I. Curriculum Vitae

Date of Birth : February 2, 1963
Place of Birth : Chania – Crete, Greece
Marital Status : Married

Academic Summary

Education

Bachelor of Science, Degree in Biology (September 1980-March 1985), University of Patras, Patras, Greece.
Doctor of Philosophy, Degree in Biology (April 1985-December 1989), University of Patras, Patras, Greece.

Positions held

January 1987-December 1989: Participation in a project supported by the Greek General Secretariat of Research and Technology, dealing with the experimental approach of the speciation theory.

July 1989-March 1990: Researcher in the Institute of Marine Biology of Crete.

April 1990-May 1993: Postdoctoral Researcher in the Institute of Molecular Biology and Biotechnology (IMBB) of Crete.

June 1993-April 1997: Postdoctoral Researcher in the European Molecular Biology Laboratory (EMBL), Heidelberg, Germany.

September 1994-March 1997: Visitor Scientist in BIOZENTRUM, University of Basel, Basel, Switzerland.

May 1997-August 1999: Associate Researcher in the Department of Biology, University of Crete.

Associate Researcher in the Department of Neurology and Sensory Organs, School of Medicine, University of Crete.

September 1999-November 2004: Lecturer of Genetics, Department of Agricultural Biotechnology, Agricultural University of Athens.

December 1999-December 2004: Visiting Researcher in the Laboratory of Population and Evolutionary Genetics, Department of Biology, University of Crete.

December 2004-April 2006: Lecturer in Human Molecular Genetics, School of Medicine, University of Crete.

September 2007-October 2013: Assistant Professor in Human Molecular Genetics, School of Medicine, University of Crete.

October 2013-September 2018: Associate Professor in Human Molecular Genetics, School of Medicine, University of Crete.

April 2016-present: Head of the Molecular Pathology and Human Genetics Section. Internal Medicine Laboratory, School of Medicine, University of Crete.

January 2019-present: Professor in Human Molecular Genetics, School of Medicine, University of Crete.

II . *Ad Hoc* reviewer in scientific journals

1. Advances in Hematology
2. American Journal of Human Biology
3. American Journal of Tropical Medicine and Hygiene
4. Archives of Medical Research
5. Arthritis and Rheumatism
6. Arthritis Research and Therapy
7. Annals of Human Genetics
8. Annals of the Rheumatic Diseases
9. Austin Journal of Clinical Immunology
10. Autoimmunity
11. African Journal of Biotechnology
12. Bioscience Reports
13. BioMed Research International
14. BMC Genetics
15. BMC Medical Genetics
16. Clinical Biochemistry
17. Clinical and Developmental Immunology
18. Clinical Experimental Immunology
19. Clinical Genetics
20. Clinical Rheumatology
21. Current Drug Metabolism
22. Current Genomics
23. Current Rheumatology Reviews
24. Diabetes Research and Clinical Practice
25. Disease Markers
26. European Journal of Human Genetics
27. European Journal of Entomology
28. European Journal of Pediatrics
29. Frontiers in Immunology
30. Gene Reports
31. Genes and Immunity
32. Genetica
33. Global Research Journal of Microbiology
34. Human Immunology
35. Human Molecular Genetics
36. Immunologic Research

37. Immunological Investigations
38. Immunology
39. Indian Journal of Medical Research
40. Interdisciplinary Perspectives on Infectious Diseases
41. International Journal of Clinical Rheumatology
42. International Journal of Immunogenetics
43. International Journal of Medical Sciences and Biotechnology
44. International Journal of Molecular Sciences
45. International Journal of Rheumatic Diseases
46. ISRN Genetics
47. Joint Bone Spine
48. Journal of Blood Disorders & Transfusion
49. Journal of Clinical Laboratory Analysis
50. Journal of Investigative Medicine
51. Journal of Medical Genetics and Genomics
52. Journal of Medicine & Biological Studies
53. Journal of Occupational Medicine and Toxicology
54. Journal of Ophthalmology
55. Journal of Oral Health Care
56. Journal of Pharmacy and Pharmacological Research
57. Lupus
58. Lung
59. Molecular and Cellular Probes
60. Molecular Biology Reports
61. Molecular Genetics & Genomics
62. Molecular Medicine
63. Nature Reviews in Rheumatology
64. Nitric Oxid: Biology and Chemistry
65. Oncology Letters
66. Pediatrics International
67. Pediatric Diabetes
68. Pediatric Rheumatology
69. PLoS ONE
70. Postgraduate Medical Journal
71. Prudence Journal of Medicine and Medical Sciences
72. Rheumatica Acta: Open Access
73. Rheumatology
74. Rheumatology International
75. Scandinavian Journal of Rheumatology
76. The Journal of Rheumatology
77. The Journal of Clinical Investigation
78. The Open Rheumatology Journal.
79. The Pharmacogenomics Journal
80. The Tohoku Journal of Experimental Medicine
81. World Journal of Gastroenterology

III. Reviewer in scientific institutes and funding organizations

1. Dutch Arthritis Association – Reumafonds, the Netherlands
2. Welcome Trust, UK
3. European University of Cyprus

IV. Member of the editorial board in scientific journals

1. African Journal of Biotechnology
2. Clinical and Developmental Immunology
3. Dataset Papers in Science
4. Dataset Papers in Medicine
5. Frontiers in Immunology
6. International Journal of Medical Genetics
7. International Journal of Medicine and Biomedical Research
8. International Scholarly Research Notices
9. Rheumatic and Musculoskeletal Diseases

V. Publications in peer-reviewed journals

* Corresponding author

1. **Goulielmos, G.N.,** Kilias, G., S.N. Alahiotis (1986). Adaptation of *Drosophila* enzymes to temperature. V. Heat shock effect on the malate dehydrogenase of *Drosophila melanogaster*. *Comp. Biochem. and Physiol*. 85 B(1): 229-234.
2. Alahiotis, S.N.**, Goulielmos, G.N.**, G. Kilias (1987). Reactions of *Drosophila* enzymes to temperature. Potential adaptive regimes. In Isozymes: Current Topics in Biological and Medical Research (Eds. C. Market, J. Scandalios and G. Whitt, New York, NY), vol 15: 153-178**.**
3. Karvountzi, E., **Goulielmos, G.N.,** Kalpaxis, D., S.N. Alahiotis (1989). Adaptation of *Drosophila* enzymes to temperature. VI. Acclimation studies using the malate dehydrogenase (MDH) and lactate dehydrogenase (LDH) systems. *J. Thermal Biol.* 14(1): 55-61.
4. **Goulielmos, G.N.** and S.N. Alahiotis (1989). Induction of the malate dehydrogenase and acetylcholinesterase by ecdysone and heat shock in *Drosophila* ovaries. *Insect Biochem.* 19(4): 393-399.
5. **Goulielmos, G.N.** and S.N. Alahiotis (1989). Interspecific hybridization between *D. melanogaster* sibling species. Isozymic patterns (in interspecific hybrids) and reproductive relationships. *Genome* 30: 146-154.
6. Killias, G., **G.N. Goulielmos**, S.N. Alahiotis (1989). Interspecific hybridization between *D. melanogaster* sibling species. Fitness components. *Hereditas* 110: 267-274.
7. Tzimagiorgis, G., Leversha, M., Chroniary, K., **Goulielmos, G.N.,** Sargent, C., Ferguson-Smith, M., N.K. Moschonas (1993). Structure and expression analysis of a novel member of the human glutamate dehydrogenase (GLUD) gene family mapped to chromosome 10p 11.2. *Hum. Genet*. 91: 433-438.
8. **Goulielmos, G.N.,** Angelicheva, D., Kapsetaki, M., Manifava, M., Moschonas N. (1993). A chromosome 10p 11.2 GT-dinucleotide polymorphism at the GLUDP5 locus. *Hum. Mol. Genet.* 2 (8): 1328.
9. Moschonas, N, **Goulielmos, G.N.,** Lubyova, B., Manifava, M., Kapsetaki M. (1993). A chromosome 10q 23 GT-dinucleotide polymorphism associated with the GLUD1 locus. *Hum. Mol. Genet*. 2 (11): 1981.
10. **Goulielmos, G.N.,** Manifava, M., Moschonas N.K. (1993). Dinucleotide-repeat polymorphism at the GLUDP2. *Hum. Mol. Genet*. 2 (12): 2202.
11. **Goulielmos, G.N.** and Zouros (1995). Incompatibility analysis of male hybrid sterility in two *Drosophila* species: Lack of evidence for maternal, cytoplasmic or transposable element effects. *Am. Nat.* 145: 1006-1014.
12. **Goulielmos, G.N.,** Gounari, F., Remington, S., Muller, S., Haner, M., Aebi, U., Georgatos, S.D. (1996). Filensin and phakinin form beaded intermediate filaments and co-assemble de novo in non-lenticular cells. *J. Cell Biol.* 132 (4): 643-655.

13. **Goulielmos, G.N.,** Remington, S., Schwesinger, F., Georgatos, S.D., Gounari F. (1996). Contribution of the structural domains of filensin in polymer formation and filament distribution. *J. Cell Sci* 109: 447-456.
14. Tavosanis, G., Llamazares, S., **Goulielmos, G.N.** and C. Gonzalez (1997). Essential role for γ-tubulin in the acentriolar female meiotic spindle of *Drosophila*. *EMBO J.* 16(8): 1809-1819.
15. Georgatos, S.D., Gounari, F., **Goulielmos, G.N.,** and U. Aebi (1997). To bead or not to bead? Lens-specific intermediate filaments revisited. *J. Cell Sci*. 110 (21): 2629-2634.
16. ***Goulielmos, G.N.,** Cosmidis, N., Loukas, M., Tsakas, S. and E. Zouros (2001). Characterization of two Alcohol Dehydrogenase (*Adh*) loci from the olive fruit fly, *Bactrocera* (*Dacus) oleae* and implications for *Adh* duplication in dipteran insects. *J. Mol. Evol*. 52 (1): 29-39.
17. Bondinas, G.P., Loukas, M.G., **Goulielmos, G.N.** and D. Sperlich (2001). The actin loci in the genus *Drosophila*: Establishment of chromosomal homologies among five palaearctic *Drosophila obscura* species by in situ hybridization. *Chromosoma* 110:441-450.
18. Bondinas, G.P., Loukas, M.G., **Goulielmos, G.N.** and D. Sperlich (2002). The actin loci in the genus *Drosophila*: Establishment of chromosomal homologies among five neoarctic *Drosophila obscura* species by in situ hybridization. *Chromosoma* 111:256-266.
19. Cosmidis, N., Pepa, V., Loukas, M., **Goulielmos, G.N.** and E. Zouros (2002). Effect of acetone feeding on Alcohol Dehydrogenase (ADH) activity in *Bactrocera oleae*. *Heredity* 89:453-459.
20. Arhontaki, K., Eliopoulos, E, ***Goulielmos, G.N,** Kastanis, P., Tsakas, S., Loukas, M. and F. Ayala (2002). Functional constraints of CuZn superoxide dismutase in species of *Drosophila melanogaster* subgroup and phylogenetic analysis. *J. Mol. Evol*. 55:745-756.
21. ***Goulielmos, G.N.**, Cosmidis, N., Theodorakopoulou, M.E., Loukas, M. and E. Zouros (2003). Tracing the history of an enzyme polymorphism: the case of alcohol dehydrogenase-2 (*Adh-2*) of the olive fruit fly *Bactrocera oleae*. *Mol Biol Evol* 20:293-306.
22. ***Goulielmos, G.**N., Loukas, M., Bondinas, G. and Zouros, E. (2003). Exploring the evolutionary history of the alcohol dehydrogenase gene (*Adh*) duplication in species of the family Tephritidae. *J. Mol. Evol*. 57:170-180.
23. Kastanis, P., Eliopoulos, E., ***Goulielmos, G.,** Tsakas, S. and Loukas, M. (2003). Macroevolutionary relationships of species of *Drosophila melanogaster* group based on mtDNA sequences. *Molecular Phylogenetics Evolution* 28:518-528.
24. ***Goulielmos, G.N.,** Eliopoulos, E., Arhontaki, K., Tserpistali, K., Tsakas, S. and Loukas, M. (2003). *Drosophila* Cu, Zn superoxide dismutase gene confers resistance to paraquat in *Escherichia coli*. *Biochem. Biophys. Res. Commun.* 308:433-438.
25. ***Goulielmos, G.N.,** Eliopoulos, E., Loukas, M. and Tsakas, S. (2004). Functional constraints of 6-phosphogluconate dehydrogenase (6PGD) protein based on sequence and structural information. *J. Mol. Evol.* 59:358-371.
26. Eliopoulos, E., ***Goulielmos, G.N.** and Loukas, M. (2004). Functional constraints of alcohol dehydrogenase (ADH) of Tephritidae and relationships with other dipteran species. *J. Mol. Evol.* 58:493-505.
27. Mantziou, G, Antoniou, A, Poulakakis, N, **Goulielmos, G,** Tsigenopoulos, C, Pinou, T, and Mylonas, M. (2005**).** Isolation and characterization of six polymorphic microsatellite markers in the fresh water turtle *Mauremys rivulata* (Testudines: Geoemydidae). *Mol. Ecol. Notes* 5:727-729.

28. Poulakakis, N., **Goulielmos, G.,** Antoniou, A., Zouros, E., and Mylonas, M. (2005). Isolation and characterization of polymorphic microsatellite markers in the wall lizard *Podarcis erhardii* (Squamata: Lacertidae). *Mol. Ecol. Notes* 5:549-551.
29. ***Goulielmos, G.N.**, Cosmidis, N., Eliopoulos, E., Loukas, M., and Zouros, E. (2006). Cloning and structural characterization of the 6-phosphogluconate dehydrogenase (*6Pgd*) locus of the medfly *Ceratitis capitata* and the olive fruit fly *Bactrocera oleae*. *Biochem. Biophys. Res. Commun*. 341:721-727.
30. Bourikas, L., Kritikos, H., Sidiropoulos, P., **Goulielmos, G.** and Boumpas, D. (2006). Concomitant onset of Grave's disease and Rheumatoid arthritis following a serious life event. A case report and review of the literature. *J. Clin. Rheumatology* 12:326-327.
31. ***Goulielmos, G.N**., Fragouli, E., Aksentijevich, I., P. Sidiropoulos, Boumpas, D.T., and Eliopoulos, E (2006). Mutational analysis of the PRYSPRY domain of pyrin and implications for familial Mediterranean Fever (FMF). *Biochem. Biophys. Res. Commun.* 345:1326-1332.
32. Vazgiourakis, V., Sidiropoulos, P., Bertsias, G., Raptopoulou, A., Koutsounaki, E., Fragouli, E., Kritikos, H., Boumpas, D.T. and ***Goulielmos, G.N.** (2007). Association of the nitric oxide synthase gene (eNOS) polymorphism with increased risk for rheumatoid arthritis - but not for systemic lupus erythematosus - in population of Crete, a southern-eastern European Greek island. *Lupus* 16:867-874.
33. Sidiropoulos, P.I., **Goulielmos, G.N.,** Voloudakis, G., Petraki, E. and D.T. Boumpas (2007). Inflammasome and rheumatic diseases: evolving concepts. *Ann Rheum Dis* 67:1382-1389.
34. Fragouli E., E. Eliopoulos, E. Petraki, P. Sidiropoulos, I. Aksentijevich, E. Galanakis, H. Kritikos, A. Repa, G. Fragiadakis, D.T. Boumpas, and ***G.N. Goulielmos** (2008). Familial Mediterranean Fever (FMF): a genetic, biochemical and structural approach of the disease in cretan population, a population of "intermediate risk". *Clin Genetics* 73:152-159.
35. Galanakis, E., Kofteridis, D., Stratigi, K., Petraki, K., Vazgiourakis, V., Fragouli, E., Mamoulakis, D., Boumpas, D.T. and ***G.N. Goulielmos** (2008). Intron 4 polymorphism of the endothelial NO synthase gene is associated with both T1 and T2 diabetes in a genetically homogeneous population. *Hum Immunol* 69:279-283.
36. Ilia S, **Goulielmos G.N.**, Samonis G, Galanakis E. (2008). Host's response in acute otitis media: evidence of genetic susceptibility. *Pediatr Infect Dis J.* 27: 929-933.
37. Koutsounaki, E., **Goulielmos, G.N**., Koulentaki, M., Choulaki, C., Kouroumalis, E., and Galanakis, E. (2008). Mannose-Binding Lectin Polymorphisms and Outcome of Patients with Chronic Hepatitis C. *J. Clin. Immun.* 28:495-500.
38. Zervou, M.I., Sidiropoulos, P., Petraki, E., Vazgiourakis, V., Krasoudaki, E., Raptopoulou, A., Kritikos, H., Choustoulaki, E., Boumpas D.T. and ***G.N. Goulielmos** (2008). Association of a TRAF1 and a STAT4 gene polymorphism with increased risk for rheumatoid arthritis in a genetically homogeneous population. *Hum Immunol* 69:576-571.
39. Zervou, M.I., D. Mamoulakis, C. Panierakis, D. T. Boumpas and ***G.N. Goulielmos** (2008). STAT4: a risk factor for Type 1 Diabetes? *Hum Immunol* 69:647-650.
40. Cosmidis, N., **G. Goulielmos**, E. Eliopoulos and M. Loukas (2008). Selection at 6-PGD locus in laboratory populations of Bactrocera oleae. *Genet Res* 90(5):379-384.
41. Kofteridis, D, E. Krasoudaki, M. Kavousanaki, M.I. Zervou, C. Panierakis, D.T. Boumpas, ***G. N. Goulielmos** (2009). STAT4 is not associated with T2 diabetes in the genetic homogeneous population of Crete. *Genetic Testing and Molecular Biomarkers* 13:281-284.
42. Bertsias GK, Nakou M, Choulaki C, Raptopoulou A, Papadimitraki E, **Goulielmos G**, Kritikos H, Sidiropoulos P, Tzardi M, Kardassis D, Mamalaki C, Boumpas DT.

(2009). Genetic, immunologic, and immunohistochemical analysis of the programmed death 1/programmed death ligand 1 pathway in human systemic lupus erythematosus. *Arthritis Rheum* 60:207-218.

43. Panierakis C., **G.N. Goulielmos**, D. Mamoulakis, S. Maraki, E. Papavassileiou, D. T. Boumpas and E. Galanakis (2009). Staphylococcus aureus nasal carriage might be associated with vitamin D receptor polymorphisms in type 1 diabetes. *Int J Inf Diseases*, 13(6):e437-43.
44. Mamoulakis D., M. Bitsori, E. Galanakis, V. Vazgiourakis C. Panierakis, ***Goulielmos, G.N.** (2009). Intron 4 polymorphism of the endothelial nitric oxide synthase gene (eNOS) is not associated with early diabetic microangiopathy in type 1 diabetes. *Int. J Immunogenet*, 36(3):153-157.
45. Kurreeman F.A.S., **G.N. Goulielmos**, B. Z. Alizadeh, B. Rueda, M. Bevova, T. R. Radstake, E. Galanakis, N. Ortego, W. Verduyn, M.I. Zervou, B. Roep, E. Urcelay, D.T. Boumpas, B. P.C. Koeleman, T. W.J.Huizinga, R. E.M. Toes, J. Martin; AADEA Group; SLEGEN Consortium (2010). The TRAF1/C5 region is associated with multiple autoimmune diseases. *Ann Rheum Dis* 69:696-699.
46. Zervou, M.I., **G.N. Goulielmos**, F. Castro-Giner, A.D. Tosca, S. Krueger-Krasagakis (2009). STAT4 gene polymorphism is associated with psoriasis in the genetically homogeneous population of Crete, Greece. *Hum Immunol*, 70(9):738-741.
47. Eliopoulos DG, Mavroudi I, Pontikoglou C, Ximeri M, Stavroulaki E, Pyrovolaki K, Velegraki M, Spanoudakis M, **Goulielmos G**, Papadaki HA. (2009). The -509C/T polymorphism of transforming growth factor-β1 is associated with increased risk for development of chronic idiopathic neutropenia. *Eur J Haematology*, 83(6):535-540.
48. Panierakis C., **G.N. Goulielmos**, D. Mamoulakis, E. Petraki, E. Papavasiliou, and E. Galanakis (2009). Vitamin D receptor gene polymorphisms and susceptibility to type 1 diabetes in Crete, Greece. *Clin Immunol*, 1332:276-281.
49. Plant D., Flynn E., Eyre S., Mbarek H., Dieudé P., Cornelis F., Rantapää Dahlqvist S., **Goulielmos G.N.**, Boumpas D.T., Sidiropoulos P., Johansen J.S., Skjødt H., Lund Hetland M., Klareskog L., Raza K., Witte T., Worthington J. (2010). Investigation of potential non-HLA rheumatoid arthritis susceptibility loci in a European cohort increases the evidence for 10 markers. *Ann Rheum Dis*, 69:1548-1553.
50. Nakou M, Bertsias G, Stagakis I, Centola M, Tassiulas I, Hatziapostolou M, Kritikos I, **Goulielmos G.N.**, Boumpas DT, Iliopoulos D (2010). Gene Network Analysis of Bone Marrow Mononuclear Cells Reveals Activation of Multiple Kinase Pathways in Human Systemic Lupus Erythematosus. *PLoS ONE*, 5:e13351.
51. ***Goulielmos G.N**., Petraki E., Vassou D., Eliopoulos E., Iliopoulos D., Sidiropoulos P., Aksentijevich I., Kardassis, D., Boumpas, D.T. (2010). The role of the pro-apoptotic protein Siva in the pathogenesis of Familial Mediterranean fever: A structural and functional analysis. *Biochem. Biophys. Res. Commun.*402:141-146.
52. Eliopoulos, E., M.I. Zervou, A. Andreou, K. Dimopoulou, G. Voloudakis, N. Cosmidis, H. Mysirlaki, V. Vazgiourakis, P. Sidiropoulos, T. Newold, D.T. Boumpas and ***G.N. Goulielmos** (2011). Association of the PTPN22 R620W polymorphism with increased risk for SLE in the genetic homogeneous population of Crete. *Lupus* 10(5):501-506.
53. Karathanasis N., Steiakaki E., **Goulielmos G.N.** and Kalmanti M. (2011). The role of the MTHFR 677 and 1298 polymorphisms in Cretan children with acute lymphoblastic leukemia. *Genetic Testing Molecular Biomarkers*, 15(1-2):5-10.
54. Zervou M.I., **Goulielmos G.N.**, Castro-Giner F., Boumpas D.T., Tosca A.D. and Krueger-Krasagakis S. (2011). A *CD40* and a *NCOA5* gene polymorphism confer susceptibility to psoriasis in a Greek population. *Hum Immunol* 72:761-765.

55. Vazgiourakis V., Zervou M.I., Choulaki C., Yilmaz N., Bertsias G., Sidiropoulos P., Plant D., Trouw L.A., Toes R.E., Melissourgaki M., Kardassis D., Yavuz S., Boumpas D.T, ***Goulielmos G.N.** (2011). A common SNP in the CD40 region is associated with Systemic Lupus Erythematosus and correlates with enhanced CD40 expression. *Ann Rheum Dis* 70:2184-2190.
56. Zervou M.I., V. Vazgiourakis, N. Yilmaz, E. Kontaki, L.A. Trouw, M. Bicakcigi, RE Toes, D.T. Boumpas, S. Yavuz and ***G.N. Goulielmos** (2011). *TRAF1/C5, eNOS, C1q*, but not *STAT4* and *PTPN22* gene polymorphisms are associated with genetic susceptibility to systemic lupus erythematosus in Turkey. *Hum Immunol* 72:1210-1213.
57. Vazgiourakis V., M.I. Zervou, E. Eliopoulos, S. Sharma, P. Sidiropoulos, B.S. Franek, E. Myrthianou, M. Melissourgaki, T. Niewold, D.T. Boumpas and ***G.N. Goulielmos** (2013). Implication of VEGFR2 in Systemic Lupus Erythematosus: A structural biological and genetic approach. *Clin Exp Rheumatol* 31(1):97-102.
58. Mantaka A., **G.N. Goulielmos**, M. Koulentaki, O. Tsagournis, A. Voumvouraki, E. Kouroumalis (2012). Polymorphisms of genes related to endothelial cells are associated with Primary Biliary Cirrhosis (PBC) patients of Cretan origin. *Hum. Immunol*, 73(8):829-835.
59. Mastrodemou S., Vazgiourakis V., Velegraki M., Pavlaki K., **Goulielmos G.N.,** and H.A. Papadaki (2012). Clonal patterns of X-chromosome inactivation in peripheral blood cells of female patients with Chronic Idiopathic Neutropenia *Haematologica* 97(12):1931-1933.
60. Pesmatzoglou, M., M. Lourou, **G.N. Goulielmos**, and E. Stiakaki (2012). DNA methyltransferase 3B (*DNMT3B*) gene promoter and interleukin-1 receptor antagonist (*IL-1Ra*) polymorphisms in childhood immune thrombocytopenia. *Clin. Dev. Immunol.*, 352059. doi: 10.1155/2012/352059.
61. Niewold TB, **Goulielmos GN**, Tikly M, Assassi S. (2012). Autoimmune disease genetics. *Clin Dev Immunol*. 2012:262858. doi: 10.1155/2012/262858.
62. Tzagkaraki E, Sofocleous C, Helen FK, Dinopoulos A, **Goulielmos G**, Mavrou A, Sofia KT, Kanavakis E. (2013). Screening of UBE3A gene in patients referred for Angelman Syndrome. *Eur J Paediatr Neurol*. 17(4):366-73
63. Trouw L.A., N. Daha, F.A.S. Kurreeman, S. Böhringer, **G.N. Goulielmos**, H.J. Westra, A. Zhernakova, L. Franke, E.A. Stahl, E.W.N. Levarht, G. Stoeken-Rijsbergen, W. Verduijn, A. Roos, Y. Li, J.J. Houwing-Duistermaat, T.W. Huizinga and R.E. Toes (2013). Genetic variants of C1q are a risk for Rheumatoid Arthritis. *Clin. Exp. Immunol.* 173(1):76-83.
64. **Goulielmos G.N.**, G. Samonis, M. Apergi, M. Christofaki, A. Valachis, M.I. Zervou, D.P. Kofteridis (2013). C1q but not Mannose-binding lectin (Mbl-2) gene polymorphisms are associated with Type 2 diabetes in the genetically homogeneous population of the island of Crete in Greece. *Hum. Immunol* 74(7):878-81.
65. Dimopoulou D.G., M.I. Zervou, M. Trachana, E. Myrthianou, J. Pratsidou-Gertsi, D. Kardasis, A. Garyfallos and ***G.N. Goulielmos** (2013). Investigation of juvenile idiopathic arthritis susceptibility loci: Results from a Greek population. *Hum. Immunol*. 74(9):1194-810.
66. Cobb J., D. Plant, E. Flynn, H. Mbarek, P. Dieudé, F. Cornelis, L. Ärlestig, S. Rantapää Dahlqvist, **G.N. Goulielmos**, D.T. Boumpas, P. Sidiropoulos, J.S. Johansen, L.M. Ørnbjerg, M. Lund Hetland, L. Klareskog, A. Filer, C.D. Buckley, K. Raza, T. Witte, R.E. Schmidt, O. FitzGerald, D. Veale, S. Eyre, Worthington. (2013). Identification of the tyrosine-protein phosphatase non-receptor type 2 (PTPN2) as a rheumatoid arthritis susceptibility locus in Europeans. *PLoS ONE* 8(6):e66456.
67. Chrabot BS, Kariuki SN, Zervou MI, Feng X, Arrington J, Jolly M, Boumpas DT, Reder AT, **Goulielmos GN**, Niewold TB (2013). Genetic Variation near IRF8 is

Associated with Serologic and Cytokine Profiles in Systemic Lupus Erythematosus and Multiple Sclerosis. *Genes and Immunity* 14(8):471-8.

68. Zervou M.I., E. Myrthianou, I. Flouri, D. Plant, D.T. G. Chlouverakis, F. Castro-Giner, P. Rapsomaniki, DT Boumpas, A. Barton, P. Sidiropoulos, ***G.N. Goulielmos** (2013). Genetic association of rheumatoid arthritis patients treated with anti-TNF medication: Results from a homogeneous Greek population. *PLoS ONE* 8(9):e74375.
69. Karathanasis N., Steiakaki E., **Goulielmos G.N**. and Kalmanti M. (2013). The Effect of RFC G80A Polymorphism in Cretan children with acute lymphoblastic leukemia and its interaction with MTHFR C677T and A1298C polymorphisms. *Int J Lab Hematology* 36(4):425-30.
70. Ilia, S., **Goulielmos, G.N.,** Samonis, G., Galanakis, E. (2013). Polymorphisms in IL-6, IL-10, TNF-α, IFN-γ and TGF-β1 genes and susceptibility to acute otitis media in early infancy. *Pediatric Infectious Diseases Journal* 33(5):518-21.
71. Burska AN, Roget K, Blits M, Soto Gomez L, van de Loo F, Hazelwood LD, Verweij CL, Rowe A, **Goulielmos GN**, van Baarsen LG, Ponchel F. (2013). Gene expression analysis in RA: towards personalised medicine. *The Pharmacogenomics Journal* 14(2):93-106.
72. Niewold T.B., **Goulielmos G.N.**, Assassi S. (2014). Autoimmune disease genetics 2013. *J. Immun. Res.* 2014:487643. doi: 10.1155/2014/487643.
73. Repa A, G.K. Bertsias, E. Petraki, D. Vassou, C. Choulaki, K. Kambas, D.T. Boumpas, **G. Goulielmos**, P. Sidiropoulos (2015). Deregulated production of interleukin-1β upon activation of the NLRP3 inflammasome in patients with familial Mediterranean fever. *Hum. Immunol* 76(7):488-95.
74. ***Goulielmos G.N**., Zervou M.I., Burska A., Ponchel F. (2016a). Questions posed by personalized medicine achievements and promises. *J Clin Res Bioeth* 7:1-3.
75. **Goulielmos G.N.,** Eliopoulos E, Karatzanis A., Zervou M.I., Emmanuel Fountakis, Stylianos Velegrakis, Trias Thireou, Emmanuel P. Prokopakis (2016b). Mutational analysis and molecular interactions of the ACVRL1 protein and implications for Rendu-Osler-Weber (ROW) syndrome. *Int. J. New Technology Research* 2:32-36.
76. ***Goulielmos G.N.,** M.I. Zervou, E. Myrthianou, A. Burska, T.B. Niewold and F. Ponchel (2016c). Genetic data: the new challenge of personalized medicine, insights for Rheumatoid Arthritis patients. *GENE* 583(2):90-101.
77. Zervou M. I., ***Goulielmos G.N.**, Francesc Castro-Giner, Rena Hiotaki, Prodromos Sidiropoulos and Sabine Krueger-Krasagakis (2016). Interleukin-12B (IL-12B) and interleukin-23R (IL-23R) gene polymorphisms do not confer susceptibility to psoriasis in a Southern European population: A case-control study. *Int. J. New Technology Research* 2:67-70.
78. Budu-Aggrey A., J. Bowes, S. Lohr, S. Uebe, M.I. Zervou, P. Helliwell, A.W. Ryan, D. Kane, E. Korendowych, E. Giardina, J. Packham, R. McManus, O. FitzGerald, N. McHugh, F. Behrens, H. Burkhardt, U. Huffmeier, P. Ho, J. Martin, S. Castañeda, **Goulielmos G.N.**, A. Reis, A. Barton (2016). Replication of a distinct PsA risk variant at the IL23R locus. *Ann Rheum Dis*, 75(7):1417-8.
79. **G.N. Goulielmos**, R.C. Chiaroni-Clarke, D.G. Dimopoulou, M.I. Zervou, M. Trachana, P. Pratsidou-Gertsi, A. Garyfallos and J.A. Ellis (2016d). Juvenile idiopathic arthritis association of PTPN22 rs2476601 SNP is specific to females in a Greek population. *Pediatric Rheumatology* 14(1):25.
80. Myrthianou E., M.I. Zervou, A. Budu-Aggrey, E. Eliopoulos, D. Kardassis, D.T. Boumpas, N. Kougkas, A. Barton, P. Sidiropoulos, ***G.N. Goulielmos** (2017). Investigation of the genetic overlap between Rheumatoid Arthritis and Psoriatic Arthritis in a Greek population. *Scandinavian Journal of Rheumatology* 46(3):180-186.
81. Latsoudis H., M.-F. Mashreghi, J. Gruen, H.-D. Chang, B. Stuhlmueller, A. Repa, I. Gergiannaki, E. Kabouraki, D. Kardassis, T. Haeupl, A. Radbruch, P.

Sidiropoulos, T.B. Niewold, D.T. Boumpas, ***G.N. Goulielmos** (2017). ALTERED EXPRESSION LEVELS OF MIR-4520A ASSOCIATED WITH FAMILIAL MEDITERRANEAN FEVER (FMF). *Journal Cellular Physiology* 232(6):1326-1336.

82. Zervou M.I., Dorschner J.M., Ghodke-Puranik Y., Boumpas D.T., Niewold T.B. and ***Goulielmos G.N.** (2017). Association of IRF5 polymorphisms with increased risk for systemic lupus erythematosus in the population of Crete, a southern-eastern European Greek island. *GENE* 610:9-14.
83. Voskarides K, Stefanou C, Pieri M, Demosthenous P, Felekkis K, Arsali M, Athanasiou Y, Xydakis D, Stylianou K, Daphnis E, **Goulielmos G**, Loizou P, Savige J, Höhne M, Völker LA, Benzing T, Maxwell PH, Gale DP, Gorski M, Böger C, Kollerits B, Kronenberg F, Paulweber B, Zavros M, Pierides A, Deltas C. (2017). A functional variant in NEPH3 gene confers high risk of renal failure in primary hematuric glomerulopathies and of microalbuminuria in the general population. *PLoS ONE* 12(3):e0174274.
84. Michail Matalliotakis M., **George N. Goulielmos**, Charoula Matalliotaki, Alexandra Trivli, Ioannis Matalliotakis, Aydin M Arici (2017). Endometriosis in adolescents and young girls: Report a series of 85 cases. *Journal of Pediatric and Adolescent Gynecology* 30(5):568-570.
85. Matalliotakis, **Goulielmos G.N.**, M. I. Zervou, C. Matalliotaki, G. Koumantakis, I. Matalliotakis (2017). Familial predisposition of endometriosis in Greece. *Journal of Endometriosis and Pelvic Pain Disorders* 9(3):184 - 187.
86. Coucoutsi C, Emmanouil G, **Goulielmos G.N.**, Sfakianaki O, Koutroubakis IE, Kouroumalis EA. (2017). Prevalence of thiopurine methyltransferase gene polymorphisms in Cretan patients with inflammatory bowel disease. *European Journal of Gastroenterology & Hepatology*, 29(11):1284-1289.
87. Matalliotakis M, **Goulielmos GN**, Kalogiannidis I, Koumantakis G, Matalliotakis I, Arici A. (2017). Extra pelvic endometriosis: Retrospective analysis on 200 cases from two different centers. *European Journal of Obstetrics & Gynecology and Reproductive Biology*, 217:34-37.
88. Mavroudi I, Eliopoulos AG, Pontikoglou C, Pyrovolaki K, Damianaki A, Koutala H, Zervou MI, Ximeri M, Mastrodemou S, Kanellou P, **Goulielmos GN**, Papadaki HA (2017). Immunoglobulin and B-cell disturbances in patients with chronic idiopathic neutropenia. *Clinical Immunology* 183:75-81.
89. Ghodke-Puranik Y, Dorschner JM, Vsetecka DM, Amin S, Makol A, Ernste F, Osborn T, Moder K, Chowdhary V, Eliopoulos E, Zervou MI, **Goulielmos GN**, Jensen MA, Niewold TB (2017). Lupus-Associated Functional Polymorphism in PNP Causes Cell Cycle Abnormalities and Interferon Pathway Activation in Human Immune Cells. *Arthritis Rheumatology* 69(12):2328-2337.
90. Matalliotakis M, Zervou MI, Matalliotaki C, Arici A, Spandidos DA, Matalliotakis I, **Goulielmos GN** (2017). Genetic association study in a three-generation family with seven members with endometriosis from Crete (Greece). *Molecular Medicine Reports* 16(5):6077-6080.
91. Thanarajasingam U, Jensen MA, Dorschner JM, Wampler Muskardin T, Ghodke-Puranik Y, Purmalek M, Eliopoulos E, Zervou MI, **Goulielmos GN**, Howard M, Kaplan MJ, Niewold TB (2017). An ELANE gene polymorphism results in the appearance of decreased NETosis, inflammatory arthritis and recurrent viral infections with parvovirus. *Arthritis Rheumatology* 69(12):2396-2401.
92. Matalliotakis M., Zervou M.I., Matalliotaki C., Rahmioglu N., Koumantakis G., Kalogiannidis I., Prapas I., Zondervan K., Spandidos D.A., Matalliotakis I. and ***Goulielmos G.N.** (2017). The role of gene polymorphisms in endometriosis. *Molecular Medicine Reports* 16(5):5881-5886.
93. Matalliotakis M., Velegrakis A., **Goulielmos G.N.,** Niraki E., Patelarou A.E., Matalliotakis I. (2017). Association of placenta previa with a history of previous

cesarian deliveries and indications for a possible role of genetic component. Domination of male sex at birth. *Balkan Journal of Medical Genetics* 20 (2), in press.

94. Zervou M.I., Dimopoulou D.G., Eliopoulos E., Trachana M., Pratsidou-Gertsi P., Spandidos D.A., Garyfallos A., ***Goulielmos G.N.** (2017). The genetics of Juvenile Idiopathic Arthritis in Greece: searching for new susceptibility loci. *Molecular Medicine Reports* 16(6):8793-8798.
95. Matalliotaki C., M. Matalliotakis, Ieromonachou P, **Goulielmos G.N.**, M.I. Zervou, A. Laliotis, D.A. Spandidos, I. Matalliotakis, A. Arici (2018). The co-existence of benign gynecological tumors with endometriosis in a group of 1000 women. *Oncology Letters* 15: 1529-1532.
96. Matalliotakis M., Zervou M.I., Eliopoulos E., Matalliotaki C., Rahmioglu N., Kalogiannidis I., Zondervan K., Spandidos D.A., Matalliotakis I. and ***Goulielmos G.N.** (2018). The role of IL-16 gene polymorphisms in endometriosis. *International Journal Molecular Medicine* 41:1469-1476.
97. Matalliotakis M., Matalliotaki C., **Goulielmos G.N.**, A. Laliotis, T. Maria, D.A. Spandidos, A. Arici, Matalliotakis I. (2018). Endometriosis–associated ovarian cancer in women with advanced endometriosis. *Oncology Letters* 15:7689-7692.
98. Hamidreza Ebrahymian, Ramazan Rezaei, Shayan Mostafaei, Saeed Aslani, **George N. Goulielmos**, Ahmadreza Jamshidi, Mahdi Mahmoudi (2018). Association study between STAT4 polymorphisms and susceptibility to systemic lupus erythematosus disease: systematic review and meta-analysis. *Meta Gene* 16:241-247.
99. Terzidou C., Trivli A., Dalianis G., Apessou D., Spandidos D.A., **Goulielmos G.N.*** (2018). Advanced choroidal melanoma with a desirable aesthetic outcome after enucleation: A case report. *Oncology Letters* 16(1):511-514.
100. **Goulielmos GN[*],** Zervou MI[*], Vazgiourakis VM, Ghodke-Puranik Y, Garyfallos A, Niewold TB (2018). The genetics and molecular pathogenesis of SLE in populations of different ancestral background. *GENE* 668:59-72.
101. Vassilopoulou L, Matalliotakis M, Zervou MI, Matalliotaki C, Spandidos DA, Matalliotakis I, Goulielmos GN (2018). *Exp Ther Med.* 16(2):1043-1051.
102. Nastazia Lesgidou, Elias Eliopoulos, **George N. Goulielmos** and Metaxia Vlassi (2018). Insights on the alteration of functionality of a Tyrosine kinase 2 variant. A Molecular Dynamics Study. *Bioinformatics* 34(17):i781-i786.
103. Plataki MN, Zervou MI, Samonis G, Daraki V, **Goulielmos GN**, Kofteridis DP (2018). The role of IL-6 gene polymorphism in Type 2 Diabetes. *Genetic Testing and Molecular Biomarkers* 22(7):448-452.
104. Koliarakis I1, Psaroulaki A, Nikolouzakis TK, N Sgantzos M, **Goulielmos G**, Androutsopoulos VP, Tsiaoussis J. (2018). Intestinal microbiota and colorectal cancer: a new aspect of research. *J BUON* 23(5):1216-1234.
105. Matalliotaki C, Matalliotakis M, Zervou MI, Trivli A, Matalliotakis I, Mavromatidis G, Spandidos DA, Albertsen HM, Chettier R, Ward K, **Goulielmos GN[*]** (2018). Co-existence of endometriosis with thirteen non gynecological co-morbidities: mutation analysis by whole exome sequencing. *Molecular Medicine Reports* 18(6):5053-5057.
106. Alexandra Trivli, Ioannis Koliarakis, Chryssa Terzidou, **George N. Goulielmos**, Charalambos S. Siganos, Demetrios A. Spandidos, Georgios Dalianis, Efstathios T. Detorakis (2019). Normal Tension Glaucoma: pathogenesis and genetics. An Updated Review. *Experimental and Therapeutic Medicine* 17:563-574.
107. Albertsen H., Matalliotaki C., Matalliotakis, M., Zervou, M.I., Mataliotakis I., Spandidos D.A., Rakesh Chettier, Kenneth Ward, **Goulielmos, G.N.[*]** (2019). Whole exome sequencing identifies hemizygous deletions in UGT2B28 and

USP17L2 genes in a three-generation family with endometriosis. *Molecular Medicine Reports*, doi: 10.3892/mmr.2019.9818, in press.

108. **Goulielmos GN***, Zervou MI, Eliopoulos E (2018). An Adult Fatal Case with a STAT1 Gain-of-function Mutation Associated with Multiple Autoimmune Diseases. *J Rheumatol*, in press.

109. Robin Mesnage, Michael N Antoniou, **George N. Goulielmos**, Aristides M. Tsatsakis (2018). Gut microbiome metagenomics to understand how xenobiotics impact human health. *Current Opinions in Toxicology*, in press.

VI. Scientific collaborations at international level

1. Member of the "External quality assessment" group for the molecular diagnosis of hereditary recurrent fevers (see European Journal of Human Genetics 2009, vol. 17, pp. 890–896).
2. Prof. Timothy Niewold (New York University, USA): the study of genetics and pathophysiology of Systemic Lupus Erythematosus (SLE).
3. Profs. Anne Barton and Jane Worthington (Manchester University, UK): the study of genetics of Rheumatoid Arthritis (RA), Psoriatic Arthritis (PsA) and personalized medicine in RA.
4. Profs. Rene Toes and Tom Huizinga (Leinden Medical School, The Netherlands): the study of the genetic basis of various autoimmune diseases.
5. Dr. Frederique Ponchel (Leeds University, UK): the study of the genetics of RA.
6. Prof. Andreas Radbruch, Drs Bruno Stuhlmüller, Hyun-Dong Chang and Mir-Farzin Mashreghi (CHARITE, Berlin, Germany): the use of new technologies for the analysis of autoinflammation.
7. Prof. Justine Ellis, Genes, Environment and Complex Disease at Murdoch Childrens Research Institute of Victoria, Australia: the study of the genetics of JIA.
8. Prof. Krina Zondervan, Nuffield Dept. of Obstetrics & Gynaecology, Wellcome Trust Centre for Human Genetics, University of Oxford, UK and Dr Pia Vahteristo, University of Helsinki, Finland: the study of the genetics of endometriosis.

VII. Funding

1. National grants:
   a. Hellenic Association of Rheumatology
   b. General Secretariat for Research and Technology (in the context of calls as "Thalis", "Aristeia", "Irakleitos", "Archimedes")
   c. ELKE – University of Crete

2. European grants:
   a. FP5 ("Quality of life and management of Living resources")
   b. FP6 (Autocure)
   c. HEALTH ("ALCOVE - Joint Action Alzheimer")
   d. FP7 (BTCure)

VIII. Molecular Medicine and Human Genetics Section, Medical School of Crete

Head: George N. Goulielmos, PhD

The bidirectional interaction between basic and clinical sciences in Medicine, have provided the impetus to the rapid development of Molecular Medicine. The nature and extent of the genetic contribution to human variation and disease is the area of interest of a related field, that of Human Genetics. Most of the genes involved in the major monogenic disorders, which follow Mendelian patterns of inheritance, have now been isolated and characterized; however, their overall population incidence in the population is relatively low. On the other hand, complex diseases such as cancer, diabetes, rheumatoid arthritis, coronary artery diseases etc result from the interaction of multiple genetic and environmental factors. This group of diseases represents the most common and the least understood human diseases. Identification of genetic factors predisposing individuals to diseases is a powerful tool since offers the opportunity for prevention or treatment of several human diseases.

George Goulielmos has expertise in the genetic study of both monogenic and complex diseases. He is devoted to the investigation of key pathogenetic events in a variety of metabolic, infectious and inflammatory diseases. The main emphasis is on investigating the naturally selected critical pathways in acute and chronic inflammations of infectious or auto-inflammatory nature. Dr. Goulielmos' long-term goal is the multidisciplinary approach to FMF, the prototype of the recurrent fever syndromes, from the epidemiologic, genetic, pathogenetic, population and structural biological point of view. The ultimate task is to depthen further the existing pathogenic mechanisms and gain insight into new molecular pathways leading to the development of FMF by using modern approaches (i.e. gene silencing, investigation of differentially expressed microRNAs, validation of these microRNAs and their target genes in FMF patients, construction of gene networks).

Moreover, Dr. Goulielmos applies human molecular genetics as a tool to understand the genetics of various complex diseases and, mainly, of the autoimmune diseases resulting from the deregulation of the immune system. In particular, his research focuses on the investigation of whether genetic polymorphisms of several genes (new or detected through Genome Wide Association Studies - GWAS), such as *eNOS, PDCD1, PTPN22, TRAF1/C5, STAT4, TLR-2, TLR-4, MBL-2, CD40, NCOA5, C1q, TGF-beta, IRF8, IRF5, IL-12B, IL-23R, PTPN2, VDR* etc play any role as predisposing or severity factors for various autoimmune diseases. The ultimate task is to extend the understanding of existing pathogenetic mechanisms by using functional genomics approaches, aiming to understand the functional significance of the detected gene polymorphisms and, finally, gain insight into new molecular pathways leading to the development of these diseases. To this end, having identified risk alleles for autoimmune diseases, the next critical step is to understand how these alleles disrupt normal immune function, thus leading to insight into fundamental mechanisms that lead to Systemic Lupus Erythematosus (SLE) and Rheumatoid Arthritis (RA), which in turn could help guide the development of novel therapies. Genetic studies conducted have demonstrated the importance of T-cell activation (*PDCD1* gene), B-cell (*CD40* gene) and the role of complement (*C1q* gene) as well as the IFN-alpha signaling (*IRF8* gene) in the development of these diseases. Primary immune cells from patients and healthy subjects are being used to study the mechanism of action of SLE and RA risk alleles. Furthermore, an RA cohort from Crete was studied in an attempt to associate various gene polymorphisms with treatment response of RA patients in anti-TNF agents. Dr. Goulielmos also studies the different patterns of immune system activation that exist between people of different ancestral background with SLE, as this should explain some of the heterogeneity in treatment responses we observe in this disease.

Dr. Goulielmos' laboratory is mapping the genetic factors that cause more autoimmune diseases apart from SLE and RA, such as Juvenile Idiopathic Arthritis (JIA), Type-1 Diabetes (T1D), Psoriasis (PS), Psoriatic Arthritis (PsA) and Bowel Inflammatory Diseases, and exploring the ways in which genetic variations alter the human immune response to result in disease. Recently, the laboratory focuses on the genetic basis of endometriosis as well and a remarkable progress has been achieved regarding the detection and confirmation of the role of

FN1, FSHB, WNT4, VEZT, IL-16, GREB1, UGT2B28 and *USP17L2* genes in the development of the disease. To this end, gene association as well as whole exome sequencing (WES) studies have been conducted, while whole genome sequencing (WGS) studies are in progress. Moreover, Goulielmos' group participates in the latest GWAS referred to endometriosis, as member of the International Endometriosis Genomics Consortium (IEGC), while new collaborations have been established with the Salt Lake City Univ. (Uhta, USA) and the Univ. of Helsinki (Finland) aiming to the clarification of the mechanisms leading to endometriosis.

G. Goulielmos has organized a very well characterized collection of cohorts from various diseases, i.e. RA, SLE, T1D, PS, PsA, JIA, Wegener's Disease, IgA Nephropathy, Acute Otitis Media, Familial Mediterranean Fever (FMF) and endometriosis, consisting of patients from the genetic homogeneous population of Crete. Moreover, he collaborates extensively with other Laboratories and is currently seeking to create a core-facility for large scale genetics within Greece. He attempts to work with other autoinflammatory diseases apart from FMF, such as TRAPS, CAPS and MKD, aiming to gain insights in the underlying pathogenetic mechanisms leading to autoinflammation and to these diseases. To this end, a new microarray chip has been developed by Goulielmos' Group aiming to the diagnosis of the 6 aforementioned autoinflammatory diseases.