

# **CURRICULUM VITAE**

**MARIA I. ZERVOU**

**Biologist, M.Sc., Ph.D**

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## **I. PERSONAL INFORMATION**

Name: **Maria I. Zervou**

Position title: Biologist M.Sc.-PhD-Scientific and Teaching Staff  
(Human Molecular Genetics).

Office Address: Department of Internal Medicine, Section of  
Molecular Pathology and Human Genetics, Medical School,  
University of Crete, Heraklion, Stavrakia 710 03, Crete, Greece

Email address: [zervou@uoc.gr](mailto:zervou@uoc.gr)  
[mzervou@med.uoc.gr](mailto:mzervou@med.uoc.gr)

Telephone: (+30) 2810394672 (office)  
(+30) 2810394626 (lab)



Google scholar profile:

<https://scholar.google.com/citations?user=LM0K7g4AAAAJ&hl=en>

Scopus profile: <https://www.scopus.com/authid/detail.uri?authorId=35486512400>

ResearchGate profile: [https://www.researchgate.net/profile/Maria\\_Zervou3](https://www.researchgate.net/profile/Maria_Zervou3)

ORCID: <https://orcid.org/0000-0001-6750-3452>

## **II. EDUCATION/TRAINING**

| INSTITUTION AND LOCATION                   | DEGREE | YEAR | FIELD OF STUDY                                |
|--------------------------------------------|--------|------|-----------------------------------------------|
| University of Crete, Department of Biology | B.Sc.  | 1999 | Biology                                       |
| University of Crete, Department of Biology | M.Sc.  | 2001 | Molecular Biology and Biotechnology of Plants |
| University of Crete, Medical School        | Ph.D.  | 2007 | Molecular Genetics of Pulmonology             |

### III. PEER REVIEWED PUBLICATIONS

# : equal contribution

\* : corresponding author

1. Lefort, F, Kybelos K.J., **Zervou M.**, Edwards, K.J., & Roubelakis-Angelakis, K.A. (2002). Characterization of new microsatellite loci from *Vitis vinifera* and their conservation in some *Vitis* species and hybrids, *Molecular Ecology Notes* 2 (1):20-21. (τρέχουσα ονομασία: *Molecular Ecology Resources*).
2. Paraskakis E., Sourvinos G., Passam F., Tzanakis N., Tzortzaki H., **Zervou M.**, Spandidos D., and Siafakas N.M. (2003). Microsatellite DNA Instability and Loss of Heterozygosity in Bronchial Asthma, *Eur Respir J.* 22: 951–955.
3. Antoniou K.M., Tzortzaki E.G., Alexandrakis M.G., **Zervou M.**, Tzanakis N., Sfiridaki K., Bouros DE, and Nikolaos M. Siafakas (2005). Investigation of IL-18 and IL-12 in induced sputum of patients with IPF before and after treatment with Interferon gamma-1b, *Sarcoidosis Vasc Diffuse Lung Dis.* 22:204-209.
4. Samara K., **Zervou M.**, Siafakas N.M., Tzortzaki E.G. (2006), Microsatellite DNA Instability in benign lung diseases, *Respir Med.* 100:202-11.
5. Alexopoulou C, I. Mitrouska, D. Arvanitis, N. Tzanakis, G. Chalkiadakis, I. Melissas, **M. Zervou**, and N.M. Siafakas. (2005), Vascular-specific growth factor mRNA levels in the human diaphragm. *Respiration* 72(6):636-641.
6. **Zervou M.I.**, E.G. Tzortzaki, D. Makris, M.Gaga, E. Zervas, E. Economidou, M. Tsoumakidou, N. Tzanakis, J. Milic-Emili and N.M. Siafakas (2006), Differences in Microsatellite DNA level between Asthma and COPD. *Eur Respir J.* 28(3):472-478.
7. Karatzanis A. D., K. Samara, **M. Zervou**, E. Tzortzaki, E. S. Helidonis, N. Siafakas, and G.A. Velegrakis (2007). Assessment for microsatellite DNA instability in nasal cytology samples of patients with allergic rhinitis, *Am J Rhinol* 21:236-240.
8. Karatzanis A D, Samara K D, Tzortzaki E, **Zervou M**, Helidonis ES, Velegrakis GA, Siafakas N (2007). Microsatellite DNA instability in nasal cytology of COPD patients. *Oncol Reports* 17(3):661-665.

9. Tzortzaki EG, Antoniou KM, **Zervou MI**, Lambiri I, Koutsopoulos A, Tzanakis N, Plataki M, Maltezas G, Bouros D, Siafakas NM (2007). Effects of antifibrotic agents on TGF-beta1, CTGF and IFN-gamma expression in patients with idiopathic pulmonary fibrosis. *Respir Med.* 101:1821-1829.
10. Economidou F, Tzortzaki EG, Schiza S, Antoniou KM, Neofytou E, **Zervou M**, Lambiri I, Siafakas NM (2007). Microsatellite DNA analysis does not distinguish malignant from benign pleural effusions. *Oncol Reports* 18(6):1507-1512.
11. **Zervou M.I.**, P. Sidiropoulos, E Petraki, V. Vazgiourakis, E. Krasoudaki, A. Raptopoulou, H. Kritikos, E. Choustoulaki, D. T. Boumpas 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. *Human Immunology*, 69:576-571.
12. Makris, D., N. Tzanakis, A. Damianaki, E. Ntaoukakis, E. Neofytou, **M. Zervou**, N.M. Siafakas, E.G. Tzortzaki (2008). Microsatellite DNA Instability and Chronic Obstructive Pulmonary Disease Exacerbations. *Eur Respir J.*, 32: 612–618.
13. **Zervou M. I.**, D. Mamoulakis, C. Panierakis, D. T. Boumpas and G. N. Goulielmos (2008). STAT4: a risk factor for Type 1 Diabetes? *Human Immunology*, 69:647-650.
14. 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.
15. **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. *Human Immunology*, Sep;70(9):738-741.
16. F.A.S. Kurreeman, G. N. Goulielmos, B. Z. Alizadeh, B. Rueda, M. Bevoa, 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 (2010). The TRAF1/C5 region is associated with multiple autoimmune diseases. *Annals of the Rheumatic Diseases*, Apr;69(4):696-699.

17. **Zervou M.I.**, Castro-Giner F, Sidiropoulos P, Boumpas DT, Tosca AD, Krueger-Krasagakis S. (2010). The Protein Tyrosine Phosphatase, Non-Receptor Type 22 R620W Polymorphism Does Not Confer Susceptibility to Psoriasis in the Genetic Homogeneous Population of Crete. *Genetic Testing and Molecular Biomarkers*. 2010 Feb;14(1):107-111.
18. 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.
19. **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.
20. Krasagakis K, Metaxari M, **Zervou M**, Stathopoulos EN, Eberle J, Kanitakis J, Georgoulas V, Krüger-Krasagakis S, Tavernarakis N, Tosca AD. Identification of the M541L sequence variation of the transmembrane KIT domain in Merkel cell carcinoma. *Anticancer Res*. 2011 Mar;31(3):807-811.
21. 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. *Annals of the Rheumatic Diseases* 70:2184-2190.
22. **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.
23. #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 (2012). Implication of VEGFR2 in Systemic Lupus Erythematosus: a combined genetic and structural biological approach. *Clin Exp Rheumatol* 2013 Jan-Feb;31(1):97-102 (#equal contribution).

24. Goulielmos G.N., Samonis G., Apergi M., Christofaki M., Valachis A., **Zervou M.I.**, Kofteridis D.P. (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.* 2013 Jul;74(7):878-881.
25. D.G. Dimopoulou, **M.I. Zervou**, M. Trachana, E. Myrthianou, J. Pratsidou-Gertsis, D. Kardassis, A. Garyfallos and G.N. Goulielmos (2013). Investigation of juvenile idiopathic arthritis susceptibility loci: Results from a Greek population. *Hum Immunol.* 2013 Sep;74(9):1194-1198.
26. **M.I. Zervou**, E. Myrthianou, I. Flouri, D. Plant, G. Chlouverakis, P. Rapsomaniki, A. Barton, D.T. Boumpas, P. Sidiropoulos and G.N. Goulielmos (2013). Lack of association of variants previously associated with anti-TNF medication response in rheumatoid arthritis patients: Results from a homogeneous Greek population. *PLoS ONE.* 2013 Sep 10;8(9):e74375.
27. 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 Immun.* 2013 Dec;14(8):471-478.
28. 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.
29. George N. Goulielmos, Elias Eliopoulos, A. Karatzanis, **Maria I. Zervou**, 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, (IJNTR)* ISSN:2454-4116 Volume 2, Issue-1, January 2016 Pages 32-36.
30. Goulielmos G.N., **M.I. Zervou**, E. Myrthianou, A. Burska, T.B. Niewold and F. Ponchel (2016). Genetic data: the new challenge of personalized medicine, insights for Rheumatoid Arthritis patients. *GENE*, Jun 1;583(2):90-101.
31. **Zervou Maria 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*, (IJNTR) ISSN: 2454-4116, Volume-2, Issue-3, March 2016 Pages 67-70.

32. Budu-Aggrey A, Bowes J, Loehr S, Uebe S, **Zervou MI**, Helliwell P, Ryan AW, Kane D, Korendowych E, Giardina E, Packham J, McManus R, FitzGerald O, McHugh N, Behrens F, Burkhardt H, Huffmeier U, Ho P, Martin J, Castañeda S, Goulielmos G, Reis A, Barton A. (2016). Replication of a distinct psoriatic arthritis risk variant at the IL23R locus. *Annals of the Rheumatic Diseases*, Jul;75(7):1417-8. doi: 10.1136/annrheumdis-2016-209290. Epub 2016 Mar 25.
33. G.N. Goulielmos, R.C. Chiaroni-Clarke, D.G. Dimopoulou, **M.I. Zervou**, M. Trachana, P. Pratsidou-Gertsis, A. Garyfallos and J.A. Ellis (2016). Association of juvenile idiopathic arthritis with PTPN22 rs2476601 is specific to females in a Greek population. *Pediatric Rheumatology*, 2016 Apr 23;14(1):25.
34. E. Myrthianou, **M.I. Zervou**, A. Budu-Aggrey, E. Eliopoulos, D. Kardassis, D.T. Boumpas, N. Koungkas, 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. (εξώφυλλο στο τεύχος του περιοδικού)
35. **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* 2017 Apr 30; 610:9-14
36. M. Matalliotakis, G. N. Goulielmos, **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.
37. 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.

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39. 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.
40. 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.
41. 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.
42. **Zervou M.I.**, Dimopoulou D.G., Eliopoulos E., Trachana M., Pratsidou-Gertsis 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.
43. C. Matalliotaki, M. Matalliotakis, Ieromonachou P, G. N. Goulielmos, **M. I. Zervou**, A. Laliotis, D. A. Spandidos, I. Matalliotakis, A. Arici (2018). Co-existence of benign gynecological tumors with endometriosis in a group of 1000 women. *Oncology Letters*, 2018 Feb;15(2):1529-1532.
44. Matalliotakis M<sup>#</sup>, **Zervou M.I.**<sup>#</sup>, 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* 2018 Mar;41(3):1469-1476 (**#equal contribution**).

45. Goulielmos, G.N.<sup>#</sup>, **Zervou M.I.<sup>#</sup>**, V. Vazgiourakis, Ghodke-Puranik Y, Garyfallos A. and Niewold T. (2018). The genetics and molecular pathogenesis of systemic lupus erythematosus (SLE) in populations of different ancestry. *GENE*, 2018 Aug 20;668:59-72 (**#equal contribution**).
46. Plataki M.<sup>#</sup>, **Zervou M.I.<sup>#</sup>**, Samonis G., Daraki V., Goulielmos G.N. and Kofteridis D. (2018). Association of the Interleukin-6 rs1800795 Polymorphism with Type 2 Diabetes Mellitus in the Population of the Island of Crete, Greece. *Genetic Testing and Molecular Biomarkers*, 2018 Jul;22(7):448-452. (**#equal contribution**).
47. Vassilopoulou L, Matalliotakis M, **Zervou MI**, Matalliotaki C, Spandidos DA, Matalliotakis I, Goulielmos GN. Endometriosis and in vitro fertilisation. *Exp Ther Med*. 2018 Aug;16(2):1043-1051.
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49. Albertsen H., Matalliotaki C., Matalliotakis, M., **Zervou, M.I.**, Matalliotakis I., Spandidos D.A., Rakesh Chettier, Kenneth Ward, Goulielmos, G.N. (2018). Whole exome sequencing identifies hemizygous deletions in UGT2B28 and USP17L2 genes in a three-generation family with endometriosis. *Molecular Medicine Reports* 2019 Mar;19(3):1716-1720.
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51. Vassilopoulou L, Matalliotakis M, **Zervou MI**, Matalliotaki C, Krithinakis K., Matalliotakis I, Spandidos DA, Goulielmos GN (2019). Defining the genetic profile of endometriosis. *Exp Ther Med.*, 17: 3267-3281.
52. Matalliotakis M, Matalliotaki C, Trivli A, **Zervou MI**, Kalogiannidis I, Tzardi M, Matalliotakis I, Arici A, Goulielmos GN. Keeping an Eye on Perimenopausal and Postmenopausal Endometriosis. *Diseases* 2019 Mar 12;7(1) 29. doi: 10.3390/diseases7010029.
53. Michail Matalliotakis, Charoula Matalliotaki, **Maria I. Zervou**, George N. Goulielmos, Aydin Arici, Demetrios A. Spandidos, Ioannis Matalliotakis

- (2019). Retrospective evaluation of pathological results among women with ovarian endometriomas versus teratomas. *Molecular and Clinical Oncology*, 10 (6):592-596.
54. Charoula Matalliotaki, Michail Matalliotakis, Nilufer Rahmioglu, George Mavromatidis, Ioannis Matalliotakis, George Koumantakis, Krina Zondervan, Demetrios A. Spandidos, George N. Goulielmos and **Maria I. Zervou\*** (2019). The role of FN1 and GREB1 gene polymorphisms in patients with endometriosis. *Molecular Medicine Reports*, 20(1):111-116.
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  57. Matalliotaki C, Matalliotakis M, Laliotis A, **Zervou MI**, Kolliarakis I, Matalliotakis I, Arici A, Goulielmos GN (2019). Epidemiological aspects of the outcomes from the treatment of endometriosis: Experience from two academic reference centers. *Experimental Therapeutic Medicine*, 2020;19(2):1079-1083.
  58. G.N. Goulielmos, M. Matalliotakis, C. Matalliotaki, E. Eliopoulos, I. Matalliotakis, **M.I. Zervou\*** (2020). Endometriosis research in the ‘omics’ era. *GENE* 741 (2020) 144545.
  59. G.N. Goulielmos, **M.I. Zervou** (2020). Risk of systemic lupus erythematosus in patients with idiopathic thrombocytopenic purpura: a population-based cohort study. *Annals of the Rheumatic Diseases*, 2022 Jul;81(7):e112.
  60. **Maria I. Zervou\***, Athena Andreou, Michail Matalliotakis, Demetrios A Spandidos, George N Goulielmos, Elias E Eliopoulos (2020). Association of the DNASE1L3 rs35677470 polymorphism with systemic lupus erythematosus, rheumatoid arthritis and systemic sclerosis: Structural

biological insights *Molecular Medicine Reports*, 2020 Dec;22(6):4492-4498.

(εξώφυλλο στο τεύχος του περιοδικού)

61. Michail Matalliotakis, Charoula Matalliotaki, **Maria I. Zervou**, Konstantinos Krithinakis, George N. Goulielmos, Ioannis Kalogiannidis (2020). Abdominal and perineal Scar endometriosis: Retrospective study on 40 cases. *Eur J Obstet Gynecol Reprod Biol.* 2020;252:225-227.
62. Alexandra Trivli, **Maria I. Zervou**, George N. Goulielmos, Demetrios A. Spandidos, Efstathios T. Detorakis (2020). Primary open angle glaucoma genetics: The common variants and their clinical associations (Review). *Molecular Medicine Reports*, 2020 Aug;22(2):1103-1110.
63. Goulielmos GN, **Zervou MI.** (2020). High risk of systemic lupus erythematosus and antiphospholipic syndrome in patients with idiopathic thrombocytopenic purpura: Genetic aspects. *Lupus* 2021 Jan;30(1):175-176.
64. Goulielmos G.N., **Zervou M.I.** (2020). Correspondence on 'Increased risk of systemic lupus erythematosus in patients with autoimmune haemolytic anaemia: a nationwide population-based cohort study' *Ann Rheum Dis*, 2020 Nov 20;annrheumdis-2020-219321. doi: 10.1136/annrheumdis-2020-219321.
65. George N. Goulielmos, **Maria I. Zervou** (2020). Comment on: Association of systemic lupus erythematosus with peripheral arterial disease: a meta-analysis of literature studies. *Rheumatology*, 2021 May 14;60(5):e187-e188.
66. Angeliki Chroni, Loukianos Rallidis, Despina Vassou, Christina Gkolfinopoulou, **Maria I. Zervou**, George N. Goulielmos, Danae Pappa, Elias Eliopoulos and Dimitris Kardassis (2020). Identification and characterization of a rare variant in apolipoprotein A-IV, p.(V336M), and evaluation of HDL functionality in a Greek cohort with extreme HDL cholesterol levels. *Arch Biochem Biophys.* 2020 Dec 15;696:108655.
67. Goulielmos G.N., **Zervou MI** (2021). Comment on: Obstetric antiphospholipid syndrome is not associated with an increased risk of subclinical atherosclerosis. *Rheumatology (Oxford)* 2021 Jun 18;60(6):e220-e221.
68. **Zervou M.I.**, Goulielmos G.N. (2021). Correspondence on “Risk of venous thromboembolism in rheumatoid arthritis, and its association with disease activity: a nationwide cohort study from Sweden” by Molander et al. *Ann.*

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69. **Zervou MI**, Goulielmos GN (2021). Comment on: The risk and trend of pulmonary embolism and deep vein thrombosis in rheumatoid arthritis: a general population-based study. *Rheumatology (Oxford)* 2021 Jul 1;60(7):e266-e267.
70. Goulielmos GN, **Zervou MI** (2021). Risk of venous thromboembolism in ankylosing spondylitis and rheumatoid arthritis: genetic aspects. *J. Rheumatol.*, 2021 Sep;48(9):1492-1493. doi: 10.3899/jrheum.210131. Epub 2021 Jun 15.
71. **Zervou MI**, Matalliotakis M, Goulielmos GN (2021). Comment on “Risk of systemic lupus erythematosus in patients with endometriosis: A nationwide population-based cohort study”. *Arch Gynecol Obstetrics*, 2022 Feb;305(2):543-544.
72. Papageorgiou L, **Zervou MI**, Vlachakis D, Matalliotakis M, Matalliotakis I, Goulielmos GN, Eliopoulos E. (2021). Demetra Application: An integrated genotype analysis web server for clinical genomics in Endometriosis. *Int. J. Mol Medicine*, 2021 Jun;47(6): 115.
73. **Zervou MI**, Goulielmos GN (2021). Comment on: Refining myositis associated with primary Sjögren’s syndrome: data from the prospective cohort ASSESS. *Rheumatology (Oxford)* 2021 Oct 2;60(10):e369-e370.
74. Eleni Kampouraki, Marilena Lourou, **Maria I Zervou**, Evangelia-Dimitra Ampazoglou, Emmanouil Yachnakis, Nikolaos Katzilakis, George N Goulielmos, Eftichia Stiakaki (2021). The role of CXCL12, TP53 and CYP1A1 gene polymorphisms in susceptibility to pediatric acute lymphoblastic leukemia. *Oncology Letters* 22: 659.
75. George N. Goulielmos, **Maria I. Zervou**, Elias Eliopoulos (2021). Comment on: Homozygous variant p. Arg90His in NCF1 is associated with early-onset interferonopathy: A case report. *Pediatric Rheumatology* 2021 Aug 16;19(1):125.
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#### **IV. An overview of the research activities**

The bidirectional interaction between basic and clinical sciences in Medicine has provided the impetus to the rapid development of Molecular Medicine. Complex diseases such as cancer, systemic lupus erythematosus, psoriasis, endometriosis, adenomyosis, diabetes, juvenile idiopathic arthritis etc, result from the interaction of multiple genetic, epigenetic and environmental factors. This group of diseases represents the most common and the least understood human diseases. A better understanding of the pathogenetic mechanisms leading to the diseases will be achieved through the identification of genetic factors increasing the predisposition of individuals to diseases, thus offering the opportunity for the prevention, an efficient treatment and the development of new therapeutic strategies. Dr Maria Zervou has an expertise in the genetic study of complex human diseases. She is devoted to the investigation of key pathogenetic events in a variety of autoimmune, metabolic, gynecological, infectious and inflammatory diseases. Thus, Dr. Zervou 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, her research focuses on the investigation of whether genetic polymorphisms of several genes (new or detected through Genome Wide Association Studies - GWAS), such as eNOS, PTPN22, TRAF1/C5, STAT4, MBL2, CD40, NCOA5, C1q, TGF-beta, VEGFR2, IRF8, IRF5, IL-12B, IL-23R etc play any role as predisposing or severity factors for various autoimmune diseases, including Systemic Lupus Erythematosus (SLE), Rheumatoid Arthritis (RA), Juvenile Idiopathic Arthritis (JIA), Type-1 Diabetes (T1D), Psoriasis (PS), Psoriatic Arthritis (PsA) and Inflammatory Bowel Diseases (IBD). In addition, Dr. Zervou explores the ways by which genetic variations alter the human immune response and result in the disease's development. The ultimate task is to understand the functional significance of the detected gene polymorphisms and, finally, gain insight into new molecular pathways leading to these diseases. Thus, genetic studies conducted have demonstrated the importance of B-cell (CD40 gene) as well as the IFN-alpha signaling (IRF8 and IRF5 genes) in the development of systemic lupus erythematosus (SLE). In the same framework, Dr. Zervou and colleagues used three-dimensional (3-D) modeling studies aiming to further understand the functional significance of various genetic polymorphisms and/or mutations regarding the diseases' development, such as VEGFR2 - SLE, ACVRL1 - Rendu-Osler-Weber (ROW) syndrome, TYK2 - JIA, PNP- SLE, ELANE - inflammatory arthritis, IL-16 - endometriosis, STAT1 - Takayasu arteritis, ADA2 - deficiency of adenosine deaminase 2, DNASE1L3 - SLE / RA / SSc, NCF1 - early onset interferonopathy and IFIH1 - T1D / AS / PS / PsA / CD. Moreover, an RA cohort from Crete was studied by Dr. Zervou and colleagues in an attempt to associate various gene polymorphisms with treatment response of RA patients in anti-TNF agents.

Last years, Dr. Zervou possessed a major role dealing with the multidisciplinary study on the genetic basis of endometriosis, by confirming the substantial role of FN1, FSHB, WNT4, VEZT, IL-16, GREB1, UGT2B28, USP17L2, FN1 and GREB1 genes in the development of the disease. Moreover, new collaborations have been established with the Salt Lake City Univ. (Uhta, USA), the Oxford University (UK) and the Univ. of Helsinki (Finland), aiming to clarify the mechanisms leading to endometriosis. To this end, gene association (case-control) as well as whole exome sequencing (WES) studies have been conducted, while whole genome sequencing (WGS) studies are in progress in an attempt to detect potentially new, interesting risk gene variants. Importantly, Dr.

Zervou participated in the biggest GWAS and meta-analysis conducted ever, referred to the genetics of endometriosis and other pain conditions and comorbidities, as a member of the International Endometriosis Genomics Consortium (IEGC). An article presenting the results of this study has been published in Nature Genetics (2023). Recently, Dr. Zervou was involved in the research activities of a group focusing on the development of various novel platforms, which are based on bioinformatics applications designed to assist medical doctors' diagnosis from the early stages of various diseases as well as to offer precision medicine insights, by using the patients' genomic data. Thus far, these bioinformatics tools have been developed for endometriosis, SLE, RA, Ankylosing Spondylitis (AS), Myasthenia Gravis (MG), Multiple Sclerosis (MS) and Inflammatory Bowel Diseases (IBD). Dr. Zervou has also developed a new gene panel aiming to the diagnosis of 6 autoinflammatory diseases, in particular Familial Mediterranean Fever (FMF), TNF Receptor-Associated Periodic Syndrome (TRAPS), Mevalonate Kinase Deficiency (MKD) and the three Cryopyrin-Associated Protein Syndroms (CAPS), Muckle-Wells Syndrome (MWS), Neonatal Onset Multisystem Inflammatory Disease (NOMID) and Familial Cryopyrin Autoinflammatory Syndrome (FCAS).