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## LABORATORY OF PHARMACOGENOMICS AND INDIVIDUALIZED THERAPY

### MEMBERS (in alphabetical order)

1. Asimina Andritsou (Graduate student) 3. Lia Barba (Graduate student) 4. Anastasios Bitsakos (Graduate student)

5.

Konstantina Chalikipoulou (PhD candidate)

6.

Kyriaki Charalambidou (Graduate student)

7.

Fotini Gerasimatou (Graduate student)

8.

Andreas Kannavos (Post-doctoral Research Scientist and Group Leader)

9.

Karyofyllis Karamperis (Graduate student)

10.

Katerina Kechagia (Graduate student)

11.

Zoe Kordou (PhD Candidate)

12. Maria Koromina (Post-doctoral Research Scientist and Group Leader)

13. Stefania Koutsilieri (PhD Candidate)

14.

Ioannis Liopetas (Graduate student)

15.

Evelyn Mendrinou (PhD candidate)

16.

Konstantinos Mitropoulos (Research Physician)

17.

Styliani Papadaki (Graduate student)

18.

George P. Patrinos (Head of Laboratory)

19.

Stavroula Siamoglou (PhD candidate)

20.

Theano Stamopoulou (Graduate student)

21.

Andreas Stathoulas (Graduate student)

22.

Apostolos Stratopoulos (PhD candidate)

23.

Sotiria Tsekoura (Graduate student)

24.

Evira Tsermpini (University Fellow

,  
Research Scientist  
and Group Leader)

25.

Evelina Zafeiri (Graduate student)

26.

Aggeliki Zakoula (Graduate student)

## **PAST MEMBERS**

- Vasiliki Chondrou (Staff scientist, Hellenic Open University, Greece)
- Marianthi Georgitsi (Assistant Professor, Aristotle University of Thessaloniki, School of Medicine, Thessaloniki, Greece)
- George Pampalakis (Assistant Professor, Aristotle University of Thessaloniki, Department of Pharmacy, Thessaloniki, Greece)
- Cristiana Pavlidis (Lecturer, Middlesex University, London, UK)
- Theodora Katsila (Researcher C', National Hellenic Research Foundation Athens, Greece)

## **STUDENT OFFICE HOURS:**

Every Tuesday: 9:00-10:30 a.m.

Every Wednesday: 9:00-10:30 a.m.

## **EDUCATION**

1998: Ph.D. in Molecular Biology, University of Athens, Greece

1993: B.Sc. in Biology, University of Athens, Greece

## CURRENT POSITIONS (in chronological order)

- **2019-TODAY: Professor** of Pharmacogenomics and Pharmaceutical Biotechnology, University of Patras Department of Pharmacy, Patras, Greece
- **2015-TODAY: Adjunct Full Professor**, United Arab Emirates University, College of Medicine, Al-Ain, United Arab Emirates
- **2010-TODAY: Member and National Representative**, CHMP Pharmacogenomics Working Party, European Medicines Agency, London, United Kingdom
- **2009-TODAY: Adjunct Faculty**, Erasmus University Medical Center, Faculty of Medicine and Health Sciences, Rotterdam, the Netherlands

## RESEARCH ACTIVITIES

### Pharmacogenomics and personalized medicine

Our group is involved in a number of projects pertaining to pharmacogenomics and response to various drugs. In particular, we are interested in assessing the effect of different pharmacogenomic markers on the variable response to HU treatment in  $\beta$ -type hemoglobinopathies patients, to Lithium treatment in bipolar disease patients, and the development of adverse effects during to 6-MP treatment in Acute Lymphoblastic Leukemia patients. We are also interested to ascertain how pharmacogenomics can contribute towards reducing the overall healthcare expenditure, by performing cost-benefit pharmacoeconomic analysis, which would facilitate integration of pharmacogenomics into clinical practice.

### Genomics of Human Populations

We are interested in examining the diversity of pharmacogenomic markers in various human populations, using genomic approaches and previously published information. Using these approaches, we have developed a worldwide database (FINDbase) where clinically relevant population-specific allele frequency data are documented and different National Genetic databases and accompanying software (ETHNOS), where all information pertaining to genetic diseases in different populations, especially from developing countries is stored.

### Family Genomics of Human Pedigrees

We have exploited whole genome sequencing approaches using known pedigree information to delineate genotype-phenotype information, to identify personalized pharmacogenomic profiles and to pinpoint genomic variants that can be putatively used as prognostic markers for human

genetic disorders.

#### Study of Human Diseases

We are currently utilizing whole-genome sequencing to study rare disorders with a genetic component, such as schizophrenia and amyotrophic lateral sclerosis to identify genomic variants that contribute to the resulting phenotype. We have recruited unique cases and candidate families for these diseases and we are actively working on identifying candidate variants for these diseases.

#### Study of the transcriptional regulation of the human fetal globin gene expression

We are interested to understand the molecular mechanisms that govern the transcriptional regulation of the human fetal globin genes in an effort to re-activate human fetal hemoglobin to treat  $\beta$ -hemoglobinopathies patients. We are combining whole genome scanning and sequencing approaches using unique families and individuals with increased fetal hemoglobin in adult life and functional studies to validate our findings.

#### Public Health Genomics

We are interested to critically appraise the impact of genetics and pharmacogenomics to society and the level of awareness over genetics, pharmacogenomics and personalized medicine in various stakeholders, namely the general public, healthcare professionals, such as physicians and pharmacists and genetic laboratories. We are also engaged into collaborative projects to identify and propose solutions to resolve practical ethical problems encountered when working in the field of genetics, including but not limited to direct-to-consumer genetic testing, sharing good practice in the field of genetics and improvement of ethical decision-making in practice.

### **PARTICIPATION IN RECENT INTERNATIONAL RESEARCH PROJECTS**

- **H2020-2015 “UPGx” (668853)**, Ubiquitous Pharmacogenomics, **Co-investigator and Work Package leader**, 2016-2021.
- **COST “CHIP\_ME” (IS1303)**, Citizen's Health through public-private Initiatives: Public health, Market and Ethical perspectives, **Member, Management and Steering Committees**, 2013-2017.

- **FP7-2012-REGPOT, “SERBORDISinn”**, Strengthening the Research Potential of IMGGE through Reinforcement of Biomedical Science of Rare Diseases in Serbia – en route for innovation, **Chairman, Steering committee**, 2013-2016.

- **GSRT-2011-COOPERATION, “eMoDiA” (SYN11-0415)**, Electronic Molecular Diagnostics Assistant, **Principal Investigator**, 2013-2015.

- **FP7-2012-HEALTH, “RD-CONNECT” (305444)**, An integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research, **Co-investigator and Governing Board member**, 2012-2019.

- **FP7-2011-REGPOT, “SEE-DRUG” (7285950)**, Establishment of a centre of excellence for structure-based drug target characterization: Strengthening the research capacity of South-Eastern Europe, **Co-investigator and Steering committee member**, 2012-2014.

- **FP7-2008-HEALTH, “GEN2PHEN” (200574)**, Genotype-to-Phenotype databases: A holistic approach, **Co-investigator and Steering committee member**, 2008-2013.

- **RPF-2006, “HU-PHARMGK” (ΠΔΕ\_046-02)**, Pharmacogenomic analysis of beta-type hemoglobinopathies patients under hydroxyurea treatment, **Principal investigator**, 2006-2009.

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## SCIENTIFIC PUBLICATIONS

- Peer-reviewed papers in international scientific journals: 213
- Books: 13
- Book chapters: 32
- Conference Proceedings: >250

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## INTERVIEWS

- **DNAdigest**  
Genomic Medicine Alliance (Part 1) (in English)  
( <http://dnadigest.org/dnadigest-interviews-genomic-medicine-alliance-part-1/> )  
Genomic Medicine Alliance (Part 2) (in English)  
( <http://dnadigest.org/dnadigest-interviews-genomic-medicine-alliance-part-2/> )
- **IONIANTV**  
EMBIA: research from the future (in Greek)  
<https://www.youtube.com/watch?v=iwhw9ZYZclQ>
- **BHMASCIENCE**  
DNA guided drug prescription (in Greek)  
<https://www.tovima.gr/2018/04/27/science/syntagografisi-me-odigo-to-dna&nbsp;>
- **WIRED**  
Ernährung angepasst an eure DNA – geht das? (In German)  
<https://www.wired.de/collection/science/ernaehrung-angepasst-eure-dna-geht-das.>
- **Biomedicine Hub Spotlight**  
<https://www.karger.com/Journal/BlogPostDetail/7dfcd53a-a88a-4886-af7b-407b613275dd>
  
- **EurekaAlert – The Global Source for Science News**  
[https://www.eurekaalert.org/pub\\_releases/2017-11/mali-db111617.php&nbsp;](https://www.eurekaalert.org/pub_releases/2017-11/mali-db111617.php&nbsp;)

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## TEACHING ACTIVITIES

**Undergraduate** □



- Cell Biology (1st semester)
- Molecular Biology – Genetics (4th semester)
- Molecular Genetics and Pharmacogenomics (4th semester)
- Bioethics and Legislation (4th and 9th semester)

### **Postgraduate**

- Molecular Diagnostics and Pharmacogenomics
- Bioinformatics

### **EDITORIAL ACTIVITIES**

- Editor-In-Chief; Translational and Applied Genomics book series (Elsevier B.V.)
- Communicating Editor, Human Mutation
- Associate editor, Personalized Medicine
- Associate Editor, Public Health Genomics
- Associate Editor, Human Genomics
- Associate Editor, Frontiers in Pharmacogenetics and Pharmacogenomics
- Associate Editor, Hemoglobin
- Associate Editor, Recent Patents in DNA and gene sequences
- Editorial Board Member, EBioMedicine
- Editorial Board Member, Pharmacogenomics
- Editorial Advisor, Encyclopedia of Life Science

### **SELECTED PUBLICATIONS**

1. Chondrou V, Stavrou EF, Markopoulos G, Kouraklis-Symeonidis A, Fotopoulos V, Symeonidis A, Vlachaki E, Chalkia P, **Patrinios GP**, Papachatzopoulou A, Sgourou A. (2018). Impact of ZBTB7A hypomethylation and expression patterns on treatment response to hydroxyurea. ***Hum Genomics*. 12(1): 45.**

2. Sukasem C, Katsila T, Tempark T, **Patrinios GP**, Chantratita W. (2018). Drug-Induced

Stevens-Johnson Syndrome and Toxic Epidermal Necrolysis Call for Optimum Patient Stratification and Theranostics via Pharmacogenomics.

**Annu Rev Genomics Hum Genet**

19

: 329-353.

3. Özdemir V, **Patrinos GP**. (2017). David Bowie and the Art of Slow Innovation: A Fast-Second Winner Strategy for Biotechnology and Precision Medicine Global Development.

**OMICS**

21(11)

: 633-637.

4. **Patrinos GP**. (2018). Population pharmacogenomics: impact on public health and drug development. **Pharmacogenomics**. 19(1): 3-6.

5. Chondrou V, Kolovos P, Sgourou A, Kourakli A, Pavlidaki A, Kastrinou V, John A, Symeonidis A, Ali BR, Papachatzopoulou A, Katsila T, **Patrinos GP**. (2017). Whole transcriptome analysis of human erythropoietic cells during ontogenesis suggests a role of VEGFA gene as modulator of fetal hemoglobin and pharmacogenomic biomarker of treatment response to hydroxyurea in  $\beta$ -type hemoglobinopathy patients.

**Hum Genomics**

11(1)

: 24.

6. Vermeulen C, Geeven G, de Wit E, Verstegen MJAM, Jansen RPM, van Kranenburg M, de Bruijn E, Pulit SL, Kruisselbrink E, Shahsavari Z, Omrani MD, Zeinali F, Najmabadi H, Katsila T, Vrettou C, **Patrinos GP**, Traeger-Synodinos J, Splinter E, Beekman JM, Kheradmand Kia S, te Meerman GJ, Ploos van Amstel JK, de Laat W. (2017). Sensitive Monogenic Noninvasive Prenatal Diagnosis by Targeted Haplotyping.

**Am J Hum Genet**

101(3)

: 326-339

7. Lakiotaki K, Kanterakis A, Kartsaki E, Katsila T, **Patrinos GP**, Potamias G. (2017). Exploring public genomics data for population pharmacogenomics.

**PLoS One**

12(8)

: e0182138.

8. Viennas E, Komianou A, Mizzi C, Stojiljkovic M, Mitropoulou C, Muilu J, Vihinen M, Grypioti P, Papadaki S, Pavlidis C, Zukic B, Katsila T, van der Spek PJ, Pavlovic S, Tzimas G, **Patrinos GP**. (2017). Expanded national database collection and data coverage in the FINDbase worldwide database for clinically relevant genomic variation allele frequencies.

**Nuclei**

**c Acids Res**

**45(D1)**

: D846-D853.

9. Balasopoulou B, Stanković B, Panagiotara A, Nikčević G, Peters BA, John A, Mendrinou E, Stratopoulos A, Legaki AI, Stathakopoulou V, Tsolia A, Govaris N, Govari S, Zagoriti Z, Poulas K, Kanariou M, Constantinidou N, Krini M, Spanou K, Ali BR, Borg J, Drmanac R, Chrousos G, Pavlovic S, Roma E, Zukic B, Patrinos GP, Katsila T. (2016). Novel genetic risk variants for pediatric celiac disease

**Hum Genomics**

10(1): 34.

10. Mizzi C, Dalabira E, Kumuthini J, Dzimiri N, Balogh I, Başak N, Böhm R, Borg J, Borgiani P, Bozina N, Bruckmueller H, Burzynska B, Carracedo A, Cascorbi I, Deltas C, Dolzan V, Fenech A, Grech G, Kasiulevicius V, Kádaši Ľ, Kučinskas V, Khusnutdinova E, Loukas YL, Macek M Jr, Makukh H, Mathijssen R, Mitropoulos K, Mitropoulou C, Novelli G, Papantoniou I, Pavlovic S, Saglio G, Setric J, Stojiljkovic M, Stubbs AP, Squassina A, Torres M, Turnovec M, van Schaik RH, Voskarides K, Wakil SM, Werk A, Del Zompo M, Zukic B, Katsila T, Lee MT, Motsinger-Rief A, McLeod HL, van der Spek PJ,

**Patrinos GP**

. (2016). A European spectrum of pharmacogenomic biomarkers: Implications for clinical pharmacogenomics.

**PLoS One**

11: e0162866

11. Lakiotaki K, Kartsaki E, Kanterakis A, Katsila T, Patrinos GP, Potamias G. (2016). ePGA: A web-based information system for translational pharmacogenomics.

**PLoS One**

. 11: e0162801.

12. Chalikiopoulou C, Tavianatou AG, Sgourou A, Kourakli A, Kelepouri D, Chrysanthakopoulou M, Kanelaki VK, Mourdoukoutas E, Siamoglou S, John A, Symeonidis A, Ali BR, Katsila T, Papachatzopoulou A, Patrinos GP. (2016). Genomic variants in the ASS1 gene, involved in the nitric oxide biosynthesis and signaling pathway, predict hydroxyurea treatment efficacy in compound sickle cell disease/ $\beta$ -thalassemia patients.

**Pharmacogenomics**

. 17: 393-403.

13. Perkins A, Xu X, Higgs DR, Patrinos GP, Arnaud L, Bieker JJ, Philipson S; KLF1 Consensus Workgroup. (2016). Krüppeling erythropoiesis: an unexpected

broad spectrum of human red blood cell disorders due to KLF1 variants.

**Blood**

. 127: 1856-1862.

14. Katsila T, Konstantinou E, Lavda I, Malakis H, Papantoni I, Skondra L, **Patrinos GP**. (2016). Pharmacometabolomics-aided Pharmacogenomics in Autoimmune Disease.

**EBioMedicine**

. 5: 40-45.

15. Pavlidis C, Lanara Z, Balasopoulou A, Nebel JC, Katsila T, **Patrinos GP**. (2015). Meta-Analysis of Genes in Commercially Available Nutrigenomic Tests Denotes Lack of Association with Dietary Intake and Nutrient-Related Pathologies.

**OMICS**

19: 512-520.

16. Karageorgos I, Giannopoulou E, Mizzi C, Pavlidis C, Peters B, Karamitri A, Zagoriti Z, Stenson P, Kalofonos HP, Drmanac R, Borg J, Cooper DN, Katsila T, **Patrinos GP**. (2015). Identification of cancer predisposition variants using a next generation sequencing-based family genomics approach.

**Hum Genomics**

. 9: 12.

17. Manolio TA, Abramowicz M, Al-Mulla F, Anderson W, Balling R, Berger AC, Bleyl S, Chakravarti A, Chantratita W, Chisholm RL, Dissanayake VH, Dunn M, Dzau VJ, Han BG, Hubbard T, Kolbe A, Korf B, Kubo M, Lasko P, Leego E, Mahasirimongkol S, Majumdar PP, Matthijs G, McLeod HL, Metspalu A, Meulien P, Miyano S, Naparstek Y, O'Rourke PP, **Patrinos GP**, Rehm HL, Relling MV, Rennert G, Rodriguez LL, Roden DM, Shuldiner AR, Sinha S, Tan P, Ulfendahl M, Ward R, Williams MS, Wong JE, Green ED, Ginsburg GS. (2015). Global implementation of genomic medicine: We are not alone.

**Science Transl. Med**

. 7: 290ps13.

18. Kampourakis K, Vayena E, Mitropoulou C, Borg J, van Schaik RH, Cooper DN, **Patrinos GP**. (2014). Key challenges for next generation pharmacogenomics.

**EMBO Rep**

, 15: 472-476.

19. Mizzi C, Mitropoulou C, Mitropoulos K, Peters B, Agarwal MR, van Schaik RH, Drmanac R, Borg J, **Patrinos GP**. (2014). Personalized pharmacogenomics profiling using whole genome sequencing. **Pharmacogenomics**, 15: 1223-1234.

20. Giardine B, Borg J, Viennas E, Pavlidis C, Moradkhani K, Joly P, Bartsakoulia M, Riemer C, Miller W, Tzimas G, Wajcman H, Hardison RC, **Patrinos GP**. (2014).

Updates of the HbVar database of human hemoglobin variants and thalassemia mutations.

**Nucleic Acids Res**

, 42: D1063-D1069.

21. Papadopoulos P, Viennas E, Gkantouna V, Pavlidis C, Bartsakoulia M, Ioannou ZM, Ratbi I, Sefiani A, Tsaknakis J, Poulas K, Tzimas G, **Patrinos GP**

(2014). Developments in FINDbase worldwide database for clinically relevant genomic variation allele frequencies.

**Nucleic Acids Res**

, 42: D1020-D1026.

22. Sosnay PR, Siklosi KR, Van Goor F, Kaniecki K, Yu H, Sharma N, Ramalho AS, Amaral MD, Dorfman R, Zielenski J, Masica DL, Karchin R, Millen L, Thomas PJ, **Patrinos GP**, Corey M, Lewis MH, Rommens JM, Castellani C, Penland CM, Cutting GR. (2013). Defining the disease-liability of mutations in the cystic fibrosis transmembrane conductance regulator gene.

**Nature Genet**

45: 1160-1167.

23. Georgitsi M, **Patrinos GP**. (2013). Genetic databases in pharmacogenomics: the Frequency of Inherited Disorders database (FINDbase). **Meth**

**ods Mol Biol**

, 1015: 321-336.

24. Borg J, Phylactides M, Bartsakoulia M, Tafrali C, Lederer C, Papachatzopoulou A, Kourakli A, Stavrou E, Christou S, Hou J, Karkabouna S, Lappa-Manakou C, Özgür Z, van IJcken W, von Lindern M, Grosveld FG, Georgitsi M, Kleanthous M, Philipsen S, **Patrinos GP**

. (2012). KLF10 gene expression is associated with high fetal hemoglobin levels and with response to hydroxyurea treatment in  $\beta$ -hemoglobinopathy patients.

**Pharmacogenomics**

13: 1487-1500.

25. **Patrinos GP**, Cooper DN, van Mulligen E, Gkantouna V, Tzimas G, Tatum Z, Schultes E, Roos M, Mons B. (2012). Microattribution and nanopublication as means to incentivize the placement of human genome variation data into the public domain. **Hum**

**Mutat**

3: 1503-1512.

3

26. Reydon TA, Kampourakis K, **Patrinos GP**. (2012). Genetics, genomics and society: the responsibilities of scientists for science communication and education.

**Per Med**

9: 633-643.

27. Mette L, Mitropoulos K, Vozikis A, **Patrinos GP**. (2012). Pharmacogenomics and public health: Implementing **Hum** populationalized

medicine.

**Pharmacogenomics**

13: 803-813.

28. Dalgleish R, Oetting WS, Auerbach AD, Beckmann JS, Cambon-Thomsen A, Devereau A, Greenblatt MS, **Patrinos GP**, Taylor GR, Vihinen M, Brookes AJ (2011). Clarity and claims in variation/mutation databasing. **Nature**

**Biotechnol**

790-792.

29:

29. Kricka LJ, Fortina P, Mai Y, **Patrinos GP**. (2011). Direct-to-consumer genetic testing: A view from Europe. **Nature Rev Genet** 12: 670.

30. Squassina A, Manchia M, Borg J, Congiu D, Costa M, Georgitsi M, Chillotti C, Ardaù R, Mitropoulos K, Severino G, Del Zompo M, **Patrinos GP**. (2011). Evidence for association of an ACCN1 gene variant with response to lithium treatment in Sardinian patients with bipolar disorder. **Pharmacogenomics**

**genomics**

12(11): 1559-1569.

31. Giardine B, Borg J, Higgs DR, Peterson KR, Philipson S, Maglott D, Singleton BK, Anstee DJ, Basak AN, Clark B, Costa FC, Faustino P, Fedosyuk H, Felice AE, Francina A, Galanello R, Gallivan MV, Georgitsi M, Gibbons RJ, Giordano PC, Harteveld CL, Hoyer JD, Jarvis M, Joly P, Kanavakis E, Kollia P, Menzel S, Miller W, Moradkhani K, Old J, Papachatzopoulou A, Papadakis MN, Papadopoulos P, Pavlovic S, Perseu L, Radmilovic M, Riemer C, Satta S, Schrijver I, Stojiljkovic M, Thein SL, Traeger-Synodinos J, Tully R, Wada T, Wayne JS, Wiemann C, Zukic B, Chui DH, Wajcman H, Hardison RC, **Patrinos GP**. (2011). Systematic documentation and analysis of human genetic variation in hemoglobinopathies using the microattribution approach.

**Nature Genet**

43: 295-301.

32. Georgitsi M, Viennas E, Gkantouna V, van Baal S, Petricoin EF, Poulas K, Tzimas G, **Patrinos GP**. (2011). FINDbase: A worldwide database for genetic variation allele frequencies updated.

**Nucleic Acids Res**

39:D926-D932.

33. Borg J, Papadopoulos P, Georgitsi M, Gutierrez L, Grech G, Fanis P, Phylactides M, Verkerk AJ, van der Spek PJ, Scerri CA, Cassar W, Galdies R, van Ijcken W, Özgür Z, Gillemans N, Hou J, Grosveld FG, von Lindern M, Felice AE, **Patrinos GP**, Philipson S. (2010). Haploinsufficiency for the erythroid transcription factor KLF1 causes Hereditary Persistence of Fetal Hemoglobin.

**Nature Genetics**

42:801-805.