**CURRICULUM VITAE**

**Name: Joanne Rachel Traeger-Synodinos**

**Date & place of birth**: 9th February 1957, London, UK.

**Nationality:** British (and Greek through marriage).

**Marital & family status**: Married, 3 children (Born 1980, 1983, 1986)

**Home address:** Danaes 2A, Ekali, Athens 14578, Greece. Home Tel: +30 210 8137270.

**Current work address**: Laboratory of Medical Genetics, National and Kapodistrian University of Athens, Choremio Research Laboratory, St. Sophia's Children's Hospital, Thivon & Levadias St., Athens 11527, Greece. Work Tel: +30 213 2013 460.

Mobile: +30 6932 766527; **email**: jtraeger@med.uoa.gr

## CURRENT POSITION HELD:

Oct 2012- present: Associate Professor of Genetics, Laboratory of Medical Genetics, School of Medicine, National & Kapodistrian University of Athens (Permanent tenure).

## PROFESSIONAL HISTORY

2006-Oct2012: Assistant Professor of Genetics, Laboratory of Medical Genetics, School of Medicine, National & Kapodistrian University of Athens (Permanent tenure).

1999-2005: Clinical Scientist, Greek National Health System, Laboratory of Medical Genetics, Athens University, Choremio Research Laboratory, St. Sophia's Children's Hospital.

1986-1999: Clinical Scientist, Greek National Health System, First Department of Paediatrics, Athens University, Choremio Research Laboratory, St. Sophia's Children's Hospital.

1983-1985: Research Associate, First Department of Paediatrics, Athens University, Choremio Research Laboratory, St. Sophia's Children's Hospital. (Scholarship from the Greek National Research Institute).

## QUALIFICATIONS AND ACADEMIC BACKGROUND

1978-1982 **Doctor of Philosophy**, (D.Phil. Oxon), University of Oxford, UK.
Title: "The synthesis of Haemoglobin E", in the Nuffield Department of Medicine, under supervision of Professor Sir David Weatherall and Dr. Bill Wood.
The thesis focused on the mechanisms underlying the reduced biosynthesis of this abnormal haemoglobin, common in SE Asia, through protein, messenger RNA and DNA studies. The findings resulted in publications in Nature and the Journal of Clinical Investigation.

1975- 1978 **B.Sc. Hons** in Physiology and Biochemistry, University of Reading, UK.

1967-1975 **High school education**: Ealing Grammar School for Girls.

GCE A-levels: Zoology, Chemistry, Physics. GCE O-levels: Biology, Chemistry and Physics, English Language, English Literature, History, Mathematics, French, Geography.

**RESEARCH INTERESTS**

Throughout my D.Phil and in all positions I have held during the last almost 30 years (which were all essentially associated with the same core departments) I have been involved in the field of molecular biology and its application to human genetics. My main research interests have focused on:

1983-present: Genetic and genomic analysis of common and rare monogenic diseases, including haemoglobinopathies (**a recognized international expert**), cystic fibrosis, phenylketonuria, familial hypercholesterolaemia, RETT syndrome, paediatric podocytopathies.

1988-present: Genotype-phenotype correlations in haemoglobinopathies, cystic fibrosis and RETT syndrome, with the aim of identifying pathophysiological mechanisms, as well as disease prognosis.

1992-present: Developing laboratory protocols for detecting genetic variation, applied also to genetic diagnostic procedures and prenatal diagnosis.

1997-present: Developing laboratory protocols for preimplantation genetic diagnosis of monogenic diseases (PGD) (**a recognized international expert**).

2005-present: Developing laboratory protocols for noninvasive prenatal diagnosis (NIPD).

2011-present: MicroRNA studies in human erythroid progenitors and terminally differentiated erythrocytes in ex-vivo cell cultures. Initial aims are to investigate whether miRNAs play a role in the pathophysiology of haemoglobinopathies. The latter may potentially support novel approaches for therapeutic interventions in patients with thalassaemia and sickle cell anaemia, a long-standing goal in the haemoglobinopathies' field. Subsequent aims include 1). The identification of normal erythroid-cell mRNA transcripts (and genes) targeted by miRNAs and the core binding sites recognized by the miRNAs; 2) The investigation of sequence variations in erythroid-specific miRNAs and the core binding sites of their associated mRNAs, the frequency of these variations in the general population and their possible influence on erythroid phenotypes in normal and disease states; 3) The investigation of likely selective pressures or constraints influencing nucleotide sequence variations in miRNAs and mRNA core binding-sites in the erythroid system, with potential extrapolation to general principles of miRNA sequence structure and function..

## TEACHING

I have over 20 years of teaching experience, initially as an affiliated non-academic member of the Athens University clinics, and subsequently in a more official capacity as an Assistant Professor of Genetics. My more recent experience is summarized below:

### Undergraduate lectures

2004-present: Elective for medical students in Human Genetics (2 hours /semester)

2006-present: Elective for medical students in Prenatal Diagnosis and High-Risk Pregnancies (2 hours /semester)

### Postgraduate lectures

Lectures in several different Master's courses run by departments in the National and Kapodistrian University of Athens Medical School and the Departments of Biology and Chemistry.

2007-present: Masters in clinical paediatrics for nurses (Molecular Genetics and Polygenic Diseases, 2 hours /semester).

2007-present: Masters in Molecular Medicine for clinicians and biological scientists (Molecular Genetics, 2 hours /semester).

2007-present: Masters in female reproduction for clinicians and biological scientists (Molecular Genetics, 2 hours /semester).

2000-present: Masters in Clinical Chemistry for chemists and biological scientists (Molecular Methods, 2 hours /semester).

2008-present: Masters in Clinical Biochemistry and Molecular Diagnosis for chemists and biological scientists (Molecular Genetics of Monogenic Diseases, 2 hours /semester).

### Dissertations and Thesis

2008 and 2011: Supervisor of 2 undergraduate dissertations (completed)

2006-present: Co-supervisor of 14 Master's dissertations (13 completed, 1 on-going)

2010-present: Co-supervisor of 4 PhD thesis projects (on-going)

### Laboratory training

In the last almost 30 years, I have trained scientists and clinicians (Greek and International) in molecular biology methods, as well as "best practice" for laboratory protocols and sample processing.

### Examiner

2005- present: as an Assistant (and now Associate) Professor I am involved in writing exam questions, marking examination papers for all the curses I teach in. In addition I regularly evaluate the written and oral presentations of undergraduate dissertations, and masters' and doctorate theses from various departments in the **University of Athens** (Medical School, Departments of Basic and Life Sciences) and other Universities throughout **Greece**. In addition I am currently evaluating 3 Doctorate Thesis submitted at Universities in the **UK** (UCL, London), the **Netherlands** (University of Leiden) and **Australia** (University of Western Australia); I will attend the viva examinations of the PhD candidates in UCL and Leiden Universities.

**International Teaching**

I am often invited to teach in international workshops, seminars etc, including

2011 ESH-ENERCA Training Course on Haemoglobin Disorders: Laboratory diagnosis and clinical management, Brussels, Belgium
Title: Prevention of Thalassaemia and Sickle Cell Disease: Antenatal diagnosis and pre-implantation diagnosis»

2010 ESHRE Campus, London, UK, March, 2010.
Title: Approaching accreditation of a PGD centre: Where to start - writing SOPs and risk assessments.

2008 European Society of Human Reproduction and Embryology, Basic Genetics for ART practitioners, Athens
Title: Basics of monogenic inheritance¨.

2008 ESHRE Campus, Brno, Czeck Republic.
Title: «Examination Process, Quality Management System and Accreditation in PGD Clinics and Laboratories.

2007 European School of Haematology (ESH), Disorders of Iron Homeostasis, Erythrocytes and Erythropoiesis, Athens
Title: Preimplantation diagnosis of thalassaemia.

2006 European School of Genetic Medicine, 4th Course in Thalassaemia and Related Disorders in the Mediterranean, University of Bologna Residential Centre, Bertinoro, Italy.
Title: Molecular Diagnosis of thalassaemia.

2005 European School of Genetic Medicine, 3rd Course in Thalassaemia and Related Disorders in the Mediterranean, University of Bologna Residential Centre, Bertinoro, Italy.
Title: Alpha-thalassaemia.

2004 European School of Genetic Medicine, 2nd Course in Thalassaemia and Related Disorders in the Mediterranean, University of Bologna Residential Centre, Bertinoro, Italy
Title: Epidemiology, molecular genetics and implications for prevention of alpha-thalassaemia in the Mediterranean».

**ORGANIZATIONAL AND ADMINISTRATIVE ACTIVITIES**

1983-1986: I played a key role assisting in the establishment of one of the first molecular biology/ molecular genetics laboratories in Greece.

1986-2009: I was a member of committees for evaluating tenders for the procurement of laboratory equipment and consumables, mainly for laboratories in the St. Sophia's Children's Hospital, the Laboratory of Medical Genetics and other research centres in Greece.

1986-present: I supervise of a small team for routine molecular diagnostics (including prenatal diagnosis) for haemoglobin disorders and thalassaemia, handling approximately 600 carrier, patient and prenatal samples annually.

1997-present: I supervise of a small team offering preimplantation genetic diagnosis (PGD) mainly for monogenic diseases (common and rare diseases), and also microarrays for molecular karyotyping (Preimplantation Genetic Screening, PGS). We are the most experienced group

in Greece and one of the most active groups in the PGD Consortium, an international special interest group under the auspices of the European Society of Human Reproduction and Embryology (ESHRE).

**Co-organiser of international meetings and workshops:**

2012 A Best Practice Meeting for Molecular Diagnosis of Haemoglobinopathies, under the auspices of the EU funded European Molecular Genetics Quality Network (EMQN), Leiden, September 2012.

2011 An ESHRE Campus symposium, in collaboration with Eurogentest, entitled "Accreditation of a Preimplantation Genetic Diagnosis Laboratory", Athens.

2010 An international symposium entitled "Preimplantation genetic Diagnosis: 20 years", Athens.

2008 A European Society of Human Reproduction and Embryology (ESHRE) Campus, entitled "Basic genetics for ART practitioners", Athens.

2008 An ESHRE Campus, entitled "Quality system management and accreditation process for PGD clinics and laboratories" Brno, Czech Republic.

2002 A Best Practice Meeting for Molecular Diagnosis of Haemoglobinopathies, under the auspices of the EU funded European Molecular Genetics Quality Network (EMQN), Manchester, UK.

## SOCIETIES AND NON-STIPENDIARY APPOINTMENTS

2012(-2014): Chair of the Steering Committee of the ESHRE PGD Consortium.

2011-present: Member of the International Advisory Committee for the ITHANET Portal (www.ithanet.eu)

2010-2012: Deputy Chair of the ESHRE PGD Consortium Steering Committee**.**

2008-present: Scientific advisor and board member of the Greek Alliance for Rare Diseases (affiliated to EURORDIS)

2004 to 2010: Steering Committee Member of the ESHRE PGD Consortium

2004-2007: Board member of the Hellenic Association of Medical Geneticists.

2004-present: Member of the European Society for Human Reproduction and Embryology (ESHRE)

1998-present: Editorial Board of HEMOGLOBIN, and from 2003 Associate Editor.

1990-present: Member of the European Society of Human Genetics (ESHG).

## RESEARCH PUBLICATIONS

To date, my research activities have lead to **> 115 peer reviewed** publications in ISI journals (impact factor >490, average IF 4.5, >1800 citations), as well as contributions to 6 books and invitations to lecture nationally and internationally.

1. **Traeger J**, Wood WG, Clegg JB, Weatherall DJ, Wasi P. Defective synthesis of HbE is due to reduced levels of βE mRNA. Nature 288: 497- 499, 1980.

2. **Traeger J,** Winichagoon P, Wood WG. Instability of βE mRNA during erythroid cell maturation in HbE homozygotes. Journal Clinical Investigation 69: 1050- 1053, 1982.

3. Kattamis C, Tzotzos S, Kanavakis E, **Synodinos J**, Metaxotou-Mavromati A. Correlation of clinical phenotype to genotype in HbH disease. The Lancet 1: 442-444, 1988.

4. **Traeger-Synodinos J**, Tzetis M, Kanavakis E, Metaxotou-Mavromati A, Kattamis C. The Corfu δβ-thalassemia mutation in Greece: Hematological phenotype and prevelance. British Journal of Haematology 79: 302- , 1991.

5. **Traeger-Synodinos J**, Kanavakis E, Tzetis M, Kattamis A, Kattamis C. Characterization of non-deletion α-thalassemia mutations in the Greek population. American. Journal of Hematology 44: 162-167, 1993.

6. Hall GW, Thein SL, Newland AC, Chisholm M, **Traeger-Synodinos J,** Kanavakis E, Kattamis C, Higgs DR.A base substitution (T->C) in codon 29 of the α2-globin gene causes α- thalassaemia. British Journal of Haematology 85: 546-552, 1993.

7. Tzetis M, **Traeger-Synodinos J**, Kanavakis E, Metaxotou-Mavromati A, Kattamis C. The molecular basis of normal HbA2 (type 2) β-thalassemia in Greece. Hematologic Pathology 8: 25-34, 1994.

8. Stefanis L. Kanavakis E, **Traeger-Synodinos J**, Tzetis M, Metaxotou-Mavromati A, Kattamis C. Hematological phenotype of the mutations IVS1-n6 (T>C), IVS1-n110 (G>A), CD39 (C>T) in carriers of beta thalassemia in Greece. Pediatric Hematology and Oncology 11: 509-517, 1994.

9. **Traeger-Synodinos J,** Kanavakis E, Kalogerakou M, Soulpi K, Missiou-Tsangaraki S, Kattamis C. Preliminary mutation analysis in the phenylanaline hydroxylase gene in Greek PKU and HPA patients. Human Genetics 94: 573-575,1994.

10. Kanavakis E, **Traeger-Synodinos J**, Tzetis M, Metaxotou-Mavromati A, Ladis B, Kattamis C. Molecular characerization of homozygous (high HbA2) β-thalassemia intermedia in Greece. Pediatric Hematology and Oncology 12: 37-45, 1995.

11. Kanavakis E, Tzetis M, Antoniadi Th, **Traeger-Synodinos J**, Doudounakis S, Adam G, Matsaniotis N, Kattamis C. Mutation analysis of 10 exons of the CFTR gene in Greek Cystic Fibrosis patients: characterization of 74.5% of CF alleles including 1 novel mutation. Human Genetics 96: 364-366, 1995.

12. Kanavakis E, Tzetis M, Antoniadi Th, **Traeger-Synodinos J**, Doudounakis S, Adam, Kattamis C. Mild cystic fibrosis phenotype in patients with the 3272-26A>G mutation. Journal of Medical Genetics 32: 406-407, 1995.

13. Tzetis M, Kanavakis E, Antoniadi Th, **Traeger-Synodinos J**, Doudounakis S, Adam G, Kattamis C. Identification of two novel mutations (296+1G>C and A46D) in exon 2 of the CFTR gene in Greek cystic fibrosis patients. Molecular and Cellular Probes, 9: 283-285, 1995.

14. Kanavakis E, **Traeger-Synodinos J**, Vrettou C, Metaxotou-Mavromati A, Lagona E, Kattamis C. The interaction of α0 thalassaemia with Hb Icaria: 3 unusual cases of haemoglobinopathy H. British Journal of Haematology 92: 332-335, 1996.

15. **Traeger-Synodinos J,** Kanavakis E, Vrettou C, Maragoudaki E, Michael Th, Metaxotou-Mavromati A, Kattamis C. The triplicated alpha globin gene locus in β-thalassaemia heterozygotes: clinical, haematological, biosynthetic and molecular studies. British Journal of Haematology 95: 467-471, 1996.

16. Mavroidis N, **Traeger-Synodinos J,** Kanavakis E, Drogari E, Kattamis C, Matsaniotis N, Day I, Humphries S. Preliminary mutation analysis in Greek Familial Hypercholesterolemia (FH) patients. including the identification of one novel mutation in the Low Density Lipoprotein-Receptor (LDL-R) gene. Human Mutation 9: 274-276, 1997.

17. Kanavakis E, **Traeger-Synodinos J,** Vrettou C, Maragoudaki E, Tzetis M, Kattamis C. Prenatal diagnosis of the thalassemia syndromes by rapid DNA analytical methods. Molecular Human Reproduction 3: 523-528, 1997.

18. **Traeger-Synodinos J**, Maragoudaki E, Vrettou C, Kanavakis E, Kattamis C. Rare β-thalassemia mutations in the Greek and Greek Cypriot populations. Hemoglobin 22: 89-94, 1998.

19. **Traeger-Synodinos J,** Mavroidis N, Kanavakis E, Drogari E, Matsaniotis N, Humphries S, Day INM, Kattamis, C. Analysis of low density lipoprotein receptor gene mutations and microsatellite haplotypes in Greek FH heterozygous children: six independant ancestors account for 60% of probands. Human Genetics 102: 343-347, 1998.

20. **Traeger-Synodinos J**, Metaxotou-Mavromati A, Kanavakis E, Vrettou C, Papasotiriou I, Michael T, Kattamis C. Alpha thalassemic hemoglobinopathy: homozygosity for the hemoglobin Agrinio alpha-globin chain variant. Hemoglobin 22, 209-215, 1998.

21. Papassotiriou I, Kister J, Griffon N, Abraham DJ, Kanavakis E, **Traeger-Synodinos J**, Stamoulakatou A, Marden MC, Poyart C. Synthesized allosteric effectors of the hemoglobin molecule: a possible mechansim for improved erythrocyte oxygen release capability in hemoglobinopathy H disease. Experimental Hematology 26: 922-926, 1998.

22. Maragoudaki E, Vrettou C, Kanavakis E, **Traeger-Synodinos J**, Metaxotou-Mavromati A, Kanavakis E. The rare silent β gene C->G mutation at 6bp 3' to the termination codon; Molecular, haematological and clinical studies in 12 Greek families. British Journal of Haematology 103: 45-51, 1998.

23. Papassotiriou I, **Traeger-Synodinos J**, Kanavakis E, Karagiorga M, Stamoulakatou A, Kattamis C. Eryhtroid marrow activity and hemoglobin H levels in HbH disease. Journal of Pediatric Hematology-Oncology 20: 539-544, 1998.

24. Papassotiriou I, **Traeger-Synodinos J,** Prome D, Kister J, Stamou E, Liakopoulou T, Stamoulakatou A, Kanavakis E, Wajcman H. Association of unstable hemoglobin variants and heterozygous β-thalassemia: example of a new variant HbAcharnes or [beta53 (D4) Ala>Thr]. American Journal of Hematology 62: 186-192, 1999.

25. Papassotiriou I, **Traeger-Synodinos J**, Vlachou C, Karagiorga M, Metaxotou A, Kanavakis E, Stamoulakatou A. Rapid and accurate quantitation of hemoglobins Barts and H using weak cation-exchange high pressure liquid chromatography: correlation with the α-thalassemia genotype. Hemoglobin 23: 203-211, 1999.

26. **Traeger-Synodinos J**, Metaxotou-Mavromati A, Karagiorga M, Papasotiriou I, Vrettou C, Stamoulakatou A, Kanavakis E. Interaction of an α+ thalassemia deletion with either a highly unstable α-globin variant (alpha2, codon 59 GGC>GAC) or a nondeletion alpha-thalassemia mutation (AATAAA>AATAAG): comparison of phenotypes illustrating " dominant " alpha-thalassemia. Hemoglobin 23: 325-337, 1999.

27. **Traeger-Synodinos J**, Harteveld K, Kanavakis E, Giordano PC, Kattamis C, Bernini LF. Hb-Aghia Sophia [alpha62 (E11) Val>o (alpha 1)], an "in-frame" deletion causing alpha thalassemia. Hemoglobin 23: 317-324, 1999.

28. Maragoudaki E, Kanavakis E, **Traeger-Synodinos J,** Vrettou C, Tzetis M, Metaxotou-Mavrommati A, Kattamis C. Molecular, haematological and clinical studies iof the -101C>T substitution in the β-globin gene promoter in 25 β-thalassemia intermedia patients and 45 heterozygotes. British Journal of Haematology 107: 699-706, 1999.

29. Vrettou C, Palmer G, Kanavakis E, Tzetis M, Antoniadi T, Mastrominas M, **Traeger-Synodinos J**. A widely applicable strategy for single cell genotyping of β-thalassemia mutations using DGGE analysis: applcation to preimplantation genetic diagnosis. Prenatal Diagnosis 19: 1209-1216, 1999.

30. Kanavakis E, Vrettou C, Palmer G, Tzetis M, Mastrominas M, **Traeger-Synodinos J.** Preimplantation genetic diagnosis in 10 couples at risk for transmitting β-thalassemia major: clinical experience including the initiation of six singleton pregnancies. Prenatal Diagnosis 19: 1217-1222, 1999.

31. Vrettou C, Kanavakis E, **Traeger-Synodinos J,** Metaxotou-Mavrommati A, Basiakos I, Maragoudaki E, Stamoulakatou A, Papassotiriou I, Katamis C. Molecular studies in β-thalassemia heterozygotes with raised HbF levels. Hemoglobin 24: 203-220, 2000.

32. **Traeger-Synodinos J**, Papassotiriou I, Metaxotou-Mavrommati A, Vrettou C, Stamoulakatou A, Kanavakis E. Distinct phenotypic expression associated with a new hyperunstable alpha globin variant (Hb Heraklion, α1cd37(C2)Pro>0): comparison to other α-thalassemic hemoglobinopathies. Blood Cells, Molecules and Diseases 26: 276-284, 2000

33. Kanavakis E, Papassotiriou I, Karagiorga M, Vrettou C, Metaxotou-Mavrommati A, Stamoulakatou A, Kattamis C, **Traeger-Synodinos J**. Phenotypic and molecular diversity of haemoglobin H disease: a Greek experience. British Journal of Haematology 111: 915-923, 2000.

34. Harteveld CL, **Traeger-Synodinos J**, Ragusa A, Fichera M, Kanavakis E, Kattamis C, Giordano P, Schilirio G, Bernini LF. Multicentric origin of Hb Constant Spring [α2 codon142 TAA>CAA]. Haematologica 86: 36-40, 2001.

35. Papassotiriou I, **Traeger-Synodinos J**, Prome D, Kister J, Vrettou C, Xaidara A, Marden M, Stamoulakatou A, Wajcman H, Kanavakis E. Hemoglobin Sitia [b128(H6) Ala>Val]: An unstable variant with a substitution in the α1β1 interface. Hemoglobin 25: 45-56, 2001.

36. Wajcman H., Lahary A, Promé D, Kister J, Riou J, Godart C, Préhu C, **Traeger-Synodinos J**, Papassotiriou I, Galactéros F. Hb Mont Saint Aignan [b128 (H6) Ala>Pro] a new unstable variant leading to chronic microcytic anemia. Hemoglobin 25: 57-65, 2001.

37. **Traeger-Synodinos J**, Papassotiriou I, Vrettou C, Skarmoutsou C, Stamoulakatou A, Kanavakis E. Erythroid marrow activity and functional anemia in patients with the rare interaction of a single functional α-globin and β-globin gene. Haematologica 86: 363-376, 2001.

38. Palmer G, **Traeger-Synodinos J**, Davies S, Tzetis M, Vrettou C, Mastrominas M, Kanavakis E. Pregnancies following blastocyst transfer in PGD cycles at risk for β-thalassaemic haemoglobinopathies. Human Reproduction 17: 25-31, 2002.

39. Kanavakis E, **Traeger-Synodinos J**. Preimplantation genetic diagnosis in clinical practice. Journal of Medical Genetics 39: 6-11, 2002. (Invited review)

40. Geraedts JP, Harper J, Braude P, Sermon K, Veiga A, Gianaroli L, Agan N, Munne S, Gitlin S, Blenow E, de Boer K, Hussey N, **Traeger-Synodinos J**, Lee S-H, Viville S, Krey Ray P, Emiliani S, Liu YH, Vermeulen S. Preimplantation genetic diagnosis (PGD), a collaborative activity of clinical genetic departments and IVF centres. Prenatal Diagnosis 22: 451-454, 2002.

41. Vrettou C, Tzetis M, **Traeger-Synodinos J**, Palmer G, Kanavakis E. Multiplex sequence variation detection throughout the CFTR gene appropriate for preimplantation genetic diagnosis in populations with heterogeneity of cystic fibrosis mutations. Molecular Human Reproduction 8: 880-886, 2002.

42. **Traeger-Synodinos J**, Papassotiriou I, Karagiorga M, Premetis E, Kanavakis E, Stamoulakatou A. Unusual phenotypic observations associated with a rare HbH disease genotype ($--^{Med}/\alpha^{TSaudi}\alpha$): implications for clinical management. British Journal of Haematology 119: 265-267, 2002.

43. Kanavakis E, Efthymiadou A, Strofalis S, Doudounakis S, **Traeger-Synodinos J**, Tzetis M. Cystic fibrosis in Greece: molecular diagnosis, haplotype frameworks, prenatal diagnosis and carrier identification amongst high-risk individuals. Clinical Genetics 63: 400-409, 2003.

44. Vrettou C, **Traeger-Synodinos J**, Tzetis M, Malamis G, Kanavakis E. Rapid screening of multiple β-globin gene mutations by real time PCR (Lightcycler): application to carrier screening and prenatal diagnosis for thalassemia syndromes. Clinical Chemistry 49: 769-776, 2003.

45. **Traeger-Synodinos J**, Vrettou C, Palmer G, Tzetis M, Mastrominas M, Davies S, Kanavakis E. An evaluation of preimplantation genetic diagnosis in clinical genetic services through three years application for prevention of β-thalassaemia major and sickle cell thalassaemia. Molecular Human Reproduction 9: 301-307, 2003.

46. Skarmoutsou C, Papassotiriou I, **Traeger-Synodinos J**, Stamou H, Ladis V, Metaxotou-Mavrommati A, Stamoulakatou A, Kanavakis E. Erythroid bone marrow activity and red cell hemoglobinization in iron sufficient β-thalassemia heterozygotes by measurement of soluble transferrin receptor (sTfR) and reticulocyte hemoglobin content (CHr): correlation with genotypes and Hb $A_2$ levels. Haematologica 88: 631-636, 2003.

47. Stamoulakatou A, Athanasiou-Metaxa M, **Traeger-Synodinos J,** Lazaropoulou C, Harteveld K, Premetis E, Tsantali H, Zorai A, Giordano P, Papassotiriou I, Kanavakis E. Rare thalassemic syndrome caused by interaction of Hb Questembert (α1 codon 131, TCT>CCT, Ser>Pro) with an α-thalassemia-2 deletion: implications for diagnosis and management. Blood Cells Molecules and Diseases 32: 118-123, 2004.

48. Kanavakis E, **Traeger-Synodinos J**, Lafioniatis S, Lazaropoulou C, Liakopoulou T, Paleologos G, Metaxotou-Mavrommati A, Stamoulakatou A, Papassotiriou I. A rare example that co-inheritance of a severe form of β-thalassemia and α-thalassemia interact in a "synergistic" manner to balance the phenotype of classic thalassemic syndromes. Blood Cells, Molecules and Diseases 32: 319-324, 2004.

49. Karagianni C, Stabouli S, Roumeliotou K, **Traeger-Synodinos J,** Kavazarakis D, Gourgiotis D, Lambrou J. Kanavakis E. Severe hypertriglyceridaemia in diabetic ketoacidosis: clinical and genetic study. Diabetic Medicine 20: 21: 380-382, 2004.

50. Vrettou C, **Traeger-Synodinos J**, Tzetis M, Palmer G, Sofocleous C, Kanavakis E. PCR for single cell genotyping in sickle cell and thalassemia syndromes as a rapid, accurate, reliable and widely applicable protocol for preimplantation genetic diagnosis. Human Mutation 23: 513-521, 2004.

51. Dimisianos G, **Traeger-Synodinos J**, Vrettou C, Papasotiriou I, Kanavakis E. A rare 33bp in-frame deletion of the $\alpha$1-globin gene causing $\alpha^+$ -thalassemia: a second observation. Hemoglobin 28(2):137-43, 2004.

52. Kavazarakis E, Stabouli S, Gouriotis D, Roumeliotou K, **Traeger-Synodinos J,** Bossios A, Fretzayes A, Kanavakis E. Severe hypertriglyceridemia in a Greek infant: Clinical, Biochemical and Genetic Study. European Journal of Pediatrics 163(8):462-6, 2004.

53. Talmaci R, **Traeger-Synodinos J**, Kanavakis E, Coriu D, Colita D, Gavrila L. Scanning of beta-globin gene for identification of beta-thalassemia mutation in Romanian population. Journal of Cellular and Molecular Medicine 8(2):232-240, 2004.

54. Schulpis KH, Tsakiris S, **Traeger-Synodinos J**, Papassotiriou I. Low total antioxidant status is implicated with high 8-hydroxy-2-deoxyguanosine serum concentrations in phenylketonuria. Clinical Biochemistry 38(3):239-42, 2005.

55. Kokkali G, Vrettou C, **Traeger-Synodinos J**, Jones GM, Cram DS, Stavrou D, Trounson AO, Kanavakis E, Pantos K. Birth of a healthy infant following trophectoderm biopsy from blastocysts for PGD of {beta}-thalassaemia major: Case report. Human Reproduction 2005 Jul;20(7):1855-9

56. Papassotiriou I, **Traeger-Synodinos J**, Marden MC, Kister J, Liapi D, Prome D, Stamoulakatou A, Wajcman H, Kanavakis E. The homozygous state for Hb Crete [beta129 (H7) Ala-->Pro] is associated with a complex phenotype including erythrocytosis and functional anemia. Blood Cells Molecules and Diseases 34(3):229-34, 2005.

57. Harper JC, Boelaert K, Geraedts J, Harton G, Kearns WG, Moutou C, Muntjewerff N, Repping S, SenGupta S, Scriven PN, **Traeger-Synodinos J**, Vesela K, Wilton L, Sermon KD. ESHRE PGD Consortium data collection V: cycles from January to December 2002 with pregnancy follow-up to October 2003. Human Reproduction 21(1):3-21, 2006.

58. Zerefos PG, Ioannou PC, **Traeger-Synodinos J**, Dimissianos G, Kanavakis E, Christopoulos TK. Photoprotein aequorin as a novel reporter for SNP genotyping by primer extension-application to the variants of mannose-binding lectin gene. Human Mutation 27(3):279-85, 2006.

59. Traeger-Synodinos J. Real-time PCR for prenatal and preimplantation genetic diagnosis of monogenic diseases. Molecular Aspects of Medicine 27(2-3):176-91, 2006. (Invited review)

60. Papassotiriou I, Stamoulakatou A, Wajcman H, Kister J, Dimisianos G, Lazaropoulou C, Kanavaki I, Vavourakis E, Kattamis A, Kanavakis E, **Traeger-Synodinos J**. Observation of a rare hemoglobin variant [Hb Lulu island, beta107(G9)Gly-->Asp, GGC-->GAC] co-inherited with a beta+-thalassemia mutation [IVS-I-110 (G-->A)] or in the heterozygous state in a Greek-Albanian family. Hemoglobin 30(4):409-18, 2006.

61. Sermon KD, Michiels A, Harton G, Moutou C, Repping S, Scriven PN, Sengupta S, **Traeger-Synodinos J**, Vesela K, Viville S, Wilton L, Harper JC. ESHRE PGD Consortium data collection VI: cycles from January to December 2003 with pregnancy follow-up to October 2004. Human Reproduction 22(2):323-36, 2007.

62. Litos IK, Ioannou PC, Christopoulos TK, **Traeger-Synodinos J**, Kanavakis E. Genotyping of single-nucleotide polymorphisms by primer extension reaction in a dry-reagent dipstick format. Analytical Chemistry 15;79(2):395-402, 2007.

63. Glynou K, Kastanis P, Boukouvala S, Tsaoussis V, Ioannou PC, Christopoulos TK, **Traeger-Synodinos J**, Kanavakis E. High-Throughput Microtiter Well-Based Chemiluminometric Genotyping of 15 HBB Gene Mutations in a Dry-Reagent Format. Clinical Chemistry 53(3):384-91 2007.

64. Kokkali G, **Traeger-Synodinos J,** Vrettou C, Stavrou D, Jones GM, Cram DS, Makrakis E, Trounson AO, Kanavakis E, Pantos K. Blastocyst biopsy versus cleavage stage biopsy and blastocyst transfer for preimplantation genetic diagnosis of beta-thalassaemia: a pilot study. Human Reproduction 22(5):1443-9, 2007.

65. Voutoufianakis S, Psoni S, Vorgia P, Tsekoura F, Kekou K, **Traeger-Synodinos J**, Kitsiou S, Kanavakis E, Fryssira H. Coinheritance of mutated SMN1 and MECP2 genes in a child with phenotypic features of spinal muscular atrophy (SMA) type II and Rett syndrome. European Journal of Paediatric Neurology 11(4):235-9, 2007.

66. Cremonesi L, Ferrari M, Giordano PC, Harteveld CL, Kleanthous M, Papasavva T, Patrinos GP, **Traeger-Synodinos J**. An overview of current microarray-based human globin gene mutation detection methods. Hemoglobin 31(3):289-311, 2007. Review.

67. Kolialexi A, Vrettou C, **Traeger-Synodinos J**, Burgemeister R, Papantoniou N, Kanavakis E, Antsaklis A, Mavrou A. Noninvasive prenatal diagnosis of beta-thalassaemia using individual fetal erythroblasts isolated from maternal blood after enrichment. Prenatal Diagnosis 27(13):1228-32, 2007.

68. Harper JC, de Die-Smulders C, Goossens V, Harton G, Moutou C, Repping S, Scriven PN, SenGupta S, **Traeger-Synodinos J,** Van Rij MC, Viville S, Wilton L, Sermon KD. ESHRE PGD consortium data collection VII: cycles from January to December 2004 with pregnancy follow-up to October 2005. Human Reproduction 23(4):741-55, 2008.

69. Iliadi AC, Ioannou PC, Traeger-Synodinos J, Kanavakis E, Christopoulos TK. High-throughput microtiter well-based bioluminometric genotyping of two single-nucleotide polymorphisms in the toll-like receptor-4 gene. Analytical Biochemistry 376(2):235-41, 2008.

70. Gibbons RJ, Wada T, Fisher CA, Malik N, Mitson MJ, Steensma DP, Fryer A, Goudie DR, Krantz ID, **Traeger-Synodinos J**. Mutations in the chromatin-associated protein ATRX. Human Mutation 29(6):796-802, 2008.

71. Goossens V, Harton G, Moutou C, Scriven PN, **Traeger-Synodinos J**, Sermon K, Harper JC. ESHRE PGD Consortium data collection VIII: cycles from January to December 2005 with pregnancy follow-up to October 2006. Human Reproduction, 23(12):2629-45, 2008.

72. Wajcman H, **Traeger-Synodinos J,** Papassotiriou I, Giordano PC, Harteveld CL, Baudin-Creuza V, Old J. Unstable and thalassemic alpha chain hemoglobin variants: a cause of Hb H disease and thalassemia intermedia. Hemoglobin 32(4):327-49, 2008. Review.

73. Douna V, Papassotiriou I, Garoufi A, Georgouli E, Ladis V, Stamoulakatou A, Metaxotou-Mavrommati A, Kanavakis E, **Traeger-Synodinos J**. A rare thalassemic syndrome caused by interaction of Hb Adana

[alpha59(E8)Gly-->Asp] with an alpha+-thalassemia deletion: clinical aspects in two cases. Hemoglobin 32(4):361-9, 2008.

74. Douna V, Liapi D, Kampourakis D, Repapinou Z, Papassotiriou I, Stamoulakatou A, Poziopoulos C, Kanavakis E, **Traeger-Synodinos J.** First observation of Hb Taybe [Codons 38/39 (-Acc) Thr-->0 (alpha1)] in Greece: clinical and hematological findings in patients with co-inherited alpha+-thalassemia mutations. Hemoglobin 32(4):371-8, 2008.

75. Douna V, Papassotiriou I, Metaxotou-Mavrommati A, Stamoulakatou A, Liapi D, Kampourakis D, Tsilimigaki A, Kanavakis E, **Traeger-Synodinos J.** Further identification of the hyperunstable alpha-globin chain variant Hb Heraklion [codons 36/37 (-CCC); Pro-->0 (alpha1)] in Greek cases with co-inherited alpha+-thalassemia mutations. Hemoglobin 32(4):379-85, 2008.

76. Douna V, Papassotiriou I, Stamoulakatou A, Metaxotou-Mavrommati A, Emmanuel Kanavakis E, **Traeger-Synodinos J**. Association of mild and severely unstable $\alpha$ chain variants: the first observation of a compound heterozygotes with Hb Setif [$\alpha$94(G1)Asp>Tyr ($\alpha$2)] AND Hb Agrinio [$\alpha$29(B10)Leu>Pro ($\alpha$2)] in a Greek family. Hemoglobin 32(6):592-5, 2008.

77. Destouni A Vrettou **C,** Antonatos D, Chouliaras G, **Traeger-Synodinos J**, Patsilinakos S, Kitsiou-Tzeli S, Tsigas D, Kanavakis E. Cell-Free DNA level in Acute Myocardial Infarction Patients during Hospitalization. Acta Cardiologica. Acta Cardiologica 64(1):51-7, 2009.

78. Wilton L, Thornhill A, **Traeger-Synodinos J,** Sermon K, Harper JC. The causes of misdiagnosis and adverse outcomes in PGD. Human Reproduction 24(5):1221-8, 2009.

79. Megremis S, Mitsioni A, Mitsioni A, Fylaktou I, Kitsiou-Tzelli S, Stefanidis C, Kanavakis E, **Traeger-Synodinos J**. Nucleotide variations in the NPHS2 gene in Greek children with steroid resistant nephrotic syndrome (SRNS). Genetic Testing and Molecular Biomarkers 13(2):249-56, 2009.

80. Litos IK, Ioannou PC, Christopoulos TK, **Traeger-Synodinos J**, Kanavakis E. Multianalyte, dipstick-type, nanoparticle-basεd DNA biosensor for visual genotyping of single-nucleotide polymorphisms. Biosensors and Bioelectronics 15;24(10):3135-9, 2009.

81. Goossens V, Harton G, Moutou C, **Traeger-Synodinos J**, Van Rij M, Harper JC. ESHRE PGD Consortium data collection IX: cycles from January to December 2006 with pregnancy follow-up to October 2007. Human Reproduction 24(8):1786-810, 2009.

82. Psoni S, Willems PJ, Kanavakis E, Mavrou A, Frissyra H, **Traeger-Synodinos J**, Sofokleous C, Makrythanassis P, Kitsiou-Tzeli S. A novel p.Arg970X mutation in the last exon of the CDKL5 gene resulting in late-onset seizure disorder. European Journal of Paediatric Neurology 14(2):188-91, 2009.

83. Harteveld CL, Kleanthous M, **Traeger-Synodinos J**. Prenatal Diagnosis of Haemoglobin Disorders: present and future strategies Clinical Biochemistry 42(18):1767-79, 2009. (Invited review)

84. Lederer CW, Basak. N, Aydinok Y Christou S, El-Beshlawy A, Eleftheriou A, Fattoum S, Felice A, Fibach E, Galanello R, Gambari R, Gavrila L, Giordano PC, Grosveld F, Hassapopoulou H, Hladka E, Kanavakis E, Locatelli F, Old JM, Patrinos GP, Romeo G, Taher A, **Traeger-Synodinos J,** Vassiliou P, Villegas A, Voskaridou E, Wajcman H, Zafeiropoulos A, Kleanthous M. An Electronic Infrastructure for Research and Treatment of the

Thalassemias and other Hemoglobinopathies: the Euro-Mediterranean ITHANET Project. Hemoglobin, Hemoglobin 33(3):163-76, 2009.

85. Phylipsen M, Amato A , Cappabianca MP, **Traeger-Synodinos J,** Kanavakis E, Basak AN, Galanello R, Tuveri T, Ivaldi G, Harteveld CL, Giordano PC. Two new β-Thalassemia deletions compromising prenatal diagnosis in an Italian and a Turkish couple seeking prevention. Haematologica 94(9):1289-92, 2009

86. Christofidou Ch, Sofocleous Ch, Vrettou Ch, **Traeger- Synodinos J**, Kekou K, Palmer G, Kokkali G, Mavrou A, Kitsiou-Tseli S, Kanavakis E. Preimplantation genetic diagnosis for X-linked and gender dependant disorders using a robust, flexible single-tube PCR protocol Reproductive BioMedicine Online, 19(3), 418–425, 2009.

87. Tsiakalou V, Petropoulou M; Ioannou P, **Traeger-Synodinos J**, Christopoulos T, Kanavakis E, Anagnostopoulos Ni, Savvidou I. Bioluminometric Assay for Relative Quantification of Mutant Allele Burden. Application to the Oncogenic Somatic Point Mutation JAK2V617F. Analytical Chemistry 81(20): 8596-602, 2009

88. Iliadi A, Makrythanasis P, Tzetis M, Tsipi M, **Traeger-Synodinos J**, Ioannou PC, Rapti A, Kanavakis E, Christopoulos TK. Association of TLR4 Single-Nucleotide Polymorphisms and Sarcoidosis in Greek Patients. Genetic Testing and Molecular Biomarkers 13(6):849-53, 2009.

89. Kitsiou-Tzeli S, **Traeger-Synodinos J**, Giannatou E, Kaminopetros P, Roma E, Makrythanasis P, Tsezou A. The c.504T>C (p.Asn168Asn) polymorphism iin the *ABCB4* gene is a predisposing factor for intrahepatic cholestasis of pregnancy in Greece. Liver International, 30(3):489-91, 2010.

90. Destouni A, Vrettou C, **Traeger-Synodinos J**, Davies S, Mastrominas M, Kanavakis E. PGD for Glycogen Storage Disease type IV: Birth of healthy twins following successful clinical application of a mutation specific protocol. Prenatal Diagnosis 30(2):180-2, 2010.

91. Psoni S, Sofocleous C, **Traeger-Synodinos J,** Kitsiou-Tzeli S, Kanavakis E, Fryssira-Kanioura H. Phenotypic and genotypic variability in four males with MECP2 gene sequence aberrations including a novel deletion. Pediatric Research 67(5):551-6, 2010.

92. Harper J, Coonen E, De Rycke M, Fiorentino F, Geraedts J, Goossens V, Harton G, Moutou C, Pehlivan Budak T, Renwick P, Sengupta S, **Traeger-Synodinos J**, Vesela K. What next for preimplantation genetic screening (PGS)? A position statement from the ESHRE PGD Consortium Steering Committee. Human Reproduction 25(4):821-3, 2010.

93. Konstantou JK, Iliadi AC, Ioannou PC, Christopoulos TK, Anagnostopoulos NI, Kanavakis E, **Traeger-Synodinos J**. Visual screening for JAK2V617F mutation by a disposable dipstick. Analytical Bioanalytical Chemistry 397(5):1911-6, 2010.

94. Goussetis E, Peristeri I, Kitra V, **Traeger-Synodinos J,** Theodosaki M, Psarra K, Kanariou M, Tzortzatou-Stathopoulou F, Petrakou E, Fylaktou I, Kanavakis E, Graphakos S. Successful long-term immunologic reconstitution by allogeneic hematopoietic stem cell transplantation cures patients with autosomal dominant hyper-IgE syndrome. Journal of Allergy and Clinical Immunology 126(2):392-4, 2010.

95. Harper JC, Coonen E, De Rycke M, Harton G, Moutou C, Pehlivan T, **Traeger-Synodinos J**, Van Rij MC, Goossens V. ESHRE PGD consortium data collection X: cycles from January to December 2007 with pregnancy follow-up to October 2008. Human Reproduction 25(11):2685-707, 2010.

96. **Traeger-Synodinos J**, Douna V, Papassotiriou I, Stamoulakatou A, Ladis V, Siahanidou T, Fylaktou I, Kanavakis E. Variable and often severe phenotypic expression in patients with the α-thalassemic variant Hb Agrinio [α29(B10)Leu→Pro (α2)]. Hemoglobin 34(5):430-8, 2010.

97. Shammas C, Papasavva T, Felekis X, Christophorou C, Roomere H, **Synodinos JT**, Kanavakis E, El-Khateeb M, Hamamy H, Mahmoud T, Shboul M, El Beshlawy A, Filon D, Hussein IR, Galanello R, Romeo G, Kleanthous M. ThalassoChip, an array mutation and single nucleotide polymorphism detection tool for the diagnosis of β-thalassaemia. Clinical Chemistry and Laboratory Medicine 48(12):1713-8, 2010.

98. Harton G, De Rycke M, Fiorentino F, Moutou C, SenGupta S, **Traeger-Synodinos J**, Harper JC. ESHRE PGD consortium best practice guidelines for amplification-based PGD Human Reproduction 26(1):33-40, 2011.

99. Harton G., Braude P., Lashwood A., Schmutzler A., **Traeger-Synodinos J**., Wilton L., Harper J.C. ESHRE PGD consortium best practice guidelines for organization of a PGD centre for PGD/preimplantation genetic screening Human Reproduction 26(1):14-24, 2011.

100. Zachaki S, Vrettou C, Destouni A, Kokkali G, **Traeger-Synodinos J**, Kanavakis E. Novel and Known Microsatellite Markers Within the β-Globin Cluster to Support Robust Preimplantation Genetic Diagnosis of β-Thalassemia and Sickle Cell Syndromes. Hemoglobin 35(1):56-66, 2011.

101. Waye JS, Nakamura-Garrett LM, Eng B, Kanavakis E, **Traeger-Synodinos J**. β(+)-Thalassemia Trait Due to a Novel Mutation in the β-Globin Gene Promoter: -26 (A>C) [HBB c.-76A>C]. Hemoglobin. 35(1):84-6, 2011.

102. Giardine B, Borg B, Higgs DR, Maglott D, Nazli Basak A, Clark B, Faustino P, Felice A, Francina A, Gallivan MVE, Georgitsi M, Gibbons RJ, Giordano PC, Harteveld CL, Joly P, Kanavakis E, Kollia P, Menzel S, Miller W, Moradkhani K, Old J, Papachatzopoulou A, Papadakis MN, Papadopoulos P, Pavlovic S, Philipsen S, Radmilovic M, Riemer C, Schrijver I, Stojiljkovic M, Thein SL, **Traeger-Synodinos J,** Tully R, Wada T, Waye J, Wiemann C, Zukic B, Chui DHK, Wajcman H, Hardison RC, Patrinos GP. Systematic documentation and analysis of human genetic variation in hemoglobinopathies using the microattribution approach. Nature Genetics 20;43(4):295-301, 2011.

103. **Traeger-Synodinos J**, Vrettou C, Kanavakis E. Prenatal, non-invasive and pre-implantation genetic diagnosis of inherited disorders: the example of the hemoglobinopathies. Expert Review in Molecular Diagnostics 11(3):299-312, 2011.

104. Megremis S, Mitsioni A, Fylaktou I, Tzeli SK, Komianou F, Stefanidis CJ, Kanavakis E, **Traeger-Synodinos J**. Broad and unexpected phenotypic expression in Greek children with steroid-resistant nephrotic syndrome due to mutations in the Wilms' tumor 1 (WT1) gene. European Journal of Pediatrics, 170(12):1529-34, 2011.

105. Morlighem JÉ, Harbers M, **Traeger-Synodinos J,** Lezhava A. DNA amplification techniques in pharmacogenomics. Pharmacogenomics, 12(6):845-60, 2011.

106. Litos IK, Ioannou PC, Christopoulos TK, Tzetis M, Kanavakis E, **Traeger-Synodinos J**. Quadruple-allele dipstick test for simultaneous visual genotyping of A896G (Asp299Gly) and C1196T (Thr399Ile) polymorphisms in the toll-like receptor-4 gene. Clinical Chimica Acta, 412(21-22):1968-72, 2011.

107. Iliadi A, Petropoulou M, Ioannou PC, Christopoulos TK, Anagnostopoulos NI, Kanavakis E, **Traeger-Synodinos J**. Absolute Quantification of the Alleles in Somatic Point Mutations by Bioluminometric Methods based on Competitive Polymerase Chain Reaction in the Presence of a Locked Nucleic Acid Blocker or an Allele-Specific Primer. Analytical Chemistry 83(17):6545-51, 2011.

108. Psoni S, Sofocleous C, **Traeger-Synodinos** J, Kitsiou-Tzeli S, Kanavakis E, Fryssira-Kanioura H. MECP2 mutations and clinical correlations in Greek children with Rett syndrome and associated neurodevelopmental disorders. **Brain and Development** 34(6):487-95, 2012.

109. Phylipsen M, **Traeger-Synodinos** J, van der Kraan M, van Delft P, Bakker G, Geerts M, Kanavakis E, Stamoulakatou A, Karagiorga M, Giordano PC, Harteveld CL. A novel α(0) -thalassaemia deletion in a Greek patient with HbH disease and β-thalassaemia trait. **European Journal of Haematology** 88(4):356-62, 2012.

110. Harper JC, Wilton L, **Traeger-Synodinos J**, Goossens V, Moutou C, Sengupta SB, Pehlivan Budak T, Renwick P, De Rycke M, Geraedts JP, Harton G. The ESHRE PGD Consortium: 10 years of data collection. Human Reproduction Update, 18(3):234-47, 2012.

111. Destouni A, Christopoulos G, Vrettou C, Kakourou G, Kleanthous M, **Traeger-Synodinos J,** Kanavakis E. Microsatellite Markers Within the α-Globin Gene Cluster for Robust Preimplantation Genetic Diagnosis of Severe α-Thalassemia Syndromes in Mediterranean Populations. Hemoglobin, 36(3):253-64, 2012.

112. Durmaz B, Ozkinay F, Onay H, Karaca E, Aydinok Y, Tavmergen E, Vrettou C, **Traeger-Synodinos J**, Kanavakis E. Genotyping of β-Globin Gene Mutations in Single Lymphocytes: A Preliminary Study for Preimplantation Genetic Diagnosis of Monogenic Disorders. Hemoglobin 36(3):230-43, 2012.

113. Papanikos F , Iliadi A, Petropoulou M, Ioannoua PC, Christopoulosb TK,, Kanavakis E, **Traeger-Synodinos J**. Lateral flow dipstick test for genotyping of 15 beta-globin gene (HBB) mutations with naked-eye detection. Analytica Chimica Acta 727: 61-66, 2012.

114. Goossens V, **Traeger-Synodinos** J, Coonen E, De Rycke M, Moutou C, Pehlivan T, Derks-Smeets IA, Harton G. ESHRE PGD Consortium data collection XI: cycles from January to December 2008 with pregnancy follow-up to October 2009. Human Reproduction, 27(7):1887-911, 2012.

115. **Traeger-Synodinos J**, Kakourou G, Vrettou C, Kanavakis E. Looking to the future: developments in preimplantation genetic diagnosis Expert Review of Obstetrics and Gynecology, 7(4): 293-295, 2012.

116. Thomaidis L, Kitsiou-Tzeli S, Kritselis E, Drandakis H, Touliatou V, MantoudisS, Leze E, Destouni A, **Traeger-Synodinos J**, Kafetzis D, Kanavakis E. Psychomotor development of children born after preimplantation genetic diagnosis and parental stress evaluation. World J Pediatr. 2012 Nov;8(4):309-16.

117. Kitsiou-Tzeli S, Deligiorgi M, Malaktari-Skarantavou S, Vlachopoulos C, Megremis S, Fylaktou I, **Traeger-Synodinos** J, Kanaka-Gantenbein C, Stefanadis C, Kanavakis E Sertoli cell tumor and gonadoblastoma in an untreated 29-year-old 46,XY phenotypic male with Frasier syndrome carrying a WT1 IVS9+4C>T mutation. Hormones (Athens). 2012 Jul;11(3):361-7.

118. Deans Z, Fiorentino F, Biricik A, **Traeger-Synodinos** J, Moutou C, De Rycke M, Renwick P, Sengupta S, Goossens V, Harton G. The experience of 3 years of external quality assessment of preimplantation genetic diagnosis for cystic fibrosis. Eur J Hum Genet. 2012 Nov 14. doi: 10.1038/ejhg.2012.244.

119. Papasavva TE, Lederer CW, **Traeger-Synodinos** J, Mavrou A, Kanavakis E, Ioannou C, Makariou C, Kleanthous M. A Minimal Set of SNPs for the Noninvasive Prenatal Diagnosis of β-Thalassaemia. Ann Hum Genet. 2013 Jan 31. doi: 10.1111/ahg.12004.

**SCIENTIFIC BOOKS**

Editor and contributor in 2; Contributor in 4

1. **Traeger-Synodinos J.** New Developments in Prenatal Diagnosis. In: The Prevention of Thalassaemia and other haemoglobinopathies, Volume 1. Eds. Agastiniotis M, Eleftheriou A, Galanello R, Old J, Petrou M, Traeger-Synodinos J. **Thalassaemia International Federation**, **2003.** (2nd edition in preparation).

2. **Traeger-Synodinos J.** New technologies for DNA mutation detection. In: The Prevention of Thalassaemia and other haemoglobinopathies, Volume 2, A Laboratory Manual. Eds. Agastiniotis M, Eleftheriou A, Galanello R, Old J, Petrou M, Traeger-Synodinos J. **Thalassaemia International Federation. Ist edition 2004; 2nd edition 2012.**

3. Kanavakis E, **Traeger-Synodinos J.** Molecular basis of thalassaemia syndromes In **Disorders** of **Iron** Homeostasis,. **Erythrocytes** and **Erythropoiesis**. Beaumont, Ph. Beris, C. Brugnara, M. Cazzola Eds. European School of Haematology. **2006**

4. **Traeger-Synodinos J,** Vrettou C, Kanavakis E Real-time PCR for prenatal diagnosis of monogenic diseases caused by single nucleotide changes – the example of the hemoglobinopathies. In **Real Time PCR (BIOS Advanced Methods)**. Ed Tevfik Dorak. Taylor & Francis Group. **2007**

5. **Traeger-Synodinos J,** Vrettou C, Kanavakis E. Rapid Detection of Fetal Mendelian Disorders: Thalassemia and Sickle Cell Syndromes. In **Prenatal Diagnosis Series: Methods in Molecular Biology,** Vol. 444, Hahn, S; Jackson, LG. (Eds.) Humana Press. **2008**

6. **Traeger-Synodinos J,** Harteveld CL. Disease services: Haemoglobinopathies. In **Molecular Diagnosis of Genetic Diseases: Methods and Protocols,** 3rd Edition. Rob Elles; Andrew Wallace Eds. Humana Press. In press.

7. **Traeger-Synodinos J,** Staessen C. Preimplantation genetic diagnosis. In **Textbook of Human Reproductive Genetics**. Cambridge University Press, In preparation.

**Invited talks at International meetings, since 2002**

| | |
|---|---|
| 2012 | 3rd Pan European Conference on Haemoglobinopathies and Rare Anaemias, October 2012, Limassol, Cyprus<br>Title: Advances in Molecular Diagnosis of Haemoglobinopathies. |
| 2012 | XXV International Symposium on Technical Innovations in Laboratory Hematology, Nice, France<br>Title: Hemoglobinopathy prevention and preimplantation genetic diagnosis (PGD). |
| 2011 | 12th International Conference on Thalassaemias and Haemoglobinopathies, Antalya, Turkey<br>Title: The Molecular Basis of Alpha Thalassaemia» |
| 2011 | Catalan Network for the Diagnosis and Clinical Management of Major Haemoglobinopathies, ENERCA, Barcelona, Spain<br>Title: New developments in preimplantation and non-invasive prenatal diagnosis» |

2011 International Symposium "Progress in management of hemoglobin disorders: new perspectives for diagnostics, prevention and care", Rome, Italy
Title: The Greek experience and PGD.

2010 Golden Helix Symposium, Genetic Analysis In Translational Medicine, Athens.
Title: Prenatal, non-invasive and preimplantation genetic diagnosis of inherited disorders: Lessons from the hemoglobinopathies»

2009 European Hemoglobinopathy Forum, Paris, France
Title: An update on the epidemiology of hemoglobinopathies in Europe.

2008 9th International Thalassemia Conference, Cairo, Egypt.
Title: Advances in approaches for non-invasive prenatal diagnosis (NIPD) and preimplantation genetic diagnosis (PGD) of thalassemia.

2008 16th Meeting of Balkan Meeting of Balkan Clinical Laboratory Federation, Athens, Greece.
Title: Molecular diagnostics and Clinical chemistry: applications, technologies, laboratory requirements and quality control.

2007 3rd Symposium Cyprus Society of Human Genetics, Nicosia, Cyprus
Title Molecular mechanisms and phenotype-genotype relationships in monogenic diseases: lessons from the haemoglobinoptahies».

2007 17th International Federation of Clinical Chemistry - FESCC European Congress of Clinical Chemistry and Laboratory Medicine Euromedlab, Amsterdam, The Netherlands.
Title: Haemoglobinopathies Identification of carriers: Screeing for what?.

2007 International Scientific Symposium: New Methods and their applications to Assisted Reproduction. Athens.
Title: Preimplantation Genetic Diagnosis for monogenic diseases: Current and emerging methods.

2006. Workshop on New Technologies for Non-Invasive Prenatal Diagnosis of Haemoglobinopathies, Limassol, Cyprus
Title: Locked nucleic acids (LNA) modifed oligonucleotides

2005 Dubai International Pathology and Genetics Conference, Dubai, United Arab Emirates.
Title; Advances in methods for DNA analysis applied to prenatal and preimplantation genetic diagnosis of the hemoglobinopathies».

2002 UK Forum on Haemoglobin Disorders, Kings College Hospital, London, UK.
Title: Preimplantation genetic diagnosis applied for β-thalassaemic haemoglobinopathies: 3 years clinical experience in Greece.

**Abstracts at scientific meetings**

International meetings: >100

Greek meetings: >95

**Patents**

A patent awarded by the Greek Patent Register (OBI www.**obi**.gr). Application Number OBI 201002924/03-09-10

Title: Method and rapid multi-allele diagnostic test for molecular diagnosis of human β-haemoglobinopathies", Ioannou P, Christopoulos TK, Traeger-Synodinos J, Papanikos F, Iliadi A, Petropoulou M, Elenis D.

**Awards**

As a member of the research team in the First Department of Paediatrics and subsequently in the Laboratory of Medical Genetics I have been given the following awards (all related to molecular genetics and molecular diagnosis of human monogenic diseases):

1. The Greek Haematology Society (1997, 1998, 2011).
2. The Greek Society of Clinical Chemistry (1998)
3. The Greek Society of Paediatrics (1986, 1987, 1991, 1992, 1993, 1994, 1995, 1996, 1997, 1998, 1999, 2000, 2001, 2007, 2009).
4. The Greek Society of Paediatric Haematology-Oncology (2010)
5. The Royal College of Obstetrics and Gynaecology (2011.
6. The 4th International Congress –The young Woman at the Rise of 21st Century (1998)

**Scholarships**

Greek National Research Foundation as an associate researcher in the First Department of Paediatrics: 1984-1985.

**FUNDED RESEARCH PROJECTS**

**EU funded projects**

1990-1994 "Thalassemia Intermedia Syndromes: Molecular, Hematological, Clinical and Therapeutic Studies", Life Sciences and Technologies for Developing Countries (STD3). Three collaborating groups, with Athens group as co-ordinator. EU contribution 683,000ECU.

• 2002-2005, entitled: "Novel genechip technology for simplified detection of molecularly heterogeneous genetic diseases: Detection of cystic fibrosis as a model" (CF-CHIP) (see http://www.nmrc.ie/projects/cf-chip/), funded under the EU Fifth Framework, Quality of Life Programme (contract no. QLK3-CT-2001-01982) for the development of new diagnostics. Ten collaborating groups, EU contribution of 1,633,300 Euro (Athens group 150,000 Euro ).

• 2006-2008, entitled: "Infrastructure for Thalassaemia Research Network" (Ithanet) funded under the EU Sixth Framework, Research Infrastructures - Communication Networks Development (contract number 026539). (see http://www.ithanet.eu/). EU contribution 1,200,000 Euro (Athens group 44,000 Euro).

**Other funding**

• **2012 John S**. **Latsis** Public Benefit **Foundation award**. Project title "microRNAs studies in normal and thalassaemic human erythroid progenitors to investigate modifiers of globin expression and potential therapeutic pathways for thalassaemia syndromes". I am Principle Investigator. (12,000 Euro).

• **2012 European Society of Human Reproduction and Embryology (ESHRE) funded project entitled**

"The ESHRE Study into the evaluation of oocyte euploidy by microarray analysis (ESTEEM) trial". A pragmatic, multicentre, randomized double-blind controlled trial with an intention-to-treat analysis of the use of preimplantation genetic screening (PGS) for aneuploidy by means of microarray comparative genomic hybridization (aCGH) for the chromosomal analysis of the polar bodies (PB) of oocytes collected after ovarian stimulation for in vitro fertilization (IVF) and with the intention to assess the genetic competence of oocytes of advanced biological age and the effect of this technique on reproductive outcome.

I am one of the Principle Investigators. I co-ordinate the research team in Athens and mediate between ESHRE and Athens. Funding will be centrally controlled by ESHRE and will cover the costs of all consumables for the microarray analysis and travel expenses for meetings.

I have also played fundamental roles in several other national (Greek) research proposals.