

European Society of Human Genetics

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Medical Genetics in the Czech Republic



Prof. Petr Goetz

consequences

History

The history of medical genetics in our country has strong roots. Mendel's pioneering work (1865) was performed in Brno (Brünn; then part of the Austrian Habsburg Monarchy). Subsequently, rediscovery of Mendel's laws at the very beginning of the last century, also substantially influenced Czech biologists. In this respect, Prof. V. R. Růžička's monograph 'Human Inheritance in Health and Disease' was published as early in 1917. Among pre-World War II founders and fearless supporters of medical genetics indisputably belongs Prof. B. Sekla, chairman of the Biological Institute located former Faculty of General Medicine in Prague. His fundamental monograph "Inheritance in Nature and Society", published in Prague in 1937, stimulated among others, the establishment of the first genetic counselling unit in Prague. In its second post-WWII edition (published in 1946) Prof.

Sekia Com

The Totalitarian power in communist Soviet Union used by Lysenko to suppress all activities in genetics, stopped a promising progress of medical genetics also in our country. The period of Lysenkism in the fifties of the last century, among other tested human and scientific moral of our geneticists. Hardly anybody passed.

However, Professor Sekla (the only one of our eminent biologists-geneticists) never accepted Lysenkism and even publicly criticized it with consequent scientific and political oppression. Nevertheless, during the Lysenkism period Czech immunogenetics achieved world-wide level with the group of Dr. Hašek, who formulated the laws of immunological tolerance at the same time (but published them in a Czech journal) as the Nobel Laureate Prof. Medawar.

Progress accelerated after the official rejection of Lysenkism in the early sixties. In 1963, the "Committee for Medical Genetics" was founded and integrated within the former Czech Society of Endocrinology of Jan Evangelista Purkyně (www.cls.cz). Two years later, an independent "Society of Medical Genetics" was established. Since mid-sixties, clinical cytogenetics has flourished and since 1963, the Cytogenetics Section of the Biological Society has developed very well. This is reflected by the organisation of international workshops. The first scientific prizes in genetics of the Czech Ministry of Health were awarded. In the early sixties Assoc. Prof. Černý, co-worker of Prof. Sekla and Assoc. Prof. Izakovič were pioneers in modern clinical genetics and cytogenetics.



Johann Gregor Mendel

After the Soviet invasion of our country, back in 1968, many geneticists were politically and/or even scientifically oppressed, while some of them were not politically tolerated as chairmen of the Society of Medical Genetics or as chairmen of Institutes/Departments of Medical Genetics. Soviet invasion also led to emigration of several prominent experimental and clinical geneticists, such as Prof. Laxova, Prof. Soudek, Prof. Kalouskova, Prof. Cervenka. Nevertheless, medical genetics has been officially acknowledged as an important clinical discipline by the Ministry of Health, which formed the basis of its development. Our speciality has been further developed and supported by dedicated long-term research state plans, with increased scientific cooperation within former COMECON countries. In 1980 a rather modern conception of genetics care has been established. This act formed a network of medical genetics departments within our country (mostly comprising counselling and cytogenetics units), with associated specialised laboratories for the detection and screening of metabolic disorders (e.g. phenylketonuria). Czech Republic was the first to introduce prenatal diagnostics in 1970. Contacts with Western colleagues remained and the local conferences under the auspices of the Czech Medical

Society of J.E. Purkyne had a broad International participation. For instance this was the only place where Eastern- and Western German geneticists could meet in person. This development logically led to the organisation of the International Conference on Prenatal Diagnosis in 1990 in Prague.

After 1990, democratic principles have been gradually restored in the Society of Medical Genetics and in the whole society since the Velvet Revolution in 1989. Increased cooperation with the Western colleagues and return of freshly trained young scientists led to the introduction of molecular genetic diagnosis and research. Both before and after the foundation of the Slovak Society of Medical Genetics at the end of the sixties, cooperation between Czech and Slovak geneticists has been very close and fruitful. Within the former Czechoslovakia a federal committee was created in order to facilitate coordination of congress organisation and joint position papers. Even after the split of Czechoslovakia in 1993, close contacts and cooperation has continued and friendly relationships have remained.



Graduating ceremony at aula magna at Charles University

Society of Medical Genetics of the Czech Medical Society of J.E.Purkyne (http://www.czechia.com/slg)

Although originally our Society was relatively homogenous and mostly formed by clinical geneticists (current members –220 members) it recently underwent diversification according to the professional interests of its members. This led to the establishment of a "Molecular Genetics Section", while close relations remained with the "Cytogenetics Section of the Biological Society of the Czech Academy of Sciences" and the "Division for Prenatal Diagnosis of the Czech Society for Gynaecology & Obstetrics". In addition, it is important to jointly address birth defects. Thus, in this respect it is necessary to mention a close collaboration with both committees of the "Division for Ultrasound Diagnostics of the Czech Society for Gynaecology &

Obstetrics". Similarly, cooperation with the "Czech Biochemical Society" has many years of lasting tradition. Recently, a new "Section for Biochemical Genetics" was established and we also have close contacts with the "Czech Society of Cellular and Gene Therapy".

The first national Congress of Medical Genetics was held in 1972 in Plzeň (Pilsen) with subsequent congresses following at 3 to 4 year intervals in different places of our country. The professional standards of those congresses were traditionally very high, even in difficult times when the organisers succeeded in keeping contact with the international developments through the participation of invited foreign speakers.

Apart from organising congresses and symposia, the Society has been active in organising the so-called "Working Days" or workshops as well as small symposia at a minimum of six per year. It is especially pertinent to mention the professional and organisational cooperation between members of the Society and the Committee for the annual renowned conferences of "Clinical and Experimental Cytogenetics", organised by the "Cytogenetics Section of the Biological Society of the Czech Academy of Sciences". Similarly, activities of the "Ultrasound Division of the Czech Society for Gynaecology and Obstetrics", serve as an excellent example of reciprocal problem solving, in the area of prenatal diagnosis. Additional cooperation includes regular conferences on the "Clinical and Biochemical Genetics of Hyperlipoproteinemia" with broad International participation and reputation.

Medical practitioners and academics from various fields of medicine (internal medicine, cardiology, diabetology, clinical biochemistry, pediatrics, neurology, oncology etc.) regularly attended all workshops. Workshops dealing with "Inherited Metabolic Diseases" date since 1970 and were held regularly every two years alternately in Czechia, Moravia and Slovakia. Clinical Genetics workshops have focused on all relevant aspects, were always meticulously organised and had broad attendance. Finally, there is an established tradition of workshops for "DNA Diagnostics" and "Oncogenetics".

In 1991 we received collective membership within the "European Society for Human Genetics". Since 1997 our Society has become a bona fide member of the "International Federation of Human Genetic Societies" and since 1998 a member of the "Society for Medical Genetics in Alps-Adria region". Members of the Cytogenetic Society are also collective members of the "European Cytogeneticists Association" (ECA).

Education, training, getting specialization

Pre-graduate education in clinical genetics started in 1967 within the frame of Pediatrics. Currently, principles of genetics, molecular genetics and human genetics are taught within the theoretical part of medical biology. Clinically oriented, weeklong courses were introduced in 1990 in the fifth year of medical studies. Besides the regular curriculum there are many optional courses concerning human genetics e.g. "Advances in Molecular Genetics", "Genetic Counselling and Cytogenetics", "Reproductive Medicine and Reproductive Genetics" or "Transplantation and Gene Therapy" which are very popular among students.

Medical Genetics, has been officially recognized as an independent medical speciality together with the Conception of Medical Genetics (organisation and implementation of genetic services in the health care system), by the Ministry of Health in 1970.

In our country the Institute for Postgraduate Medical Education (founded in 1953) is specialised on postgraduate education and in qualifying attestations of physicians and pharmacists.

One of the departments of the Institute is the Department of Medical Genetics. Activities of this Department represent postgraduate education of specialists in medical genetics, as well as, general education in medical genetics. Conditions for getting specialization (so called "qualifying attestation") in Medical Genetics for M.Ds include: 2-3 years of work (at least) at various departments of general hospitals after having a qualifying exam in one of the general medical specializations (internal medicine, surgery, gynecology/obstetrics, pediatrics etc). This is followed by a 3-year work at Departments/Institutes of medical genetics, where the graduate attends at

least 3 training courses and has to pass practical and theoretical qualifying exam on medical genetics. Conditions for getting specialization (qualifying attestation) in methods of medical genetics, for graduates of

Conditions for getting specialization (qualifying attestation) in methods of medical genetics, for graduates of non-medical faculties (biologists, biochemists, agronomists etc), are similar.

There are two such specializations: in clinical cytogenetics and in molecular genetics, these include a 3-year work (at least) at the Departments/Institutes of medical genetics, passing at least 3 training courses and passing practical and theoretical qualifying exams. Finally, there is a similar institute for postgraduate medical education for technical assistants and/or nurses with secondary education.

Institutions and services

A network of 39 Institutes/Departments of Medical Genetics ensures genetic services for our 10,3 million Twelve of them are mostly state population. institutions providing complex genetic services (counselling, cytogenetics, molecular genetics etc), while the rest are mostly private establishments that are active in genetic counselling and/or laboratory diagnostics, which usually cooperate with other institutions. University Institutes are also responsible mainly for pregradual and partly for postgradual, education. Within academic institutions, there are Centres with national competence and quality assurance. Such reference Centres include the Cystic Fibrosis Centre, Neurogenetics Centre, Centre of Tumor Cytogenetics, Department of Epidemiology and Tumor Genetics, Institute of Prenatal and Fetal Medicine, Centre of Molecular Biology and Gene therapy, Centre of Reproductive Medicine, Institute of Hematology and Blood Transfusion etc. Broad



Prague Castle and VItava River

International activities are also performed at the "Institute of Inherited Metabolic Diseases" and the "Laboratory of Mitochondrial Diseases".

The spectrum of provided genetic services is comparable with other European countries: genetic counselling, prenatal diagnosis (including, developing preimplantation diagnosis), nationwide neonatal screening for phenylketonuria and congenital hypothyroidism, Registry of Congenital Anomalies, cytogenetics, genetic testing, methods of IVF etc.

We follow International ethical recommendations for medical genetics (several step counselling in the course of testing, written informed consent, privacy and prevention of misuse of genetic information etc). However, not all ethical guidelines are legislatively based. Internal and external quality assurance schemes are ensured in close collaboration with EMQN, ECA, and CF Thematic Network etc.

Within the genetic services network of our country we operate 67 full-time medical geneticists, more than the two thirds of them are fully qualified, while the remaining are under training. The overall geneticist per general population ratio is approximately 1: 156 000. The overall genetic university graduates – i.e. "technical assistants" (graduates of non-medical faculties) per population ratio is 1: 130 000, for laboratory workers with secondary education 1: 64 000 and, for nurses 1: 192 000.

Research

In 2004 there were 99 genetic research grant projects funded by the National Health Service, the university and basic science grant agencies. Our geneticists participate also at 8 international grant projects (HHMI, EU 5FP / 6FP projects). Research is concentrated on complex and longitudinal genetic studies and genetic care on prenatal and postnatal development of individuals and families with the risk of diseases and developmental errors. There is also close collaboration with research genetic projects of the Czech Academy of Sciences in the frame of the Czech Centre for Integrated Genomic.

Prof. Petr Goetz, MD, PhD Chairman of the Czech Society of Medical Genetics

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Welcome to the Board Members



President Leena Peltonen, M.D., Ph.D., professor

Department of Molecular Medicine and Medical Genetics, National Public Health Institute and University of Helsinki, FINLAND

Professor Leena Peltonen is among leading molecular geneticists world-wide. She is one of the pioneers in the use of genetically isolated populations in the genetics-based identification of disease genes. Using study samples from Finland her research group has characterized the genetic background of numerous monogenic diseases and more recently identified disease loci in several common diseases. She has also been pivotal in numerous international research efforts and scientific networks. She has been the

member of the international HUGO Council, the member of the UNESCO Bioethics Committee and in the Board of Directors of the American Society of Human Genetics. She has returned to Helsinki in July 2002 to lead the Finnish Center of Excellence of Disease Genetics as well as one of three recently established large multinational genomics programs of the European Community, GENOMEUTWIN. Dr. Peltonen has received several international science awards, she has produced over 370 scientific publications and 57 review articles and supervised 52 Ph.D. theses.

New Executive Board Members



President-Elect Andres Metspalu



Secretary General Helena Kääriäinen



Deputy Secretary General Thomas Meitinger



Tresurer Andrew Read

New Board Members were also elected as follows



Jacques S. Beckmann

Jacques Beckmann received his M. Sc. degree in chemistry at the Free University of Brussels, in 1968, and his Ph.D. degree (1973) in biochemistry at the Weizmann Institute of Science, Rehovot. He subsequently spent two post-doctoral stages, respectively, at Edinburgh University (on gene regulation in eukaryotes) and at the University of California San Diego (on yeast tRNA splicing). He then joined the Dept. of Plant Genetics and Breeding of the Agricultural Research Organization in Bet-Dagan, Israel. In the 1980s, together with Prof. M. Soller from the Hebrew University, they pioneered the use of marker-assisted genetic improvement in plants and animals, focusing on Quantitative Trait Loci (QTLs). His interest shifted in 1990 to human genetics with a move to Paris, where he held successively senior research positions at the CEPH, Généthon (Evry), and finally the Centre National de Génotypage (CNG, Evry), where he was Deputy-Director. During those years he

collaborated with Prof. D. Cohen, J. Weissenbach, M. Lathrop, J. Dausset and others and contributed significantly to the elaboration of genetic, physical and gene maps of the human genome. In august 2000 he joined as Full Professor the Department of Molecular Genetics at the Weizmann Institute of Science in Rehovot. Prof. Beckmann was also involved in, and led, the positional cloning of a number of disease loci. His interests have recently shifted to pharmacogenetics and, in collaboration with Prof. J. Sussman, to a

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new domain at the interface between genetic diversity and proteomics. Prof. Beckmann, who was recently appointed as head of the Service de Génétique Médicale at the University Hospital in Lausanne, has published over 200 scientific reviewed articles in molecular genetics, genetics and genomics and has served on the editorial boards of a number of scientific journals.



Dian Donnai

Professor Dian Donnai is professor of Medical Genetics in the University of Manchester and until recently was Clinical Director of the North West Regional Genetics Service. This is one of the largest and most comprehensive genetic services in the UK serving a population of 5 million. Recently the centre was successful in bids to the Department of Health and was designated as one of the two National Genetics Reference Laboratories and a Genetic Knowledge Park with >£7 million invested over 5 years. The Reference Lab and Knowledge Park research programmes include many aspects of service development and delivery for rare disorders as well as public engagement and education themes and links

with patient organisations and with industry. By background she was trained in paediatrics and genetics and within genetic services concentrates on dysmorphology. Her research has been into the mechanisms underlying birth defect syndromes and into genetic services. She is a member of several national committees involved with genetics policy and regulation and was President of the Clinical Genetics Society. She was also a member of Scientific Programme Committee of the ESHG. She is Editor of *Clinical* Dysmorphology and on the Editorial Board of Clinical Genetics.



Nurten A. Akarsu

Nurten A. Akarsu (Ankara, Turkey) MD., PhD (1960): is a medical geneticist and an Associate Professor in the Faculty of Medicine, Hacettepe University. She has been running a gene mapping facility in the Research Center of Pediatric Hematology Unit in Hacettepe University since 2000.

Her main research interests are human pedigree analysis, genetic modelling and mapping of various human disorders. Her main contributions resulted in a number of striking studies, concerning the identification of genetic localizations and/or genes of limb malformations and congenital glaucoma. She also successfully participated in collaborative linkage studies, such as preaxial polydacty, Robinow syndrome, congenital extraocular muscular fibrosis and bipolar disorders. However, for the last five years she has primarily focussed on genetics of psychiatric disorders. Her studies are well awarded from a number of both national and interjernational competetive grant foundations.

Dr. Akarsu completed her medical training at the Ankara University Medical Faculty, Ankara, Turkey. After working for four years as a general practitioner, she entered a Ph.D program in Medical Genetics and received her PhD from the Department of Medical Biology and Genetics, Ankara University Medical Faculty. During her PhD training she received her first inspiration on gene mapping studies in Erasmus University summer school on Genetic Epidemiology and Advanced Statistical Methods, Rotterdam, the Netherlands. Soon after getting her PhD she worked with Prof. Mansoor Sarfarazi in mapping and gene identification projects, focussed on particularly synpolydactyly and congenital glaucoma during over 2 years postdoctoral research period in the University of Connecticut Health Center, CT, USA.

She is an executive board member of the Association of Medical Genetics of Turkey (http://www.tamgen.org). In this society, she dedicatively works in programming medical genetics speciality education. She is also striving for a more formal recognition of the non-medical laboratory experts, molecular geneticists and cytogeneticists in Turkey. She is also a member of the American Society of Human Genetics (ASHG), European Society of Human Genetics (ESHG) and Turkish Medical Association.

Prof. Alessandra Renieri

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THE BOARD OF THE SOCIETY (May 2004-May 2005)

EXECUTIVE BOARD

President Vice-President President-Elect Secretary-general Deputy secretary-general Treasurer Prof Leena Peltonen (2008) Prof Veronica van Heyningen (2007) Dr Andres Metspalu (2009) Prof Helena Kaariainen (2007) Prof Thomas Meitinger (2007) Prof Andrew Read (2007)

BOARD MEMBERS

Term ends at the general assembly in the year shown in brackets

Dr. Nurten Akarsu Prof. Stylianos Antonarakis Prof. Jacques Beckmann Dr Alexis Brice Prof. Karen Brondum-Nielson Prof. Dian Donnai Dr Thoas Fioretos Dr Lina Florentin Prof. Thierry Frebourg Prof. Shirley Hodgson Ankara (2009) Geneva (2005) Lausanne (2009) Paris (2008) Copenhagen (2006) Manchester (2009) Lund (2007) Athens (2005) Rouen (2006) London (2005) Prof. Helena Kaarianen Prof. Nicolas Levy Prof. Milan Macek Prof. Gert Matthijs Prof. Pier Franco Pignatti Prof. Alessandra Renieri Dr Eduardo Tizzano Prof. GertJan van Ommen Dr Christos Yapijakis Helsinki (2006) Marseille (2008) Prague (2007) Leuven (2008) Verona (2008) Siena (2009) Barcelona (2006) Leiden (2006) Athens (2007)

LIAISON MEMBERS

Dr. Ségolène Aymé Prof Han Brunner Chair, PPPC Chair, SPC

Prof Jean-Jacques Cassiman IFHGS Dr Domenico Coviello Chair, Education Com.

Election of President-Elect & Board Members 2005

The Society Board intends that the President-Elect and four Board members will again be elected by postal ballot of all the members, rather than by voting in person at the Annual General Meeting, to increase participation of all members. Members of the society who wish to stand for election are requested to read the job descriptions of the duties of the President and Board Members detailed on the back of the enclosed nomination form. The Statutes of the Society state that the Board should be broadly representative of nationalities and specialties. A list of current Board members is on the Society's web site www.eshg.org (Term of office ends at the general assembly in the year shown in brackets) The ballot form showing the names of members wishing to stand for election will be sent to eligible voting members in the next mailing and a date given by which it should be returned. The result will be announced at the Annual General Meeting.

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Membership Renewal - Information for 2005

It is now time to ask you to renew your subscription for 2005. You will shortly be receiving a form to renew your membership, showing the different classes available. Please note that if you take out a full (Regular) membership you may have an electronic subscription to the Journal as well as a paper copy. The Journal will not be delivered to you from December 2004 until we receive your renewal form, so please do not delay when you receive your renewal request.

ESHG HISTORICAL INTEREST GROUP

At its June 2004 meeting, ESHG council and membership approved this, and Professor Peter Harper has agreed to act as its initial coordinator. Members of the steering group currently also include: Jaakko Ignatius (Oulu); Milan Macek (Prague); Christos Yapidzakis (Athens); Helena Kaariainen (Turku).

As a first task the group plans to collect as much information as possible about the formation and early years of ESHG itself. If you have relevant letters, photos or any other information, please be in touch with Peter Harper (HarperPS@cf.ac.uk) or any other member of the group. We hope that there will be enough material of interest to present at next year's meeting.

What does EMQN stand for?

Over 400 molecular genetic testing laboratories across Europe are aware that EMON means the European Molecular Genetics Quality Network. At this time of year they expect an EMQN package to drop through the mail box with DNA samples to genotype as an external guality assessment sometimes known (EQA, as laboratory proficiency) exercise. EMQN offers an EQA for most of the common diagnostic services for single gene conditions plus some tests for rarer disorders and most recently a technical scheme to assess the quality of DNA sequencing.

In a typical scheme laboratories genotype the EQA sample (which is given with referral information like a normal clinical case). They clinical report return their usual to an international expert panel that asks "is the genotype correct and the data interpretation guidelines?" consistent with Individual laboratories are not identifiable but they can compare their performance against others in the scheme in a general table and consider individual comments on their report from the experts.

EMQN was supported by the European Commission framework for research programme in 1998 and is looking forward to playing a full part in Eurogentest the new Framework Programme 6th network led from Leuven in Belgium. The Eurogentest programme will allow the EMQN management group to bring together all the main national molecular genetic EQA schemes in Europe in a forum to discuss the possibility of harmonising procedures and standards. In over 20 European countries EMQN links to network laboratories through national partners nominated by human genetics societies.

EQA is an important part of quality assurance. EMQN's aim is to encourage laboratories to participate regularly either through their national EQA scheme or EMQN as appropriate.

EMQN invites all molecular genetic testing laboratories to register with the network via the website (www.emqn.org) and talk to us about their EQA needs.

We look forward to hearing from you.

Rob Elles co-ordinator Simon Patton executive administrator (office@emqn.org)

European Molecular Genetics Quality Network c/o National Genetics Reference Laboratory Manchester, UK.



Figure 1: Growth in participation by laboratories in EMQN EQA schemes 1997-2004

2nd International Workshop on Genetics, Medicine & History

11th-12th May, 2005, Brno, Czech Republic

The first international workshop in Birmingham in 2003 was a great success, so we are planning a further one for May 2005. Since European Society for Human Genetics (ESHG) meets in Prague then (May 6th-10th), we have booked the new conference centre in Mendel's beautiful Abbey in Brno, for a satellite meeting.

For anyone who has not visited this birthplace of genetics, the workshop will be a splendid opportunity; we plan some cultural events as well as the opportunity to present material and have discussions with other interested people. An outline programme is attached; details will depend on contributions submitted and also on whether we are successful in obtaining funds for invited speakers. The structure of the workshop will be informal, to allow maximum discussions and interaction.

Brno can be reached by train from Prague; for those not attending the main ESHG meeting it is also an easy drive from Vienna. A midday start is planned to allow people to travel from Prague on the morning of 11th May.

Numbers will be limited to 80 (the capacity of the restored Mendel Refectory), so all those interested should be in contact early. Priority will be given to those giving a presentation (or poster). Costs are being finalised, but we aim to keep them as low as possible.

Contact details: HarperPS@Cardiff.ac.uk or Professor Peter Harper, Institute of Medical Genetics, University of Wales College of Medicine, Heath Park, Cardiff CF14 4XN, UK.



Munich 2004



Society Website