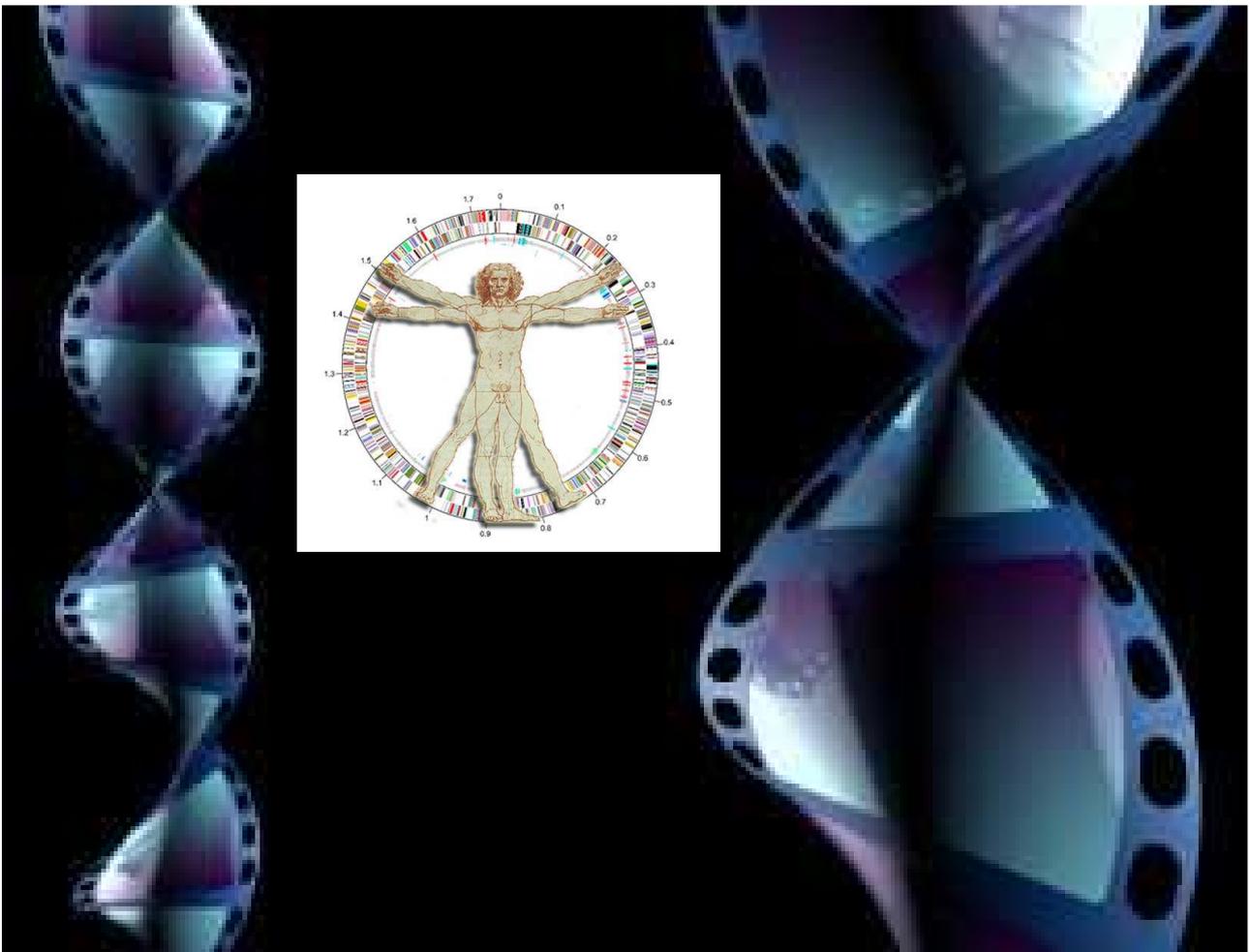


Fourth International Workshop on Genetics, Medicine and History

Principal Theme

Early History of Human Molecular Genetics

Gothenburg, Sweden, June 11-12, 2010



Organised by the Genetics and Medicine Historical Network
in conjunction with the annual conference of European Society of Human Genetics.

Funded by the European Society of Human Genetics and the Wellcome Trust, UK.

Organising Committee

- Prof. Christos Yapijakis, Department of Neurology, University of Athens Medical School, Athens, Greece (Organiser)
- Prof. Toine Pieters, Department of Medical Humanities (Metamedica), VU University Medical Center, Amsterdam, The Netherlands
- Prof. Andrew Read, Department of Human Genetics, University of Manchester, Manchester, United Kingdom
- Prof. Peter Harper, Institute of Medical Genetics, Cardiff University, Cardiff, United Kingdom

Acknowledgements

The organising committee and the Genetics and Medicine Historical Network would like to thank the Wellcome Trust and the European Society of Human Genetics for financial support.

We are also most grateful to Mirjam Uebelhör and Veit Watzal, ESHG Congress Office, Vienna Medical Academy, Dr Jerome del Picchia, of the ESHG Office Vienna and Jo Richards, of the Institute of Medical Genetics and the Wales Gene Park, Cardiff for their help and support in the organization of the workshop.

CONTENTS

INTRODUCTION	2
PROGRAMME	3
ABSTRACTS	5 - 20
LEAD AUTHORS (following order of Programme)	
Christos Yapijakis	5
Soraya de Chadarevian	6
Jan Witkowski	7
Tom Maniatis	8
Andrew Read	9
Patrick Lestienne	10
Judith Friedman	11
Ludmila Pollock	12
Dimitris Loukopoulos	13
Constantinos Deltas	14
Heike Petermann	15
Sue Povey	16
Mary-Claire King	17
Peter S Harper	18
Bengt-Olle Bengtsson	19
Walter Bodmer	20
LIST OF PARTICIPANTS	21 - 25
GENETICS AND MEDICINE HISTORICAL NETWORK	26

INTRODUCTION

Fourth International Workshop on

Early History of Human Molecular Genetics

Gothenburg, Sweden, June 11-12, 2010

The Fourth International Workshop on the Early History of Human Molecular Genetics is an evolving part of a series of workshops organized by the Genetics and Medicine Historical Network as satellite meetings of the annual conference of the European Society of Human Genetics. The underlying aim of these workshops is to bring geneticists together with historians, philosophers and other social scientists.

The first three workshops (Birmingham 2003, Brno 2005, Barcelona 2008) were very successful judging from attendance and the discussions they fuelled.

In the last half century, we have witnessed the transformation of human genetics from a marginal field of medicine into the core of contemporary molecular medicine. The concept of the biological basis of genetic disease was steadily built in the past decades allowing the simultaneous use of novel findings in clinical practice, at an unprecedented pace compared to other scientific developments. Genetics has increasingly dominated medical practice and a great amount of related hopes, hypes and fears are currently challenging mankind.

How did it all begin? The original foundations of molecular biology and the pioneering work of biomedical scientists will be under focus in this Workshop. The personal experiences of some key people who participated in the advancement of the field of human molecular genetics will be the core material. Witnesses of the explosion of genetic knowledge and its application in clinical practice will be invited to share with other Workshop participants their own memories, sentiments, struggles, failures and successes.

The international workshop intends to address and circumscribe the main historical events of the pioneering era of early human molecular genetics. The protagonists themselves will be at center stage in discussion of the main advances that shaped the field. In addition, historical, philosophical, social analysis of this scientific revolution will take place.

Professor Christos Yapijakis
(organiser)

Early History of Human Molecular Genetics

Gothenburg, Sweden, June 11-12, 2010

Friday, June 11th

9.30-10.30am Coffee and registration

10.30-12.30pm

Session 1: From basic molecular biology to human molecular genetics

Christos Yapijakis (Athens, Greece): Welcome and introductory presentation: Ancestral concepts of human genetics and molecular medicine in Epicurean philosophy.

Soraya de Chadarevian, (Los Angeles, USA): Hemoglobin and human molecular genetics.

Jan Witkowski (Cold Spring Harbor, USA): Cold Spring Harbor and the beginnings of human molecular genetics.

12.30-1.30pm Lunch

1.30-3.30pm

Session 2: From DNA analysis to human genetic disease

Tom Maniatis (New York City, USA): Recombinant DNA technology and human molecular genetics.

Andrew Read (Manchester, UK): Technology and the development of clinical molecular genetics.

Patrick Lestienne (Bordeaux, France): The mitochondrial genome: historical aspects.

3.30-4.00pm Coffee

4.00-5.30pm

Session 3: From pedigrees to the human genome

Judith Friedman (Victoria, Canada): A brief history of the theory of anticipation in hereditary disease.

Ludmila Pollock (Cold Spring Harbor, USA): Documenting the history of the Human Genome Project. An international data repository.

5.30-6.15pm

Discussion: How can we best preserve the history of human molecular genetics?

7.30pm Workshop Dinner

Saturday June 12th

9.00-10.30am

Session 4: Genetic testing and prenatal diagnosis

Dimitris Loukopoulos (Athens, Greece):

Thalassaemia: genetic testing and prenatal diagnosis

Constantinos Deltas (Nicosia, Cyprus):

Founder mutations, heterozygous advantage and thalassaemia in Cyprus.

Heike Peterman (Muenster, Germany):

The 'special' situation of genetic testing and prenatal diagnosis in Germany. The influence of history.

10.30-11.00am Coffee

11.00-1.00pm

Session 5: Gene mapping and isolation

Sue Povey (London, UK):

The Human Gene Mapping Workshops 1973-1991.

Mary-Claire King (Seattle, USA):

The revolution in understanding human molecular evolution: Work in the laboratory of Allan Wilson, 1964-1991.

Peter Harper (Cardiff, UK):

Historical lessons from Huntington's disease.

1.00-1.45pm Lunch

1.45-3.30pm

Session 6: From human genetics to genetic medicine

Bengt-Olle Bengtsson (Lund, Sweden):

Film from the 1956 First International Human Genetics Congress, Copenhagen.

Walter Bodmer (Oxford, UK):

The beginnings of clinical cancer genetics.

Planning of future workshops, and conclusion of Workshop.

Author(s): **CHRISTOS YAPIJAKIS**

**Lead author's
contact details:** University of Athens
Medical School
74 Vas. Sofias
11528
Athens, Greece

Email: cyapijakis_ua_gr@yahoo.com

Title: **ANCESTRAL CONCEPTS OF HUMAN GENETICS AND
MOLECULAR MEDICINE IN EPICUREAN
PHILOSOPHY**

Abstract:

Human genetics and molecular medicine are scientific fields that evolved during the last century. Nevertheless, less known is the fact that over two millennia ago mankind had grasped the concepts of the molecular basis of life in health and disease, as well as the basic laws of heredity. It was the influence of Epicurean philosophy that led some exceptional people of the ancient Hellenistic and Roman eras in understanding human nature and proposing some notions that were discovered as facts only recently. The founder of this humanistic philosophy was Epicurus of Athens (341-270 BC) who combined the atomic physics of Democritus with the naturalistic ethics of Aristotle. He maintained that eternal atoms continuously combine by necessity and chance forming worlds, mountains and evolving living organisms. He proposed that any given interaction of atoms within a molecular structure confers new qualities to the molecule. Unlike Aristotle who believed that only males contributed in heredity, Epicurus suggested that males and females equally contributed hereditary material to their progeny. According to the Roman Lucretius (95-45 BC), Epicurus described dominant, recessive and co-dominant types of inheritance. The Epicurean Greek physician Asclepiades (124-40 BC) suggested that diseases are caused by alteration of form or position of a patient's molecules. He introduced the acute and chronic distinction of disorders, and the psychological support of patients. One of his followers, the Greek physician Soranus (1st/2nd century AD), known as the father of gynecology and pediatrics, described congenital malformations as well as hereditary conditions such as mental disorders.

Author(s): **SORAYA DE CHADAREVIAN**

**Lead author's
contact details:** University of California Los Angeles
Department of History
6265 Bunche Hall
90095-1473
Los Angeles
USA

Email: chadarevian@history.ucla.edu

Title: **HEMOGLOBIN AND HUMAN MOLECULAR
GENETICS**

Abstract:

Considering the title of the workshop one main question springs to mind: was it human genetics that at a certain point in history turned molecular or was it molecular genetics that eventually expanded from the study of viruses and bacteria to include humans? Were both processes at play? Or is there a completely different story to tell? How do practitioners present the history of their field and what is at stake? I will consider these questions while focusing on hemoglobin as a versatile tool in the early history of human molecular genetics and placing it at the intersection of anthropological, medical, biological and evolutionary concerns.

Author(s): **JAN WITKOWSKI**

Lead author's contact details: Cold Spring Harbor Laboratory
Banbury Center
1 Bungtown Road
NY 11724
Cold Spring Harbor
USA

Email: witkowsk@cshl.edu

Title: **COLD SPRING HARBOR LABORATORY AND THE BEGINNINGS OF HUMAN MOLECULAR GENETICS**

Abstract:

Cold Spring Harbor Laboratory made significant contributions to human molecular genetics and the human genome project. But these contributions were made not through the Laboratory's research, but through its meetings and courses programs. The latter began in 1947 with the Phage course, when Salvador Luria and Max Delbruck taught the techniques that were at the cutting edge of genetics research of that time. Genomics-related courses began with the "Making and Using DNA Microarrays" course beginning in 1999, while the 2010, courses include "Genetics of Complex Human Diseases", "Computational & Comparative Genomics" and "Integrative Statistical Analysis of Genome Scale Data". However, in my presentation, I will concentrate on the impact of the "Genome Mapping and Sequencing" meetings which began in 1988. These meetings provided not only a forum for the exchange of data and ideas, they also helped promote a sense of community among researchers working on the genomes of diverse organisms, pursuing technological developments or studying biological problems. The meetings span 23 years and provide a series of snapshots of how the field of human molecular genetics and genomics has changed in that period.

Author(s): **TOM MANIATIS**

Lead author's contact details: Columbia University College of Physicians and Surgeons
Biochemistry and Molecular Biophysics
701 West 168th Street
10032
New York
USA

Email: tm2472@columbia.edu

Title: **RECOMBINANT DNA TECHNOLOGY AND HUMAN MOLECULAR GENETICS**

Abstract:

In the mid 1970's scientific approaches and reagents used to study bacterial and phage genetics were employed to develop DNA cloning methods. Advances in bacterial and phage genetics, restriction/modification systems, plasmid biology and phage life cycles during the previous 25 years provided the tools and concepts necessary for these advances. Recombinant DNA methods were used to isolate, sequence and study specific eukaryotic mRNAs (cDNA cloning) and genes (genomic DNA cloning), which led to the identification of mutations that cause human genetic diseases. Ultimately, these tools in conjunction with advances in DNA sequencing technology led to the determination of the sequence of the human genome. Among the earliest advances were the cloning and determination of the linkage arrangement of human alpha and beta globin genes, and the identification of deletion and single base mutations that cause the genetic disease thalassemia. The history of the development of recombinant DNA methods will be discussed, with an emphasis on the transition from prokaryotic molecular genetics to major advances in human genetics.

Author(s): **ANDREW READ**

Lead author's contact details: St Mary's Hospital
Dept of Genetic Medicine
Hathersage Road
M13 0JH
Manchester
UK

Email: Andrew.Read@man.ac.uk

Title: **TECHNOLOGY AND THE DEVELOPMENT OF CLINICAL MOLECULAR GENETICS**

Abstract:

I will review the development of molecular genetics services in the UK during the 1980s and early 1990s, from the struggle to establish pioneer services to the acceptance of a DNA laboratory as an essential part of a clinical genetics service. The early applications of molecular genetics depended on restriction fragment length polymorphisms scored by Southern blotting. For most diseases the initial work was based on gene tracking using RFLPs loosely linked to unknown genes. Useful predictions depended on family studies. This mandated a very close partnership between scientists and clinicians, and naturally located the work in clinical genetics centres where family-based approaches were well developed.

As the major disease genes were cloned and sequencing became more routine, work for most diseases moved from gene tracking to direct mutation testing. This reduced the need for family studies and loosened the link between the referring clinician and the laboratory. PCR simplified procedures and allowed individual laboratories to tackle a wider range of diseases. It also removed the dependence on probes, release of which had to be negotiated with the originator. These developments could have paved the way for large-scale commercial laboratories to take over the bulk of the work but, interestingly, in the UK this never happened.

Author(s): **PATRICK LESTIENNE**

**Lead author's
contact details:** U 889 INSERM
Université Bordeaux 2
146 rue Léo Saignat
33076 Bordeaux. France

Email: Patrick.Lestienne@inserm.fr

Title: **THE MITOCHONDRIAL GENOME: HISTORICAL
ASPECTS**

Abstract:

The cell's energy is mostly provided by mitochondria. Several experiments described here, showed that their DNA encode a part of the respiratory chain complexes. Their finding began with yeast mutants unable to achieve respiration. Studies on cells from vertebrate revealed their presence in the early 1960 by S. and M. Nass, and in mitochondria on yeast by G. Schatz. The finding of restriction enzymes enabled to discover their maternal inheritance, to reveal its extreme polymorphism and to trace human evolution since about 200,000 years. During the early 1980, the team headed by F. Sanger sequenced the human mtDNA from placenta, and some of his associates found a mitochondrial specific genetic code. The endosymbiotic theory of evolution could be sustained by the analysis of the mtDNA from lower organisms, whose high mtDNA gene content have now migrated towards the nucleus in vertebrates. This reductive evolution enabled a higher copy number of mtDNA per mitochondria, which, depending on their origin, may be as high as one thousand per cell. This feature also enabled to sequence exhumed remains.

More recently, mtDNA alterations were disclosed to cause diseases, among them Kearns-Sayre syndrome of which I was involved in.

Author(s): **JUDITH FRIEDMAN**

Lead author's contact details: 906 10160-115 Street
T5K 1T6
Edmonton, Alberta
Canada

Email: jfriedma@uvic.ca

Title: **A BRIEF HISTORY OF THE THEORY OF ANTICIPATION IN HEREDITARY DISEASE**

Abstract:

Mid-nineteenth century French alienists (Lucas 1874-50, Morel 1857, 1860) recognized that certain families appeared to be degenerating over time. Nervous temperament in one generation could lead to epilepsy in the next until within four generations sterility and early death ended the family line. This notion of degenerate or progressive heredity was taken up by many important figures in psychiatry (Maudsley 1879, Clouston 1883, Tredgold 1908), medicine (Nordau 1895, Zeigler 1905), and heredity (Darwin 1859, 1868).

The British ophthalmologist Edward Nettleship noticed that some diseases were not inherited according to the patterns of heredity set forth by Galton, Weismann, or Mendel, and coined the term 'anticipation' to describe diseases which appeared earlier, and often in a more severe state, in succeeding generations (Nettleship 1905, 1909, 1910). Anticipation was embraced by some (Mott 1911, 1912) but rejected by others (Pearson 1912) and it was hotly contested throughout the first half of the 20th century.

The British psychiatrist and geneticist Lionel Penrose argued that the appearance of anticipation was caused by experimental artefacts and statistical fallacies and that any variation in age of onset and severity of disease in such diseases was caused by allelic modification (Penrose 1948). These ideas were swiftly taken up by human and medical geneticists in the post-war period and anticipation all but disappeared from the literature.

Anticipation was not considered seriously again until the mid-1980s (Höweler 1986, 1989) but was not widely accepted until after the discovery of its underlying genetic mechanism, expanding trinucleotide repeats, in 1991.

Author(s): **LUDMILA POLLOCK**

Lead author's contact details: Cold Spring Harbor Laboratory Library & Archives
One Bungtown Road
11724
Cold Spring Harbor, NY, USA

Email: pollock@cshl.edu

Title: **HUMAN GENOME PROJECT - INTERNATIONAL DATA REPOSITORY**

Abstract

The Human Genome Project was one of the great scientific enterprises of the 20th century. The materials of the HGP, which will bring great benefits to humanity, need to be exhaustively documented. A proposal to create an International Data Repository of such materials was presented at a meeting at Cold Spring Harbor Laboratory and discussed with an international group of scientists, historians, librarians and archivists.

Some of the technical roots and early years of the HGP have been described in books, scientific journals, and periodicals. With some exceptions, these have been popular rather than scholarly accounts. While some key documents have been archived, most have not. Researchers must find original documents which are scattered throughout the world in research institutes, government agencies, foundations, companies and scientists' own personal collections. Documents are disappearing each year as scientists and government officials retire, companies change ownership or leadership, and the world moves on. Accounts of major meetings, lab notes, communication through letters and e-mails, photos and videos, and digital documents still remain unavailable to researchers and the public because they have not been organized, cataloged or digitized.

The international Advisory Committee in support of this project was formed and CSHL is the lead institution. We strongly believe in the importance of creating a comprehensive repository for dissemination of the history of the HGP, regardless of the physical or electronic location of these materials, printed or digital born. The project's mission, goals, scope, timeline, and a model for the project will be presented.

Author(s): **DIMITRIS LOUKOPOULOS**

Lead author's contact details: Biomedical Research Foundation
Academy Of Athens Department, Hematology
4, Soranou Ephessiou
11527
Athens, Greece

Email: dloukop@bioacademy.gr

Title: **THALASSEMIA. GENETIC TESTING AND
PRENATAL DIAGNOSIS**

Abstract:

Thalassemia major is a severe, long-lasting, costly, and virtually incurable condition. Its' impact, both social and financial, is enormous in the Mediterranean and other countries further East, where it occurs in high frequency; it is now expanding across the North-Western world through economic immigration. As yet, there is no radical treatment; even worse, the ever increasing number of patients will inevitably block delivery of optimal treatment to those who actually survive. For the time being, effective prevention through carrier identification and prenatal diagnosis is the only possible solution and is already successfully applied in several countries, while others follow this example. The process has developed through various phases and required solution of a number of organizational, legal, financial and technical problems; the availability of prenatal diagnosis proved a major factor for its acceptance at large, because it allowed couples at risk to acquire non-thalassemic children. In the seventies, prenatal diagnosis required acquisition of fetal blood (through a fetoscope at the 20th week of pregnancy) and globin biosynthesis aiming to show that the fetus synthesized an amount of beta-chains excluding thalassemia major [simple when the risk was beta-0/beta-0, but difficult to interpret when the risk involved beta (+) thalassemic genes]. Later on (in the eighties) prenatal diagnosis was offered on DNA obtained at the 10th week of pregnancy by trophoblast biopsy (and rarely amniocytes) by an array of molecular techniques, initially by RFLPs across the beta-globin cluster and recently by direct PCR identification of the defect. Pre-implantation diagnosis is now emerging as an alternative possibility.

Author(s): **CONSTANTINOS DELTAS**

**Lead author's
contact details:** University of Cyprus
 Biological Sciences
 Aretousas 1
 2045
 Strovolos, Cyprus

Email: Deltas@ucy.ac.cy

Title: **FOUNDER MUTATIONS, HETEROZYGOUS
ADVANTAGE AND THALASSAEMIA IN CYPRUS**

Abstract:

The Cyprus population used to compose a limited gene-pool for many thousands of years, something that along with probable genetic drifts fostered multiple founder phenomena to show up. Prime examples are the following:

- 1) Only five β -globin defects account for >98% of β -thalassaemia chromosomes (three account for >90%). β -thal mutations used to have a frequency of 1/5-1/7 until recently and is presently estimated around 1/8 among Greek-Cypriots.
- 2) The high frequency of the F508del mutation of the CFTR gene (responsible for Cystic Fibrosis), in a small village at south-east of Nicosia, where 1/14 is a carrier. It has come down through history that most probably a distant ancestor, presumably since the Frankish period was the founder of a community on a feud of that region during the 16th century. These people had survived the massacre that had followed the occupation of Famagusta by the Ottomans in 1571.
- 3) The nearly 20% frequency of mutation F479L in the MEFV gene, among patients who inherit the Familial Mediterranean Fever. This same mutation and the accompanied common haplotype is identical to the haplotype bearing the mutation among Armenians, who are known to have migrated to Cyprus in refuge from persecutions by the Seljuks Turks during the 12th century.

Additional mutation founders will be discussed in view of the recent application of molecular approaches in Cyprus and a special attention will be given on the issue of Malaria and thalassaemia in Cyprus, in the light of discoveries regarding porotic hyperostosis in ancient skeletal remains.

Author(s): **HEIKE PETERMANN**

Lead author's contact details: WWU Muenster
Institute for Ethics, History and Theory of Medicine
Von-Esmarch-Str. 62
48149
Muenster, Germany

Email: heike.petermann@uni-muenster.de

Title: **THE 'SPECIAL' SITUATION OF GENETIC TESTING AND PRENATAL DIAGNOSIS IN GERMANY: REMARKS ON THE INFLUENCE OF HISTORY**

Abstract:

The situation in Germany is quite different to that in Europe and the USA. The legal regulation on prenatal diagnosis and genetic testing are 'special'. In law and in fact this is determined by the act on abortion (§§ 218, 1976, then 1995), on protection of embryos (Embryonenschutzgesetz, 1990, then 2001) and on genetic testing (Gendiagnostikgesetz, 2009). Those regulations are stricter than in other countries. F.e. preimplantation diagnosis (PID) is forbidden.

What are the reasons for those strict legal regulations?

Since 1945 we have in Germany a lively debate on questions of genetic testing and prenatal diagnosis. This is dominated on the one side by the organisation of disabled persons (Aktion Mensch) and on the other hand by persons claiming their right of self-determination and also freedom of research. Till today this discussion is going on and the arguments of both sides are reflected in a heterogeneous legislation.

Some of the reasons are

- The reference to the Third Reich till today works as discussion stopper
- Personal continuities in science and politics
- Consequences of KZ medical experimentation in the 1940ies
- Necessity to break with Third Reich
- Increasing Focus on ethical implications of diagnosis and therapy

The situation in Germany is mainly determined by historical experiences and reasons. But there is decreasing knowledge about the situation in the Third Reich (f.e. eugenics and euthanasia). Therefore it is hardly possible to discuss without any reservations about genetic testing and prenatal diagnosis in Germany.

Author(s): **SUE POVEY, IAN W CRAIG**

Lead author's contact details: UCL
Genetics, Evolution and Environment
Wolfson House 4 Stephenson Way
NW1 2HE
London
UK

Email: s.povey@ucl.ac.uk

Title: **THE HUMAN GENE MAPPING WORKSHOPS
1973-1991**

Abstract:

Although the theoretical basis of human genetic mapping was well developed by 1955 and a few linkage groups had been discovered the first placement of a human gene on an autosome was made only in 1968. From the first Human Gene Mapping workshop in New Haven to HGM11 in London enthusiasts came together at regular intervals to try to fill the vast uncharted stretches of human chromosomes with genes and other genetic markers. In a few frantic days each time all available information was integrated and each participant took home proudly the latest often hand-drawn map of every chromosome, final and more considered reports emerging about a year later. Friendships and collaborations emerged and persist to this day. The talk will outline the story of these meetings and their contribution to human genetics.

Author(s):

MARY-CLAIRE KING

**Lead authors
contact details:**

Dept of Medicine (Medical Genetics) and Genome
Sciences
Health Sciences Room K-160
University of Washington School of Medicine
Seattle WA 98195-7720

Email:

mcking@u.washington.edu

Title:

**THE REVOLUTION IN UNDERSTANDING
HUMAN MOLECULAR EVOLUTION: WORK IN
THE LABORATORY OF ALLAN WILSON, 1964-
1991.**

Abstract

Author(s): **PETER S. HARPER**

**Lead author's
contact details:** Cardiff University
Medical Genetics
Heath Park
CF14 4XN
Cardiff
Wales

Email: HarperPS@cf.ac.uk

Title: **HISTORICAL LESSONS FROM HUNTINGTON'S
DISEASE**

Abstract:

Huntington's disease has provided a paradigm for human inherited disease since its first recognition. Not only was it a striking early example of human autosomal dominant mendelian inheritance and the anomalies in this needing to be explained, but it also figured strongly in the history of eugenics. The process of mapping and subsequently of isolating the responsible gene helped to set patterns of research for the positional cloning of other major disease genes, while resolution of the molecular mechanisms involved contributed to solving the problem of anticipation and the anomalous inheritance patterns observed many years before. The disorder has both illustrated and in part resolved practical and ethical issues in late onset disease prediction, while currently it shows how our new molecular understanding of genetic disorders can be applied to the search for effective therapeutic measures.

Author(s): **BENGT-O BENGTSSON**

Lead author's contact details: Genetics, Department of Biology,
Lund University,
Sweden

Email: Bengt_Olle.Bengtsson@cob.lu.se

Title: **THE 1948 CONGRESS OF GENETICS IN
SWEDEN: PEOPLE AND POLITICS**

Abstract:

In July 1948, the Eighth International Congress of Genetics was held in Sweden. After an ambitious pre-congress tour to plant-breeding institutes in southern Sweden, the main program was held in Stockholm with a brief visit to Uppsala.

During all outdoor activities Nils Nybom, then a young student and later professor in horticultural genetics, filmed the participants with his little hand-held camera. From this material he produced an edited film of remarkable quality, which today is owned by the Mendelian Society in Lund. The film is a unique document of an important event, showing prominent (and forgotten) colleagues of genetics debating, conversing and exchanging pleasantries, for example Tschermak, Fisher, Haldane, Auerbach, Dobzhansky, Levan, Essen-Möller and many more. All headed by H. J. Muller, who acted as the congress' president.

The film has been digitalized and gently re-edited. In its present version it lasts 30 minutes and in my commentary to it I will discuss the treatment of German scientists, the starting fight between geneticists and Lysenkoists, and the way human genetics was presented during the congress.

Author(s): **WALTER BODMER**

Lead author's contact details: Cancer & Immunogenetics Laboratory
Weatherall Institute of Molecular Medicine
John Radcliffe Hospital
Oxford OX3 9DS

Email: walter.bodmer@hertford.ox.ac.uk

Title: **THE BEGINNINGS OF CANCER MOLECULAR GENETICS**

Abstract:

The origins of cancer molecular genetics come from the idea that cancer is an evolutionary somatic genetic process. The ground work for this was laid in the early years of the 20th century, but it was not until after the development of reliable cytogenetics, and recombinant DNA technology in the 1970s that the first specific evidence for genetic changes at the molecular level became available. There were at least three major directions of development. The first was the discovery of dominant oncogenes by Varmus and Bishop, and the application of this to human tumours. The second was the recognition by Janet Rowley that the Philadelphia chromosome found in CML was a 9/22 translocation, and then the identification of the bcr/abl combination at the break point, following the mapping of the abl oncogene to chromosome 9. Finally came the application of DNA polymorphisms to gene mapping, both at the somatic level by LOH analysis, and through positional cloning of the genes for Retinoblastoma, Familial Adenomatous Polyposis (FAP) and other clearly inherited cancer family syndromes. Following Knudsen's ideas on the relation between germ line and somatic changes, the identification of the familial cancer genes, especially for colorectal cancer, led to major advances in the characterisation of somatic genetic changes in sporadic cancers. At the same time, the ability to predict those at risk for, for example, FAP led to the need to establish cancer family clinics and to cancer genetics as a major part of adult clinical human genetics."

LIST OF PARTICIPANTS

Dvora Abeliovich

Dept. Human Genetics, Hadassah Hebrew University Hospital, Ein Kerem,
Jerusalem, 91120, Israel
dvoraha@cc.huji.ac.il

Egbert Bakker

LUMC, Clinical Genetics, Albinusdreef 2, 2300 RC, Leiden, The Netherlands
e.bakker@lumc.nl

Bengt O. Bengtsson

Genetics, Department of Biology, Lund University, Sweden
bengt_olle.bengtsson@cob.lu.se

Walter Bodmer

Cancer & Immunogenetics Laboratory, Weatherall Institute of Molecular Medicine
John Radcliffe Hospital, Oxford OX3 9DS
walter.bodmer@hertford.ox.ac.uk

Angus Clarke

Institute of Medical Genetics, Cardiff University, Heath Park, Cardiff, CF14 4XN.
United Kingdom
ClarkeAJ@cardiff.ac.uk

Soraya de Chadarevian

University of California Los Angeles, Department of History, 6265 Bunche Hall,
90095-1473, Los Angeles, USA
chadarevian@history.ucla.edu

Constantinos Deltas

University of Cyprus, Biological Sciences, Aretousas 1, 2045, Strovolos, Cyprus
Deltas@ucy.ac.cy

Michael Dougherty

The American Society of Human Genetics, Education, 9650 Rockville Pike, 20814
Bethesda, United States
mdougherty@ashg.org

Nils Fietje

The Wellcome Trust, Medical Humanities, Euston Road, NW1 2BE, London
n.fietje@wellcome.ac.uk

Judith Friedman

906 10160-115 Street, T5K 1T6, Edmonton, Alberta, Canada
jfriedma@uvic.ca

Judith Hall

University of British Columbia/BC Children's Hospital, Medical Genetics & Pediatrics,
4480 Oak Street, Room L408, V6H 3V4, Vancouver, BC, Canada
jhall@cw.bc.ca

Peter Harper

Institute of Medical Genetics, Cardiff University, Heath Park, Cardiff, CF14 4XN.
United Kingdom
HarperPS@cardiff.ac.uk

Jennifer Haynes

Wellcome Trust, Library, 183 Euston Road, NW1 2BE, London
j.haynes@wellcome.ac.uk

Mary-Claire King

Dept of Medicine (Medical Genetics) and Genome Sciences, Health Sciences Room K-160, University of Washington School of Medicine, Seattle WA 98195-7720
mcking@u.washington.edu

Patrick Lestienne

University of Bordeaux 2, Unit 889 INSERM, 146 Rue Léo saignat, 33076, Bordeaux, France.
Patrick.Lestienne@inserm.fr

Dimitris Loukopoulos

Biomedical Research Foundation, Academy of Athens Department, Hematology 4, Soranou Ephessiou, 11527 Athens, Greece
dloukop@bioacademy.gr

Tom Maniatis

Columbia University College of Physicians and Surgeons, Biochemistry and Molecular Biophysics, 701 West 168th Street, 10032 New York, USA
tm2472@columbia.edu

Søren Nørby

Rolighedsvej 6, Frederiksberg C, DK-1958, Denmark
SIRN@dadlnet.dk

Tayfun Ozcelik

Bilkent University, Molecular Biology & Genetics, Bilkent, 06800, Ankara, Turkey
tozcelik@bilkent.edu.tr

Heike Petermann

WWU Muenster, Institute for Ethics, History and Theory of Medicine, Von-Esmarch-Str. 62, 48149 Muenster, Germany
heike.petermann@uni-muenster.de

Toine Pieters

Department of Metamedica, VU Medical Center, van der Boechorstr. 7, Postbox 7057 1007 MB Amsterdam, Netherlands

a.pieters@bio.vu.nl

Mila Pollock

Cold Spring Harbor Laboratory, 1 Bungtown Road, Cold Spring Harbor, NY 11724, United States

pollock@csih.edu

Sue Povey

UCL, Genetics, Evolution and Environment, Wolfson House 4 Stephenson Way, NW1 2HE, London, UK

s.povey@ucl.ac.uk

Andrew Read

St Mary's Hospital, Dept of Genetic Medicine, Hathersage Road, M13 0JH, Manchester, UK

Andrew.Read@man.ac.uk

Alan Rushton

Hunterdon Medical Center, Pediatrics, 2100 Wescott Drive, 08822, Flemington United States

arrdoc@aol.com

Karina Sasin

GENE Association, oś. Zwycięstwa, Poznań, Poland

karinasasin@gmail.com

Marianne Schwartz

Rolighedsvej 6, DK-1958, Frederiksberg C, Denmark

janne_schwartz@yahoo.dk

Liz Shaw

Wellcome Trust, Medical Humanities, Euston Road, NW1 2BE, London
e.shaw@wellcome.ac.uk

Alda Sousa

Department of Population Studies, ICBAS , University of Porto, Lg Abel Salazar, 2
4099-003, Porto, Portugal
absousa@icbas.up.pt

Carina Wallgren-Petterson

Department of Medical Genetics, Folkhälsan and University of Helsinki, P.O. Box
211, Helsinki, 00251, Finland
carina.wallgren@helsinki.fi

Jan Witkowski

Cold Spring Harbor Laboratory, Banbury Center, 1 Bungtown Road, NY 11724, Cold
Spring Harbor, USA
witkowsk@cshl.edu

Christos Yapijakis

Department of Neurology, University of Athens Medical School, Vas Sophias 74,
Athens, 11528, Greece
cyapijakis_ua_gr@yahoo.com

Lung-Chih Yu

National Taiwan University, P.O. Box 23-106, 106 Taipei, Republic of China
yulc@ntu.edu.tw

Genetics and Medicine Historical Network

This international network was established in 2002 to encourage historical activities relating to genetics and medicine, to promote the preservation of records and other material and to link those interested, whether geneticists, historians or others. Current activities include:

- A website www.genmedhist.org
- An electronic newsletter to keep members in touch
- Regular workshops and historical sessions as broader meetings
- The *Human Genetics Historical Library* of books in the field
- Archiving of scientific records of key workers in human and medical genetics
- An oral history programme of interviews with retired and older human geneticists
- A series of peer reviewed historical articles in the journal *Human Genetics*
- Further details can be obtained from Jo Richards, richardsje2@cf.ac.uk