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SECOND MEMORIAL IDA BIANCO AND EZIO SILVESTRONI

INTERNATIONAL SYMPOSIUM

Progress in management of hemoglobin disorders: new perspectives for diagnostics, prevention and care

Nobile Collegio Chimico Farmaceutico
Universitas Aromatariorum Urbis
Via in Miranda 10
Fori Imperiali

Rome, 4th November 2011

A.N.M.I. ONLUS
Centro Studi delle Microcitemie,
Rome - Italy
Director: Dr. Antonio Amato

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Creteil, France

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Leiden, The Netherlands

A. Streetly
London, UK

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Athens, Greece

M. Kleanthous
Nicosia, Cyprus

C. Hoppe
Oakland CA, USA

C. Borgna
Ferrara, Italy

M.D. Cappellini
Milan, Italy

G. Ferrari
Milan, Italy

G. Lucarelli
Rome, Italy

Greetings to

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blod
Prevention, cure and research for Thalassemia

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FACOLTÀ DI MEDICINA E CHIRURGIA - UNIVERSITÀ DEGLI STUDI "TOR VERGATA",
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Centro di Studi della Microcitemia di Roma
Direttore: Dott. Antonio Amato

International Symposium

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Roma 4 novembre 2011
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Via in Miranda, 10 - Roma

Patrocini Concessi
Patrocinio Regione Lazio,
Patrocinio Comune di Roma
Patrocinio EURISPES

Patrocini Richiesti
Patrocinio Ministero della Salute
Patrocinio Provincia di Roma

TARGET: MEDICI SPECIALISTI IN EMATOLOGIA

MODALITÀ DI EROGAZIONE: RESIDENZIALE

TEAM DI PROGETTO

Responsabile Scientifico
Dr. Antonio Amato – Direttore Centro Studi Microcitemia di Roma (Anmi Onlus) - Roma

IL RAZIONALE DEL TEMA:

Il Convegno si propone di mettere in luce le più significative esperienze di prevenzione delle talassemie a livello mondiale, confrontare i programmi di prevenzione in atto o in divenire presso popolazioni con aspetti culturali, etici e religiosi differenti e infine mettere in evidenza alcuni tra gli interventi più innovativi nell'ambito del trattamento delle talassemie, tra cui l'evoluzione e le prospettive della terapia genica.
L'Incontro avrà tra i relatori il Prof. Giordano (Olanda), il Prof. Harteveld (Olanda), il Prof. Fiorelli (Italia), il Prof.

Wajcman (Francia), la Prof.ssa Streetly (UK), il Prof Traeger Synodinos (Grecia), la Prof.ssa Kleanthous (Cipro), la Prof.ssa Hoppe (USA), la Prof.ssa Cappellini (Italia), la Prof.ssa Ferrari (Italia) e il Prof. Lucarelli (Italia). La Presidenza è affidata al Prof. Isacchi (Roma TVG).
L'evento si presenta come una assise di grande rilevanza scientifica, unico nel suo genere in Italia, per il quale verrà dato ampio spazio a livello nazionale ed internazionale; gli atti ed i materiali prodotti costituiranno oggetto di pubblicazione web ed in letteratura scientifica.

OBIETTIVI FORMATIVI
Aggiornare sui Progressi nello Studio delle Patologie Emoglobiniche con approfondimenti in Campo Diagnostico, Preventivo e Terapeutico

Obiettivo dell'evento: Linee guida – Protocolli – Procedure – Documentazione clinica

ObiettIvo

competenze tecnico-professionali - Conoscere e valutare le principali esperienze di prevenzione delle patologie emoglobiniche a livello mondiale, considerando elementi di efficienza ed efficacia nelle diverse condizioni culturali, etiche e religiose delle popolazioni nei confronti delle quali i protocolli vengono attuati. Acquisire competenze nell'ambito delle tecniche diagnostiche più comunemente utilizzate per talassemie ed emoglobinopatie, dei loro sviluppi e dei procedimenti innovativi che consentono il riconoscimento e la diagnosi dei difetti genetici rari o ancora ignoti (nuovi difetti genetici). Avere conoscenza del progresso realizzato nella terapia delle patologie emoglobiniche mediante le nuove tecniche di monitoraggio delle malattie talassemiche ed i protocolli terapeutici in ambito clinico e gli effetti in termini di salute e qualità e durata di vita dei pazienti. Aprirsi alle prospettive future aggiornando le proprie conoscenze sui progressi nell'applicazione del TMO e della terapia genica, quali procedimenti per la guarigione definitiva da talassemia ed anemia drepanocitica.

competenze di processo - Permettere l'acquisizione delle conoscenze di laboratorio dei nuovi procedimenti diagnostici in termini di tecniche biochimiche per la diagnostica di base e di tecniche di biologia molecolare per la diagnostica di II° livello. In ambito più strettamente sanitario, analizzare le metodiche di nuova applicazione nel campo della medicina perinatale e prenatale, gli sviluppi dei protocolli clinici utilizzati per la ferrochelazione ed il trattamento delle complicanze delle emoglobinopatie ed ancora le prospettive attualmente offerte per la guarigione mediante il TMO e la terapia genica (stato dell'arte nell'allestimento dei protocolli applicativi nell'uomo).

competenze di sistema - In ambito diagnostico e preventivo integrare le competenze di medici e biologi per consentire lo sviluppo di piani di prevenzione territoriale per le malattie talassemiche, mettendo in rete l'operatività in campo diagnostico con la medicina del territorio e la specialistica materno-infantile. In ambito clinico aggiornare le conoscenze sulle nuove applicazione diagnostico-terapeutiche per poter offrire ai pazienti le migliori prospettive in termini di vita e salute.

Tipo materiale durevole rilasciato ai partecipanti: Libro degli Abstract, accesso on-line ai proceedings del Simposio pubblicati sul sito www.silvestronibiancomemorial.org

Ore formative: 7

Crediti conseguiti: 7

N. partecipanti : 50 partecipanti di cui 20 biologi e 30 medici specialisti in ematologia, patologia clinica, medicina preventiva

Corso Gratuito

A.N.M.I. O.N.L.U.S.
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Direttore: Dott. Antonio Amato

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Patrocinio EURISPES

Patrocini Richiesti
Patrocinio Ministero della Salute
Patrocinio Provincia di Roma

08.00 Registration desk open

08.45 **G. Isacchi**: Introduction
Saluti delle Autorità

09.00 Introductory Lecture - **G. Fiorelli**: Archaeology of the human globin genes.

SESSION 1: DIAGNOSTICS - Chairs: **A. Colosimo** / ?
09.20 **C. Harteveld:** New approaches in diagnostics for Thalassemia.
09.50 **H. Wajcman:** Advances in the methods for identification and characterization of abnormal hemoglobins.
10.20 Discussion

10.40 Coffee break

SESSION 2: PREVENTION - Chairs: **P.C. Giordano** / A. Amato

11.00 **P.C. Giordano:** Primary and secondary prevention of hemoglobinopathies in non-endemic areas.
11.15 **A. Streetly:** The national antenatal screening programme for sickle cell and thalassaemia in England.
11.45 **J. Traeger Synodinos:** Primary prevention in Greece and PGD.
12.15 **M. Kleanthous:** Prevention in Cyprus and non-invasive prenatal diagnosis.
12.45 **C. Hoppe:** Newborn screening of SCD in the USA.
13.15 Discussion

13.30 Lunch

SESSION 3: CLINICS - Chairs: **M.D. Cappellini** / L. Pagano

14.45 **C. Borgna:** Life expectancy in patients with TM: work in progress.
15.30 **M.D. Cappellini:** Treatment of thalassemia: what's the news?
16.00 Discussion

16.15 **Silvestroni and Bianco International Award - First Edition**
SESSION 4: FUTURE - Chairs: B. Dallapiccola / S. Storti
16.45 **G. Ferrari**: Gene therapy in beta-thalassemia
17.15 **G. Lucarelli:** Hemopoietic stem cell transplantation in hemoglobinopathies: from thalassemia to sickle-cell.
17.45 Discussion

18.00 Conclusions
18.30 ECM test

Abstract

Primary and secondary prevention of the hemoglobinopathies: Trials and implementations in endemic and non-endemic countries. - P. C. Giordano

The most common recessive hereditary disorder in human once restricted to the tropical and subtropical belt of the old world is today a matter of concern at the global level. What was a rare disease sporadically observed in non-endemic countries because of ancient migrations, sickle cell disease and thalassemia major have dramatically increased in the last four decades due to the intensive economic and political migrations. While in the same period well organized primary prevention has reduced the natural incidence in most South-European countries, the birth of severely affected children has grown significantly in the immigration countries of Northern Europe because of poor or absent prevention. Newborn screening, already present for secondary (morbidity) prevention of metabolic disease, has been expanded to hemoglobinopathies in several countries. However, even assuming a perfectly organized follow up of all affected births and reported carriers, less then half of the couples at risk will in this way be reached for an informed reproductive choice at the prospective level. Other more efficient tools like pre-conception and early pregnancy carrier screening, although mostly available on demand, are still not implemented at the national level in most immigration areas and are sometime even discouraged by conservative GP's and public health authorities. More efforts are needed to change these attitudes and to provide information to healthy carriers and to couples at risk who have the right to know and to be assisted in their reproductive choice.

New approaches in diagnostics for Thalassaemia - C. Harteveld

The thalassaemias are a diverse group of disorders of haemoglobin synthesis, all of which result from a reduced output of the alpha- or beta-chains of the adult haemoglobin. The molecular defects underlying these disorders are both deletions and point mutations in the alpha- or beta-globin genes or gene-clusters. To detect point mutations causing alpha- or beta-thalassaemia, direct sequencing is the method of choice to detect the widest spectrum of molecular defects. However, PCR based strategies like ASO-probe hybridization and ARMS are also frequently used by many labs world wide.

The most established approach in DNA diagnostics to screen for the most common deletion defects causing alpha-thalassaemia or beta-thalassaemia is gap-PCR, because the method makes use of equipment already available in most diagnostic laboratories, is inexpensive and fast. For less common rearrangements in the alpha- and beta-globin gene clusters, Multiplex Ligation-dependent Probe Amplification (MLPA) has become a standard tool, besides Southern blotting and cytogenetic methods. Multiplex Ligation-dependent Probe Amplification is a technology based on ligation of probe-pairs hybridized to a region of interest to detect deletions or duplications by quantitative PCR and fragment analysis. Due to the implementation of MLPA for the detection of copy number variation in the alpha- and beta-globin gene clusters, more and more new types of deletions were detected. But also the discovery of duplications of the complete alpha-globin gene cluster including the Major Conserved Regions shed light on some unexpectedly severe cases of beta-thalassaemia Intermedia in beta-thalassaemia carriers.

However, by using MLPA the exact breakpoints of these novel deletions and duplications remain unknown. Knowledge of breakpoint sequences might give more insight in the molecular mechanisms giving rise to these rearrangements and may facilitate primer design for gap-PCR to screen for certain common population specific deletions. For this purpose a custom fine-tiling array for high resolution breakpoint determination was designed to perform array Comparative Genome Hybridization (aCGH). The oligonucleotides cover the complete alpha- and beta-globin gene clusters including the neighboring regions. Based on the results breakpoint primers were designed to perform gap-PCR and breakpoint sequencing.

Primary prevention in Greece and preimplantation genetic diagnosis (PGD)

Joanne Traeger-Synodinos, DPhil (Oxon)
Medical Genetics, Athens University Medical School, St. Sophia's Children' Hospital, Athens 11527, Greece

In Greece the hemoglobinopathies have an estimated carrier frequency of 16%, which includes ~7% b-thalassemia, 8%

a-thalassemia (7% a+-thal; 1% a0-thal) and ~1% sickle cell. Based on the annual birth-rate, it is calculated that annually there are about 640 pregnancies at-risk for β-thal and sickle cell syndromes, and a further 120 at-risk for a-thalassemia (although, with only 1% a0-thal carriers, the risk for severe forms of a-thalassemia are rare in Greece). A programme of hemoglobinopathy screening was initiated in the mid-1970's and continues today under the auspices of the National Thalassemia Prevention Center, in the Laikon Hospital, Athens, complemented by molecular diagnostic services offered by the Laboratory of Medical Genetics, Athens University.

Primary screening for hemoglobinopathies is voluntary. It usually targets couples prior to, or very early in, pregnancy and it is based on a complete blood count, hemoglobin analysis (qualitative and quantitative), iron and ferritin measurements. Molecular analysis is applied to support definitive diagnosis, especially relevant in atypical cases, and also fundamental for the application of prenatal diagnosis in carrier couples. Molecular methods must address a wide spectrum of mutations underlying both b-thalassemia and a-thalassemia. In Greece, over 30 different b-globin gene mutations (including Hb S) have been observed, of which 6 account for >90% of disease alleles. In a-thalassemia, more than 15 different mutations have been observed, including 2 a+ deletions, 3 a0 deletions and >10 nondeletion a-thalassemia alleles.

At the Laboratory of Medical Genetics, Athens University, molecular diagnosis for hemoglobinopathies, including conventional prenatal diagnosis (based on analysis of trophoblast or amniocytes) uses methods such as amplification refractory mutation system (ARMS)-PCR, denaturing gradient gel electrophoresis (DGGE), gap-PCR, real-time PCR, sequencing and STR analysis. Newer methods such as High Resolution Melting Analysis (HRMA) are under development. The laboratory receives approximately 600-700 samples annually for molecular analysis, including 60-80 samples for conventional prenatal diagnosis.

Preimplantation genetic diagnosis (PGD) represents an alternative procedure to conventional prenatal diagnosis, with the advantage of avoiding termination of affected pregnancies. As a specialized form of PND it is particularly appropriate for couples with an unsuccessful reproductive history and/or requiring assisted reproduction. In the last 12 years, the Laboratory of Medical Genetics has developed and applied PGD protocols for many monogenic disorders, mainly the b-hemoglobinopathies. The number of couples expressing interest in PGD has been increasing steadily and the laboratory performs about 60 PGD cycles annually. Since 1997 over 300 couples have been counseled for PGD and almost 600 clinical cycles have been applied, from which >160 healthy babies have been born.

PGD requires close collaboration between experts in assisted reproductive techniques (ART) and genetics, and involves many stages. Genetic analysis can be performed on polar bodies (biopsied day 1 post-insemination), 1-2 blastomeres (biopsied day 3 post-insemination) or 5-10 trophectoderm cells (biopsied on day 5 from blastocysts). Whatever the stage of biopsy, the quantity of sample available for genetic analysis is very limited, and is often only a single cell. The limited sample (usually a single cell) is considered the most technically challenging aspect of PGD, compounded by the often sub-optimal quality of the embryo and/or cell biopsied. For monogenic diseases PGD involves methods which are almost exclusively based on PCR. Protocols have to be rapid, to produce a result within about 24-72 hours, (depending on stage of biopsy and day of embryo-transfer). Prior to clinical application, the protocol must be stringently optimized to address innate limitations of single-cell PCR (total PCR failure and allelic drop-out, ADO), and sample contamination. PCR failure, although undesirable, will not lead to an unacceptable misdiagnosis. On the other hand, ADO and contamination may lead to serious misdiagnosis. To support accurate genotyping and to monitor contamination it is recommended that optimized PGD-PCR protocols are based on multiplex and fluorescent PCR, analysing the parental mutation(s) along with several linked markers across the disease-associated locus. Stringent laboratory and clinical procedures are applied at all times, to support the transfer of unaffected embryos.

PGD for the selection of a histocompatible sibling to facilitate a bone marrow transplant in a thalassemia major patient is considered appropriate in a number of societies (where ethical controversies associated with donor-sibling selection have been resolved). However, the ultimate success i.e. the birth of an unaffected histocompatible baby is limited in practice: only 25% of two siblings/embryos will be HLA-matched, which combined with the chance that only 75% of these embryos will be unaffected for the hemoglobinopathy, means that only 18.8% of all embryos fertilized in any cycle will be suitable for transfer. With overall implantation and pregnancy delivery rates around 30%, the overall success rate for PGD-HLA matching rarely surpasses about 10-15% for any cycle initiated. Before embarking on this reproductive option all couples should be clearly counselled and informed about this. Despite the potential success several hundred clinical cycles have been reported to date

In conclusion, prevention of hemoglobinopathies in Greece, since 1978, has supported a big reduction in the birth of

affected newborns, with currently only around 10-12 new cases born annually. To some extent the few new cases are likely attributed to an ever widening number of private genetic laboratories performing carrier detection and prenatal diagnosis, along with the presence of new immigrants in Greece who may not be informed of their potential risk, presenting issues which need to be addressed.

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4. Theodoridou S, Alemayehou M, Prappas N, et al, Carrier screening and prenatal diagnosis of hemoglobinopathies. A study of indigenous and immigrant couples in northern Greece, over the last 5 years. Hemoglobin. 2008;32(5):434-9.
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Advances in the methods for identification and characterization of abnormal hemoglobins"
Henri Wajcman, Kamran Moradkhani

The two main questions are: when should an abnormal hemoglobin be fully characterized and what are the methods to use. At this date, more than 1100 variants have been reported in the HbVar database and if the frequent ones are easily recognized by a confrontation of some of their hematological, epidemiological, or biochemical characteristics, a precise identification of rare variants may be more difficult and requires a study at the molecular level using DNA or protein sequencing. The obvious circumstance in which a variant needs to be fully characterized is when it is observed in the context of a hematological disorder such as chronic hemolytic anemia, polycythemia or methemoglobinemia. The second main reason to characterize any variant is when it is found, even in a clinically normal individual, during a premarital screening or a genetic counselling, when the other partner carries a thalassemic trait, or HbS. For example if some variant with a HbD mobility is observed in a couple in which the second partner carries HbS, it is essential to know if the variant is Hb D Punjab, which associated to HbS leads to a severe sickle cell syndrome or some harmless HbD, such as Hb D-Iran or Korle Bu. Due to the high frequency of the HbS allele, it is not exceptional to see an HbS allele carrying a second mutation, which may or not modify the sickling properties and the biochemical behavior. An identical situation exists when one of the partner has a thalassemic trait and the other carries a variant.
The methods which are today used as routine for the diagnosis of a Hb variant are cation exchange-HPLC and capillary electrophoresis (CE) using instruments specially adapted to Hb analysis. Each of these methods has its own advantages and inconvenients. As an example HbE elutes at the same position as HbA2 in CE-HPLC and is clearly separated in CE. Concerning rare variants, a much larger number of data is available in the literature for HPLC than for CE. Elution profiles of the common variants are well established in both systems. CE
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Hemopoietic stem cell transplantation in hemoglobinopathies: from thalassemia to sickle-cell - Guido Lucarelli
Sickle Cell Anemia (SCA) remain a disease with high risk of morbidity and early death. Although medical treatments are life-extending, end-organ damage could not be avoided in most patients over time. In last years migrations of population with SC trait are increased, also in Italy, with the increase number of pediatric SCD.
Allogeneic haematopoietic stem cell transplantation (HSCT) is the only curative treatment for SCA. We report our experience concerning 13 geno-identical HSCT for SCA-patients prepared with the same myeloablative conditioning regiment consisting of Busulfan, Cyclophosphamide and rabbit ATG.
All patients had sustained engraftment. One patient became a stable mixed chimera with 25% of donor cells four years after transplantation. One patient died one year after transplantation. The probability of survival, SCA-free survival, and transplant related mortality at five years after transplant were 92%, 92%, and 7%, respectively. All twelve surviving patients remained free of any SCA-related events after transplantation.

Curriculum Vitae

DR ANTONIO AMATO

Il dott. Antonio Amato è nato a Catanzaro l'8 aprile 1960.
Si è laureato in Medicina e Chirurgia presso l'Università Cattolica del Sacro Cuore di Roma nell'ottobre 1984.
Ha conseguito la specializzazione in Ematologia Generale, Clinica e Laboratorio nel 1987 presso la stessa Università.
Ha conseguito diploma di perfezionamento in "Gestione Manageriale di Aziende Sanitarie" presso l'Università "La Sapienza" di Roma nel 1997 ed in "Fitoterapia Clinica" presso l'Università della Tuscia – Viterbo nel 2004.
Dal 1989 svolge attività professionale in qualità di Dirigente Medico del Centro Studi Microcitemia di Roma (ANMI Onlus) occupandosi di aspetti diagnostici e clinici delle talassemie ed emoglobinopatie.
Dal 2004 ha assunto l'incarico di Direttore dello stesso Centro.

CATERINA BORGNA

Born in Siena on Dec 2, 1946. Medical Degree magna cum laude at the University of Pavia, July 1971 Educational Council for Foreign Medical Graduates (ECFMG) 1972 Stage in Pediatric Hematology at Stanford University, California from March 1972 to the end of February 1973. Medical Fellow in Pediatric Hematology at Stanford University, California from March 1973 to February 1974. Stage in Pediatric Hematology and Hepatology, University of Paris-Sud, France from January to November 1975 Specialty Degree in Pediatrics and Hematology at the University of Pavia, Italy From October 1978 is Assistant Professor at the Pediatric Dept of the University of Pavia, Italy In November and December 1981 works at the Fred Hutchinson Cancer Research Center di Seattle (USA) where she performs the first bone marrow transplant for thalassemia in one of her Italian patients. In 1982, as the winner of a fellowship of the Associazione Italiana per la Ricerca contro il Cancro spends three months al Fred Hutchinson Cancer Research Center di Seattle (USA) In 1986 moves to Verona with the position of University Researcher From 1994 becomes Associate Professor of Pediatrics at the University of Ferrara, Italy, From 2001 is Full Professor and Chief of Pediatrics at the University of Ferrara

DR.SSA MARIA DOMENICA CAPPELLINI

Attività Didattica:
Docente: Corso di Medicina Interna (V-VI anno), Laurea in Medicina e Chirurgia
Coordinatore: Corso di Fisiopatologia Medica (Med 09), III anno, Laurea in Biotecnologie Mediche
Coordinatore: Corso di Applicazioni Biotecnologiche in Medicina II (Med 09), III anno, Laurea in Biotecnologie Mediche
Coordinatore: Corso di Fisiopatologia Medica (Med 09), II anno, Laurea in Tecniche di Laboratorio Biomedico
Docente (1.5 CFU) di Medicina Interna (Med 09) nell'ambito del corso di Patologia e farmacologia, Laurea in Assistenza Sanitaria (fino aa 2004)
Docente (1 CFU) Corso di Fisiopatologia:dalla cellula alla clinica, I anno, Laurea Magistrale in Biotecnologie Mediche e Medicina Molecolare
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Direttore: Scuola di Specializzazione in Ematologia II dal 2002
Docente: Dottorato di Medicina Molecolare
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Docente: Scuola di specializzazione in Ematologia I
Docente: Scuola di Specializzazione in Genetica Medica

Svolge inoltre :
a. esercitazioni pratiche agli studenti del V-VI anno di corso di medicina e Chirurgia
b. attività professionalizzante agli studenti del IV anno di corso in Medicina e Chirurgia
Dal 2002 al 2005:
Relatore di: 7 tesi di laurea in Medicina e Chirurgia

3 tesi di laurea in Biotecnologie Mediche Quinquennale
3 tesi di laurea in Biotecnologie Mediche Triennale
2 tesi di dottorato in Medicina Molecolare
Membro della Commissione programmazione della Facoltà di Medicina e Chirurgia fino al 30.9.05

Attività Assistenziale
Affidamento incarico dirigenziale al personale medico universitario convenzionato:
Responsabilità organizzativa "Centro Anemie Congenite" (dal 2000) 1° Divisione di Medicina Generale – Ospedale Maggiore Policlinico IRCCS, Milano
Responsabile UOS "Anemie Congenite" (dal Dicembre 2004) Dipartimento Emergenza Urgenza – Ospedale Maggiore Policlinico IRCCS, Milano Dipartimento Medicina Interna – Università di Milano

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Membro della Società Italiana di Ematologia
Membro della Società Europea di Ematologia
Presidente del Comitato Scientifico dell'Associazione Talassemici e Drepanocitici Lombardi
Membro del Comitato scientifico del TIF (Thalassemia International Federation, commissione WHO)
Membro del Board Editoriale di Haematologica
Membro della Commissione Tecnico Scientifica V Piano Sangue e Plasma (art.5 L.R. 05/2005) Regione Lombardia
Referee per le seguenti riviste scientifiche: British Journal of Hematology; Hemoglobin; Lancet; Journal of Pediatric.

Grants:
Ricerca corrente Ospedale Maggiore Policlinico IRCCS:
2002:Studio del ruolo di geni modificatori nell'espressione fenotipica delle Sindromi Talassemiche
2003: Studio del ruolo di geni modificatori nell'espressione fenotipica delle Sindromi Talassemiche (continuazione)
2004: Malattie multiorgano da difetti ereditari del Globulo Rosso: dalla prevenzione alla cura
2005: Malattie multiorgano da difetti ereditari del Globulo Rosso: dalla prevenzione alla cura (continuazione)
FIRB 2001 finanziato 2003: Mappa proteomica dell'eritrocita :implicazioni cliniche e terapeutiche (coordinatore scientifico della ricerca) Totale progetto: 814.000 euro
Ricerca Finalizzata 2002 "Relazione genoma /proteoma nelle patologie ereditarie del globulo rosso: implicazioni diagnostiche e terapeutiche (coordinatore scientifico della ricerca) Tot. 250.000 euro
PRIN 2002: Approccio proteomico al danno ossidativo della membrana eritrocitaria nlla beta-talassemia e nella drepanocitosi 41.800 Euro
PRIN 2004: Studio dell'espressione della AHSP (alpha-haemoglobin stabilizing protein) e della ferrochelatasi durante il differenziamento eritroide 44.400 Euro
Progetti Malattie rare ISS 2005: Definizione delle basi biochimiche e Molecolari delle Porfirie Acute: implicazioni diagnostiche e molecolari 80.000 euro
Progetto a Concorso 2005. Fondazione Policlinico, Mangiagalli, Regina Elena IRCCS: Diagnosi e trattamento degli eventi acuti nelle Sindromi Falcemiche. 50.000 euro

Interessi di ricerca: ha sempre svolto ricerca applicativa in ambito clinico con particolare riguardo per patologie ereditarie del globulo rosso estendendo i propri interessi di ricerca alle basi molecolari di tali patologie.
In particolare si è occupata della caratterizzazione molecolare delle sindromi talassemiche, dei difetti enzimatici eritrocitari , e di Porfirie. L'esperienza acquisita nell'ambito della diagnostica di patologie ereditarie mediante tecniche dibiologia molecolare, oltre ad avere portato all'identificazione di nuove mutazioni nei geni di cui si è occupata,responsabili di malattia, ha consentito di mettere a punto tecniche facilmente trasferibili alla medicina molecolare. Il laboratorio del Centro Anemie Congenite, di cui la sottoscritta è responsabile, presso l'Ospedale Maggiore Policlinico di Milano, è riferimento nazionale per la diagnosi molecolare delle Talassemie Intermedie e delle

Porfirie.
Ha sviluppato inoltre tecniche di biologia cellulare per lo studio in vitro dei meccanismi di regolazione dell'eritropoiesi con particolare riguardo allo switch dell'emoglobina e all'induzione di emoglobina fetale con prospettive di applicazione terapeutica per le sindromi talassemiche e la drepanocitosi.
Ha svolto ricerche cliniche in campo cardiologico, pneumologico ed endocrinologico nelle sindromi talassemiche in età adulta, per le quali non vi sono esperienze precedenti se non limitate all'età pediatrica. A questo proposito e' coautrice con A.Cohen (University of Pennsylvania) e J.Porter (University College, London) di linee guida per la cura del paziente talassemico adulto ed è delegato del TIF (in official relations with WHO) per l'implementazione di tali linee guida nei paesi del Mediterraneo e del Medio Oriente.
Dal 2002 è membro dell'Advisory Board (Novartis) per lo sviluppo di un nuovo chelante orale del ferro (ICL670) del quale è stata Principal Investigator per i seguenti protocoll: ICL 105; !CL 107; ICL 109. Tutti i protocolli sono stati approvati dal Comitato Etico dell'Ospedale Maggiore Policlinico presso cui sono state condotte le sperimentazioni previa regolare stipula di convenzione.

Pubblicazioni Scientifiche
1. N. 280 lavori originali **su riviste Peer-reviewed recensite in Medline**
2. N. 70 lavori scientifici **su riviste nazionali**
3. N. 35 capitoli su libri **internazionali e nazionali**
4. >500 atti ed abstracts **di congressi nazionali ed internazionali**
5. N. 100 Relazioni ad invito **a congressi nazionali ed internazionali**

Elenco lavori originali su riviste Peer-reviewed recensite in Meline
Triennio 2002-2005
1 Fiorelli G.,De Feo T.M.,Duca L.,Tavazzi D.,Nava I., Fargion S.,Cappellini M.D. Red blood cell antioxidant and iron status in alcoholic and non alcoholic cirrhosis Eur.J. Clin.Invest. 32,21-7,2002 IF: 1.907
2 Cighetti G.,Duca L., Bortonel., Sala S., Nava I., Fiorelli G., Cappellini M.D Oxidative status and malondialdehyde in beta-thalassemia patients Eur. J.Clin. Invest. 32,55-60,2002 IF: 1.907
3 Tavazzi D., Martinez di Montemuros F., Fargion S., Fracanzani A.L., Fiorelli G., Cappellini M.D. Levels of uroporphyrinogen decarboxylase (URO-D) in erythrocytes of italian porphyria cutanea tarda patients. Cell. Mol. Biol. 48, 27-32, 2002 I.F:1.747
4 Cappellini M.D.,Martinez di Montemuros F., Di Pierro E., Fiorelli G. Hematologically important mutations: acute intermittent porphyria Blood Cell Mol. Dis. 28, 5-12, 2002 I.F. 1.772
5 Ceci A., Bainardi P., Felisi M., Cappellini M.D., Carnelli V., De Sanctis V et al. (The Italian registry for the controlled use of Deferiprone) Safety and effectiveness of deferiprone after three years follow up in an italian population at large.
Brit. J.Haematol. 118,330-6,2002 I.F.3.052
6 Cappellini M.D. The adult thalassemic patient Haematologica: 87; 125,2002 I.F.:3.453
7 Karimi M., Yarmohammadi H.,Farjadian S., Zinali S., Moghaddam Z., Cappellini M.D., Giordano P.C. Beta-thalassemia intermedia from southern Iran:IVS-II-1(G-A) is the prevalent thalassemia intermedia allele. Hemoglobin:, 26,147-54, 2002 I.F.0.570
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9 Canavese C, Gabrielli D, Guida C, Cappellini MD. Nephrologists and porphyrias. G Ital Nefrol. 2002 Jul-Aug;19(4):393-412. 10 Karimi M, Martinez di Montemuros F, Danielli MG, Farjadian S, Afrasiabi A, Fiorelli G, Cappellini MD. Molecular characterization of glucose-6-phosphate dehydrogenase deficiency in the fars province of Iran. Haematologica. 2003 Mar;88(3):346-7. I.F.:3.453
11 Martinez di Montemuros F, Di Pierro E, Patti E, Tavazzi D, Danielli MG, Biolcati G, Rocchi E, Cappellini MD. Molecular characterization of porphyrias in Italy: a diagnostic flow-chart. Cell Mol Biol (Noisy-le-grand). 2002 Dec;48(8):867-76. I.F:1.747
12 Martinez di Montemuros F, Tavazzi D, Patti E, Cappellini MD. Gene symbol: UROD. Disease: Porphyria, cutanea tarda.
Hum Genet. 2003 Sep;113(4):368-9 I.F. 4.022

13 Dongiovanni P, Valenti L, Fracanzani AL, Cappellini MD, Fargion S, Taioli E. TNFalpha promoter polymorphisms in Italian patients with porphyria cutanea tarda. Dig Liver Dis. 2003 Aug;35(8):596-7. I.F.:1.463
14 Martinez di Montemurous F, Tavazzi D, Patti E, Cappellini MD Human gene mutations. Gene Symbol: UROD. Disease: Porphyria, cutane tarda. Hum Genet. 2004 Jan 114(2):221 I.F. 4.022
15 Di Pierro E, Moriondo V, Cappellini MD. Human gene mutations. Gene Symbol: FECH. Disease: Porphyria, eritropietic.
Hum Genet. 2004 Jan 114(2):221 I.F. 4.022
16 Di Pierro E, Moriondo V, Cappellini MD. Gene Symbol: FECH. Disease: Porphyria, erythropoietic. Hum Genet. 2004 May;114(6):609 I.F. 4.022
17 Di Pierro E, Moriondo V, Cappellini MD. Gene Symbol: FECH. Disease: Porphyria, erythropoietic.
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18 Di Pierro E, Roselli EA, Cappellini MD. Gene symbol: HBMS. Disease: Porphyria, acute intermittent.
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24 Di Pierro E, Patti E, Cappellini MD Gene symbol: HMBS. Disease: Porphyria, acute intermittent. Hum Genet 2004 Jul 15(2)170.
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25 Patti E, Di Pierro E. Cappellini MD Gene symbol: PPOX. Disease: variegate porphyria. Hum Genet. 2004 Jul; 115(2):170 I.F. 4.022
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Haematologica. 2004 Oct 89 (10): 1179-86 I.F.:3.453
27 Borgna-Pignatti C, Rugolotto S, De Stefano P, Zhao H, Cappellini MD, Del Vecchio GC, Romeo MA, Forni GL, Gamberoni MR, Ghilardi R, Piga A, Cnaan A. Survival and complications in patients with thalassemia major treated with trasfusione and deferoxmine Haematologica 2004 oct. 89 (10):1187-93 I.F.:3.453
28 Castelli R, Graziaei G, Karimi M, Cappellini MD. Intrathoracic masses due to extramedullary hematopoiesis. Am J Med. Sci. 2004 Nov, 328 (5):299-303 I.F.:1.404
29 Cappellini MD: Iron-chelating therapy with the new oral agent ICL670 (Exjade (®)) Best Pract Res Clin Haematol. 2005 Jun, (2):289-98
30 Derchi G, Forni GL, Formisano F, Cappellini MD, Galanello R, D'Ascola G, Bina P, Magnano C, Lamagna M. Effecacy and safety of sildenafil in the treatment of severe pulmonary hypertension in patients with hemoglobinopathies. Hematologica 2005, 90 (4):452-458. I.F.:3.453
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intermittent. Hum. Genet. (2005) 116:533-545. I.F. 4.022
34 Di Pierro E, Brancaleoni V, Cappellini MD. Human gene mutations. Gene symbol: HMBS. Disease: Porphyria acute intermittent. Hum. Genet. (2005) 116:533-545. I.F.4.022
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36 Agostoni PG, Cerino M , Palermo P, Magini A , Bianchi , Bussotti P, Fiorelli G , Cappellini MD Exercise capacity in patients with beta-thalassaemia intermedia.Br J Haematol Oct, 131 (2): 27 IF 3.267

Totale IF: 103.637
Totale IF corretto per lettere: 62.271

GIULIANA FERRARI

1985: Laurea in Scienze Biologiche, Università di Milano.
1986: Post-doctoral fellow, The Wistar Institute of Anatomy and Biology, Philadelphia, PA, U.S.A.
1989-1992: Assistente Ricercatore, Laboratorio di Ematologia, Istituto Scientifico H.S.Raffaele, Milano.
1992-1997: Assistente Ricercatore, Laboratorio di Espressione Genica, DIBIT, Istituto Scientifico H.S.Raffaele, Milano.
1996-2000: Project leader, HSR-Telethon Institute for Gene Therapy (TIGET), Istituto Scientifico H.S.Raffaele, Milano.
2001-presente: Capo Unità, Unità di Trasferimento Genico in Cellule Staminali, HSR-TIGET, Istituto Scientifico H.S.Raffaele, Milano.
2006- presente: Professore Associato di Biologia Molecolare, Università Vita-Salute S.Raffaele, Milano.
2008-presente: Coordinatore dell'Area Ricerca di Base, HSR-TIGET, Istituto Scientifico H.S.Raffaele, Milano.

GEMINO FIORELLI

Nato a Pergola (Pesaro e Urbino) l' 8/2/1929
Laureato in Medicina e Chirurgia IL 16/11/1953 presso l'Università di Roma
Dal 1/11/1980: Professore Ordinario di Medicina Interna, Università degli Studi di Milano
Dal 1/11/1991 al 30/9/2004: Direttore dell'Unità Operativa di Medicina Interna 1^ presso l'Ospedale Policlinico di Milano IRCCS
Dal 1/11/1998 al 30/9/2001: Direttore del Dipartimento di Medicina Interna, Università di Milano
Dal 20/1/1998 al 30/9/2004: Direttore della I Scuola di Specializzazione in Medicina Interna, Università degli Studi di Milano.
Dal 1/10/2004: Professore Emerito dell'Univeristà di Milano
Ha svolto attività didattica nel Corso di Medicina Interna e ha tenuto lezioni nelle Scuole di Specializzazioni (Medicina Interna e Ematologia) dell'Università di Milano e anche presso altre Università.
Ha dato significativi contributi scientifici nell'ambito della ricerca clinica come documentato da oltre 300 pubblicazioni scientifiche.
Ha partecipato a Congressi Nazionali ed Internazionali con relazioni su argomenti inerenti le tematiche di ricerche sviluppate nel corso degli anni.
E' stato Presidente della Società Italiana di Medicina Interna - Sezione Regionale Lombarda nel triennio 1993/1995.
E' stato Presidente della Società Italiana di Alcologia - Sezione Regionale Lombarda nel triennio 1995/1998.
E' stato membro del Direttivo della Società Italiana di Medicina Interna (1998-2001).
E' Membro di diverse Società Mediche Nazionali ed internazionali.
Le tematiche di ricerca riguardano:
a) Deficit di G6PD eritrocitaria con riferimento alla eterogeneità biochimica e molecolare e alle manifestazioni cliniche.
b) Sindromi talassemiche con particolare riguardo alla eterogenità molecolare e agli aspetti clinici (sovraccarico di ferro, epatopatie, ecc…)
c) Emocromatosi ereditaria con riferimento agli aspetti epidemiologici e alla patologia epatica da sovraccarico di ferro.

P.C. GIORDANO

Graduated as chemistry engineer in Italy was appointed at the Institute of Human Genetics at Leiden University (The Netherlands) in 1966. Became involved in research projects on protein structure and function, hemoglobin expression, pathology and genetics. Was concerned with hemoglobinopathy diagnostic from 1984 and defended in 1998 a PhD thesis entitled "Hemoglobinopathies in The Netherlands, diagnostics, epidemiology and prevention". As Head of the Dutch reference laboratory for hemoglobinopathy diagnostics and associated professor, acquired specializations in Clinical Biochemical Genetics and in Clinical Molecular Genetics. As vice-president of the Dutch multi-ethnic patient's association, scientific adviser of the Dutch foundation for quality diagnostic and of the Dutch Public Health Council focused his interest on preventive medicine in immigrant populations. Emeritus from Mei 2010, continues his activities as advisor, associated editor and member of several editorial boards.

CORNELIS L. HARTEVELD

(PhD), Molecular and Biochemical Geneticist

Dr Cornelis L. Harteveld is molecular and biochemical geneticist at the department of Human and Clinical Genetics, Leiden University Medical Center. He obtained his bachelor of science in biochemical chemistry from the Anthony van Leeuwenhoek Institute in Delft in 1987 and graduated in Molecular Genetics at Leiden University in 1991. He did his PhD on the Molecular Basis of alpha-thalassaemia in 1998 and is presently working on hemoglobinopathies as an Assistant-Professor, at the department of Human Genetics at the academic hospital Leiden. Dr. Hartevelds principle areas of research and diagnostics are phenotype-genotype correlation of hemoglobinopathies and the technical improvement of diagnostics. He is responsible for the diagnosis of carriers and patients and for prenatal diagnosis for thalassemia Major and Sickle Cell disease in the Dutch Reference Laboratory for Hemoglobinopathies within the department of Human and Clinical Genetics at the Leiden University Medical Center in Leiden, The Netherlands.

CAROLYN HOPPE

Carolyn Hoppe is a hematologist/oncologist at CHO and a clinical scientist at CHORI with a focus in translational research in sickle cell disease. Her interest in studying genetic modifiers of sickle cell disease began early in her fellowship, while working in the laboratory of Dr. Elizabeth Trachtenberg at CHORI. A recipient of the American Society of Hematology Fellow Scholar Award, she performed her initial studies investigating HLA associations with stroke risk in children with sickle cell anemia (SCA).

Dr. Hoppe was awarded the Doris Duke Charitable Foundation Clinical Scientist Development Award (DDCF CSDA) to extend her work on genetic modifiers of stroke to a larger population of children with SCA. She successfully sought approval from the Stroke Prevention Trial for Sickle Cell Anemia (STOP) and the Cooperative Study of Sickle Cell Disease (CSSCD) executive committees for use of over 360 archived DNA samples.

In addition to her initial studies investigating HLA markers of stroke, Dr. Hoppe has expanded her research to include other potential genetic modifiers of stroke in children with SCD. Through her continued collaboration with Dr. Henry Erlich in the Department of Human Genetics at Roche Molecular Systems (RMS), and Dr. Janelle Noble at CHORI, she has applied novel genotyping assays to identify potential risk-conferring genes in SCD.

Dr. Hoppe has been a team member of the Northern California Comprehensive Sickle Cell Program for the past five years. As the medical director of the CHORI Hemoglobinopathy Laboratory and the California State Newborn Screening for Hemoglobinopathies Follow-up Program, Dr. Hoppe has expanded the services provided by this program to include molecular diagnostics. More recently, Dr. Hoppe and Dr. Frans Kuypers have jointly established the CHORI Laboratory Diagnostics Center and have recently received funding to serve as the central genotype-phenotype laboratory for the CSCC-Collaborative Database Project.

GUIDO LUCARELLI

Name : Guido Lucarelli

Born : 6.2.1934 ; Lucca,

Degree : Medicine, July 1959, University of Bologna
Assistant in Medicine : 1960-1963, University of Parma
N.H.I. – Public Health
Service International
Postdoctoral Research
Fellow : 1963-1964, Tuft's University, Boston
Fellow in Hematology : 1964-1965, Tuft's University, Boston
Professor in Hematology : July 1967, University of Parma
Chief of Hematology : 1965-1970, University of Parma
Visiting Professor : 1978, Seattle, U.S.A.
Chief in Hematology : 1971 - 2001, Hospital of Pesaro
Director : 2002 - International Project on Transplantation in Thalassemia
Scientific Director : 2003 - 2007- International Center for Transplantation in Thalassemia and Sickle Cell Anemia, Mediterranean Institute of Hematology (IME), Roma
Director Transplant Program (active) : 2005, International Center for Transplantation in Thalassemia and Sickle Cell Anemia, Mediterranean Institute of Hematology (IME), Roma
DirectorTransplant Program (active) : 2005, International Center for Transplantation in Thalassemia and Sickle Cell Anemia, Mediterranean Institute of Hematology (IME), Roma
Member of ASH 1993 : Ham-Wasserman Lecture
Past Fields of interest In research : Regulatory mechanism of fetal and neonatal Erythropoiesis
Experimental Fetal Liver Transplantation
Human Fetal Liver Transplantation
Current fields of interest In research : Clinical: Bone Marrow Transplantation in Thalassemia and Sickle Cell Anemia
Persistent Mixed Chimerism in Thalassemia

ALLISON STREETLY OBE

Dr Streetly is the Programme Director of the NHS Sickle Cell and Thalassaemia Screening Programme. She is a Consultant in Public Health. Since 2001 she is been seconded to the National Screening Committee (NSC). She set up the NHS Sickle Cell & Thalassaemia Screening Programme which completed implementation of newborn screening in 2006 and antenatal screening in 2008. She has also supported the for the development of a network of centres for the care of children with sickle cell disorders to ensure that babies identified by the screening process are enrolled into effective care programmes. The programme steering group is Chaired by the Archbishop of York Dr John Sentamu and close involvement of the voluntary sector and users has always been a key aspect of the programmes working as has community engagement work more generally.
Dr Streetly studied medicine and social and political sciences at Cambridge University and completed her clinical training at St Thomas' Hospital. She trained in Public Health at the London School of Hygiene and Tropical Medicine and SE London, Kent and East Sussex.
Roles have included:
Co-ordinating a national audit of the newborn bloodspot screening programmes in England (1994-1998) which to the establishment of the Newborn Bloodspot Screening Programme Centre.
Advising the National Confidential Enquiry into Patient Outcome and Death (2008) including member of expert advisory group on audit of sickle and thalassaemia deaths.
Membership:
Medical advisor to the Sickle Cell society and screening advisor to the UKTS.
Fellow of the Faculty of Public Health.
Member of the Fetal, Maternal & Child Health sub-group of the NSC of the UK
Member of the Diagnostic and Screening Panel of the Health technology Assessment Programme.

JOANNE RACHEL TRAEGER-SYNODINOS
Date & place of birth: 9th February 1957, London, UK.
Nationality: British (and Greek through marriage).
Current work address: Laboratory of Medical Genetics, National & Kapodistrian University of Athens, Choremio Research Laboratory, St. Sophia's Children's Hospital, Thivon & Levadias St., Athens 11527, Greece. Tel: +30 10 746 7461 ; Fax: +30 10 779 5553 **email**: jtraeger@med.uoa.gr

HIGHER EDUCATION
1975- 1978 **B.Sc. Hons** in Physiology and Biochemistry,

University of Reading,UK
1978-1982 **Doctor of Philosophy**, (D.Phil. Oxon)

Entitled "The synthesis of Haemoglobin E", in the Nuffield Department of Medicine, under Sir Professor David Weatherall,
University of Oxford, UK.

WORK EXPERIENCE
On completion of my doctorate, I moved to Greece (to accompany my husband), where my academic contacts in the field of haemoglobinopathies led immediately to a position:
1983-1985 Research associate of the Choremio Research Laboratory,

First Department of Paediatrics, Athens University, located in the
St. Sophia's Children's Hospital. Supported by a research
scholarship from the (Greek) National Research Institute.
1986-2005 Clinical Scientist at the St. Sophia's Children's Hospital,

(largest paediatric hospital in Greece), allowing continued collaboration with the Choremio Research Laboratory and Athens University, and additionally involvement in clinical laboratory services, including haematology and molecular diagnostics.
2006-present Assistant Professor of Genetics, Laboratory of Medical Genetics,

National & Kapodistrian University of Athens, Choremio Research
Laboratory, St. Sophia's Children's Hospital.

RESEARCH INTERESTS
I have been involved in the field of molecular biology and its application to human genetics for more almost 30 years, including
Genetic analysis of common and rare monogenic diseases, including haemoglobinopathies, cystic fibrosis, phenylketonuria, familial hypercholesterolaemia, RETT syndrome, pediatric podocytopathies.
Genotype-phenotype correlations in the above disorders, applied for disease prognosis, patient management and genetic counselling.
the development of analytical protocols for detecting genetic variation and genetic diagnostic procedures, mainly for molecular genetics applications
improving and developing approaches for prenatal diagnosis, including alternative approaches such as non-invasive prenatal diagnosis (NIPD) and preimplantation genetic diagnosis (PGD).

PEER REVIEWED PUBLICATIONS

I have >95 publications in peer reviewed scientific journals. Those more closely related to the scientific field related to this project include:

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.

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Honorary Assistant (Department of Hæmatology [Pr. Dacie], Royal Post-Graduate Medical School, Hammersmith Hospital, London) (1973)
Visiting Assistant Professor (Department of Biochemistry [Pr. Jones], University of Oregon, Portland) (1976),
Visiting Assistant Professor Department of Hematology [Pr. Nagel], Albert Einstein College of Medicine, New York (1984)
Laboratory of Molecular Biology [Pr. Perutz], MRC, Cambridge (1974)

Teaching Activity:

Faculty of Medicine Cochin-Port Royal: several lectures on protein biochemistry and hemoglobin disorders

1977-1979. Lecturer in Biochemistry at the Faculty of Medicine of Lomé (Togo) (within the cooperative program between the Faculty of Medicine Cochin and the University of Benin)

Lectures within many specialized curses organized by the Faculty of Medicine of Créteil.

Clinical activity:
Involved about 20% of the time in the clinical activity for diagnosis of hemoglobin disorders (Laboratory of Molecular Genetics, Henri Mondor Hospital)

Editorial Activity:

Editor-in-Chief of the journal **HEMOGLOBIN** since 1998

Scientific Secretary of the Proceedings of the French Academy of Science **(CR BIOLOGIES)** since 2005

Member of the Editorial Board of the Balkan Journal of Medical Genetics (Skopje, Macedonia)

Database Activity:

Curator of the **HbVar** database on abnormal hemoglobins

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