SURNAME: TZETIS
NAME: MARIA
NATIONALITY: GREEK AND CANADIAN
HOME ADDRESS: PAPARIGOPOULOU 45, ATHENS 15343, GREECE.
TEL: (301) 600 3673, mobile +306977656415
WORK ADDRESS: DEPT OF MEDICAL GENETICS, NATIONAL KAPODISTRIAN UNIVERSITY OF ATHENS, MEDICAL SCHOOL, ST. SOPHIA'S CHILDREN'S HOSPITAL, ATHENS 11527.
TEL. / FAX: (301) 779 5762 **email:** mtzetis @med.uoa.gr
Current position: Professor in Genetics, Department of Medical Genetics, Medical School, National and Kapodistrian University of Athens.

EDUCATION

UNIVERSITY:

1975-1979: B.Sc. Biology, McMaster University, Dept. of Biology, Ontario, Canada.
1989-1992: D.Phil. Thesis entitled "The molecular basis of normal HbA2 (type 2) β-thalassemia in Greece" University of Athens, Medical School.

POSITIONS:

- **1980-1982:** Research Assistant, Ontario Cancer Institute, Ontario, Canada.
- **1982-1984:** Research Assistant, Dept. of Medical Genetics, University of Toronto, Canada.
- **1984-2004:** Research Fellow, First Dept. of Pediatrics, National & Kapodistrian University of Athens, St. Sophia's Children's Hospital, Athens 11527.
- **2004- 2009:** Lecturer in Genetics, Department of Medical Genetics, Medical School, National & Kapodistrian University of Athens.
- **2009-2016:** Assistant Professor, Department of Medical Genetics, Medical School, National & Kapodistrian University of Athens.
- **2016-present:** Associate Professor of Genetics, Department of Medical Genetics, Medical School, National & Kapodistrian University of Athens

SUMMARY OF RESEARCH ACTIVITIES

1982-84: As research assistant to Professor H. Willard, I was involved in the preparation of mouse/ human cell hybrids for use in experiments to investigate the mechanisms of X-inactivation and X-chromosome mapping.
1984-present:Characterization of the molecular basis of the thalassemia syndromes in Greece, mainly unusual forms of β-thalassemias (β-silentI and II) and δ-thalassemias and application to carrier and prenatal diagnosis and phenotype/genotype correlation.
1992-present: Characterization of the mutations in the CFTR gene underlying cystic fibrosis in Greek CF patients and in addition haplotype analysis with the aim of offering prenatal diagnosis, carrier screening to family-members of affected individuals (both already initiated), and assessing phenotype/ genotype correlation.
1993: Characterization of the microsatellite haplotypes in Greek Wilson disease families (with application for preclinical and prenatal diagnosis) and mutation identification causative for the disease. Member of the EUROWILSON consortium funded by Wilson Disease: Creating a European Clinical Database and designing randomised controlled clinical trials. (Sixth Framework Programme, Project No. LSHM-CT2004 503430).
1997: Molecular diagnosis for atypical CFTR cases such as male infertility, bronchiectasis and pancreatitis.
1998: Expression studies (RT-PCR) of splicing mutations in the CFTR gene using nasal epithelial cells from CF patients. Genotype/ Phenotype correlation
1998: Molecular methodology for mutation identification in single cells with the aim for developing Pre-implantation Genetic Diagnosis (PGD) for Thalassemia syndromes and Cystic Fibrosis.
2000: Study of the promoter region of the UGT1A1 gene in patients with Gilbert syndrome.

2001: Development of a novel microarray methodology for the study of SNPs in the promoter region of the TNF-a gene, a1AT, TGFbeta1. Their association with obstructive pulmonary disease, bronchiectasis, idiopathic osteoarthritis and sarcoidosis in Greek patients.
2003: Development of **NanoChip** technology (Nanochip Molecular Biology Workstation-NMW1000 NanoChip™) for the screening of the most common mutations of the CFTR and ATP7A genes.
2003-present: Case-Control Study of mutations in the ESR1, ESR2, AR and UGT1A1 genes in women with different stages of Breast cancer. Case/ control studies on the genetic risk factors predisposing patients to stroke. Study of patients with eye disorders: Retinitis Pigmetosa, Startgard's, Leber Congenital Amaurosis. Mutation analysis of *RPE65, ABCA4* and *RHO* genes. Study of Neurofibromatosis type 1 (NF1) and development of targeted next generation sequencing panel for the study of RASopathies. Diagnostic and research application of array Comparative Genome Hybridization (aCGH) (AGILENT oligo-arrays) in cases of MR/DD, autism and schizophrenia, Acute Lymphoblastic Leukemia.

COOPERATION WITH SCIENTIFIC CENTERS ABROAD

1. Institute of Molecular Biology, Cagliary, Sardegna, Italy.
2. Centro de Genetica Humana, Instituto Nacional, Lisboa, Portugal.
3. Centre of Human Genetics, University of Leuven, Belgium
4. University of Toronto, Sick Kids Hospital; Cystic Fibrosis Mutation Database/ Genetic Analysis Consortium (CFMDB: www.genet.sickkids.on.ca/cftr)
5. Cystic Fibrosis Expression Network/ Working Group on CFTR Expression http://central.igc.gulbenkian.pt/cftr/VR/index.html
6. Eurowilson: European clinical database for the purpose of collecting clinical information and molecular diagnosis for patients with Wilson disease and designing randomized controlled clinical trials. www.eurowilson.org
7. *«A Consensus Conference organized by the European Cystic Fibrosis Society (www.ecfsoc.org), with the partnership of the European Society of Human Genetics (www.eshg,org), and the EuroGentest Network of Excellence (www.eurogentest.org)»* , Garda, Italy, 23-24, 2007

PhD Supervision: National & Kapodistrian University of Athens, 27 PhDs

FUNDED RESEARCH PROJECTS

EU: 3 projects ***1.***Cystic Fibrosis and Pulmozyme therapy, EPET II, 1994-1998; ***2.*** Development of new biotechnological approaches for the diagnosis, prevention and treatment of Hereditary and acquired diseases of Greece, EPET II, 1994-1998; ***3.*** CF CHIP: Novel genechip technology for simplified detection of molecularly heterogeneous genetic diseases: Detection of Cystic Fibrosis as a model, Project funded by the European Union Fifth Framework Programme: Quality of Life and Management of Living Resources, Project number: QLK3-CT-2001-01982. Duration: 36 months. Starting Date: 21 February 2001, http://www.nmrc.ie/projects/cf-chip/)
4. EYD EPANEK 2014-2020: Research Program, Competitiveness-Entrepreneurship-Innovation. RESEARCH-CREATE-INNOVATE, Title: Induced Pluripotent Stem Cells for the cellular treatment of osteoarthritis (acronym: iStemTheOS), project code: T1EDK-00128 (NKOA: total: 244.766,65 euro), starting date: 17/10/2018, Expiration date: 17/10/2021 (36 months), Principal researcher for the Medical School (NKUA) (ref. Research: 15361)

Invited Lectures (to international conferences: 12/ Abstracts at international meetings: **136**
Abstracts Greek meetings: 50

Summary of (foreign language) publications

Total number: 124 (ISSN/SCI), 6 (not in ISSN/SCI), 74 (journal supplements), 2 (chapters in books). (**TOTAL: 206**)
Total IF: of 124 ISSN/ ISCI publications: **496.676/ Average:** 4.14 (ISSN/ SCI)
***h* index**: 32 (Scopus), 39 (Google Scholar)/ **Citations:** 4372 (Scopus), 6207 (Google Scholar