THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS

2013

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Message from the President of the Board of Directors

Christos Eliades

2012 was a landmark year, in that it materialized the dream of all previous CING Boards of Directors and the Scientific staff… Hard work has laid down the foundations of the School of Molecular Medicine, the inauguration of which on 17/10/2012 was marked by the presence and speech of Nobel Prize winner Prof. James Watson.

The CING, just like other world-wide known centers of excellence, is now steadily standing on three pillars: education – research – treatment. Our fellow Human, patients, were and will always be at the center of our activities, and the inauguration of the School of Molecular Medicine opens new doors for research, always at the service of Humanity, of patients. This patient-centric vision is why we also hired extra medical staff, to dramatically shorten the waiting list time, thus improving patient treatment conditions and quality of life.

At the same time, the CING excelled in breakthrough research that got it under the spotlights of the global scientific community… the publication of the team under Prof. Philippos Patsalis on the non-invasive pre-natal method for detecting the Down syndrome received global acclaim… By the same token, the same CING team received €1,6M grant (part of a bigger multi-country €11M grant) for the creation of a printer-size machine that will be able to detect the Down syndrome and other genetic diseases as well as another €2,5M grant from Europe's prestigious ERC program for the development of non-invasive prenatal diagnosis tests for almost all known genetic disorders. Of course there are other research achievements which cannot be listed in the context of a short message… suffice to say however, that the CING grant success ratio, is well above average.

It was always my firm belief, that the greatest treasure that little Cyprus has was/is its human 'capital'. The discovery of natural gas reserves is a gift from Mother Nature; however, the real treasure of this island is its people, and the CING has demonstrated that regardless of its small size, little Cyprus can play on the global scene.. Cyprus has the enviable title of having the world-wide highest per capita ratio of university graduates, and that's its greatest asset. It's this human capital that makes me believe that Cyprus will be able to leave behind the gloom of the economic crisis, just like it did in 1974, and will again be facing the future with confidence and determination.

Message

Message from the Chief Executive Medical Director

Philippos C. Patsalis

The Cyprus Institute of Neurology & Genetics continues to operate as a Centre of Excellence. Every year, the provision of medical and biomedical services is increased and upgraded to satisfy the needs of our patients and society. In parallel, we seek and obtain competitive national, European and international grants and carry out innovative and technologically advanced research projects. Also, operating as an academic centre, we are very proud that the postgraduate education we provide has been enhanced even further with the operation of our recently established postgraduate school, the Cyprus School of Molecular Medicine. Service, Research and Education co-exist to create a positive feedback system, whereby education links with research to provide tangible results which are then applied in the service sector of CING to the maximum benefit of patients. We continue upgrading and modernizing the state-of-the-art infrastructure and work of the Institute, making it even more competitive, with a leading role in the region and an international reputation. We are committed that we will continue to work hard, with zeal, passion and determination, to keep The Cyprus Institute of Neurology & Genetics at the front line of the international scientific arena and close to our patients.

About us

The Cyprus Institute of Neurology & Genetics

The Cyprus Institute of Neurology & Genetics (CING) is a bi-communal, non-profit, private, research, medical and academic Institution.

The Vision of the CING is to function as a National Centre of Excellence and a Regional Referral Centre in the areas of Neurology, Genetics, Biomedical and Medical Sciences.

The Mission of the CING is to develop and provide specialized clinical and laboratory services, develop and pursue advanced innovative research and provide postgraduate education in the areas of Neurology, Genetics, Biomedical, Medical and other similar and related Sciences.

CING's ultimate aim is to improve and upgrade the quality of life of patients and strengthen its international role in the areas of its specialties.

CING Core Values

Our five core values, summarized by the term "ΑΚΕΚΟ" (Αριστεία, Κοινωνική Προσφορά, Επαγγελματισμός, Καινοτομία, Ομαδικότητα) guide every decision we make as an institution and as individuals. They are the foundation for our business strategy and are reflected in every aspect of our work life.

- Αριστεία (Excellence) – We each strive to be the best in our own field by continuous education and training and combine services, research and education in order to assist CING to become a global center of excellence.
- Κοινωνική Προσφορά (Social Service) – We care about our patients and we are committed to listen and respond to their needs and to constantly improve our services aiming to a better quality of life.
- Επαγγελματισμός (Professionalism) – At all times we act with humility, integrity, honesty and high ethics, providing quality service, being reliable and responsible.
- Καινοτομία (Innovation) – We continuously search for new ways to improve everything we do.
- Ομαδικότητα (Teamwork) – We work together with support, encouragement and respect to achieve our mission.

HR Excellence in Research

HR EXCELLENCE IN RESEARCH

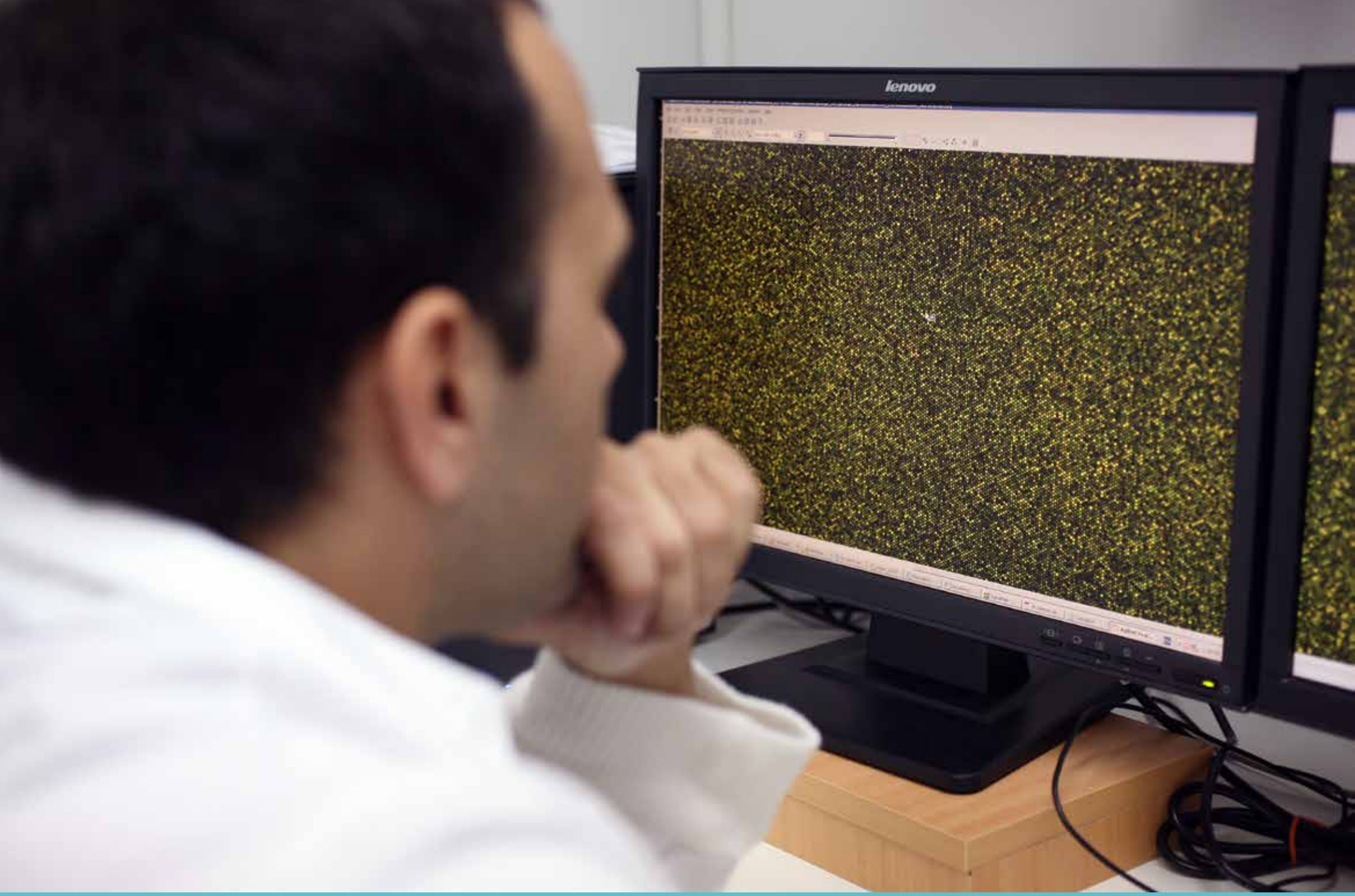
The European Commission, on 20 November 2012, acknowledged The Cyprus Institute of Neurology & Genetics for the progress it has made in its Human Resource Strategy for Researchers implementing The European Charter for Researchers and The Code of Conduct for the Recruitment of Researchers (Charter & Code) and officially awarded CING the "HR Excellence in Research" logo.

The Charter & Code consists of 40 general principles and requirements which specify the roles, responsibilities and entitlements of researchers as well as of employers and/or funders of research. It provides a framework for the career management of researchers and promotes open and transparent recruitment and appraisal procedures.

The award of the "HR Excellence in Research" logo by the European Commission is an important distinction for The Cyprus Institute of Neurology & Genetics. It identifies the CING as provider and supporter of a stimulating and favourable working environment. It also gives a new boost in attracting funding from European and International sources, attracting research talents and in general enhancing the quality of research conducted at CING.

Health & Safety and Quality Office

Maria Theocharidou, BSc, MSc


lenovo

Scientific Research

The Cyprus Institute of Neurology & Genetics maintains the largest and most technologically developed research infrastructure in Cyprus and the neighboring area in the sectors of Neurology, Genetics, Biomedical and Medical Sciences.

The Institute makes important contributions towards basic and translational research through research grants awarded and industry funding. CING research scientists are characterized by their enthusiasm and devotion to the initial mission of the Institute “to develop and pursue advanced research”, resulting in the Institute becoming an important regional research resource.

Over the last few years, the number of research grants obtained from funding organizations in Cyprus and abroad has increased, i.e. there are approximately 50 ongoing research grants with a total funding of approximately €8.000.000.

The results of the successful completion of research programs are depicted in the number of publications in international peer review scientific journals and books. The publications in high impact factor journals of peer reviewed articles are approximately 45 per year. Moreover, more than 35 national and international awards for scientific work have been granted to CING by various scientific organizations and authorities in Cyprus and abroad.

The Cyprus Institute of Neurology & Genetics obtains the majority of competitive grants of the country in the health sector and provides very competitive and pioneering biomedical research. CING has established more than 100 international research collaborations with universities and research institutes abroad.

The Cyprus Institute of Neurology & Genetics, through its subsidiary company, CING Innovations Ltd, supports the creation of spin-off companies for the development of novel ideas of its scientific staff, into commercially viable products/services.

Academic Programs & Training Activities

The Cyprus Institute of Neurology & Genetics has developed the facilities and an atmosphere enabling education and training activities within the Institute. CING promotes academic programs and studies as well as training of doctors, scientists and students.

As of September 2012, CING provides academic postgraduate degrees to students from Cyprus and abroad in the specialized fields of Medical Genetics and Molecular Medicine, through the two MSc and the two PhD programs that are currently offered by the Cyprus School of Molecular Medicine (CSMM). CSMM is a postgraduate school of CING, approved by the Ministry of Education and Culture.

Previously and until the establishment of the CSMM, CING has been collaborating with universities in Cyprus and abroad for the completion of PhD program studies. Fifty doctoral theses resulting in the award of the PhD title have been completed and some more are still working towards their PhD studies in collaboration with universities such as the University of Cyprus, University of Athens, University of London, Imperial College of London, University of Bristol. CING has also been recognized by European universities for the completion of the final thesis of Biology degree students.

A substantial number of neurologists from Cyprus and abroad have completed their specialization training at CING and continue specializing in Clinical Neurology and Electromyography. Also, a training program, approved and accredited by the Cyprus Ministry of Health, is offered for the acquisition of the one year medical specialty in Neurology. CING also provides three-month period up to two years education and specialization to graduate students, doctors and scientists from Cyprus and abroad in specialized fields of genetics.

In addition to the above, CING organises lectures on a weekly basis, as well as national and international scientific meetings, conferences and courses in Cyprus and abroad. Since 2004, CING has been recognized by the European School of Genetics Medicine as a regional centre of education in the sector of Medical Genetics and to date, 10 European congresses have been organized at the CING.

The Cyprus Institute of Neurology & Genetics is considered to be the most advanced tertiary medical academic centre in our country in the health sector.


THE CYPRUS INSTITUTE OF
NEUROLOGY & GENETICS
Random segregation of chromosomes
and independent assortment of alleles
Loci on different chromosomes will assort independently
Red is maternal
Blue is paternal
What happens with the linked/syntenic locus Z?
Syntenic locus Z
Non-recombinant gamete
Recombinant gamete
CYPRUS SCHOOL
of molecular medicine

Medical & Biomedical

Specialized Services

The Cyprus Institute of Neurology & Genetics provides a wide range of highly specialized clinical and laboratory medical and biomedical services to all Doctors, Clinics and Hospitals in the Government and Private sector, offering diagnostics for common and rare diseases to the Cypriot community and to countries of the region. CING also provides forensic services to the Government and Private sector.

CING provides approximately 70.000 examinations, clinical and laboratory services per year. The cost of these examinations is approximately €7.000.000, per year. CING is the only Institute in Cyprus that has the technology, know-how and expertise to provide more than 90% of these examinations.

The Institute has established collaborations with centers and universities of the neighboring area and provides diagnostic services as a Regional Referral Center.

Several services offered by CING are accredited or certified thereby ensuring their high quality. All CING laboratories currently participate in international external quality control schemes. Furthermore, the Departments and Clinics are working towards accreditation with the International Standards, CYS EN 15189/2007 and 17025/2005. It is expected that within the year 2013 CING Departments and Clinics will be accredited.

CING plays a leading role in Cyprus in the fields of Neurology, Genetics, Biomedical and Medical Sciences. It has introduced and applied clinical and laboratory services with great success, and has undertaken the responsibility of several national diagnostic and screening programs in these sectors.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ZEISS

The Muscular Dystrophy Research Trust of Cyprus (MDRTC), renamed in 1995 as “Cyprus Foundation for Muscular Dystrophy Research”, was established in 1987 by the Cyprus Muscular Dystrophy Association, Lady Langley (wife of the then Commander of the British Bases in Cyprus) and Prof. Lefkos T. Middleton (first Medical Director of CING). The Cyprus Institute of Neurology & Genetics was created in 1990 by MDRTC.

CING the initial stages (1990-1995)

The Cyprus Institute of Neurology & Genetics commenced operations in 1990 in a space of 300m^2 at the Makarios III Hospital in Nicosia with basic equipment and personnel numbering approximately 20 individuals (scientific and administration). This space was generously provided by the Cyprus Government, who also donated the land for building CING’s new premises. The United States Government donated to CING, through the Cyprus office of the United Nations High Commission for Refugees, an amount for running the Institute during the years 1990-1995, as well as for the establishment of its new premises. Using part of this initial one-off funding, CING established the Departments and Clinics at Makarios III Hospital, employed a small number of employees and purchased basic equipment. These small scientific groups (Departments and Clinics) managed to attract the first external competitive research grants and published the first scientific papers of the Institute.

CING new premises (1995-2013)

In 1995, CING moved to its own purpose-built premises made up of an area of approximately 10.000m^2. Due to the lack of a medical center in Cyprus with the infrastructure and know-how to provide highly specialized laboratory and clinical services, the Cyprus Government offered to fund CING annually, in return for the provision of specialized clinical and diagnostic services to Government Hospitals. This annual funding has been the major backbone of the CING budget for all these years, and has helped it to survive financially and expand further. Since 1995, CING has been providing highly specialised services and the most competitive and pioneering biomedical research aiming towards early detection and prevention of disease. Moreover, it provides postgraduate education which is closely associated to research and services.

CING today

Today, the Institute is equipped with the latest high-tech laboratory and clinical equipment. Its personnel numbers more than 180 individuals, plus 40-70 students of its postgraduate school, Cyprus School of Molecular Medicine, which commenced operations in September 2012. CING also hosts a further 20 individuals as Visiting Scientists. For the achievement of its mission, six Clinics and eight Departments are operating in the areas of their specialty.

CING is one of the very few innovative organizations in Cyprus that has developed a critical mass, and contributes actively in the research and development of new knowledge. CING has available appreciable human potential, laboratory infrastructure unique for Cyprus, excellent relations and collaborations with countries of the Middle East, Northern Africa, Europe and America, and is successfully competing at the national and international level.

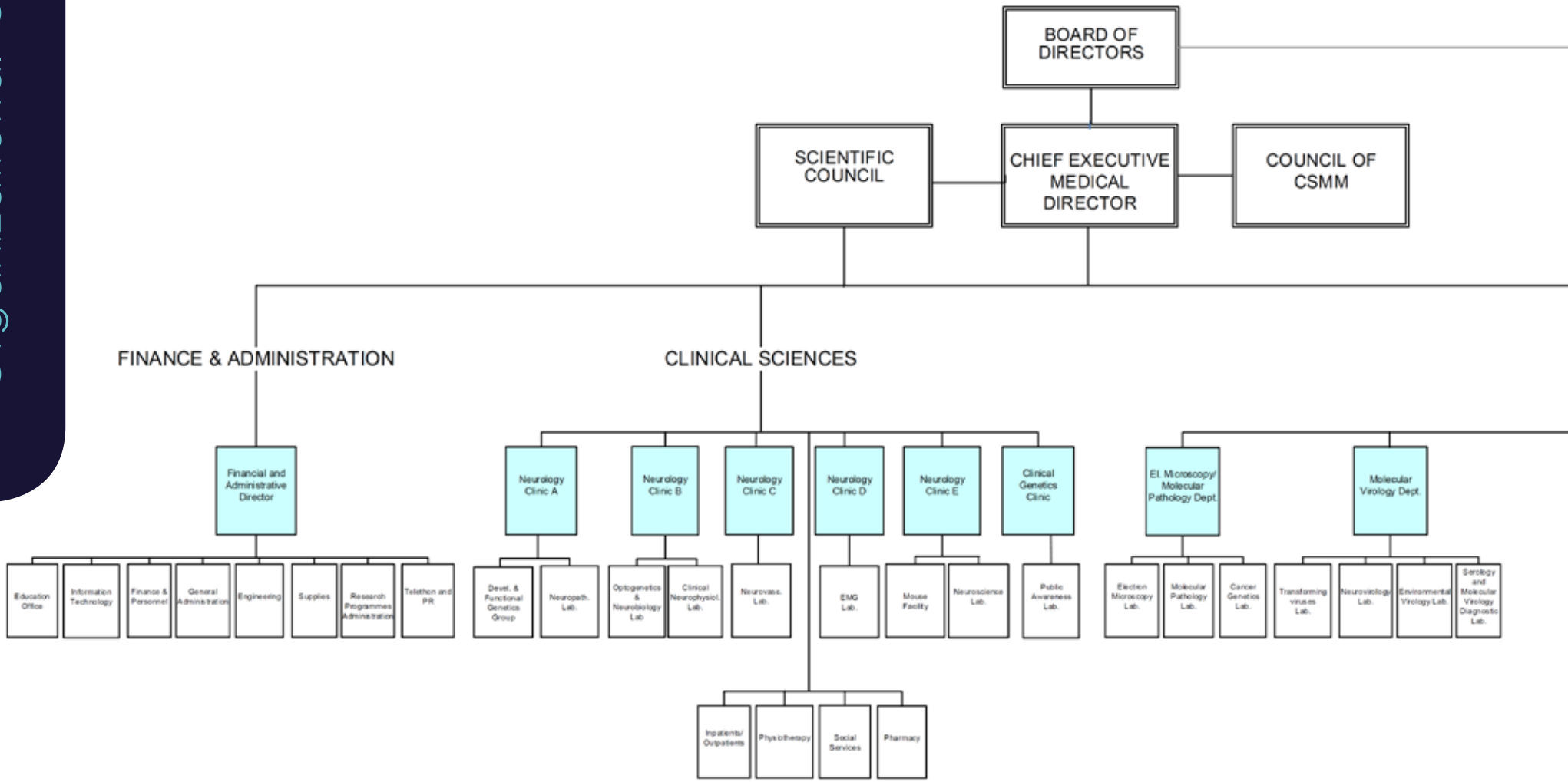
CING receives international recognition and plays an active and essential role as a national, regional and international center of excellence for the provision of high quality services, research and education. The dynamic success of the Institute is based on the development of medical services and research programs which are directly related to the needs of Cyprus and the countries of the region.

CING is a medical and biomedical translation center, which is probably the most successful model nowadays as it combines academic, service and research activities in one center, providing major medical benefits to the people, society and the country. CING was one of the first, and today one of the best, examples in Europe during the last 22 years where postgraduate education, research and service have been successfully combined.

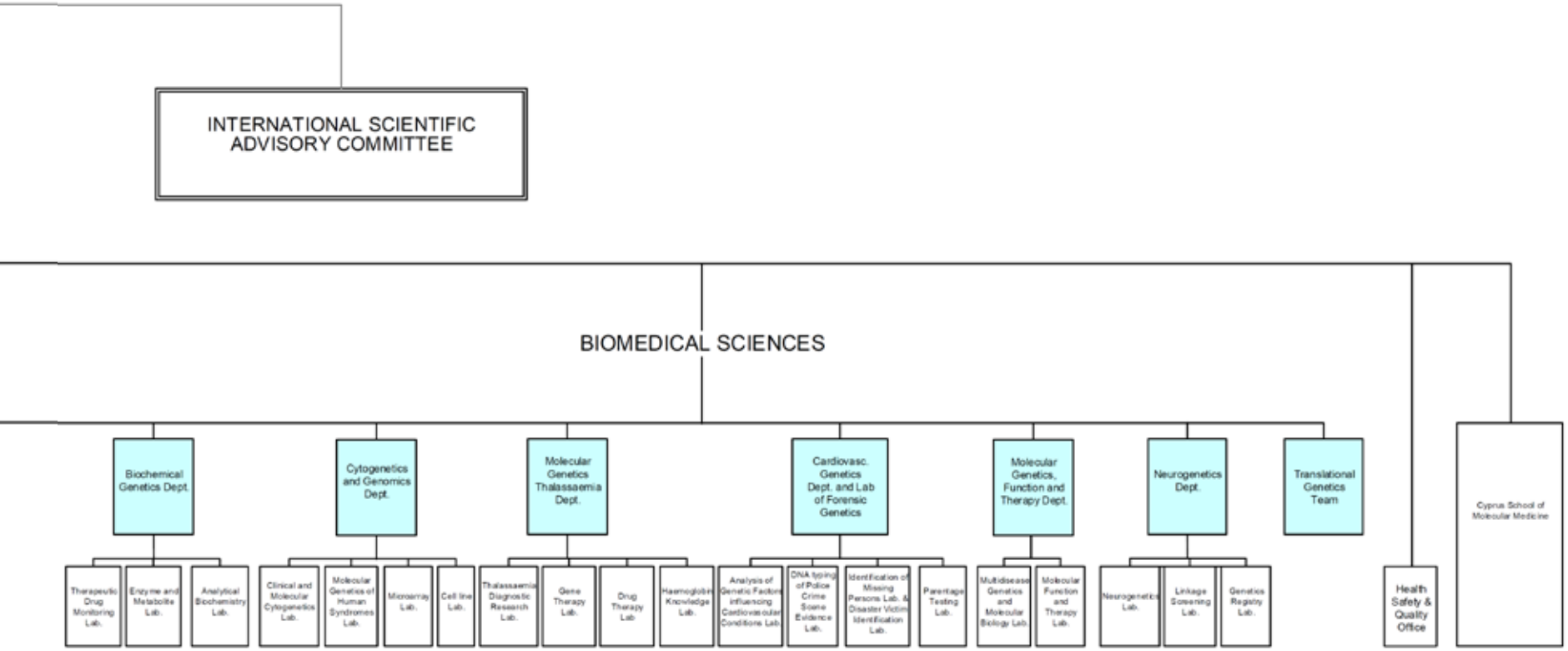
Brief History of

The Cyprus Institute of Neurology & Genetics

Organizational Chart



THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS



Organization & Structure

Board of Directors

The Board of Directors consists of 20 members. Ten members, including the President, are representatives of the Cyprus Government (eight are appointed by the Council of Ministers, one by the Ministry of Finance and one by the Ministry of Health). Nine members are representatives of patient associations, the CING Scientific Council, the United States Government, the British Bases in Cyprus and the Turkish-Cypriot community. One member is the CING Chief Executive Medical Director. The Board of Directors exercises the following powers and executes the following duties: a) administers and controls the affairs of the Institute and all its property, and in general deals with all related matters, b) acts in regard to the above matters and property in a way that best promotes the interests and aims of the Institute, c) exercises all other activities and undertakes acts and actions which are conducive or essential, for the achievement of the aims of the Institute.

- Eliades Christos, President *(Cyprus Government Representative)*
- Papageorghiou Tellos, Vice President *(CING Scientific Council Representative)*
- Voskos Panicos, Treasurer *(Cyprus Government Representative)*
- Patsalis Philippos, Secretary *(CING Chief Executive Medical Director)*
- Angastiniotis Michael *(Pancyprian Antianemic Association Representative)*
- Charalambous Pambos *(Cyprus Government Representative)*
- Constantinou George *(CING Scientific Council Representative)*
- Costeas Paul *(US Government Representative)*
- Demetriou Andreas *(Cyprus Government Representative)*
- Djavit Ahmed *(Cyprus Government Representative)*
- Hami Mustafa *(Turkish-Cypriot Representative)*
- Ioannou Ioannis *(Cyprus Government Representative)*
- Kaimakliotis Ioannis *(US Government Representative)*
- Kalakouta Olga *(Ministry of Health Representative)*
- Stylianou Stelios *(Muscular Dystrophy Association Representative)*
- Stylianou Voula *(Ministry of Finance Representative)*
- Varoglu Ahmet *(Turkish-Cypriot Representative)*
- Zackheos Sotos *(Cyprus Government Representative)*
- Vacant position *(Cyprus Government Representative)*
- Vacant position *(British Bases Representative)*

Chief Executive Medical Director

Philippos Patsalis is appointed to serve as the Chief Executive Officer of The Cyprus Institute of Neurology & Genetics. He heads all medical, scientific and other activities of the Foundation and exercises initiatives in all matters pertaining to the overall direction of the Institute, in accordance with the Foundation's Memorandum of Association.

Philippos Patsalis is also the Chief Executive Director of the Cyprus School of Molecular Medicine. He has the overall supervision of the operation of the School. He oversees all external relations and is responsible for promoting the expansion of the Cyprus School of Molecular Medicine.

Philippos C. Patsalis, BSc, MA, MPh, PhD, HCLD

Scientific Council

The Scientific Council consists of the Chief Executive Medical Director who is the Chairman of the Scientific Council, the Financial and Administrative Director and the Heads of Departments and Clinics of the Institute.

The Scientific Council is in charge of the academic and scientific work of the Institute both in the field of medical and laboratory services and in the area of research and post-graduate training. More particularly it, a) approves the academic research and training programs, and medical and laboratory services provided, b) submits to the Board of Directors proposals for the allocation of the budget and the Institute's requirements concerning medical and other equipment, c) makes suggestions to the Board of Directors regarding the establishing or abolishing of medical and scientific teams, laboratories, Departments or Clinics, d) sets up committees from among its members and may delegate to such committees, on any terms and conditions it deems fit, any of its powers, e) carries on any activities, and carries out any duties granted to it or imposed on it by the Articles of Association or the Institute's Regulations and f) regulates its own proceedings subject to the provisions of the Articles of Association and the Institute's Regulations.

Scientific Council Members

- Patsalis C. Philippos, (Chairman) BSc, MA, MPh, PhD, HCLD
- Phylactou A. Leonidas, (Secretary) BSc, PhD
- Cariolou A. Marios, BSc, PhD
- Christodoulou G. Christina, BSc, MSc, DEA, Dipl. Virol., PhD
- Christodoulou Kyproula, BSc, MSc, PhD
- Christophidou-Anastassiadou Violetta, MD *(in attendance)*
- Drousiotou Anthi, BSc, PhD, ARCSc
- Flouros Marios, BSc, MHA, FCA
- Kleanthous Marina, BSc, PhD
- Kleopa A. Kleopas, MD
- Kyriacou Kyriacos, BSc, PhD, FRMSoc
- Kyriakides Theodoros, MB, ChB, BSc (Hons), FRCP (Lon)
- Pantzaris Marios, MD
- Papacostas Savvas, BSc, MA, MD, FAAN
- Sismani Carolina, BSc, PhD
- Zamba-Papanicolaou Eleni, MD


THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS

International Scientific Advisory Committee

The International Scientific Advisory Committee (ISAC) consists of 10 internationally-renowned scientists appointed by the CING Board of Directors. The ISAC has the following competencies: a) to advise the Board of Directors on matters of scientific policy, b) to provide assistance to the Scientific Council on scientific matters and c) to carry out assessments and evaluations on any matter forwarded to it by the Board of Directors or the Scientific Council. Today, the ISAC consists of the following nine members:

Prof. John Christodoulou, Director
Western Sydney Genetics Program & Sydney Medical School
The Children's Hospital at Westmead
University of Sydney
Australia

Prof. V.M. Der Kaloustian, Professor of Pediatrics and Human Genetics
Clinical Director, Division of Medical Genetics
McGill University Health Center
Montreal, Canada

Prof. Douglas Higgs, Director of the Molecular Haematology Unit and Hon. Consultant Haematologist
MRC Molecular Haematology Unit
Institute of Molecular Medicine
John Radcliffe Hospital
University of Oxford
Oxford, U.K.

Prof. Peter Karayiannis, Reader in Molecular Virology
Department of Medicine, Hepatology Section
Division of Medicine
Imperial College London
London, U.K.

Prof. James R. Lupski, Cullen Professor of Molecular and Human Genetics and Professor of Pediatrics
Baylor College of Medicine Houston
Texas, U.S.A.

Prof. Dimitris P. Mikhailidis, Academic Head of Department
Department of Clinical Biochemistry (Vascular Disease Prevention Clinics)
Royal Free Hospital
Royal Free University, College School of Medicine
University College London
London, U.K.

Prof. Konstantin Miller, General Secretary of the European Cytogenetics Association
Director of Cytogenetics
Institute of Human Genetics
Hannover Medical School
Hannover, Germany

Prof. Jahn M. Nesland, Vice President of International Academy of Pathology
Director of Division of Pathology
The Norwegian Radium Hospital
University of Oslo
Oslo, Norway

Prof. Timothy Pedley, Chairman
The Neurological Institute
Columbia University
New York, U.S.A.

External Evaluation of the CING

In 2012, an External Evaluation Team, consisting of seven world renowned scientists, members of CING's International Scientific Advisory Committee, conducted an on-site external evaluation. The evaluation was based on high international standards. We are very proud and satisfied that the recommendations made by the External Evaluation Team have been successfully addressed. The results of the evaluation were impressive and showed the continuing upgrade and progress of the Institute.

The evaluation of our Institute provides an external peer review evaluation of our standards, which helps us to achieve our long and short term goals. Evaluation is a very useful tool for continuing self-improvement.

Telethon

Telethon is an international charitable campaign that began in the United States and subsequently extended to Europe. The countries in Europe that have adopted and successfully organize TELETHON are France, Italy, Belgium, Switzerland, Luxemburg and Cyprus.

The main aims of TELETHON are:

- To increase public awareness of the problems of patients who suffer from neurological, neuromuscular and genetic diseases.
- To project the scientific work carried out at the CING.
- To financially support the research activities of the CING on neurological, neuromuscular and genetic diseases.
- To financially support the Cyprus Muscular Dystrophy Association, the efforts of which are focused on the continuous improvement of the quality of life of its members.

The Cyprus TELETHON includes many fundraising events that take place throughout the year.

The Cyprus TELETHON is being organized annually by The Cyprus Institute of Neurology & Genetics in collaboration with the Cyprus Muscular Dystrophy Association, since 1994.


TELETHON

CYPRUS SCHOOL of molecular medicine

Chief Executive Director

Professor Philippos C. Patsalis, BSc, MA, MPh, PhD, HCLD

Dean

Professor Leonidas A. Phylactou, BSc, PhD

Education Office
Tel: (+357) 22 392 840 / Fax: (+357) 22 392 845
Email: csmm@cing.ac.cy

Activities

The Cyprus Institute of Neurology & Genetics has established a postgraduate school, named the Cyprus School of Molecular Medicine (CSMM) available to students with research interests applicable to the Institute's activities. The School is organized as a distinct entity within CING and welcomed its first intake of students for the academic year 2012-13 in September 2012.

The CSMM offers four programs of study leading to two MSc and two PhD degrees, respectively.
The programs offered as of September 2012 are:

1. Master in Science (MSc) in Molecular Medicine
2. Master in Science (MSc) in Medical Genetics
3. Doctor of Philosophy (PhD) in Molecular Medicine
4. Doctor of Philosophy (PhD) in Medical Genetics

As a Center of Excellence in basic and applied research in biomedical and clinical sciences aiming to combine services, research and education so as to ultimately improve the quality of life of people, the CING has paved the way for a new generation of scientists. The Cyprus School of Molecular Medicine, housed within the CING premises provides students with a unique educational experience combining taught courses and research conducted in specialized state-of-the-art laboratories, in the areas of neurology, genetics and biomedical sciences.

The advanced curriculum, highly qualified academic staff and state-of-the-art infrastructure facilities combined with the acceptance of the most competitive students culminate in the awarding of the highest quality postgraduate degrees. The Cyprus School of Molecular Medicine has set high standards, prerequisites and qualifications for accepting its students; these high standards are retained throughout students' studies and graduation. All Programs offered are addressed to students with high academic and research excellence.

The Cyprus School of Molecular Medicine was established to act as a catalyst towards the aims of CING and to give students unique education in the areas of neurology, genetics and biomedical sciences. The CSMM innovative postgraduate programs cover a wide spectrum of interesting disciplines and are organized around teaching courses and research in highly specialized laboratories.

Objectives of the School

The Cyprus School of Molecular Medicine provides opportunities for postgraduate education, training and exposes students to a competitive research environment. The School supports and enforces international standards of excellence for its students. The School's objectives are:

- To establish an educational center of excellence for postgraduate programs of international standing and reputation while also attracting and welcoming applications from international students
- To attract and educate students who can engage in competitive work and to provide them with immediate accession to the Cyprus market and academia, so that they can contribute to the socioeconomic landscape of Cyprus
- To produce high quality research output from students' projects (PhD programs) that will contribute towards the improvement of the quality of human life in Cyprus and worldwide
- To challenge students with a wide variety of concepts and approaches and enforce international standards of excellence in the fields of Medicine and Biomedical Sciences
- To offer exceptional curricula for its students which will provide the theoretical and applied knowledge necessary to achieve international caliber doctoral research
- To cooperate with high level international research and education centers and to promote cooperation and understanding through education, research and innovation
- To attract excellent local and foreign students through the international visibility of the School's faculty, staff, and students
- To develop effective communication skills for all its students and to help the students exercise these skills in a competitive environment
- To promote the School as a center of excellence for students and scholars from abroad

MSc Programs

The MSc programs are organized around teaching courses, (including tutorial sessions for each course on a weekly basis) and a research or a library project. Successful students will have to pass all course examinations and the MSc Thesis Examination or the library project report to be awarded an MSc degree. The CSMM offers a 12-month MSc program in full-time mode studies and a 24-month MSc program in part-time mode. The programs are organized, run and reviewed by the CSMM after the approval by the Academic Committee.

PhD Programs

The PhD programs are organized around teaching courses, (including tutorial sessions for each course on a weekly basis) and a research project (thesis work). Successful students will have to pass all course examinations, the PhD thesis examination and have at least one first author publication in a peer-reviewed journal to be awarded a PhD degree. The CSMM offers a four-year doctoral program of study which can be extended to six years for the completion of the thesis work. The programs are organized, run and reviewed by the CSMM after approval by the Academic Committee.

Applications

The application process for the CSMM takes place from January to April and is announced through the CING's and CSMM's websites at

www.cing.ac.cy/csmm

Inauguration Ceremony with the Nobel Laureate Dr James D. Watson

The Inauguration Ceremony of the Cyprus School of Molecular Medicine took place with great success within a celebratory atmosphere where in excess of 1.000 guests gathered at the premises of The Cyprus Institute of Neurology & Genetics on 17 October 2012.

The Inauguration Ceremony held a historic stigma as the Nobel Laureate and internationally renowned Geneticist, Dr James D. Watson visited Cyprus for the first time as a guest of the CING specifically for the Ceremony. Dr Watson gave the lecture of academic excellence, "From the Double Helix to the Curing of Cancer".

Finance

Personnel

Finance and Personnel

- Vera Kazandjian, BA, MBA - Financial and Personnel Services Manager
- Anna Michaelidou, BSc, MSc
- Maro Shiakalli
- Elena Photiou

Supplies

- Amal Vlachou, BSc, MPH - Purchasing and Store Manager
- John Ioannou
- Eleni Pashiali

Administrative Support

- Andreas Papadouris, BSc, MBA - Administrative Services Manager
- Jenny Andreou
- Stavros Andreou
- Polycarpos Chryssafiades
- Maria Damianou
- Fredericos Efstathiou
- Eleni Eliadou
- Christina Hadjiyianni
- Elena Kyriacou
- Maria Loizou, BA, MBA
- Maria Mouzourkou
- Elena Polycarpou

Research Programmes Office

- Elena Ioannidou, BSc, MBA - Research Programmes Officer
- Andri Charalambous, BA

Telethon Office

- Kypros Karanikis, Dip (HM) - Telethon and PR Manager
- George Vatiliotis

Engineering

- Stylianos Stylianou, BEng, MBA - Senior Engineer
- Neophytos Polycarpou
- Constantinos Papadopoulos
- Christakis Georgiou

Information Technology

- Charalambos Hadjinicolaou, BSc, MSc, CITM - IT Manager
- Kristiana Anninou, BSc, MSc
- Aristos Aristodemou, BSc
- Andreas Miltiadous

Education Office

- Marinos Voukis, BSc, MBA - Education Office Manager
- Maria Ellina
- Andria Ioakem, BA, MA, IDM
- Eleftheria Ioannou
- Maria Lagou, BSc

& Administration Department

Marios Flouros, BSc, MHA, FCA

Financial and Administrative Director

Tel.: (+357) 22 392 722 / (+357) 22 358 600
Fax: (+357) 22 358 238
E-mail: flourosm@cing.ac.cy

Activities

The Finance and Administration Department is responsible for the financial and administrative matters of the Institute. Its mission is to support the service, research and education activities of the Institute through the provision of high quality administrative services to all the spectrum and levels of the Institute hierarchy, its patients, its students and the public. The Finance and Administration Department comprises of eight sections:

- General Administration
- Finance and Personnel
- Supplies
- Engineering
- Information Technology
- Research Programmes Office
- Telethon and PR
- Education Office

Clinical Sciences

Outpatients & Inpatients

Theodoros Kyriakides, MB, ChB, BSc (Hons), FRCP (Lon)

Co-ordinator

Tel.: (+357) 22 392 740 / (+357) 22 358 600
Fax: (+357) 22 392 786
E-mail: theodore@cing.ac.cy

Personnel

Inpatients

- Judith Toufexis
- Rayna Deneva
- Mariana Hanghiuc
- Krassimira Stoimenova
- Olga Stylianou
- Dimitrinka Charalambous
- Kyriaki Charalambous
- Svetlana Kaplani
- Galini Chroidou
- Electra Askalidou
- Lilyana Gencheva
- Ekaterine Chkheidze
- Katerina Demetriou
- Rafaella Pitsillide
- Niki Papaioannou
- Lalyn Kapris

Outpatients

- Eftychia Gagglia
- Eleni Papadopoulou
- Marina Chryseliou
- Maria Symeou
- Maria Yennari
- Eliana Savvidou

Physiotherapy

- Annita Ormiston, MCSP
- Irini Zannettou, BSc
- Maria Charalampous, BSc

Social Services

- Marina Pavlou, BSc

Pharmacy

- Eleni Kkolou, BSc
- Andry Ploutarchou, BSc

Activities

The medical care includes follow up appointments at the Outpatients Clinic and, when necessary, admission to the neurological ward. Furthermore, the department provides specialized laboratory diagnostic services in neurophysiology, neurovascular disorders and neuropathology.

Physiotherapy services are also provided to patients with neuromuscular and other disabling disorders. The social services (on site and at home) also provide advice and support to patients and their families.

The department houses orthopaedic, respiratory gastroenterology and endocrinology clinics for a more comprehensive care of patients. A psychologist and a dietician are available on a weekly basis.

The Clinical Neurology department examines, investigates and treats more than 6,000 patients annually with a steadily increasing number of referrals.

Neurology Clinic A

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Personnel

Neuromuscular Laboratory

- Rebecca Papacharalambous, ATEI
- Elena Panayiotou-Worth, BSc, MSc

Activities

The study and treatment of neuromuscular diseases has been the original aim of the CING and this continues to be the main priority of this Clinic. It serves as a national tertiary referral centre for neuromuscular disorders and houses the only neuropathology laboratory in Cyprus that processes muscle and nerve biopsies. A wide variety of neuromuscular disorders are investigated and treated including muscular dystrophy, inflammatory myopathies, inherited and acquired neuropathies, amyotrophic lateral sclerosis and others. A multi-disciplinary approach to therapy is followed. Demyelinating disease including Multiple sclerosis is another interest of the Clinic although a large number of challenging general neurology patients is also seen. The Clinic also established and functions the Neuromuscular Laboratory.

Research Interests

- The study of the effect of complement C1Q on disease phenotypeTTRMet30 peripheral neuropathy using a variety of transgenic mouse models
- The study of modifier genes in disease severity in Multiple sclerosis
- The study of reactive oxidative species in the pathophysiology of Duchenne muscular dystrophy
- An investigation of apoptosis as disease mechanism in mitochondrial encephalomyopathies

Selected Publications

Kyriakides T, Pegoraro E, Hoffman EP, Piva L, Cagnin S, Lanfranchi G, Griggs RC, Nelson SF. SPP1 genotype is a determinant of disease severity in Duchenne muscular dystrophy: predicting the severity of Duchenne muscular dystrophy: implications for treatment. Neurology 2011;77:1858; author reply 1858-1859.

Kyriakides T, Papacostas S, Papanicolaou E, Bagdades E, Papathanasiou ES. Sleep hypoventilation syndrome and respiratory failure due to multifocal motor neuropathy with conduction block. Muscle & nerve 2011;43:610-614.

Kyriakides T, Angelini C, Schaefer J, Sacconi S, Siciliano G, Vilchez JJ, Hilton-Jones D, European Federation of Neurological S. EFNS guidelines on the diagnostic approach to pauci- or asymptomatic hyperCKemia. European journal of neurology : the official journal of the European Federation of Neurological Societies 2010;17:767-773.

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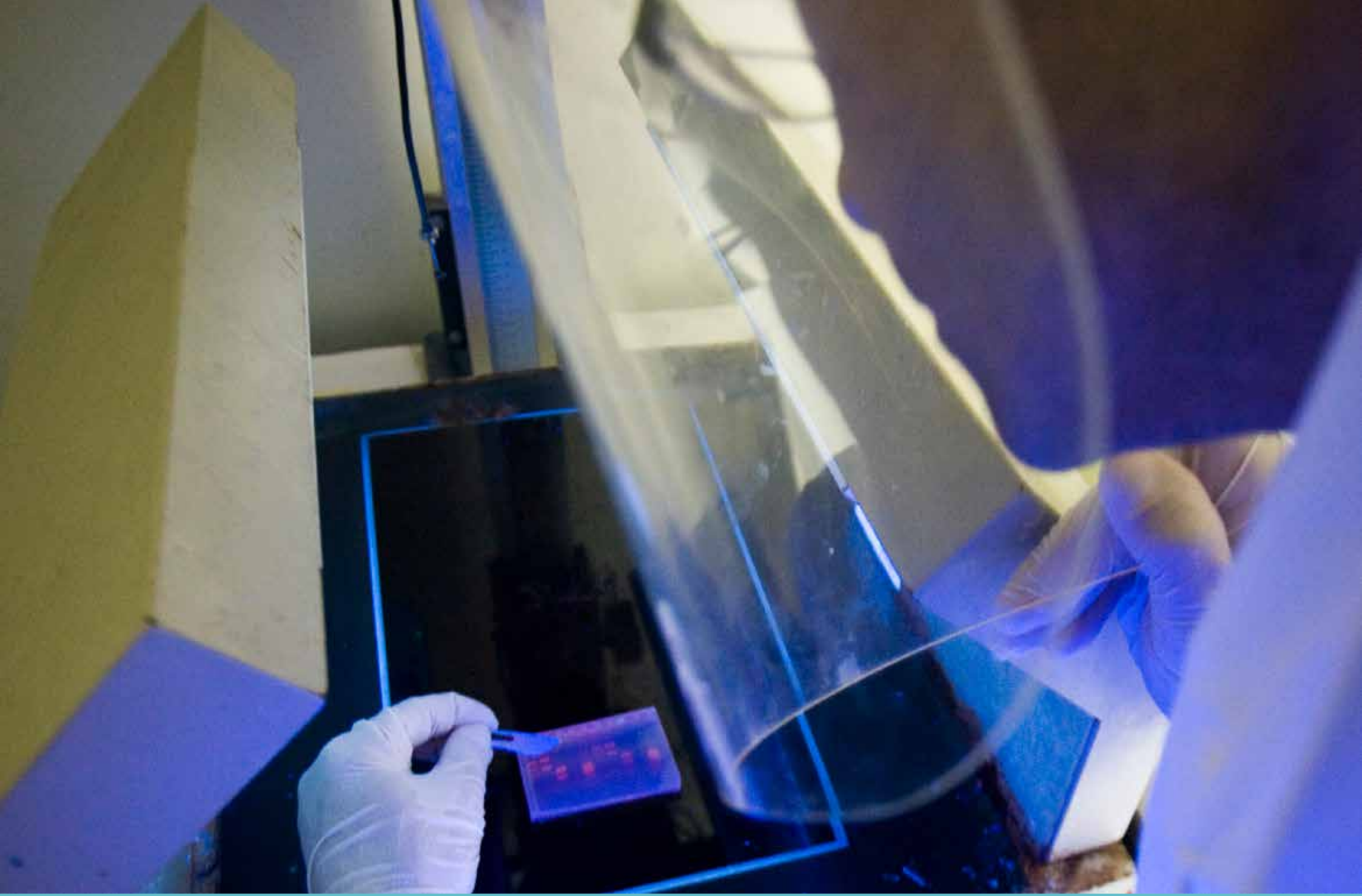
Kleopa KA, Zamba-Papanicolaou E, Alevra X, Nicolaou P, Georgiou DM, Hadjisavvas A, Kyriakides T, Christodoulou K. Phenotypic and cellular expression of two novel connexin32 mutations causing CMT1X. Neurology 2006;66:396-402.

Kleopa KA, Drousiotou A, Mavrikiou E, Ormiston A, Kyriakides T. Naturally occurring utrophin correlates with disease severity in Duchenne muscular dystrophy. Human molecular genetics 2006;15:1623-1628.

Sivakumar K, Kyriakides T, Puls I, Nicholson GA, Funalot B, Antonellis A, Sambuughin N, Christodoulou K, Beggs JL, Zamba-Papanicolaou E, Ionasescu V, Dalakas MC, Green ED, Fischbeck KH, Goldfarb LG. Phenotypic spectrum of disorders associated with glycyl-tRNA synthetase mutations. Brain : a journal of neurology 2005;128:2304-2314.

Kleopa KA, Kyriacou K, Zamba-Papanicolaou E, Kyriakides T. Reversible inflammatory and vacuolar myopathy with vitamin E deficiency in celiac disease. Muscle & nerve 2005;31:260-265.



Neurology Clinic B

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Personnel

Neurophysiology Laboratory

- Yiolanda Christou, MD
- Chloe Antoniou, BSc, PhD
- Eleftherios S. Papathanasiou, PhD
- Michele Galganski-Cleanthous, MS, REEGT
- Panayiota Myrianthopoulou, BS
- Andria Stylianou , BSc

Activities

Epilepsy; we evaluate patients with difficult and treatment-resistant forms of seizure disorders. Patients undergo thorough evaluation and, in selected cases, video-EEG recording to assess whether they could improve or become seizure free with surgery. Patients may be offered alternative treatments such as the Ketogenic Diet or implantation of a Vagus Nerve Stimulator, a method that controls seizures and improves quality of life. Patients may participate in clinical trials with new anti-epileptic medications. Specialized neuropsychological evaluations include Wada tests to assess the location of memory and language in the brain. Selected patients are either referred abroad, or operated locally by collaborating surgeons from the United States.

Behavioral Neurology; A subspecialty which deals with dementias, such as Alzheimer's disease, other neurodegenerative disorders, and behavioral symptoms resulting from brain disease. Patients are offered systematic investigations and treatment and they have the chance to participate in clinical trials with newer medications. They are followed by a multidisciplinary team that includes nursing, physiotherapy, speech pathology, psychology and social work.

The Clinic established and operates the Clinical Neurophysiology Laboratory which evaluates the function of the brain and spinal cord with exams such as electroencephalography and video-EEG for epilepsy, Evoked Potentials for sensory organ function, sleep studies and multiple sleep latency tests for sleep-related problems, excessive daytime sleepiness, or respiratory symptoms at night. The newly established laboratory for neurobiology and optogenetics offers basic research and hosts Masters and PhD students for their research.

Education offers opportunities for involvement from secondary school to post doc level. Secondary school students may spend their science week or their summers at the Clinic and become familiar with the basic and clinical aspects of Neurology. University students may complete brief educational and research projects. Master and Doctoral students may conduct their research thesis or dissertation on topics relevant to the Clinic's activities and research interests. The Clinic offers up to one year of post-graduate training in Neurology in collaboration with the Clinical Sciences Section. The Clinic Head holds academic and teaching appointments at the University of Cyprus, the University of Rochester Medical Center in New York, and the newly established School of Medicine of London's St. George's University branch in Nicosia.

Research Interests

- Psychological, Cognitive and quality of life issues in patients with epilepsy
- Clinical Trials on the efficacy and safety of new anti-epileptic and dementia drugs
- Neurophysiology of Multiple Sclerosis
- Development of seizure prediction algorhythms using physiological parameters
- Animal models of Alzheimer's disease
- Application of Optogenetic methodologies in the evaluation and therapeutic applications for epilepsy

Selected Publications

Votsi C, Zamba-Papanicolaou E, Georghiou A, Kyriakides T, Papacostas S, Kleopa KA, Pantzaris M, Christodoulou K. Investigation of SCA10 in the Cypriot population: further exclusion of SCA dynamic repeat mutations. Journal of the neurological sciences 2012;323:154-157.

Stavrinides P, Constantinidou F, Anastassiou I, Malikides A, Papacostas S. Psychosocial adjustment of epilepsy patients in Cyprus. Epilepsy & behavior : E&B 2012;25:98-104.

Kyriakides T, Papacostas S, Papanicolaou E, Bagdades E, Papathanasiou ES. Sleep hypoventilation syndrome and respiratory failure due to multifocal motor neuropathy with conduction block. Muscle & nerve 2011;43:610-614.

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Papacostas S, Malikides A, Petsa M, Kyriakides T. Ten-year mortality from Creutzfeldt-Jakob disease in Cyprus. Eastern Mediterranean health journal = La revue de sante de la Mediterranee orientale = al-Majallah al-sihhiyah li-sharq al-mutawassit 2008;14:715-719.

Papacostas SS, Myrianthopoulou P, Dietis A, Papathanasiou ES. Induction of central-type sleep apnea by vagus nerve stimulation. Electromyography and clinical neurophysiology 2007;47:61-63.

Papathanasiou ES, Papacostas SS, Charalambous M, Eracleous E, Thodi C, Pantzaris M. Vertigo and imbalance caused by a small lesion in the anterior insula. Electromyography and clinical neurophysiology 2006;46:185-192.

Neurology Clinic C

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Personnel

- Eleni Leonidou, MD
- Anastasia Lambrianidou, PhD

Activities

A national Multiple Sclerosis (MS) Clinic was established at The Cyprus Institute of Neurology & Genetics in 1989, when it became apparent that the prevalence of MS in Cyprus was high and approaching that of northern European populations.

Epidemiological research at the Institute has shown a high prevalence in the Greek Community with 175 cases per 100.000. The Clinic cares for over 1000 patients with access to inpatient, neurophysiology and physiotherapy facilities. Drug therapy, including the administration of corticosteroids, interferons, mitoxantrone and the newer monoclonal antibodies is supervised by the clinic.

The Clinic, with a special interest in movement disorders, follows many Parkinson's disease patients. Since 2004, the Clinic is collaborating with a surgical centre abroad, for surgical treatment of Parkinson's disease (Deep Brain Stimulation). Twenty-six patients have been operated until today and are followed by the Clinic. Since 2012 the Clinic in collaboration with a neurosurgical team, they have operated the first Cypriot patient with Deep Brain Stimulation in Cyprus.

With special interests in chronic intractable pain, the Clinic has started since 2001, in collaboration with the same group of neurosurgeons, to place spinal cord neurostimulators as a permanent and effective treatment for this severe disabling condition.

The Clinic established and functions the Neurovascular Laboratory. The laboratory serves as a reference laboratory for the study of the extracranial carotid arteries, the extracranial vertebral arteries and the intracranial arteries (the only laboratory in our country that offers the last service). Carotid artery atherosclerosis is the major cause of ishaemic brain damage (ischaemic stroke), so the study of the carotids with ultrasound offers significant clinical data for both primary and secondary stroke prevention. The laboratory offers unique and experienced diagnostic service in arterial dissection, both in the territory of the internal carotid and vertebral arteries. Transcranial study of the intracranial brain arteries offer diagnostic services for intracranial arterial stenosis, intracranial aneurysms, arterial vasospasm following subarachnoid haemorrhage and also transcranial monitoring for emboli detection as well as for arterial reserve study (CO2 reactivity test).

Research Interests

- Development of databases for epidemiological and clinical data for Multiple Sclerosis and Stroke
- Development of automated computer programmes to identify and evaluate the arterial wall thickness and plaque delination in carotid atherosclerotic disease
- Qualitative study of the carotid wall changes and carotid atherosclerotic plaques and the evaluation of future events (stroke and carotid stenosis)
- Quantitative and qualitative study of the demyelinating plaques in the brain and the evaluation of prognostic factors in Multiple Sclerosis prognosis
- Nutrition and Multiple sclerosis (MS), the development of a novel oral neutraceutical formulation with polyunsaturated fatty acids and vitamins for the treatment of relapsing remitting MS
- The role of viral infection in Multiple Sclerosis
- The role of micro-RNA molecules as a prognostic marker in clinically isolated syndrome (CIS)
- The role of specific genes in Multiple Sclerosis nature and prognosis

Selected Publications

Loizou C, Pantzaris M, Pattichis C, Seimenis I. Brain MRI Image normalization in texture analysis of multiple sclerosis. Journal of Biomedical Graphics and Computing 2013;3.

Loizou C, Kyriacou EC, Seimenis I, Pantzaris M, Petroudi S, Karaolis MA, Pattichis CS. Brain white matter lesion classification in multiple sclerosis subjects for the prognosis of future disability. Intelligent Decision Technologies Journal (IDT) 2013;7:3-10.

Votsi C, Zamba-Papanicolaou E, Georghiou A, Kyriakides T, Papacostas S, Kleopa KA, Pantzaris M, Christodoulou K. Investigation of SCA10 in the Cypriot population: further exclusion of SCA dynamic repeat mutations. Journal of the neurological sciences 2012;323:154-157.

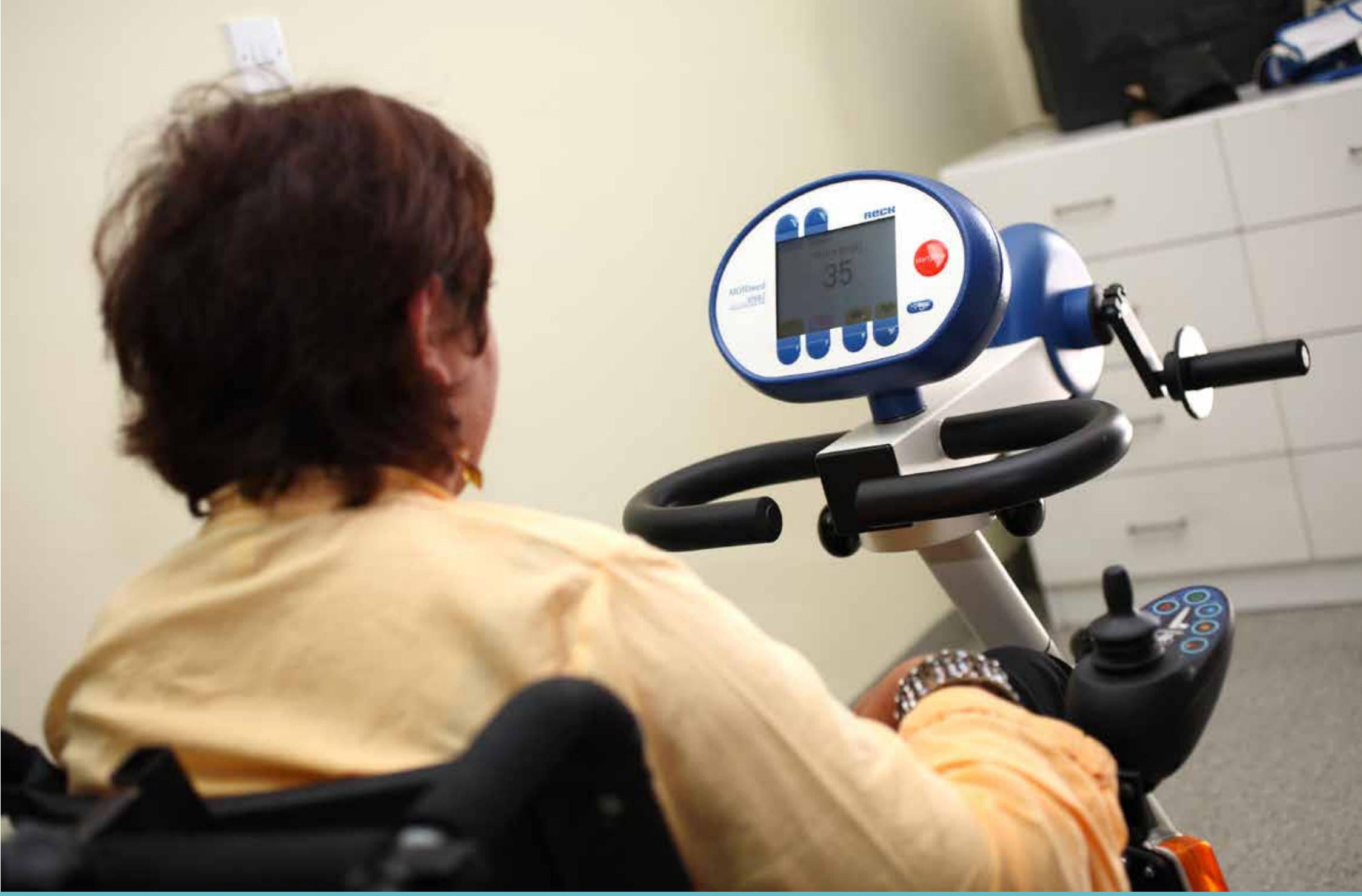
Petroudi S, Loizou C, Pantziaris M, Pattichis C. Segmentation of the common carotid intima-media complex in ultrasound images using active contours. IEEE transactions on bio-medical engineering 2012;59:3060-3069.

Pantzaris M, Loukaides G, Ntzani E, Patrikios I. A novel oral nutraceutical formula (PLP10) for the treatment of relapsing remitting multiple sclerosis: a randomized double-blind, placebo-controlled proof-of-concept clinical trial. , Clinical Nutrition suppliments 2012;17:266, 2012.

Loizou CP, Petroudi S, Pattichis CS, Pantziaris M, Kasparis T, Nicolaides A. Segmentation of atherosclerotic carotid plaque in ultrasound video. Conference proceedings : Annual International Conference of the IEEE Engineering in Medicine and Biology Society IEEE Engineering in Medicine and Biology Society Conference 2012;2012:53-56.

Christodoulou L, Loizou CP, Spyrou C, Pantziaris M, Kasparis T. Full-Automated Medical Imaging System for Segmentation and Detection of Carotid Plaque and Carotid Artery Lumen from Ultrasound Images. Biomedizinische Technik Biomedical engineering 2012.

Loizou CP, Murray V, Pattichis MS, Seimenis I, Pantziaris M, Pattichis CS. Multiscale amplitude-modulation frequency-modulation (AM-FM) texture analysis of multiple sclerosis in brain MRI images. IEEE transactions on information technology in biomedicine : a publication of the IEEE Engineering in Medicine and Biology Society 2011;15:119-129.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Neurology Clinic D

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Personnel

EMG Laboratory

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- Michele Galganski-Cleanthous, MS, REEGT
- Alexandros Heraclides, PhD
- Panayiota Myrianthopoulou, BS
- Eleftherios S. Papathanasiou, PhD

Activities

Clinic D provides outpatient clinic services in the fields of neurogenetic diseases including Spinocerebellar Ataxias (SCA), Friedreich's Ataxia, Charcot-Marie-Tooth Polyneuropathies (CMT), Huntington's disease, Myotonic Dystrophy and Spinal Muscular Atrophy (SMA). These services also cover a number of neuromuscular disease patients including Myasthenia Gravis (MG) and Motor Neuron Disease (MND). A special Botulinum Toxin treatment clinic is offered to a number of patients with focal dystonias, sialorrhea and spasticity.

The Clinic functions the EMG Laboratory. The EMG laboratory provides services for the investigation of the peripheral nervous system including neuropathies, radiculopathies, plexopathies, myopathies and neuromuscular junction defects. Some of the services are Nerve Conduction Studies, Blink Reflex, Jaw Jerk Reflex, Masseter Inhibitory Reflex, Repetitive stimulation, Mixed nerve, Inching technique, Autonomic testing, H-reflex, Electromyography, Quantitative EMG and Single fibre EMG.

Research Interests

Clinic D is interested and involved in a number of research projects on neurogenetic diseases including:

- Charcot-Marie-Tooth polyneuropathies
- Friedreich's Ataxia
- Spinocerebellar Ataxias
- Hereditary motor neuronopathy type Jerash
- FSHMD
- Spinal Muscular Atrophy
- Quantitative EMG & Single Fibre

Selected Publications

Votsi C, Zamba-Papanicolaou E, Georghiou A, Kyriakides T, Papacostas S, Kleopa KA, Pantzaris M, Christodoulou K. Investigation of SCA10 in the Cypriot population: further exclusion of SCA dynamic repeat mutations. Journal of the neurological sciences 2012;323:154-157.

Papathanasiou ES, Zamba-Papanicolaou E. A comparison between disposable and reusable single fiber needle electrodes in relation to stimulated single fiber studies. Clinical neurophysiology : official journal of the International Federation of Clinical Neurophysiology 2012;123:1437-1439.

Zamba-Papanicolaou E, Koutsou P, Daiou C, Gaglia E, Georghiou A, Christodoulou K. High frequency of Friedreich's ataxia carriers in the Paphos district of Cyprus. Acta myologica : myopathies and cardiomyopathies : official journal of the Mediterranean Society of Myology / edited by the Gaetano Conte Academy for the study of striated muscle diseases 2009;28:24-26.

Papathanasiou ES, Zamba-Papanicolaou E. Differential orbicularis oculi involvement in neuromuscular junction dysfunction. Journal of clinical neurophysiology : official publication of the American Electroencephalographic Society 2008;25:293-298.

Nicolaou P, Georghiou A, Votsi C, Middleton LT, Zamba-Papanicolaou E, Christodoulou K. A novel c.5308_5311delGAGA mutation in Senataxin in a Cypriot family with an autosomal recessive cerebellar ataxia. BMC medical genetics 2008;9:28.

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Zamba-Papanicolaou E, Christodoulou K, Christodoulou C, Kyriakidesl T, Middleton LT. Hereditary motor neuronopathies. Revue neurologique 2002;158:1220-1224.

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AEP General
VENP Final Mont
100 dB
Standard Headset
5 Hz
SL : -- dB
Stim : 100 dB
Avg: 16
0.5 uV
2 ms/D
Avg: 168
0.5 uV/D
2 ms/D
Input
Odd/Even
Mean
Diff
Run1
Left
Averaging
Pause/Stop
Erase
Print
BenQ
Exit
Enter

Neurology Clinic E

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Personnel

Neurology Clinic E

- Yiolanda Christou, MD
- Margarita Olymbiou, MSc *(PhD canditate)*
- Styliana Kyriakoudi, MSc *(PhD canditate)*

Neuroscience Laboratory

- Irene Sargiannidou, PhD
- Alexia Kagiava, PhD
- Natasa Schiza, MSc *(PhD canditate)*

Mouse Facility

- George Lapathitis, BSc, MSc, PhD
- Christos Karaiskos, BSc, MSc
- Georgia Philippou
- Constantina Andreou

Activities

Neurology Clinic E provides specialized services for a broad spectrum of neurological disorders focusing on neuromuscular disorders and electromyography. The Clinic provides care for outpatients and inpatients with myasthenia gravis, inherited and acquired peripheral neuropathies and myopathies, motor neuron disease, and other degenerative, autoimmune and metabolic neurological diseases, including inherited and acquired demyelinating disorders of the peripheral and central nervous system. We also perform regularly nerve conduction studies and electromyography for adults and children. The Clinic Head also oversees the activities of the Mouse Facility.

Our research group in the Neuroscience Laboratory has been active both in clinical as well as basic research with emphasis on translational aspects of neuroscience and generating models of neurological disease. We generate and study cellular and mouse models of inherited neuropathies caused by gap junction mutations and gene therapy applications are being designed. The study of molecular mechanisms in inherited and autoimmune disorders of ion channels and related molecules has been another focus of our activities. Furthermore, we have started the investigation of gap junction pathology in post-mortem human brain from multiple sclerosis patients, as well as in Experimental Autoimmune Encephalomyelitis mouse models generated in our lab.

Research Interests

- Generating cellular and animal models of inherited neuropathies
- Studying mechanisms of axonal degeneration in disease models of neuropathy
- Generating disease models of multiple sclerosis
- Studying pathological mechanisms in multiple sclerosis brain
- Developing new therapeutic approaches with gene replacement for inherited neuropathies and leukodystrophies
- Studying disorders of nerve and brain excitability
- Investigating molecular mechanisms of neuronal dysfunction caused by ion channel and cell adhesion molecule defects
- Investigation of genetic and molecular mechanisms of neurodegenerative disorders such as Parkinson’s disease and Motor Neuron disease
- Clinical and genetic investigation of families with inherited neuropathies and other neurogenetic disorders
- Clinical and epidemiological study of myasthenia in Cyprus and investigation of new treatments

Selected Publications

Markoullis K, Sargiannidou I, Schiza N, Hadjisavvas A, Roncaroli F, Reynolds R, Kleopa KA. Gap junction pathology in multiple sclerosis lesions and normal-appearing white matter. Acta neuropathologica 2012;123:873-886.

Markoullis K, Sargiannidou I, Gardner C, Hadjisavvas A, Reynolds R, Kleopa KA. Disruption of oligodendrocyte gap junctions in experimental autoimmune encephalomyelitis. Glia 2012;60:1053-1066.

Irani SR, Pettingill P, Kleopa KA, Schiza N, Waters P, Mazia C, Zuliani L, Watanabe O, Lang B, Buckley C, Vincent A. Morvan syndrome: clinical and serological observations in 29 cases. Annals of neurology 2012;72:241-255.

Kleopa KA. The role of gap junctions in Charcot-Marie-Tooth disease. The Journal of neuroscience : the official journal of the Society for Neuroscience 2011;31:17753-17760.

Vavlitou N, Sargiannidou I, Markoullis K, Kyriacou K, Scherer SS, Kleopa KA. Axonal pathology precedes demyelination in a mouse model of X-linked demyelinating/type I Charcot-Marie Tooth neuropathy. Journal of neuropathology and experimental neurology 2010;69:945-958.

Irani SR, Alexander S, Waters P, Kleopa KA, Pettingill P, Zuliani L, Peles E, Buckley C, Lang B, Vincent A. Antibodies to Kv1 potassium channel-complex proteins leucine-rich, glioma inactivated 1 protein and contactin-associated protein-2 in limbic encephalitis, Morvan's syndrome and acquired neuromyotonia. Brain : a journal of neurology 2010;133:2734-2748.

Sargiannidou I, Vavlitou N, Aristodemou S, Hadjisavvas A, Kyriacou K, Scherer SS, Kleopa KA. Connexin32 mutations cause loss of function in Schwann cells and oligodendrocytes leading to PNS and CNS myelination defects. The Journal of neuroscience : the official journal of the Society for Neuroscience 2009;29:4736-4749.

Sargiannidou I, Ahn M, Enriquez AD, Peinado A, Reynolds R, Abrams C, Scherer SS, Kleopa KA. Human oligodendrocytes express Cx31.3: function and interactions with Cx32 mutants. Neurobiology of disease 2008;30:221-233.


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RACK 5
RACK 4
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Clinical Genetics Clinic

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Personnel

- George Tanteles, MD, MRCPCH
- Elena Spanou Aristidou, BSc, MS
- Turem Delikurt, BSc, MSc *(PhD canditate)*

Activities

Our team has expertise in the diagnosis, management and genetic counseling of patients and people at risk for a wide variety of genetic conditions and/or constellation of medical problems. We provide clinical diagnostic and counseling services for the whole spectrum of genetic conditions at any age, for any system, including prenatal genetics, dysmorphology, neurogenetics, neuropsychiatric genetics, ophthalmic and cancer genetics.

The Clinical Genetics Team consists of two clinical geneticists, two genetic counselors, a postdoctoral research fellow and a genetics nurse. Clinics are held at both The Cyprus Institute of Neurology & Genetics and the Archbishop Makarios III Hospital in Nicosia.

The Clinic serves as a reference centre for the whole of the island. Families are referred by healthcare and other professionals as well as through self-referrals. Typically, reasons for referral include abnormal prenatal diagnosis results, birth defects, chromosomal anomalies, connective tissue disorders, craniosynostoses, developmental delay/mental retardation, hypotonia, inborn errors of metabolism, skeletal dysplasias and tumour disorders. Individuals with family members who have a genetic condition are also referred to clarify their risk for inheriting and/or passing on the genetic condition.

Our group is also active in clinical as well as applied research, working in close collaboration with other Departments and Clinics within CING as well as other healthcare professionals from Cyprus and abroad.

The Clinic is a major training centre for specialist registrars, nurses, scientists and for professionals from other disciplines. Members of the Clinic are also active in various local and international committees which foster the exchange of relevant experience, policies and practices between relevant stakeholders acting in the field of clinical genetics.

Research Interests

Research activities include projects on:

- Epidemiology of genetic disorders in Cyprus
- Rare mental retardation syndromes
- Neuropsychiatric disorders
- Rare Diseases (syndromes and other genetic diseases) specific to the Cypriot population, both in general and in isolated communities
- Genetic and linguistic factors in specific patterns of speech development
- Genodermatoses in Cyprus as part of a European network of collaboration
- Ethical and anthropological studies on the reflection of applications of genetics within the Cypriot society
- Development of models for raising public awareness in a culturally specific manner

Selected Publications

Dixit A, McKee S, Mansour S, Mehta SG, Tanteles GA, Anastasiadou V, Patsalis PC, Martin K, McCullough S, Suri M, Sarkar A. 7q11.23 Microduplication: a recognizable phenotype. Clinical genetics 2013;83:155-161.

Sismani C, Anastasiadou V, Kousoulidou L, Parkel S, Koumbaris G, Zilina O, Bashiardes S, Spanou E, Kurg A, Patsalis PC. 9 Mb familial duplication in chromosome band Xp22.2-22.13 associated with mental retardation, hypotonia and developmental delay, scoliosis, cardiovascular problems and mild dysmorphic facial features. European journal of medical genetics 2011;54:e510-515.

Neocleous V, Skordis N, Portides G, Efstathiou E, Costi C, Ioannou N, Pantzaris M, Anastasiadou V, Deltas C, Phylactou LA. RET proto-oncogene mutations are restricted to codon 618 in Cypriot families with multiple endocrine neoplasia 2. Journal of endocrinological investigation 2011;34:764-769.

Sismani C, Kitsiou-Tzeli S, Ioannides M, Christodoulou C, Anastasiadou V, Stylianidou G, Papadopoulou E, Kanavakis E, Kosmaidou-Aravidou Z, Patsalis PC. Cryptic genomic imbalances in patients with de novo or familial apparently balanced translocations and abnormal phenotype. Molecular cytogenetics 2008;1:15.

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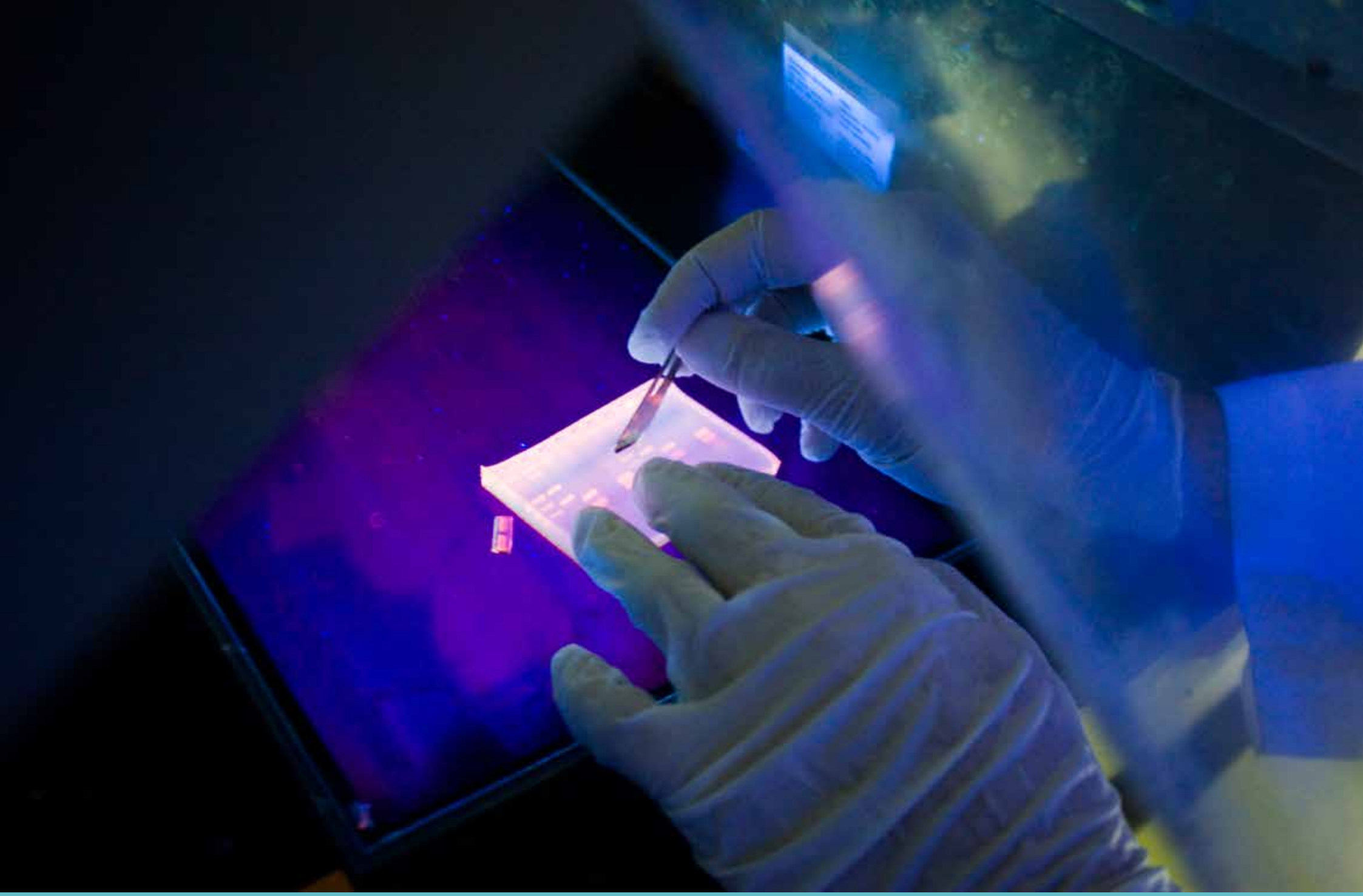
Neocleous V, Portides G, Anastasiadou V, Phylactou LA. Determination of the carrier frequency of the common GJB2 (connexin-26) 35delG mutation in the Greek Cypriot population. International journal of pediatric otorhinolaryngology 2006;70:1473-1477.

Neocleous V, Aspris A, Shahpenterian V, Nicolaou V, Panagi C, Ioannou I, Kyamides Y, Anastasiadou V, Phylactou LA. High frequency of 35delG GJB2 mutation and absence of del(GJB6-D13S1830) in Greek Cypriot patients with nonsyndromic hearing loss. Genetic testing 2006;10:285-289.

Hadjisavvas A, Papasavva T, Loizidou M, Malas S, Potamitis G, Christodoulou C, Pavlides G, Papamichael D, Klonis C, Nasioulas G, Anastasiadou V, Kyriacou K. Novel germline mutations in the APC gene of Cypriot patients with familial and sporadic adenomatous polyposis. Clinical genetics 2006;69:404-409.


POWERTEC

Biomedical Sciences



Department of
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Activities

The Department of Biochemical Genetics acts as the reference centre in Cyprus for the laboratory investigation and monitoring of inherited metabolic disorders (inborn errors of metabolism) and neuromuscular diseases. There are more than 500 known inherited metabolic disorders which cumulatively affect approximately 1 in 500 newborns. Many of these conditions are severely disabling and some are fatal, but an increasing number of them can be treated, provided they are diagnosed promptly. The contribution of the laboratory investigation to the diagnosis of inherited metabolic disorders is crucial since most of these disorders present with non-specific symptoms and are difficult to diagnose clinically.

The Department of Biochemical Genetics has established a wide range of highly specialized biochemical and molecular tests for the diagnosis and monitoring of inherited metabolic disorders including amino acid, organic acid and acylcarnitine analysis. For a list of the services provided please visit our site www.cing.ac.cy.

The Department receives samples from the government and private sector as well as neighboring countries. Over the years many diagnoses were made and in many cases they resulted in the prevention of death or mental retardation.

The Department of Biochemical Genetics also provides therapeutic drug monitoring of immunosuppressive drugs..These measurements are crucial to the success of the transplantation and the prevention of graft rejection.

The Department of Biochemical Genetics is certified by the European Research Network for evaluation and improving of screening, diagnosis and treatment of inherited disorders of metabolism (ERNDIM) and takes part in External Quality Control and Proficiency Schemes. The Department is expected to get accredited according to ISO15189 in 2013.

Research Interests

- The epidemiology and molecular characterization of inherited metabolic disorders in Cyprus
- The deciphering of the cellular events implicated in disease pathology in selected inborn errors of metabolism with an emphasis on lysosomal storage disorders

Selected Publications

Georgiou T, Ho G, Vogazianos M, Dionysiou M, Nicolaou A, Chappa G, Nicolaides P, Stylianidou G, Christodoulou J, Drousiotou A. The spectrum of mutations identified in Cypriot patients with phenylalanine hydroxylase deficiency detected through neonatal screening. Clinical biochemistry 2012;45:588-592.

Drousiotou A, DiMeo I, Mineri R, Georgiou T, Stylianidou G, Tiranti V. Ethylmalonic encephalopathy: application of improved biochemical and molecular diagnostic approaches. Clinical genetics 2011;79:385-390.

Georgiou T, Chuang JL, Wynn RM, Stylianidou G, Korson M, Chuang DT, Drousiotou A. Maple syrup urine disease in Cypriot families: identification of three novel mutations and biochemical characterization of the p.Thr211Met mutation in the E1alpha subunit. Genetic testing and molecular biomarkers 2009;13:657-664.

Kleopa KA, Drousiotou A, Mavrikiou E, Ormiston A, Kyriakides T. Naturally occurring utrophin correlates with disease severity in Duchenne muscular dystrophy. Human molecular genetics 2006;15:1623-1628.

Georgiou T, Stylianidou G, Anastasiadou V, Caciotti A, Campos Y, Zammarchi E, Morrone A, D'Azzo A, Drousiotou A. The Arg482His mutation in the beta-galactosidase gene is responsible for a high frequency of GM1 gangliosidosis carriers in a Cypriot village. Genetic testing 2005;9:126-132.

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Tishkoff SA, Varkonyi R, Cahinhinan N, Abbes S, Argyropoulos G, Destro-Bisol G, Drousiotou A, Dangerfield B, Lefranc G, Loiselet J, Piro A, Stoneking M, Tagarelli A, Tagarelli G, Touma EH, Williams SM, Clark AG. Haplotype diversity and linkage disequilibrium at human G6PD: recent origin of alleles that confer malarial resistance. Science 2001;293:455-462.

Drousiotou A, Stylianidou G, Anastasiadou V, Christopoulos G, Mavrikiou E, Georgiou T, Kalakoutis G, Oladimeji A, Hara Y, Suzuki K, Furihata K, Ueno I, Ioannou PA, Fensom AH. Sandhoff disease in Cyprus: population screening by biochemical and DNA analysis indicates a high frequency of carriers in the Maronite community. Human genetics 2000;107:12-17.

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Activities

The Department of Cardiovascular Genetics pursues research on identifying genetic and environmental factors that contribute to the development of cardiovascular disease. In this field it has developed diagnostic assays for thrombophilia and familial hypercholesterolaemia (FH). In collaboration with Cape Town University the Department has investigated the effect of a series of genes on athletic performance and endurance in ironman triathletes. The Department is currently at the stage of initiating collaborative genetic studies on Thoracic Aortic Aneurisms (TAAs) with both local and foreign Centers of Excellence.

The Laboratory of Forensic Genetics (LabFoG) uses state of the art DNA-based typing methodologies to study evidence from civil, criminal, mass disaster and missing persons investigations. The LabFoG, from 1996 - 2012, was actively involved in the attempts to identify, through the analysis of skeletal remains, Cypriot missing persons since the tragic events of 1958-1964 and 1974. The Laboratory has also pursued research which focused on skeletal remains from archaeological sites. More recently, the LabFoG is pursuing research that concentrates on the primary and secondary transfer of DNA as it applies to criminal investigations. The LabFoG maintains the National Criminal DNA database for the Cyprus Police Authorities. Since 1999, the Laboratory of Forensic Genetics, is a Member of the European Network of Forensic Science Institutes DNA Working Group.

Research Interests

- Study of candidate genes and environmental factors involved in cardiovascular disease
- MALDI-TOF assisted SNP genotyping
- The role of genes in athletic performance
- MALDI-TOF in Forensic Genetics and Cardiovascular Disease
- Transfer of cells in Forensic Genetics

Selected Publications

Diakou M, Miltiadous G, Xenophontos SL, Manoli P, Cariolou MA, Elisaf M. Spectrum of LDLR gene mutations, including a novel mutation causing familial hypercholesterolaemia, in North-western Greece. European journal of internal medicine 2011;22:e55-59.

Loizidou MA, Cariolou MA, Neuhausen SL, Newbold RF, Bashiardes E, Marcou Y, Michael T, Daniel M, Kakouri E, Papadopoulos P, Malas S, Hadjisavvas A, Kyriacou K. Genetic variation in genes interacting with BRCA1/2 and risk of breast cancer in the Cypriot population. Breast cancer research and treatment 2010;121:147-156.

Xenophontos S, Hadjivassiliou M, Karagrigoriou A, Demetriou N, Miltiadous G, Marcou I, Elisaf M, Mikhailidis DP, Cariolou MA. Low HDL cholesterol, smoking and IL-13 R130Q polymorphism are associated with myocardial infarction in Greek Cypriot males. A pilot study. The open cardiovascular medicine journal 2008;2:52-59.

Parson W, Niederstatter H, Lindinger A, Gill P, Group EDW. Y-STR analysis on DNA mixture samples--results of a collaborative project of the ENFSI DNA Working Group. Forensic science international Genetics 2008;2:238-242.

Irwin J, Saunier J, Strouss K, Paintner C, Diegoli T, Sturk K, Kovatsi L, Brandstatter A, Cariolou MA, Parson W, Parsons TJ. Mitochondrial control region sequences from northern Greece and Greek Cypriots. International journal of legal medicine 2008;122:87-89.

Saunders CJ, Xenophontos SL, Cariolou MA, Anastassiades LC, Noakes TD, Collins M. The bradykinin beta 2 receptor (BDKRB2) and endothelial nitric oxide synthase 3 (NOS3) genes and endurance performance during Ironman Triathlons. Human molecular genetics 2006;15:979-987.

Saunders CJ, de Milander L, Hew-Butler T, Xenophontos SL, Cariolou MA, Anastassiades LC, Noakes TD, Collins M. Dipsogenic genes associated with weight changes during Ironman Triathlons. Human molecular genetics 2006;15:2980-2987.

Miltiadous G, Xenophontos S, Bairaktari E, Ganotakis M, Cariolou M, Elisaf M. Genetic and environmental factors affecting the response to statin therapy in patients with molecularly defined familial hypercholesterolaemia. Pharmacogenetics and genomics 2005;15:219-225.


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- Georgia Herodotou

Activities

The Department of Cytogenetics and Genomics provides diagnostic services, research as well as education in the area of Cytogenetics and Genomics. It is accredited by the College of American Pathologists (CAP) and meets the European quality control standards in Cytogenetics and Molecular Genetics.

The Department consists of the following laboratories: Clinical & Molecular Cytogenetics, Molecular Genetics of Human Syndromes, Microarrays, and Cell Lines. It serves as a referral center for prenatal and postnatal diagnosis of chromosomal disorders (e.g. Down syndrome), Fragile X syndrome, as well as many other genetic diseases and syndromes which cause intellectual disability, multiple congenital anomalies, developmental delay, abnormal sexual differentiation and many other conditions.

One of the Department's main focuses is to investigate at high resolution the whole human genome through microarray technology (array-CGH) for the identification of small and cryptic copy number changes and copy number variations, which cause many genetic syndromes and conditions. In addition, the Department maintains the specialized facility for the establishment and cryopreservation of immortal cell lines of patients for future diagnostic and/or research purposes. It carries out very competitive research programs in the area of human genomics and publishes its scientific outcome in international peer review scientific journals.

Research projects receive funding from the Cyprus Research Promotion Foundation, the 5th, 6th and 7th European Union Framework Programs and other international organizations. The Department is involved in several research programs and has collaborators in Universities and Research Institutes in Europe and the US.

The Department is involved in the provision of education by providing lectures and laboratory training to scientists, medical doctors and students. Furthermore, PhD students have graduated or are currently carrying out their doctoral degree.

Research Interests

- Application of high-resolution microarrays in prenatal diagnosis
- Development of exon specific array for the detection of microdeletions and microduplications in genes of the chromosome X
- Genetic investigation of patients with X-linked Intellectual Disability (XLID)
- Genomic investigation and characterization of unknown and rare genetic and neurological syndromes in the Cyprus population with high resolution microarrays
- Investigation and characterization of the clinical significance of Copy Number Variations in the human genome
- Genetic Investigation of autism

Selected Publications

Kasnauskiene J, Ciuladaite Z, Preiksaitiene E, Matuleviciene A, Alexandrou A, Koumbaris G, Sismani C, Pepalyte I, Patsalis PC, Kucinskas V. A single gene deletion on 4q28.3: PCDH18--a new candidate gene for intellectual disability? European journal of medical genetics 2012;55:274-277.

Sismani C, Anastasiadou V, Kousoulidou L, Parkel S, Koumbaris G, Zilina O, Bashiardes S, Spanou E, Kurg A, Patsalis PC. 9 Mb familial duplication in chromosome band Xp22.2-22.13 associated with mental retardation, hypotonia and developmental delay, scoliosis, cardiovascular problems and mild dysmorphic facial features. European journal of medical genetics 2011;54:e510-515.

Evangelidou P, Sismani C, Ioannides M, Christodoulou C, Koumbaris G, Kallikas I, Georgiou I, Velissariou V, Patsalis PC. Clinical application of whole-genome array CGH during prenatal diagnosis: Study of 25 selected pregnancies with abnormal ultrasound findings or apparently balanced structural aberrations. Molecular cytogenetics 2010;3:24.

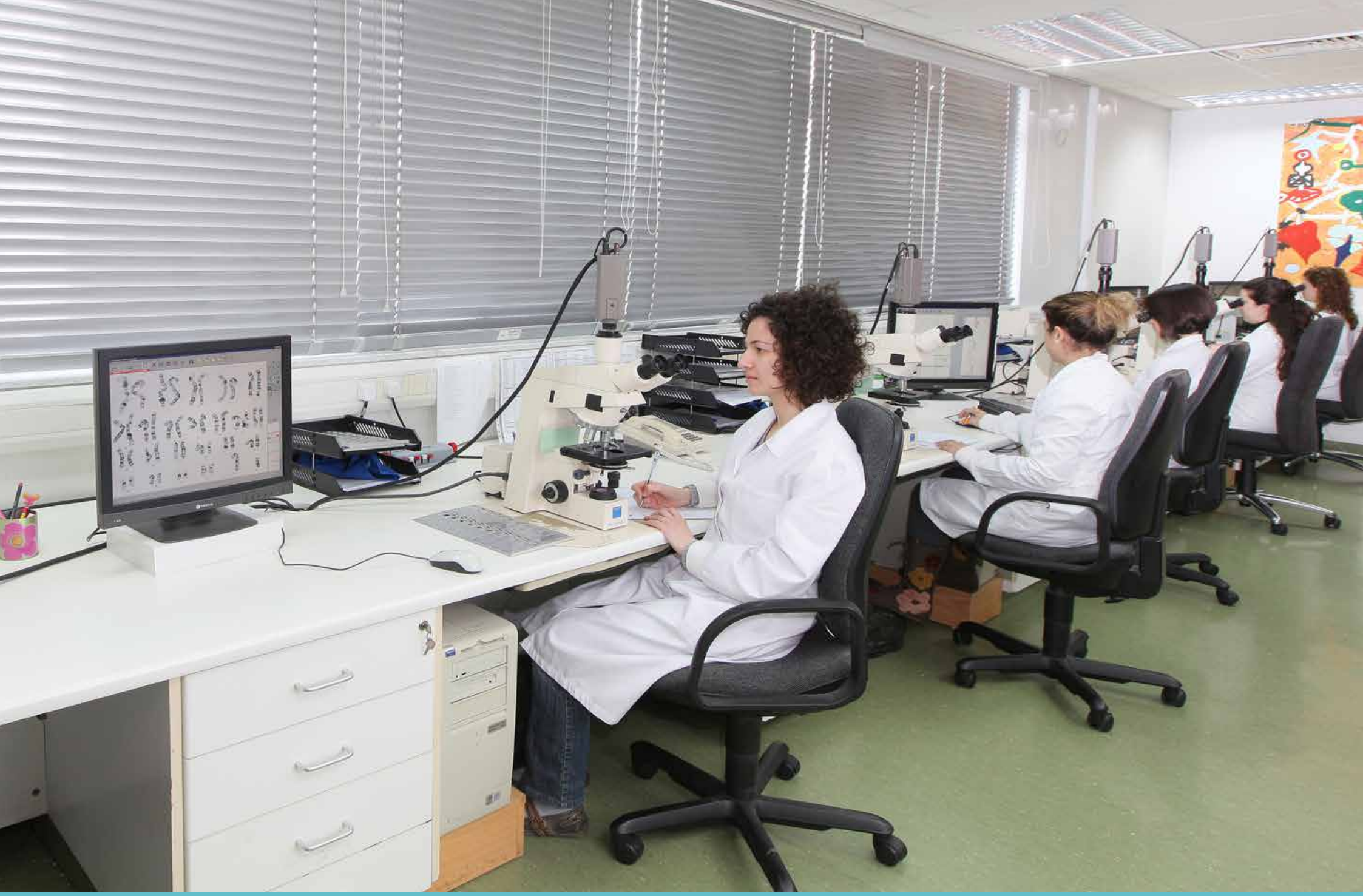
Bashiardes S, Kousoulidou L, van Bokhoven H, Ropers HH, Chelly J, Moraine C, de Brouwer AP, Van Esch H, Froyen G, Patsalis PC. A new chromosome x exon-specific microarray platform for screening of patients with X-linked disorders. The Journal of molecular diagnostics : JMD 2009;11:562-568.

Sismani C, Kitsiou-Tzeli S, Ioannides M, Christodoulou C, Anastasiadou V, Stylianidou G, Papadopoulou E, Kanavakis E, Kosmaidou-Aravidou Z, Patsalis PC. Cryptic genomic imbalances in patients with de novo or familial apparently balanced translocations and abnormal phenotype. Molecular cytogenetics 2008;1:15.

Kousoulidou L, Mannik K, Sismani C, Zilina O, Parkel S, Puusepp H, Tonisson N, Palta P, Remm M, Kurg A, Patsalis PC. Array-MAPH: a methodology for the detection of locus copy-number changes in complex genomes. Nature protocols 2008;3:849-865.

Patsalis PC, Evangelidou P, Charalambous S, Sismani C. Fluorescence in situ hybridization characterization of apparently balanced translocation reveals cryptic complex chromosomal rearrangements with unexpected level of complexity. European journal of human genetics : EJHG 2004;12:647-653.

Sismani C, Syrrou M, Christodoulou K, Hamel B, Chelly J, Yntema HG, van Bokhoven H, Tzoufi M, Georgiou I, Patsalis PC. A gene for nonsyndromic X-linked mental retardation (MRX77) maps to Xq12-Xq21.33. American journal of medical genetics Part A 2003;122A:46-50.



Department of Electron Microscopy/ Molecular Pathology

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- Christiana Demetriou *(PhD candidate)*
- Kleitos Socratous, MSc (*PhD candidate)*
- Maria Kakkoura, *(PhD candidate)*

Activities

The Department operates the only Transmission Electron Microscope available in Cyprus. It offers a centralized ultrastructural diagnostic service in histopathology and deals with a range of human diseases, including neuromuscular, kidney and skin disorders as well as the diagnosis of undifferentiated neoplasms.

The Department also offers a range of Molecular Genetic tests for diagnosing predisposition to common types of familial cancer, such as breast and colorectal. A spectrum of predisposition genes are analysed including: BRCA1 and BRCA2 genes which predispose to hereditary breast and ovarian cancer; APC and MYH genes that are linked to Familial Adenomatous Polyposis (FAP) syndrome and mismatch repair genes (MLH1, MSH2, PMS2 and MSH6) that predispose to Hereditary Non-Polyposis Colorectal Cancer syndrome known as HNPCC.

An integral part of delivering the services outlined above is the Cancer Genetics Clinic which has been established at CING in collaboration with the Clinical Genetics Clinic.

The Department also performs mutation analysis of somatic DNA for establishing a pharmaco-genomics profile that determines the selection of patients targeted treatment.

The Department participates in the European Molecular Genetics Quality Network (EMQN) assessment schemes, for evaluating the quality and standards of the services that are being offered, since 2000.

Research Interests

- Familial cancer genetics: Characterize pathogenic mutations in cancer predisposition genes in Cypriot families
- Epidemiology of breast cancer: The Department is coordinating a National epidemiological study on breast cancer in order to identify risk factors which modify breast cancer risk in the Cypriot population
- Functional studies on the role of the BRCA genes: BRCA unclassified mutations that have been identified in the Cypriot families are being cloned, expressed and functionally evaluated in cell cultures, in order to understand their clinical significance
- Breast Cancer Proteomics and Biomarker Discovery: Investigation of the mechanisms of breast cancer progression, using proteomics with the ultimate aim of identifying novel biomarkers in Cypriot breast cancer patients
- Mitochondrial myopathies: In collaboration with the Department of Neuropathology we are investigating the morphological and mitochondrial DNA defects that characterize Cypriot patients with mitochondrial myopathies

Selected Publications

Demetriou CA, Hadjisavvas A, Loizidou MA, Loucaides G, Neophytou I, Sieri S, Kakouri E, Middleton N, Vineis P, Kyriacou K. The mediterranean dietary pattern and breast cancer risk in Greek-Cypriot women: a case-control study. BMC cancer 2012;12:113.

Loizidou MA, Hadjisavvas A, Ioannidis JP, Kyriacou K. Replication of genome-wide discovered breast cancer risk loci in the Cypriot population. Breast cancer research and treatment 2011;128:267-272.

Sutton CW, Rustogi N, Gurkan C, Scally A, Loizidou MA, Hadjisavvas A, Kyriacou K. Quantitative proteomic profiling of matched normal and tumor breast tissues. Journal of proteome research 2010;9:3891-3902.

Loizidou MA, Cariolou MA, Neuhausen SL, Newbold RF, Bashiardes E, Marcou Y, Michael T, Daniel M, Kakouri E, Papadopoulos P, Malas S, Hadjisavvas A, Kyriacou K. Genetic variation in genes interacting with BRCA1/2 and risk of breast cancer in the Cypriot population. Breast cancer research and treatment 2010;121:147-156.

Hadjisavvas A, Loizidou MA, Middleton N, Michael T, Papachristoforou R, Kakouri E, Daniel M, Papadopoulos P, Malas S, Marcou Y, Kyriacou K. An investigation of breast cancer risk factors in Cyprus: a case control study. BMC cancer 2010;10:447.

Gale DP, de Jorge EG, Cook HT, Martinez-Barricarte R, Hadjisavvas A, McLean AG, Pusey CD, Pierides A, Kyriacou K, Athanasiou Y, Voskarides K, Deltas C, Palmer A, Fremeaux-Bacchi V, de Cordoba SR, Maxwell PH, Pickering MC. Identification of a mutation in complement factor H-related protein 5 in patients of Cypriot origin with glomerulonephritis. Lancet 2010;376:794-801.

Loizidou MA, Michael T, Neuhausen SL, Newbold RF, Marcou Y, Kakouri E, Daniel M, Papadopoulos P, Malas S, Hadjisavvas A, Kyriacou K. DNA-repair genetic polymorphisms and risk of breast cancer in Cyprus. Breast cancer research and treatment 2009;115:623-627.

Zouvani I, Aristodemou S, Hadjisavvas A, Michael T, Vassiliou M, Patsias C, Pierides A, Ioannou K, Kyriacou K. Incidence of thin basement membrane nephropathy in 990 consecutive renal biopsies examined with electron microscopy. Ultrastructural pathology 2008;32:221-226.


Rotor-Gene Q

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Activities

The Department of Molecular Genetics, Function & Therapy (MGFT) carries out research and services in specific biomedical areas in order to achieve its mission in accordance with the wider mission of the CING. Regarding research, the Department is interested in identifying the molecular causes of diseases, in studying the function of molecules which are involved in diseases and in the development of novel genetic approaches which may form the basis of therapy of diseases.

Particularly, the use of regulatory RNA in research and the involvement of RNA in disease pathogenesis are priority research fields in MGFT. Moreover, there is interest in developing and exploiting genetic and biological tools for the appropriate implementation of the research projects. The Department develops and applies, also, approaches (mainly genetic) for the diagnosis of several diseases which affect the people of Cyprus and the wider area. Some of these approaches are unique and provide novel findings about these diseases. MGFT participates in several quality control schemes for its services.

Research Interests

- The involvement of RNA in diseases
- The use of regulatory RNA as genetic tools for the study and development of therapeutic approaches of diseases
- The study of the molecular pathogenesis of Myotonic Dystrophy and the restoration of the disease phenotype
- Investigation of the pathway which leads to the formation of skeletal muscle cells
- Induction of myogenesis in diseases
- Induction of reversal of muscle cell differentiation and reactivation of muscle cells
- Development of approaches for efficient delivery of genetic material in cells
- Identification of genetic defects in diseases which affect the population of Cypriots
- Development of new diagnostic methods

Selected Publications

Neocleous V, Skordis N, Shammas C, Efstathiou E, Mastroyiannopoulos NP, Phylactou LA. Identification and characterization of a novel X-linked AVPR2 mutation causing partial nephrogenic diabetes insipidus: a case report and review of the literature. Metabolism: clinical and experimental 2012;61:922-930.

Mastroyiannopoulos NP, Nicolaou P, Anayasa M, Uney JB, Phylactou LA. Down-regulation of myogenin can reverse terminal muscle cell differentiation. PloS one 2012;7:e29896.

Koutsoulidou A, Mastroyiannopoulos NP, Furling D, Uney JB, Phylactou LA. Expression of miR-1, miR-133a, miR-133b and miR-206 increases during development of human skeletal muscle. BMC developmental biology 2011;11:34.

Mastroyiannopoulos NP, Shammas C, Phylactou LA. Tackling the pathogenesis of RNA nuclear retention in myotonic dystrophy. Biology of the cell / under the auspices of the European Cell Biology Organization 2010;102:515-523.

Neocleous V, Ioannou YS, Bartsota M, Costi C, Skordis N, Phylactou LA. Rare mutations in the CYP21A2 gene detected in congenital adrenal hyperplasia. Clinical biochemistry 2009;42:1363-1367.

Lee YB, Bantounas I, Lee DY, Phylactou L, Caldwell MA, Uney JB. Twist-1 regulates the miR-199a/214 cluster during development. Nucleic acids research 2009;37:123-128.

Hjiantoniou E, Anayasa M, Nicolaou P, Bantounas I, Saito M, Iseki S, Uney JB, Phylactou LA. Twist induces reversal of myotube formation. Differentiation; research in biological diversity 2008;76:182-192.

Mastroyiannopoulos NP, Feldman ML, Uney JB, Mahadevan MS, Phylactou LA. Woodchuck post-transcriptional element induces nuclear export of myotonic dystrophy 3' untranslated region transcripts. EMBO reports 2005;6:458-463.


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Activities

The Molecular Genetics Thalassaemia Department provides diagnostic services, research and education in the field of thalassaemia and other haemoglobinopathies.

The services include molecular and prenatal diagnosis of all forms of haemoglobinopathies, preimplantation genetic diagnosis (PGD) for thalassaemias and other rare diseases, non-invasive prenatal diagnosis (NIPD) test for the detection of the Y chromosome in the maternal plasma of pregnant women for severe X-linked disorders and NIPD for RHD status in Rhesus-negative pregnant women.

The research activities of the Department comprise the development and improvement of diagnostic tests for haemoglobinopathies, the development of NIPD methods for thalassaemia, studies for finding new HbF inducers for the drug therapy of thalassaemia, pharmacogenetic and pharmacogenomic studies for β-thalassaemia patients and studies aiming to establish a safe, therapeutically effective and clinically applicable gene therapy for β-thalassaemia. As coordinator of the FP6 ITHANET project we pioneered the use e-Infrastructure tools for the thalassaemia community and continue to develop the ITHANET portal as a community tool for the thalassaemias and haemoglobinopathies.

The Department provides training on laboratory aspects, summer internships and supervises BSc, MSc and PhD theses.

Research Interests

- Improvement of thalassaemia diagnostic services: development of new tests for PGD of rare diseases, development of novel NIPD methods for β-thalassaemia, and the determination of the molecular basis of high HbF in Cyprus
- Development of e-Infrastructure tools for thalassaemia: establishing the ITHANET portal as a scientific community portal for the thalassaemias, providing comprehensive database and interactive tools for the thalassaemias
- Drug therapy for thalassaemia: identification and development of novel chemical compounds with HbF-inducing activity for the treatment of thalassaemia and for pharmacogenetic and -genomic studies to determine patient responses to drugs
- Gene therapy for β-thalassaemia: developing improved vectors for more efficient and mutation-specific gene therapy of β-thalassaemia

Selected Publications

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 beta-Thalassaemia. Annals of human genetics 2013;77:115-124.

Destouni A, Christopoulos G, Vrettou C, Kakourou G, Kleanthous M, Traeger-Synodinos J, Kanavakis E. Microsatellite markers within the alpha-globin gene cluster for robust preimplantation genetic diagnosis of severe alpha-thalassemia syndromes in Mediterranean populations. Hemoglobin 2012;36:253-264.

Borg J, Phylactides M, Bartsakoulia M, Tafrali C, Lederer C, Felice AE, Papachatzopoulou A, Kourakli A, Stavrou EF, Christou S, Hou J, Karkabouna S, Lappa-Manakou C, Ozgur Z, van Ijcken W, von Lindern M, Grosveld FG, Georgitsi M, Kleanthous M, Philipsen S, Patrinos GP. KLF10 gene expression is associated with high fetal hemoglobin levels and with response to hydroxyurea treatment in beta-hemoglobinopathy patients. Pharmacogenomics 2012;13:1487-1500.

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Activities

The Molecular Virology Department (MVD) was established in 1995. Our aim is to combine serological and molecular methods for specifi c and precise diagnosis of viral infections. Today, as Diagnostic Services, we provide tests to diagnose more than 20 viruses causing clinical symptoms in children, immunosupressed patients, pregnant women etc. We offer qualitative and wherever relevant quantitative diagnostic tests for viruses including Herpesviruses, enteroviruses, adenoviruses, parvoviruses, Hepatitis B virus, Hepatitis C virus, Human Papillomavirus, BK virus, Toxoplasma etc. Each year, we perform hundreds of diagnostic tests evaluating the presence of antiviral antibodies and viral genomes.

Our Department is considered the Reference Laboratory for the Detection and Surveillance of enteroviruses since 1996. Routinely and also in emergency cases we are able to detect and type enteroviruses circulating in the population in order to prevent epidemics. For similar reasons, we prepared in-house protocols for respiratory viruses, Noroviruses, Adenoviruses and Rotaviruses. We consider it another major achievement of our Department to assist health offi cials to identify viral agents and offering support in preparedness to prevent epidemics.

We pay particular attention to maintain the highest possible standards through internal and external quality control assessments and we therefore participate in multiple QCMD proficiency tests on a yearly basis.

Research Interests

We focus on the role of viruses in neurological syndromes with emphasis on Multiple Sclerosis (MS). We studied the possible role of herpesviruses in the onset and progression of MS. This work was funded by a competitive grant awarded by the American National Multiple Sclerosis Association. Our interests in MS are further pursued through the creation and study of a mouse model of MS (Theiler's murine encephalomyelitis virus infected mouse model).

- In the field of Neurovirology, the Departmental interests focus on looking at molecular mechanisms involved in neurotropic viruses passing from the peripheral to the central nervous system (CNS) and the interactions of these neurotropic viruses can have with cells of the CNS eg. oligodendrocytes
- The Department is also involved in studying HPV infections in Cyprus. Our interest is focused on viral interactions with host proteins
- Our department is the reference laboratory for enteroviruses in Cyprus. Surveillance of viruses and monitoring of their circulation/genetic evolution is possible to identify potential strains to cause epidemics and assist in the development of prevention strategies
- In Environmental Virology, we investigate the presence and spreading of enteroviruses/polioviruses, adenoviruses, respiratory viruses These studies allow for: the surveillance of viral epidemics and the study of variants and emerging viruses

Selected Publications

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Activities

The Neurogenetics Department (ND) provides molecular genetic services, carries out research programmes and offers postgraduate education and training.

ND specialized services include diagnostic, pre-symptomatic and carrier testing as well as prenatal diagnosis, for a wide range of neurogenetic diseases such as amyloidosis (FAP), Huntington disease (HD), ataxia (FRDA, SCA1, SCA2, SCA3/MJD, SCA6, SCA7, SCA8, SCA10, SCA12, SCA17, DRPLA, AOA1), Charcot-Marie-Tooth disease (CMT1A, CMT1B, CMT1D, CMT1E, CMT1F, CMTX1, CMT4A, CMT4E, CMT2A, CMT2D, CMT2E, CMT2I, CMT2J, CMT2K, ARCMT2), hereditary neuropathy with liability to pressure palsies (HNPP), spinal muscular atrophy (SMA), myotonic dystrophy (DM1), amyotrophic lateral sclerosis (ALS1, ALS10, ALS6), Parkinson disease (PARK8) and hereditary spastic paraplegia (SPG3A, SPG4, SPG31, SPG44).

ND translational research is focused on the identification of disease associated genes/mutations and risk factors, elucidation of the molecular mechanisms leading to pathogenesis and disease and investigation towards the development of more effective therapies for the benefit of the patient.

ND educational activities include teaching of the postgraduate course “MG103: Methodologies and technologies applied in Medical Genetics” for the MSc/PhD programmes of the Cyprus School of Molecular Medicine, supervision/mentoring of MSc and PhD students, supervision of BSc projects as well as specialized trainings and internships.

Research Interests

- Identification of novel genes/mutations in ataxia
- Identification of novel genes/mutations in neuropathy
- Investigation of the molecular mechanisms involved in the development of ataxia
- Investigation of the molecular mechanisms involved in the development of neuropathies
- Investigation of Type 2 Diabetes susceptibility loci in the Cypriot population

Selected Publications

Nicolaou P, Cianchetti C, Minaidou A, Marrosu G, Zamba-Papanicolaou E, Middleton L, Christodoulou K. A novel LRSAM1 mutation is associated with autosomal dominant axonal Charcot-Marie-Tooth disease. European journal of human genetics : EJHG 2013;21:190-194.

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- Elisavet Papageorgiou, PhD *(Visiting Scientist)*
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Activities

Translational Genetics Team is compiled by a dynamic group of scientists and is dedicated to carry out research beyond the state-of-the-art, as well as postgraduate level education. The Team utilizes the latest technological tools and molecular methodologies to study the genomics and methylomics of the fetal human embryo at the 1st, 2nd and 3rd trimester. The Team also focuses in developing new methodologies for Non-Invasive Prenatal Diagnosis for genetic disorders by utilizing fetal derived DNA, and other, biomarkers present in maternal blood to decipher the fetal genome and identify alterations in early stages of gestation. These very competitive research programs are especially facilitated by close national and international collaborations with leading industry and academic institutions. The great significance of the Team's accomplishments is illustrated by the high impact of their publications, and the establishment of the NIPD Genetics Ltd, an Institute spin-off biotech company. In addition, research findings were filed as a patent application for a novel method for Non-Invasive Prenatal Diagnosis of fetal aneuploidies in 2011.

FP7 European Research Project - ANGELab

A major competitive EU research grant of €11 million has been jointly obtained by the research team of Ph. Patsalis of CING and NIPD Genetics. Approximately €1,6m will come to Prof. Patsalis Team. The FP7 European research grant is a joint effort to combine the biological science and microsystem engineering to develop a novel machine the size of a printer, which will function as a complete laboratory, a technology known as lab-on-a-chip. The machine, called ANGELab, will carry out non-invasive prenatal diagnosis of Down Syndrome and other genetic disorders.

ERC Advanced Grant - NIPD

The European Research Council (ERC) awarded a very significant funding of Research Excellence, the ERC Advanced Grant of €2,5m to Prof. Ph. Patsalis. The research proposal is based on the discovery, understanding and complete characterization of control of all genes of the human fetus. Then, the complete fetal genome sequence will be re-assembled as it is heavily fragmented in maternal blood. With the successful completion of this research, Patsalis' team will be in a position to offer non-invasive prenatal diagnosis to all pregnant women for many known genetic disorders.

By virtue of its focus on research, the team is actively involved in training programs offered by the Cyprus School of Molecular Medicine, and accommodates research projects by several graduate students. Furthermore, several previous PhD students have successfully completed their theses and have become accomplished members of the scientific community.

Research Interests

- Study the genomics and methylomics of the fetal human embryo
- Identification of fetal biomarker for the diagnosis and prevention of genetic disorders
- Development of Non-Invasive Prenatal Diagnosis for genetic disorders
- Use of Next Generation Sequencing technologies for identifying pathogenic mutations in fetal DNA

Selected Publications

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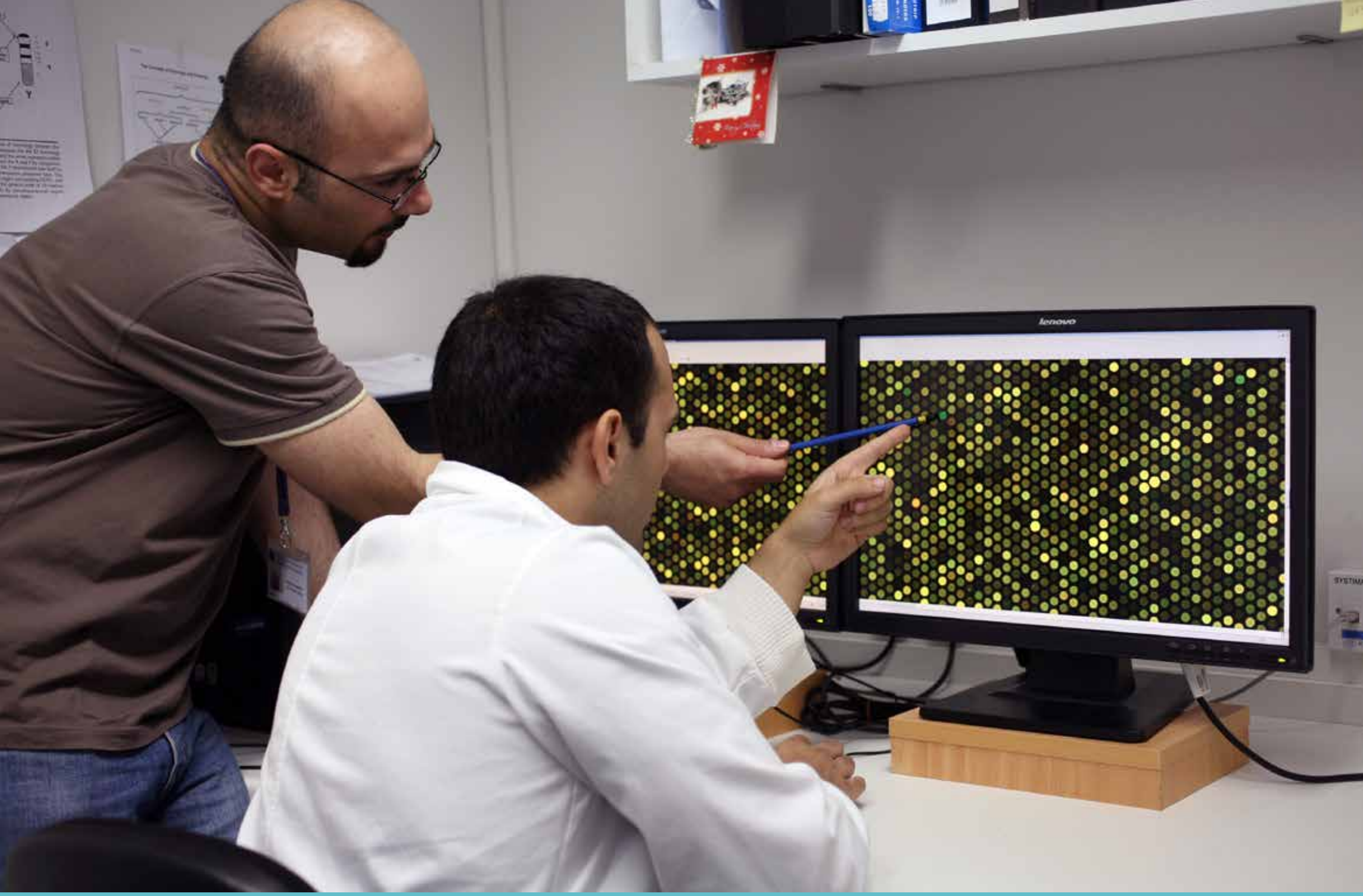
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