Warszawski Uniwersytet Medyczny

Laboratorium Zakładu Genetyki Medycznej

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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: [redacted]

Date of birth: 25.01.1979

Address: ul. [redacted] 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: [redacted]

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.

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Name and surname of the patient: Wiktor Koniew

PESEL: [redacted]

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.

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