



Genekor
Committed to Biotechnological Innovation

Genekor Medical S.A. | 52, Spaton Ave., 15344, Gerakas, Athens, Greece ,G.E.MI. nr: 0007856001000
email: info@genekor.com, www.genekor.com | Tel. (+30) 210 6032138 Fax. (+30) 210 6032148

RESULTS REPORT

Sample information

Name :	Date Received :
Medical ID :	Date of Report :
Date of Birth :	Req. Physician :
Material :	Barcode :
	Referral <i>CYP2C9, CYP2C19, ABCB1</i>

CYP2C9, CYP2C19, ABCB1

Results

Gene	Result	Activity
CYP2C9	*1/*2	Moderate
CYP2C19	*1/*1	Normal
ABCB1	Heterozygous	Moderate

Methodology

Total genomic DNA was extracted from the submitted sample. Analysis for the presence of the **CYP2C9*2** (rs1799853, c.430C>T, p.R144C) and **CYP2C9*3** (rs1057910, c.1075A>C, p.I359L) polymorphisms in the *CYP2C9* gene, **CYP2C19*2** (rs4244285, c.681G>A, p.P227=) and **CYP2C19*3** (rs4986893, c.636G>A, p.W212*) polymorphisms in the *CYP2C19* gene, and the **C3435T** (rs1045642) polymorphism in the *ABCB1* gene was performed using a targeted sequencing approach (**KAPA HyperPlus, Roche**). Sequencing was performed using the **Ion GeneStudio S5 Prime System Next-Generation Sequencing platform (DNBSEQ-T7)**.

The **CYP2C9** and **CYP2C19** genes play a critical role in the metabolism of numerous clinically important drugs, including **omeprazole, diazepam, clopidogrel, warfarin, and siponimod**. Patients who are **poor CYP2C19 metabolizers** (carriers of the *2 or *3 alleles) have a reduced ability to convert clopidogrel into its active form. This may result in reduced platelet inhibition, potentially decreasing treatment effectiveness and increasing the risk of thrombotic complications.

The **CYP2C9** gene is primarily involved in the metabolism of anticoagulants such as **warfarin**, although there is evidence suggesting a possible role in clopidogrel metabolism as well. CYP2C9 genotyping may be particularly useful in patients receiving combined **anticoagulant and antiplatelet therapy**, potentially contributing to treatment individualization.

In addition, the **ABCB1** gene plays an important role in the absorption and transport of clopidogrel. The **C3435T (rs1045642)** polymorphism, which is frequently observed in the heterozygous state, may influence drug bioavailability. Specifically, the homozygous **TT genotype** has been associated with lower platelet inhibition and reduced concentrations of both clopidogrel and its active metabolite.

Finally, genotyping of **CYP2C9** and **CYP2C19** may also be relevant before initiating **warfarin** therapy, as it provides information regarding the patient's metabolic capacity. This information may contribute to adjustment of the initial dose, potentially reducing the risk of bleeding or thrombotic events and improving treatment efficacy.

Note: Each molecular analysis has an inherent error rate of approximately **0.5–1%**. This may be attributable to rare events and factors associated with sample preparation and analysis.



Electronically Signed by

- George Nasioulas, PhD Molecular Biologist, Scientific Director, AMKA:26025301255



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Name	Barcode
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Bibliography

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URL:<https://www.knmp.nl/downloads/pharmacogeneticrecommendations-may-2020.pdf>
5. <https://www.pharmgkb.org>
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