medicine, University of Ljubljana, Ljubljana, Slovenia

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