



Warszawski Uniwersytet  
Medyczny

**Laboratorium Zakładu  
Genetyki Medycznej**

*ul. Pawińskiego 3c,  
02-106 Warszawa,  
tel.: 022/572 06 95, fax: 022/572 06 95*

Nr. ewid. KIDL 2831



Uniwersytet Warszawski

**Instytut Genetyki i  
Biotechnologii**

*ul. Pawińskiego 5A  
02-106 Warszawa  
tel. (22) 592-22-39, fax. (22) 592 22 44*

Warszawa 31.03.2014

## Result of DNA analysis

The analysis was performed in the Department of Medical Genetics, Medical University of Warsaw in scientific collaboration with the Institute of Genetics and Biotechnology, University of Warsaw. Person for correspondence: Katarzyna Tońska, PhD, DSc, tel. +48 22 592 22 39, +48 22 592 22 41, e-mail: kaska@igib.uw.edu.pl

**Name and surname of the patient:** Tomasz Koniew

**PESEL:** ██████████

**Date of birth:** 25.01.1979

**Address:** ul. ██████████ 41-800 Zabrze

**Sex:** male

**Sample number:** M271

**Physician in charge:** Marta Lipowska, M.D., PhD, Biruta Kierdaszuk, M.D.

**Clinical diagnosis:** Mitochondrial myopathy

**Type of biological material:** peripheral blood, muscle sample

**Analysis performed:**

Mitochondrial DNA analysis:

RFLP test for m.3243A>G, m.8344A>G, m.8993T>G, m.8993 T>C point mutations was performed.

PCR test for the common deletion was performed using primers covering region: 8224-13501. Long-PCR test for mtDNA deletion was performed using primers covering region: 6730-16545. For all the above mentioned tests DNA isolated from peripheral blood as well as from a muscle sample was used.

*POLG* gene analysis:

*POLG* gene analysis was performed by sequencing coding region of the gene. DNA isolated from peripheral blood was used for this analysis.

**Results:**

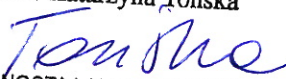
Multiple mtDNA deletions present in the muscle sample (at a low level).

Heterozygous *POLG* variants: T251I, P587L, K1191N

**Interpretation and recommendations:**

Molecular analysis confirmed multiple mtDNA deletions. *POLG* gene (nuclear) analysis revealed three heterozygous variants: T251I, P587L, K1191N. According to the literature, T251I, P587L variants are frequently present on the same chromosome.

Molecular analysis of patient's parents' DNA confirmed that the patient is a compound heterozygote, with T251I, P587L mutations on one, and K1191N on the other chromosome.

11069 dr hab. Katarzyna Tońska  
  
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**Name and surname of the patient:** Halina Koniew

**PESEL:** ██████████

**Date of birth:** 20.11.1956

**Sex:** female

**Sample number:** M353

**Physician in charge:** Agnieszka Tomaszewska, M.D., PhD, Biruta Kierdaszuk, M.D.

**Clinical diagnosis:** *POLG* gene mutation in patient's son

**Type of biological material:** peripheral blood

**Analysis performed:**

*POLG* gene analysis:

*POLG* gene regions covering variants: T251I, P587L, K1191N were analyzed by sequencing.

**Results:**

Heterozygous *POLG* variant: K1191N

**Interpretation and recommendations:**

*POLG* gene (nuclear) analysis revealed the presence of the recessive K1191N variant.

11069 dr hab. Katarzyna Tońska  
*Tońska*  
DIAGNOSTA LABORATORYJNY



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**Name and surname of the patient:** Wiktor Koniew

**PESEL:** ██████████

**Date of birth:** 6.05.1951

**Sex:** male

**Sample number:** M352

**Physician in charge:** Agnieszka Tomaszewska, M.D., PhD, Biruta Kierdaszuk, M.D.

**Clinical diagnosis:** *POLG* gene mutation in patient's son

**Type of biological material:** peripheral blood

**Analysis performed:**

*POLG* gene analysis:

*POLG* gene regions covering variants: T251I, P587L, K1191N were analyzed by sequencing.

**Results:**

Heterozygous *POLG* variants: T251I, P587L present

**Interpretation and recommendations:**

*POLG* gene (nuclear) analysis revealed two heterozygous variants: T251I and P587L. According to the literature, T251I, P587L variants are frequently present on the same chromosome and are recessive.

11069 dr hab. Katarzyna Tońska  
*Katarzyna Tońska*  
DIAGNOSTA LABORATORYJNY

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