Large-scale nationwide genomics projects: The Genome of Greece (GoGreece) initiative

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EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

CHMP Pharmacogenomics Working Party (PGWP)

Disclaimer

Declared conflict of interests: None

The opinions expressed in this presentation do not reflect the policies and position of the European Medicines Agency

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How an ancient temple relates to the translation of Genomics into Medicine?

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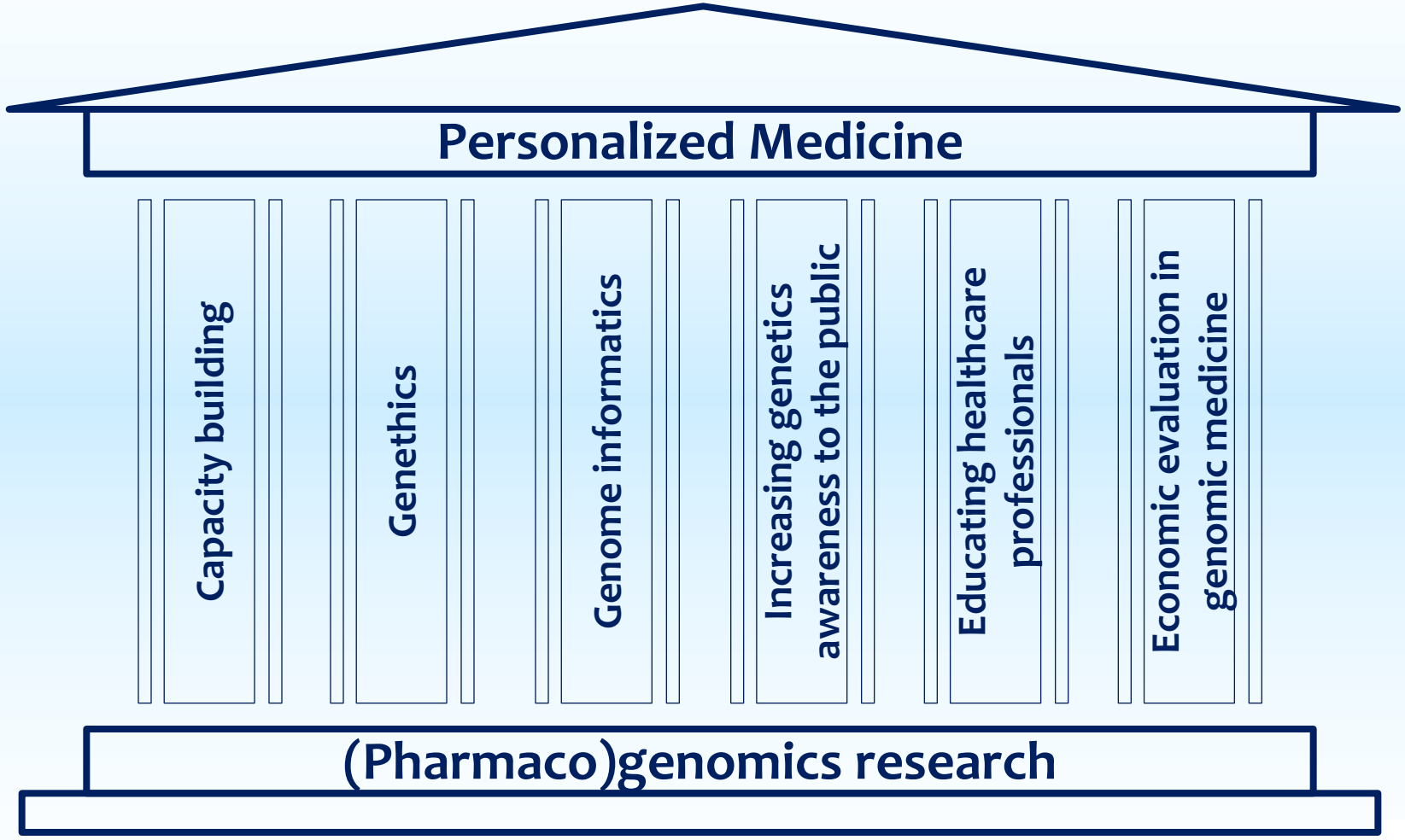
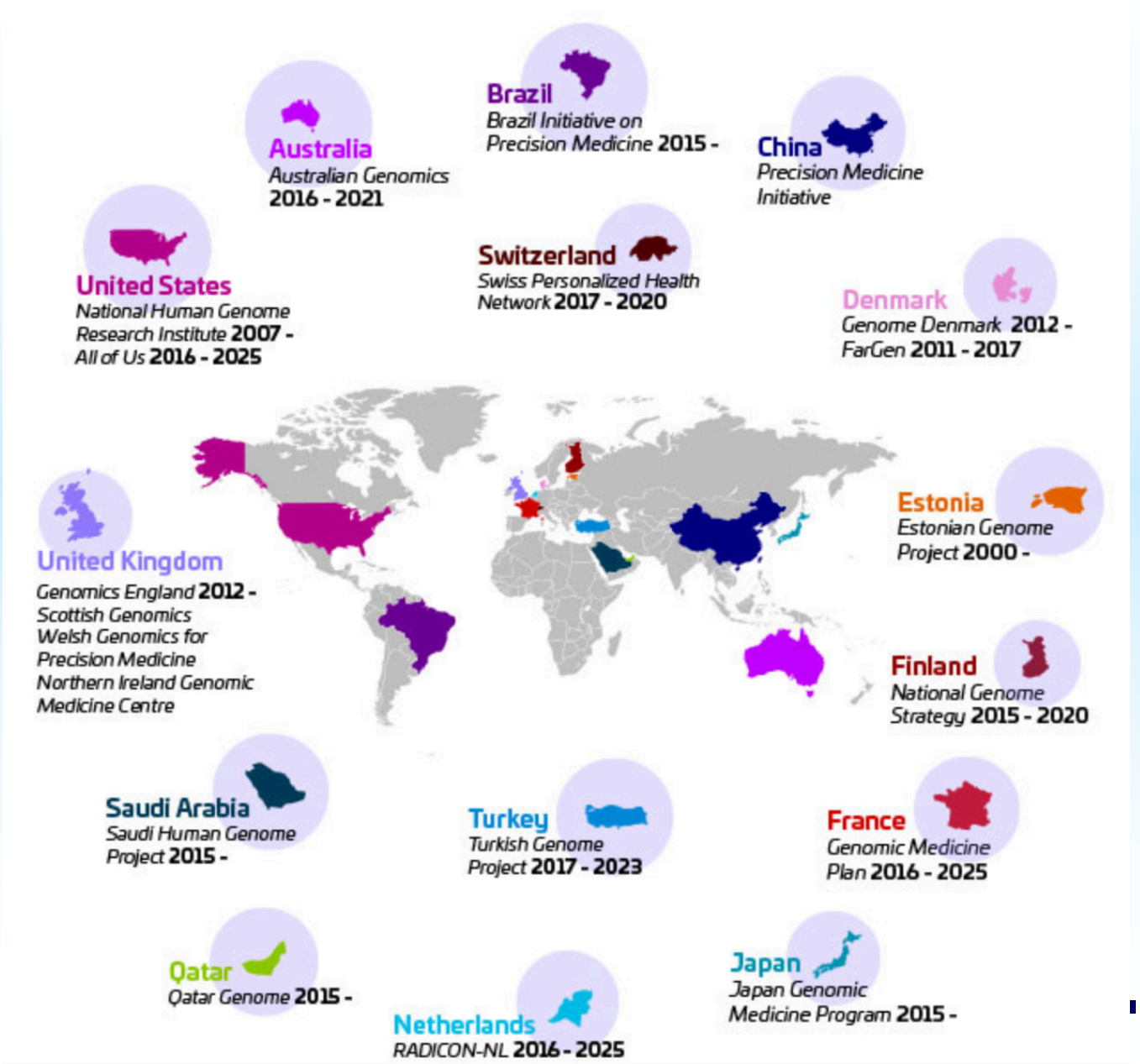


Existing National Human Genome projects



The Genome of Greece (GoGreece) initiative

A long-term large-scale National Genomics project focused on the implementation of Personalized Medicine in Greece

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The “GoGreece” initiative: Aims and goals - I

a. Determine the allelic architecture of the Greek population, especially focusing on rare, pathogenic or not, variants that are characteristic for the Greek population,
b. Characterize the unique allelic spectrum of isolate populations in Greece, such as the islands, mountainous regions, as well as sub-population groups such as Greek Roma and Pomaks,
c. Establish the pathogenic variant spectrum of common and rare genetic diseases in the Greek population,
d. Catalyze the clinical implementation of Pharmacogenomics in the Greek healthcare sector,

The “GoGreece” initiative: Aims and goals - II

e. Develop and maintain a comprehensive knowledgebase to report the extant genetic heterogeneity in the Greek population, as far as pathogenic variants and polymorphic alleles are concerned,
f. Address important ethical, legal and societal issues pertaining to the implementation of Personalized Medicine interventions in Greece,
g. Enrich the genomics education of healthcare professionals and biomedical scientists in Greece, and
h. Raise genomics awareness of the general public.

WP1: Governance and Coordination

WP2: Sample collection
- ✓ Logistics
- ✓ IRB approvals

WP3: Genetic analysis
- ✓ Monogenic diseases/modifier genes
- ✓ Polygenic disorders
- ✓ Pharmacogenomics
- ✓ Genetics of isolated populations

WP4: Genome Informatics
- ✓ Translational tools and EHR
- ✓ Greek National Genetic Database

WP7: Biobanking

WP9: General Public Engagement
- ✓ Informational portal for genetic diseases in Greece
- ✓ Awareness Days and Exhibitions
- ✓ Training school students (2MoBiL)

WP5: Ethical Legal and Societal Issues
- ✓ Ethics
- ✓ Stakeholder analysis

WP6: Pricing and Regulation
- ✓ Guidelines for genome-informed drug prescription in Greece
- ✓ Database with drugs with a Personalized Medicine indication in drug labels in Greece
- ✓ Health Technology Assessment for the reimbursement of Personalized Medicine interventions in Greece

WP8: Healthcare Professional Education
- ✓ Establishment of e-learning modules
- ✓ Organization of onsite training for continuous medical education
- ✓ National Personalized Medicine Conference
- ✓ National Personalized Medicine scientific journal

WP10: Dissemination and Communication
- ✓ Project visibility (Scientific articles and presentations in international conferences)
- ✓ Sustainability and fund raising (Crowdfunding, Charitable entities, grant applications)

2010

Project initiated

2012

First Greek individual sequenced

2020

Phase I: First 1000 Greek individuals to be sequenced

The “Genome of Greece” initiative in numbers

The “Genome of Greece” initiative in numbers

4

Research Centers Involved so far

- Laboratory of Pharmacogenomics and Individualized Therapy, University of Patras, PATRAS (Coordinating Center)
- National and Kapodistrian University of Athens (ATHENS)
- ANALYSI Biomedical Laboratories (THESSALONIKI)
- Cretan Genomics Center (RETHYMNO, CRETE)

The “Genome of Greece” initiative in numbers

>1500

Recruited subjects

61

Sequenced individuals by WGS

734

Sequenced individuals by PGx panel resequencing

109

Sequenced individuals by WES

The “Genome of Greece” initiative in numbers

9

Projects initiated

11

Scientific articles produced

5

Theses Defended

4

Conferences Organized

“Go Greece” – Preliminary findings I

Characterization of a **novel genetic locus** that is associated with sporadic **Amyotrophic Lateral Sclerosis** in Greek patients.

Mitropoulos *et al. Human Genomics* (2017) 11:30
DOI 10.1186/s40246-017-0126-2

Human Genomics

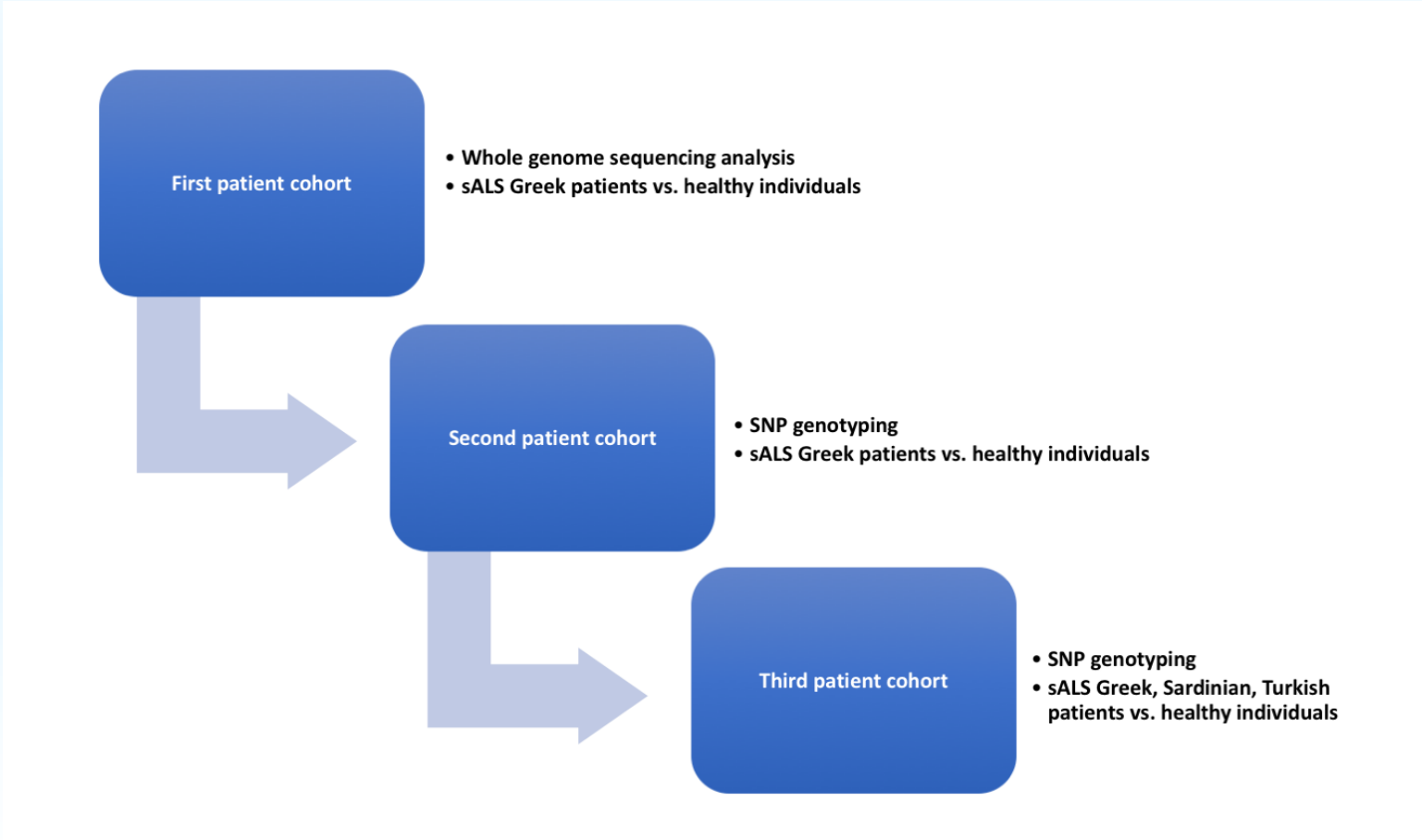
PRIMARY RESEARCH Open Access

Genomic variants in the *FTO* gene are associated with sporadic amyotrophic lateral sclerosis in Greek patients

Konstantinos Mitropoulos[1†], Eleni Merkouri Papadima[2†], Georgia Xiromerisiou[3], Angeliki Balasopoulou[2], Kyriaki Charalampidou[2], Vasiliki Galani[2], Krystallia-Vassiliki Zafeiri[2], Efthymios Dardiotis[3], Styliani Ralli[3], Georgia Deretzi[4], Anne John[5], Kyriaki Kydonopoulou[6], Elpida Papadopoulou[6], Alba di Pardo[7], Fulya Akcimen[8], Annalisa Loizedda[9,10], Valerija Dobričić[11], Ivana Novaković[11,12], Vladimir S. Kostić[11], Clint Mizzi[2], Brock A. Peters[13,14], Nazli Basak[8], Sandro Orrù[9], Evangelos Kiskinis[7], David N. Cooper[15], Spyridon Gerou[6], Radoje Drmanac[13], Marina Bartsakoulia[2], Evangelia-Eirini Tsermpini[2], Georgios M. Hadjigeorgiou[3], Bassam R. Ali[5], Theodora Katsila[2] and George P. Patrinos[2,5*]

“Go Greece” – Preliminary findings I

Characterization of a **novel genetic locus** that is associated with sporadic **Amyotrophic Lateral Sclerosis** in Greek patients.



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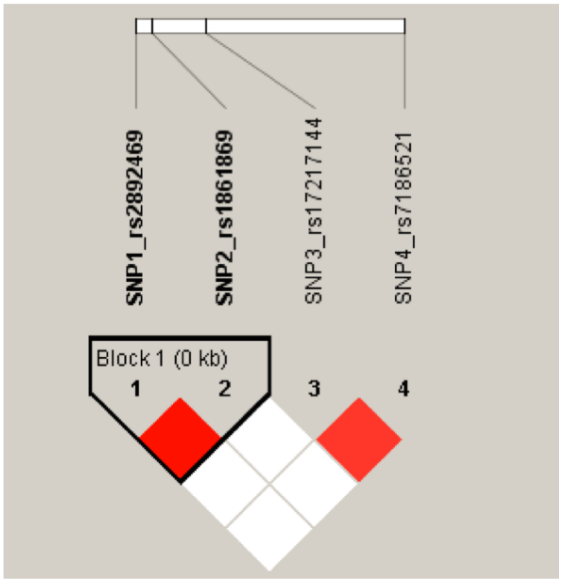
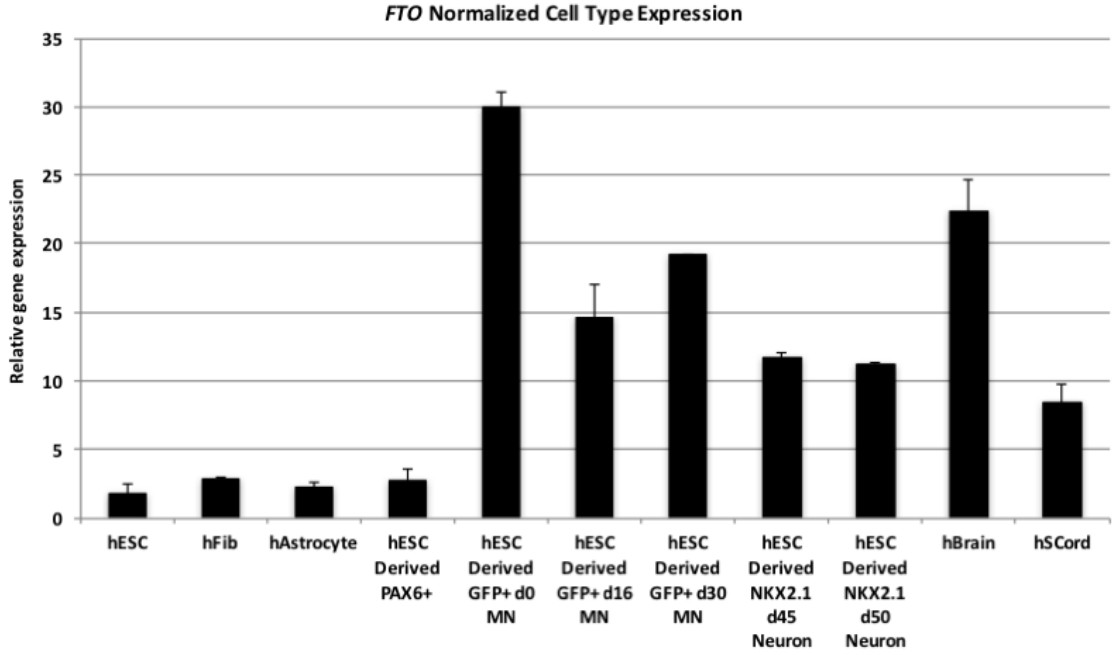
Mitropoulos et al., *Hum Genomics*, 2018

“Go Greece” – Preliminary findings I

Characterization of a **novel genetic locus** that is associated with sporadic **Amyotrophic Lateral Sclerosis** in Greek patients.

HaploView v4.2 analysis (Greek Cohort)





“Go Greece” – Preliminary findings II

Characterization of a **novel genetic locus** that is associated with **Celiac disease** in Greek patients.

Balasopoulou *et al. Human Genomics* (2016) 10:34
DOI 10.1186/s40246-016-0091-1

Human Genomics

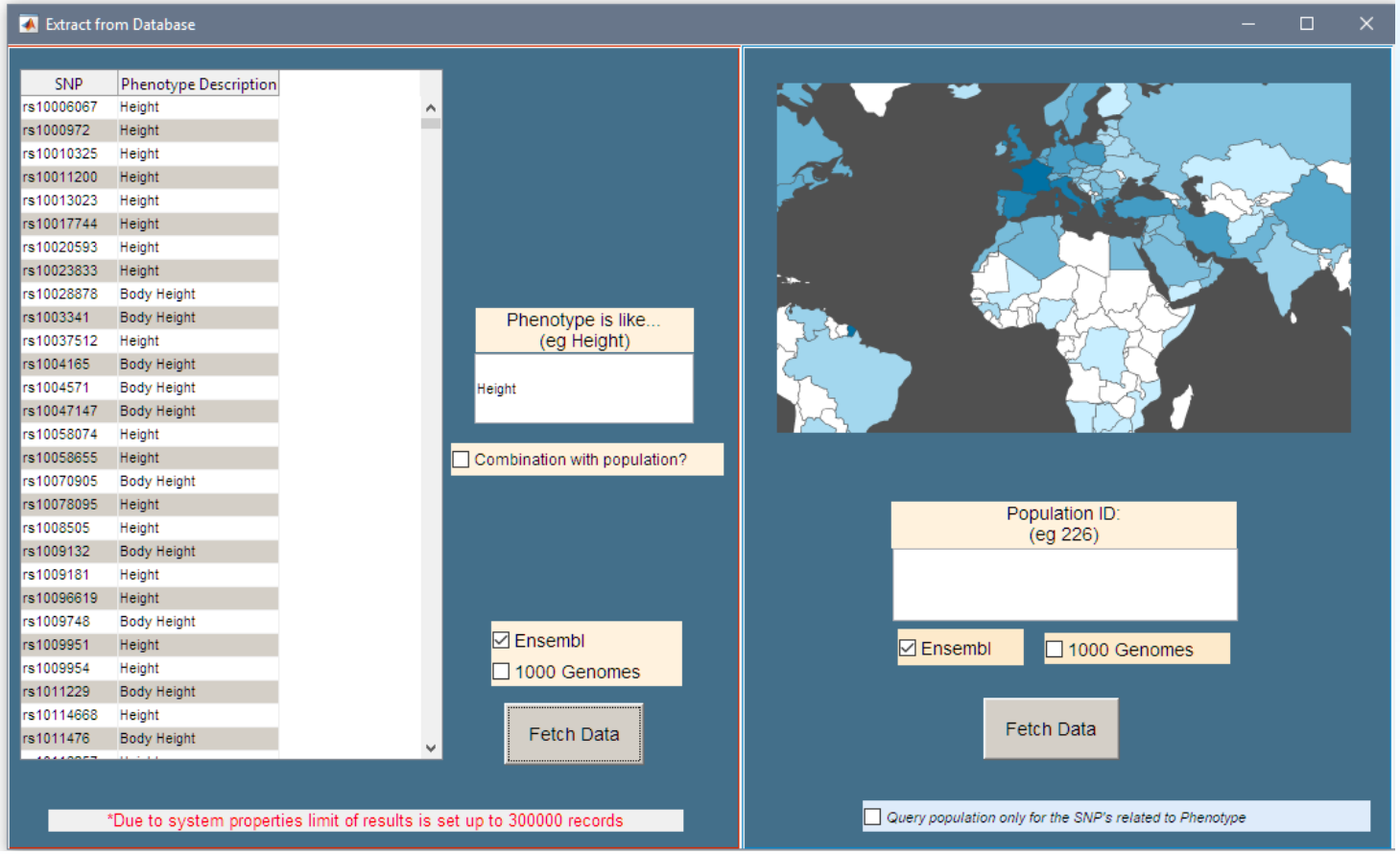
PRIMARY RESEARCH Open Access

Novel genetic risk variants for pediatric celiac disease

Angeliki Balasopoulou[1†], Biljana Stanković[2†], Angeliki Panagiotara[1], Gordana Nikčevic[2], Brock A. Peters[3,4], Anne John[5], Effrosyni Mendrinou[1], Apostolos Stratopoulos[1], Aigli Ioanna Legaki[1], Vasiliki Stathakopoulou[1], Aristoniki Tsolia[1], Nikolaos Govaris[1], Sofia Govari[1], Zoi Zagoriti[1], Konstantinos Poulas[1], Maria Kanariou[6], Nikki Constantinidou[6], Maro Krini[7], Kleopatra Spanou[6], Nedeljko Radlovic[8], Bassam R. Ali[5], Joseph Borg[9], Radoje Drmanac[3,4], George Chrousos[7], Sonja Pavlovic[2], Eleftheria Roma[7], Branka Zukic[2], George P. Patrinos[1,5]

“Go Greece” – Preliminary findings III

Identification of more than **80 genomic variants** that are **specific for the Greek population**, as compared to the Caucasian population.



Loukatou et al., *in preparation*

“Go Greece” – Preliminary findings III

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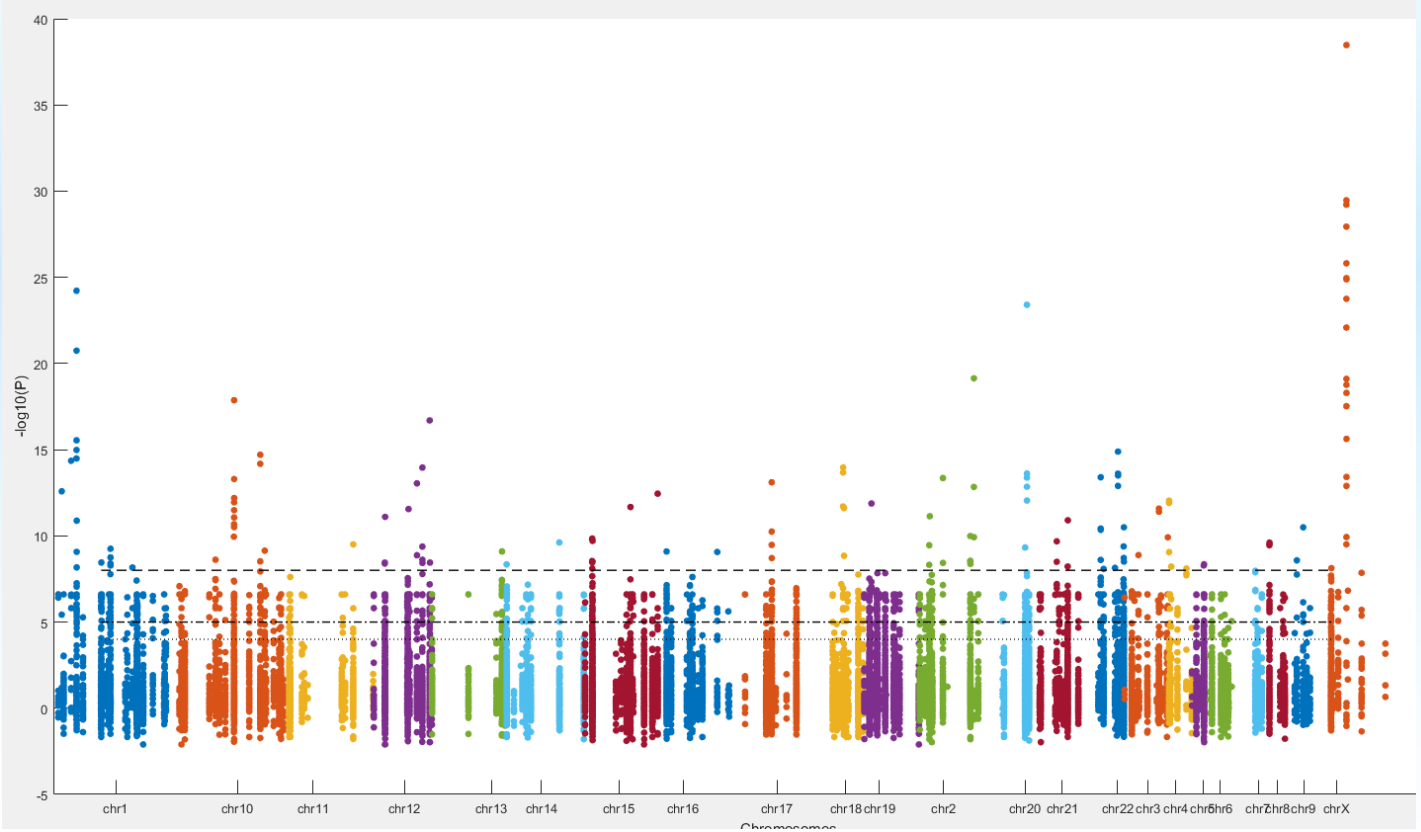


"Go Greece" – Preliminary findings IV

Assessing the level of **genomics knowledge of healthcare professionals and clinicians** in Greece.

Research Article

For reprint orders, please contact: reprints@futuremedicine.com

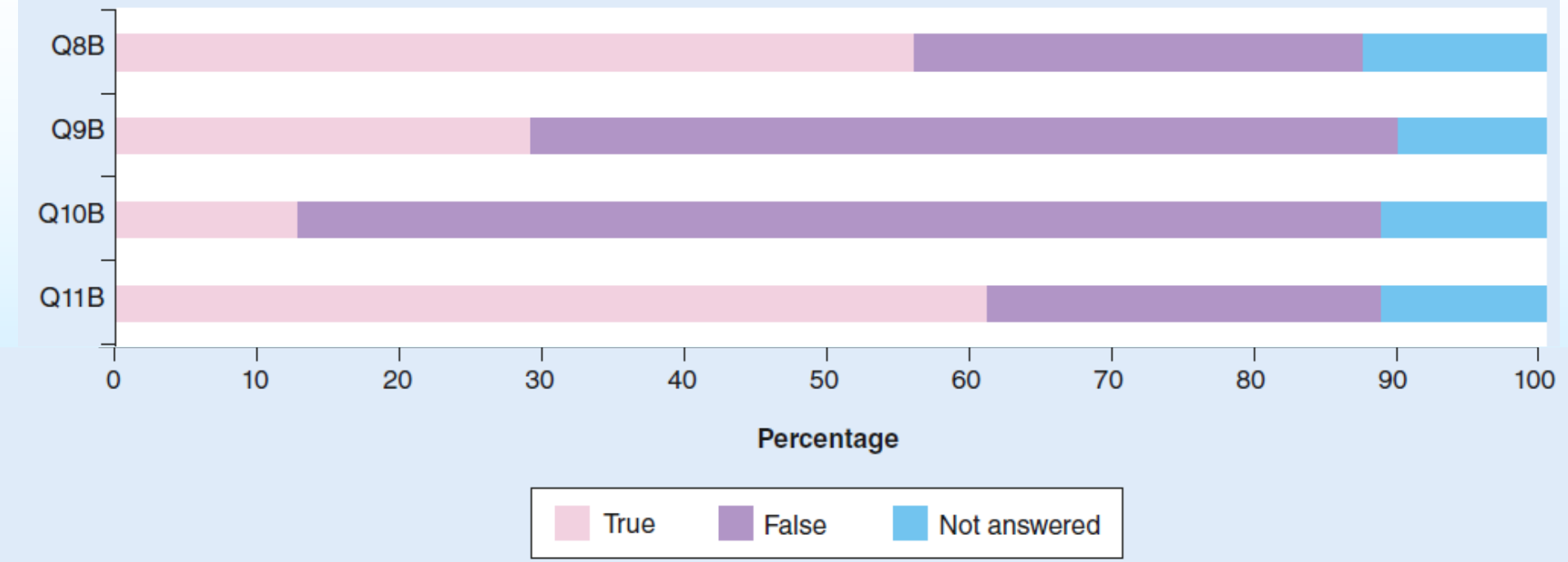
Personalized Medicine

Critical appraisal of the views of healthcare professionals with respect to pharmacogenomics and personalized medicine in Greece

10.2217/PME.13.92 © 2014 Future Medicine Ltd *Personalized Medicine* (2014) 11(1), 15–26 ISSN 1741-0541

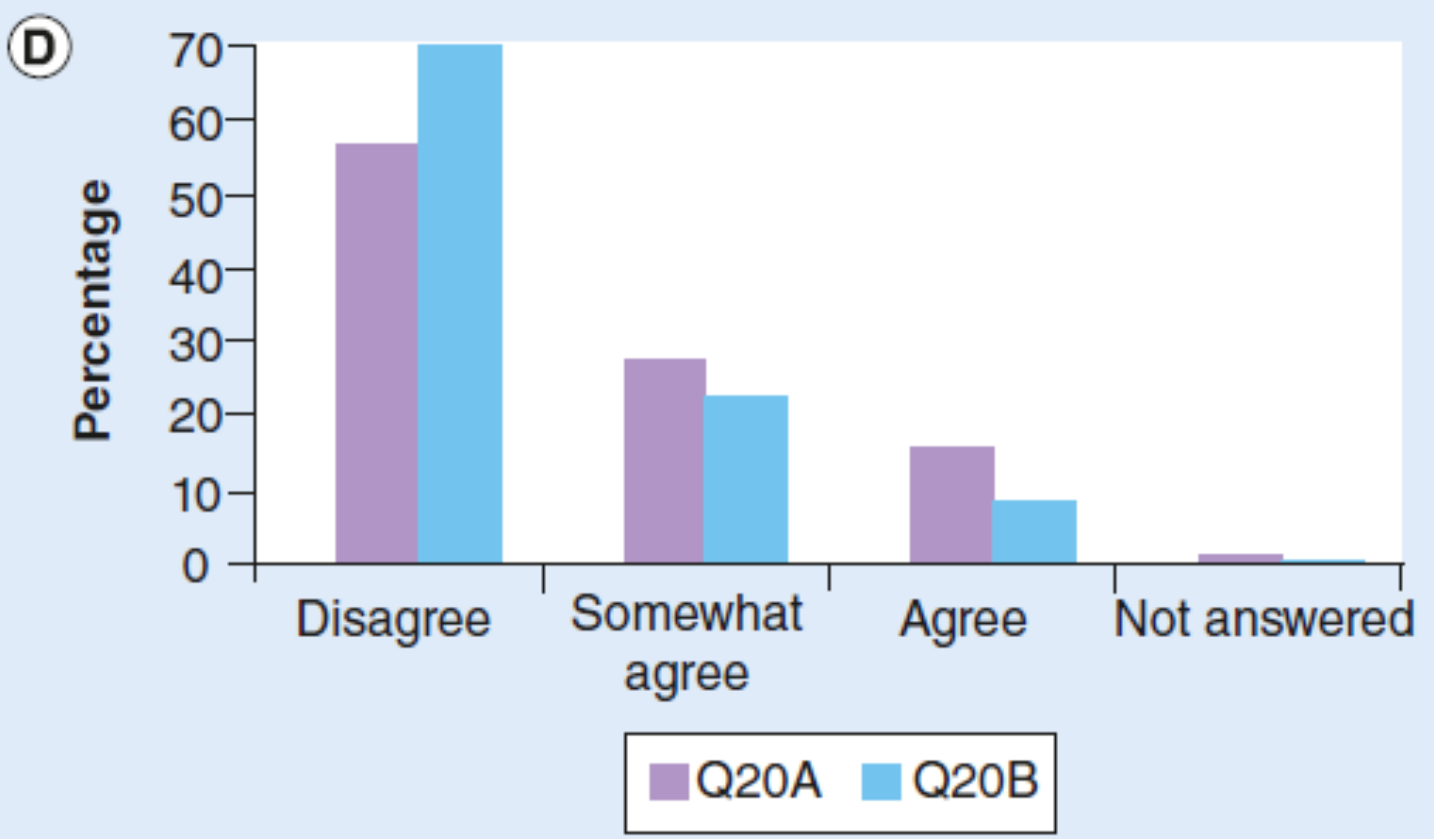
Aim: To ascertain their views and opinions on various issues pertaining to genomics, personalized medicine and their impact to society

Critical appraisal of the views of healthcare professionals on Genomics and Personalized Medicine in Greece



Q8B: Approximately 99.9% of the human DNA sequence is identical
Q9B: Humans have 48 chromosomes
Q10B: A only pairs with C and G only pairs with T
Q11B: Inherited diseases are caused by changes in the genetic material

Critical appraisal of the views of healthcare professionals on Genomics and Personalized Medicine in Greece



Q20A, Q20B: My undergraduate studies at the University provided me with sufficient knowledge on pharmacogenomics and personalized medicine

Mai et al., *Pers Med* 2014;11(1):15-26

“Go Greece” – Preliminary findings V

Assessing the **level of genomics awareness of the general public** in Greece

RESEARCH ARTICLE

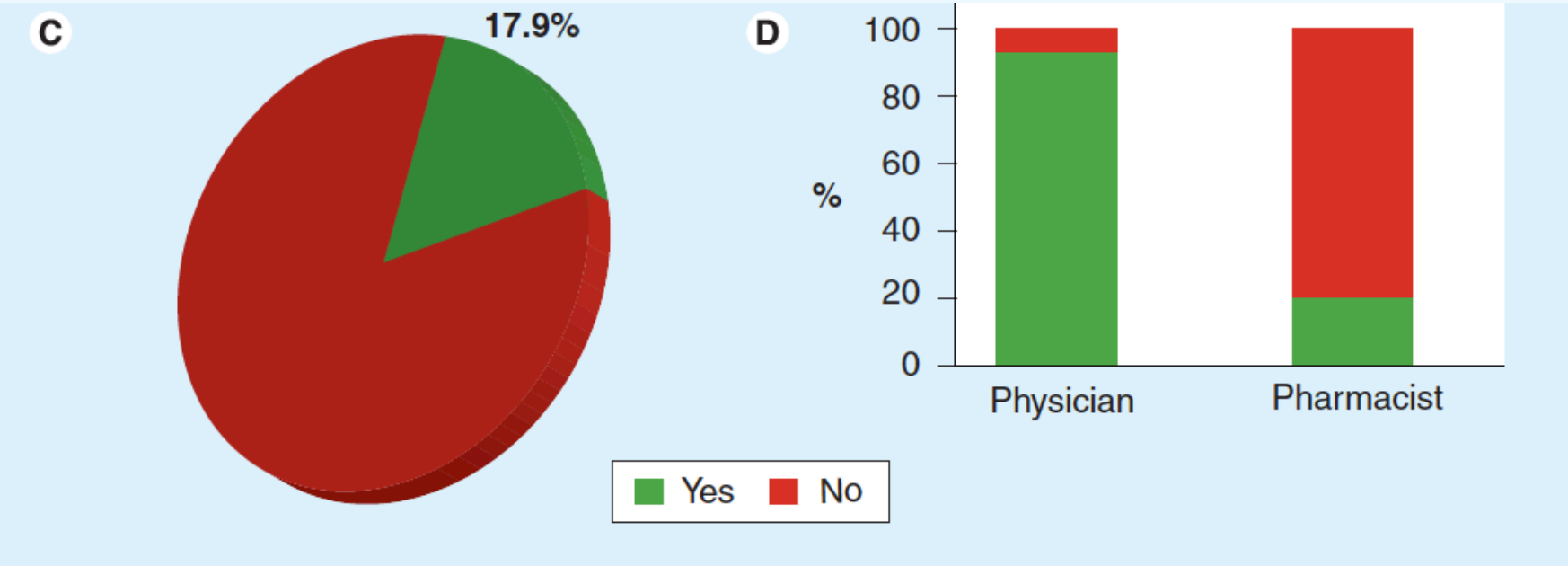
A critical view of the general public’s awareness and physicians’ opinion of the trends and potential pitfalls of genetic testing in Greece

Yuan Mai*, Theodora Koromila*, Aggeliki Sagia*, David N Cooper, Georgios Vlachopoulos, George Lagoumintzis, Panagoula Kollia, Konstantinos Poulas, Vlassios Stathakopoulos & George P Patrinos†

†Author for correspondence: University of Patras, School of Health Sciences, Department of Pharmacy, University Campus, GR-265 04, Rion, Patras, Greece
Tel.: +30 261 096 9834
gpatrinos@upatras.gr

“Go Greece” – Preliminary findings V

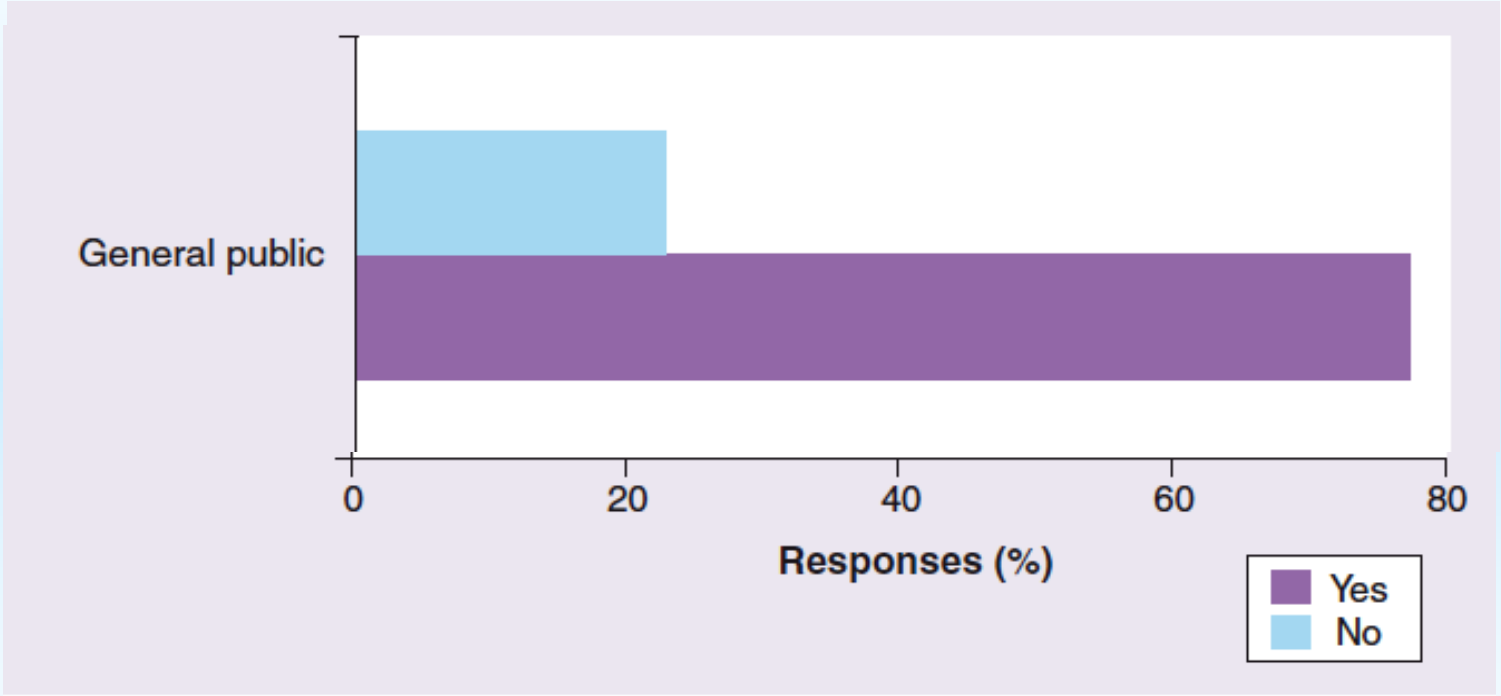
Assessing the **level of genomics awareness of the general public** in Greece



C. Should the genetic tests be sold using the DTC model?
D. Who should prescribe genetic tests to the patients?

“Go Greece” – Preliminary findings V

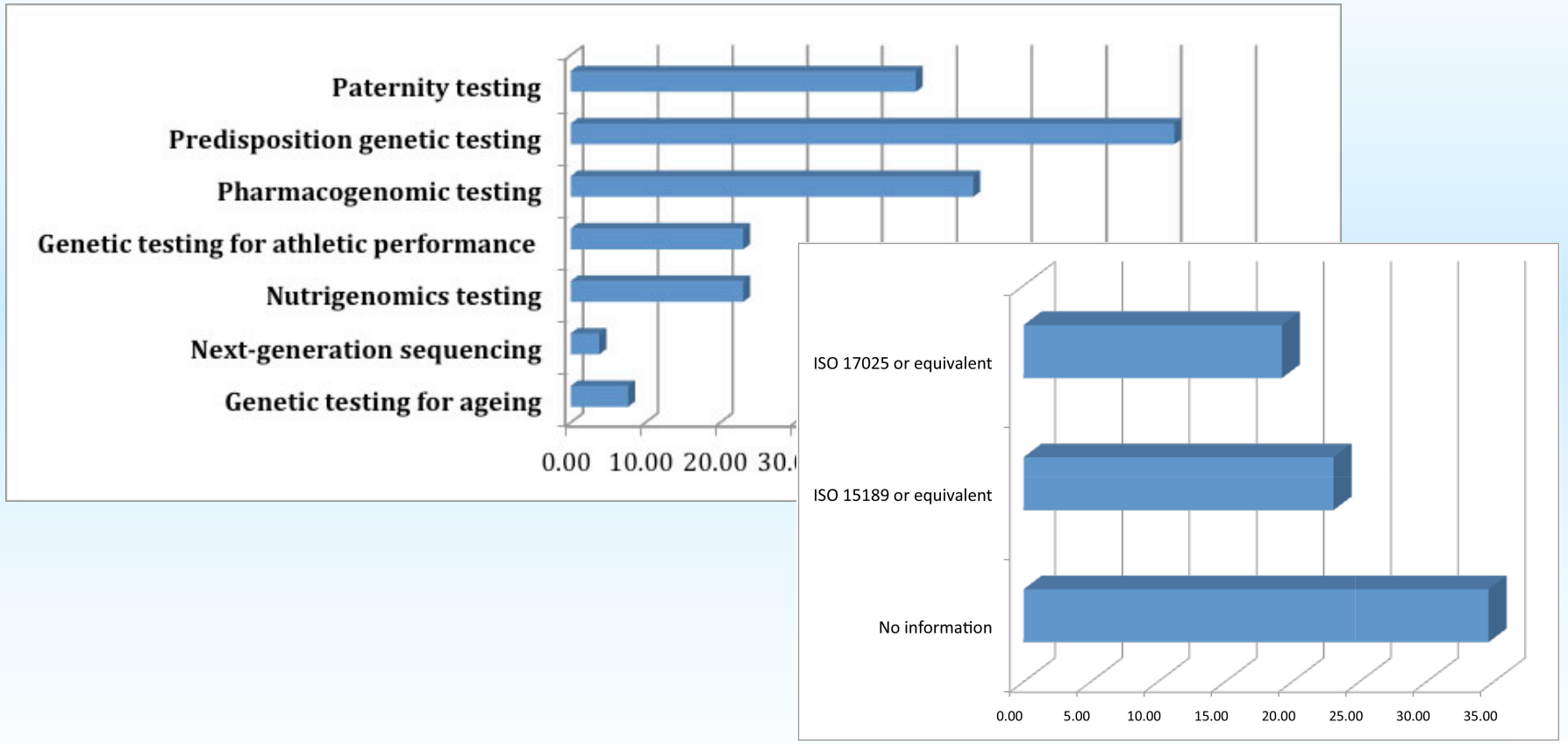
Assessing the **level of genomics awareness of the general public** in Greece



Do you believe that a personalized diet based on your genetic profile could be beneficial to you and your overall health and lifestyle?

“Go Greece” – Preliminary findings VI

Assessing the types of **genetic services provided by private genetic laboratories** in Greece



Kechagia et al., *Public Health Genomics* 2014

“Go Greece” – Preliminary findings VII

Disentangling the **views, opinions and stance points of the various stakeholders** engaged in Personalized Medicine in Greece

Public Health Genomics

Public Health Genomics 2014;17:280–286
DOI: 10.1159/000365896

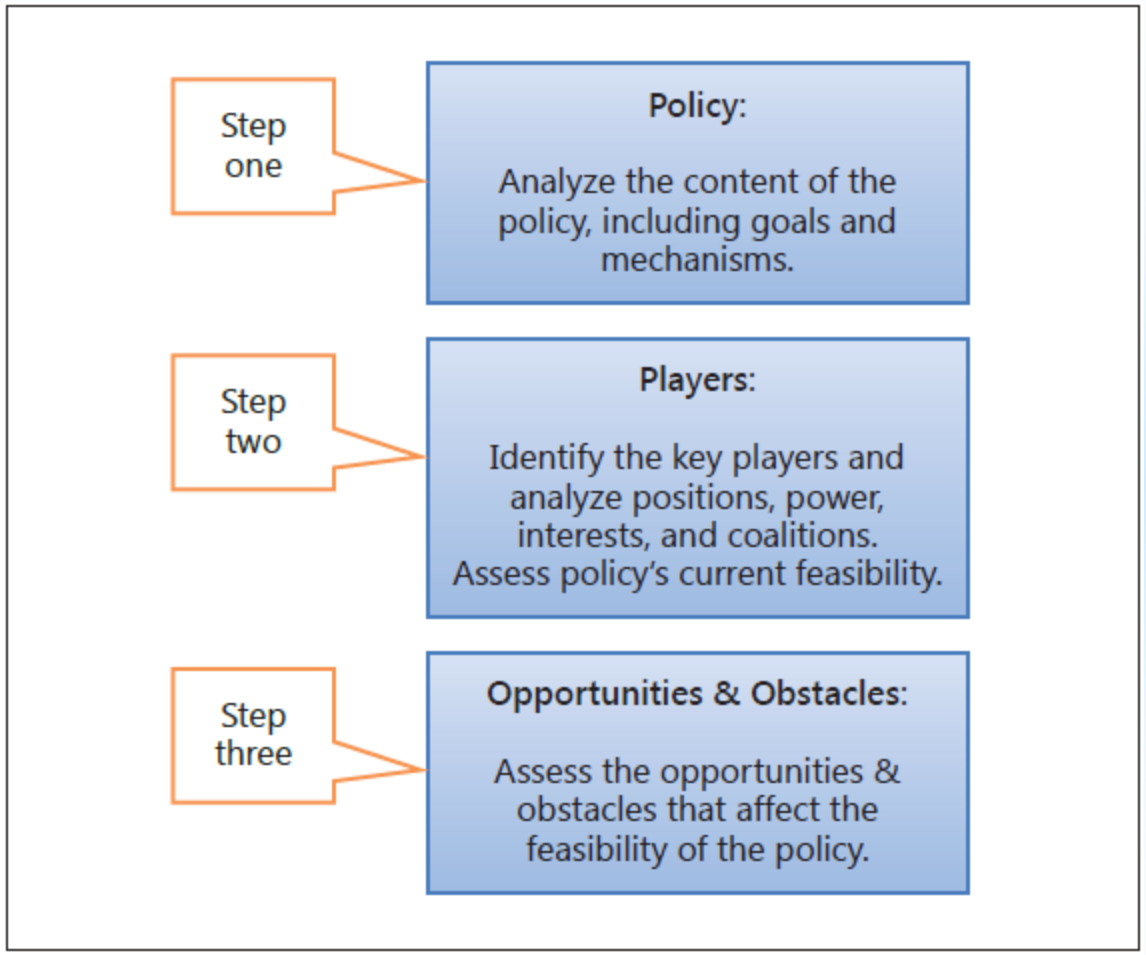
Published online: September 9, 2014

Stakeholder Analysis in Pharmacogenomics and Genomic Medicine in Greece

Christina Mitropoulou[a] Yuan Mai[b] Ron H. van Schaik[a] Athanassios Vozikis[c]
George P. Patrinos[b]

[a]Department of Clinical Chemistry, Erasmus University Medical Center, Rotterdam, The Netherlands;
[b]Department of Pharmacy, School of Health Sciences, University of Patras, Patras, and [c]Economics Department, University of Piraeus, Piraeus, Greece

“Go Greece” – Preliminary findings VII



“Go Greece” – Preliminary findings VII

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ΕΞΑΤΟΜΙΚΕΥΜΕΝΗ ΙΑΤΡΙΚΗ

(PERSONALIZED MEDICINE)

The first scientific journal in the field of Personalized Medicine in Greece, published in 2019

www.permed.gr

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The Greek National Personalized Medicine Conference

UNIVERSITY OF PATRAS

https://conferences.permed.gr/2019/

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GoldenHelix ACADEMY

By

The GoldenHelix FOUNDATION

Aim: www.academy.goldenhelix.org

To establish an e-learning platform with modules on Personalized Medicine-related topics, such as Public Health Genomics, Genome informatics, Economics and Health Technology Assessment in Genomics, Genethics, *etc*, leading to Certificates and Diplomas, specifically aimed for the needs of stakeholders in Greece.

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The **Golden Helix Academy** is part of the educational activities of the Golden Helix Foundation. The Golden Helix Academy provides training in the field of Genomic and Precision Medicine offering different type of training as outline below:

GoldenHelix ACADEMY

E-learning training modules

Introduction to Bioinformatics for clinical genome interpretation

Economic Evaluation in Genomic Medicine

Pharmacogenomics in Clinical Care and Drug Discovery

On-site training courses

On-site Course: Bioinformatics and Clinical Genomics

On demand on-site training

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Mobile Molecular Biology Laboratory (*2MoBiL*)

- Portable Molecular Biology Laboratory for the training of school students in the field of Molecular Genetics and Personalized Medicine,
- Accompanied by plastic models, comics and online games for primary school students,
- Ideal for the interactive learning of students as well as educators [using the train-the-trainers (3T) approach]
- Awarded the Kafatos Labbox price in 2018.

Acknowledgements

The GoldenHelix FOUNDATION

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