GenMedHist: Newsletter No.18, November 2014

Bulletin of the Genetics and Medicine Historical Network

Contents

Introduction

The publication of Newsletter 18 of *GenMedHist: Bulletin of the Genetics and Medical Historical Network* finds plans for the Sixth International Workshop on Genetics, Medicine, and History well underway. Dr. Heike Petermann and Dr. Sam Maddra, two members of the organizing committee, would like to invite Network members to join us in Glasgow for the workshop which will be held 5-6 June 2015 as a satellite meeting just prior to the joint meeting of the European Society of Human and the British Society for Genetic Medicine. The topics for what promises to be an exciting workshop are "The Evolution of Human Gene Mapping" and the "Oral History of Human Genetics in Europe." Proposals for presentations at the meeting are welcome. Please see the Call for Abstracts/Papers of the Workshop for further information.

Several projects on the history of genetics and medicine are highlighted in the current issue. Dr. Peter Harper's article takes people back to the origins and early years of the Genetics and Medicine Historical Network. Karen Pierce, the Cataloguing Librarian for the Human Genetics Historical Library located at Cardiff University, has news of new donations for the collection and a grant generously made by European Society of Human Genetics to help with cataloguing materials. She has also helpfully included a tutorial to aid those interested in accessing this unique collection for research purposes. Prof. Dr. med. Joerg Schmidtke announcement that the Institute of Human Genetics of the Hannover Medical School is opening its historical library to the public by appointment is exciting news for those researching medical genetics in Germany.

Michal Simunek updates readers on historical projects on the history of medical genetics and genetics in Czechoslovakia and the opening of a new exhibition on the history of genetics in Brno. He would also like to invite readers to the Mendel Forum to be held 6-8 March 2015 in Brno. Both observers and presenters are welcome. For further information, please see the Invitation and Call for Papers for the 2015 Mendel Forum.

Long-time readers of the newsletter may recall the announcement that in 2006 Dr. Arno Motulsky had donated his papers to the American Philosophical Society Library which contains collections of papers donated by several prominent geneticists (*GMHN Newsletter* No. 10 and No. 11). Charles Greifenstein of the APS has written an account Dr. Motulsky's life and career and highlights some of the interesting materials that can found within this collection.

It is with sadness that we mark the passing of André Boué (1925-2012), Joëlle Boué (1926-2014), and Lore Zech (1923-2013). Dr. Simone Gilgenkrantz has kindly shared an account of the Boués' life and research with readers of the *Bulletin*. An article on Lore Zech appears among the recent publications.

News of new research in the field comes from conference reviews written by Marion Schmidt and Mila Pollock. For the first time, the *Bulletin* includes a list of new publications that might interest readers along with the monographs that appear 'On the bookshelf.'

News of ongoing projects or upcoming events that Network members would like to see included in our next newsletter may be sent to me at jfriedma@uvic.ca. Similar news may be shared more immediately by posting a notice on our affiliated listserv genmedhist-l@jiscmail.ac.uk.

Judith Friedman

Welcome to Glasgow

Set against a backdrop of outstanding Victorian architecture, Glasgow is one of Europe's most exciting destinations, combining the vibrancy and sophistication of a great international city with the friendliness of its people and a sense of style which is second to none. It is the largest city in Scotland and was recently voted one of the top ten cities in the world.

Glasgow grew from a small rural settlement on the River Clyde to become one of the largest seaports in Britain. Expanding from the medieval bishopric and royal burgh, and the later establishment of the University of Glasgow in the 15th century, it became a major centre of the Scottish Enlightenment in the 18th century. From the 18th century the city also grew as one of Great Britain's main hubs of transatlantic trade with North America and the West Indies.

Today Glasgow is a world renowned cultural capital, with an exceptional portfolio of free to enter museums and art galleries, including such gems as the Kelvingrove Art Gallery & Museum, the Burrell Collection and the Museum of Modern Art along with the University of Glasgow's own Hunterian Museum and Art Gallery (Scotland's oldest public museum). Moreover the city is home to the architectural treasures of Charles Rennie Mackintosh (1868-1928), whose fame draws visitors from far and wide, and his distinctive style can be appreciated in many of the city's buildings and museums.

As you might expect from a city of Glasgow's stature, international bands, singers and comedians regularly perform in the various venues dotted around the city. Theatregoers also have an abundance of choice with the Scottish National Orchestra, Ballet and Opera companies all being based in the city and an array of theatres such as the Kings and the world famous Pavilion. At night the city really comes alive and the restaurants and cafes encompass the entire spectrum of great food and drink, whether your choice is traditional Scottish fayre, European, Asian or the very latest in fusion cuisine.

Glasgow is twinned with eight different cities around the world, including Havana, Bethlehem, and Nuremberg, the host city of the last workshop on the history of human genetics.

The University of Glasgow

Founded in 1451, Glasgow is the fourth-oldest university in the English-speaking world. Over the last five centuries and more it has constantly worked to push the boundaries of what's possible, fostering the talents of seven Nobel laureates.

It includes among its alumni, the father of economics Adam Smith (1723-1790), Scotland's architect of devolution Donald Dewar (1937-2000) and renowned physicist and engineer William Thomson, Lord Kelvin or

1st Baron Kelvin (1824-1907). In 1933 the University welcomed theoretical physicist and philosopher of science Albert Einstein (1879-1955) to give a lecture on the origins of the general theory of relativity. Scotland's first female medical graduates completed their degrees at Glasgow in 1894. Also the world's first ultrasound images of a foetus were published by physician and Glasgow Professor of Midwifery Ian Donald (1910-1987) in 1958.

Today, the University of Glasgow is a member of the prestigious Russell Group of leading UK research-led universities and has an annual research income of more than £181,000,000.

The most influential geneticists of Glasgow University were:

Guido Pontecorvo (1907-1999), who liked to be known by his nickname, Ponte, was the University's first Professor of Genetics, 1955 to 1968, and has been described as "one of the founding fathers of modern genetics". He endowed prizes and scholarships for students at the University and the Genetics Building was named for him in 1994.

James (Jim) Harrison Renwick (1926-1994) was a Titular Professor of Genetics at the University from 1967-1968 and made a fundamental contribution to modern genetics. For a period of nearly 20 years up to the early 1970s, he pioneered the use of genetic markers to map disease genes on human chromosomes, seeing this field develop from its infancy.

Malcolm Andrew Ferguson-Smith (b. 1931) is an award winning graduate of the University who was Professor of Medical Genetics from 1973 until 1987 and a celebrated geneticist who contributed significantly to gene mapping. In the 1970s he established a Regional Genetics Service for the West of Scotland which provided the opportunity for continuing clinical genetics research and the development of prenatal diagnosis and screening for Down Syndrome and Spina Bifida.
(Source: http://www.gla.ac.uk/services/archives/collections/university/medicine/genetics/)

Sam Maddra (University of Glasgow), Heike Petermann (University of Münster)

Images reproduced Courtesy of the University of Glasgow

Call for Abstracts/Papers

Sixth International Workshop on the History of Human Genetics Glasgow, UK (Scotland), June 5-6, 2015

Principal Themes: 'Human Gene Mapping' and 'Oral History of Human Genetics'

Call for Abstracts/Papers:
We would like to invite proposals for the Sixth International Workshop on the History of Human Genetics which will take place in Glasgow, UK (Scotland), 5-6 June 2015. The topics for this workshop are 'Human Gene Mapping' and the 'Oral History of Human Genetics'.

Proposals for presentations (**250 words maximum**) should be submitted via the ESHG homepage: www.eshg.org. The deadline for proposals is **January 9, 2015**.

The History of Human Gene Mapping

Gene mapping in Drosophila began over a century ago, but human gene mapping is more recent, beginning with the linkage between haemophilia and colour-blindness on the X chromosome by Bell and Haldane in 1937 and the first autosomal linkage by Jan Mohr in Copenhagen in 1951.

In 1973 the first Workshop on Human Gene Mapping took place at Yale University, organised by Francis Hugh Ruddle (1929-2013). He was inspired during his post-doctoral training in 1960 and 1961 by the work of Guido Pontecorvo (1907-1999) at Glasgow, who had demonstrated that genes could be mapped in somatic cells.

As a result of this and subsequent workshops around 2000 genes were assigned to specific chromosomal locations before the Human Genome Project was launched in 1989. The data were stored in a publicly available database allowing the sharing of human genome information.

Guido Pontecorvo with Frank Ruddle and Francis Crick in June 1978. University of Glasgow Archive Services, Papers of Guido Pellegrino Arrigo Pontecorvo, GB248 UGC 198/10/1/1/26

How did it all begin? And what was the evolution from 1973 up to the start of the Human Genome Project? The focus of this workshop will be on the pioneering work involved in human gene mapping itself, but also on the biography of the involved scientists. Those associated with this work from different countries are invited to share their experiences and memories with the participants. Presentations on the historical, philosophical, social analysis of aspects of the scientific revolution of gene mapping are also welcome.

The Oral History of Human Genetics in Europe

Oral histories provide an invaluable source of information for those studying the history of human genetics. During this workshop there will be a discussion on carrying out interviews with contemporary witnesses and other aspects of the oral history of human genetics. Conducting an interview requires careful planning both before and after the interview itself. The process of the transcription of interviews as well as ethical and legal aspects of oral histories will also be topics for discussion.

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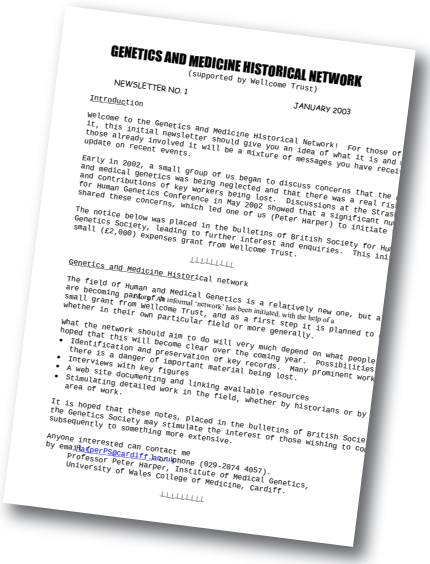
Genmedhist – The Genetics And Medicine Historical Network - How Did It Begin?

It is now 12 years since the Genetics and Medicine Historical Network was founded in 2002– not long perhaps in historical terms, but already the early details are becoming hazy (to me at least) so our editor's request that I should write a short note about its early history is a timely one.

Luckily the origins and progress of the Network have been documented to a considerable extent in its successive newsletters, which have served as a useful reminder while writing this, so perhaps an image of Newsletter 1, from January 2003, is a good starting point (figure 1). 'Homespun' is probably the best description of its appearance, by comparison with recent issues of the newsletter, but at least it was a start.



GENETICS AND MEDICINE HISTORICAL NETWORK

(supported by Wellcome Trust)

NEWSLETTER NO. 1

JANUARY 2003

Introduction

Welcome to the Genetics and Medicine Historical Network! For those of it, this initial newsletter should give you an idea of what it is and those already involved it will be a mixture of messages you have recei update on recent events.

Early in 2002, a small group of us began to discuss concerns that the and medical genetics was being neglected and that there was a real ris and contributions of key workers being lost. Discussions at the Stras for Human Genetics Conference in May 2002 showed that a significant nu shared these concerns, which led one of us (Peter Harper) to initiate

The notice below was placed in the bulletins of British Society for Hu Genetics Society, leading to further interest and enquiries. This ini small (£2,000) expenses grant from Wellcome Trust.

Genetics and Medicine Historical network

The field of Human and Medical Genetics is a relatively new one, but a are becoming particularly an historical network has been initiated, with the help of a small grant from Wellcome Trust, and as a first step it is planned to whether in their own particular field or more generally.

What the network should aim to do will very much depend on what people hoped that this will become clear over the coming year. Possibilities

- Identification and preservation of key records. Many prominent work
- there is a danger of important material being lost.
- Interviews with key figures
- A web site documenting and linking available resources
- Stimulating detailed work in the field, whether by historians or by area of work.

It is hoped that these notes, placed in the bulletins of British Socie the Genetics Society may stimulate the interest of those wishing to co subsequently to something more extensive.

Anyone interested can contact me by email MaPerPS@cardiff.ac.uk phone (029-2074 4057).

Professor Peter Harper, Institute of Medical Genetics, University of Wales College of Medicine, Cardiff.

Figure 1: The 'homespun' appearance of GenMedHist's Newsletter 1.

Before this newsletter appeared though, quite a lot of background work had gone on. A number of us across Europe and in North America had become worried that the early history of human and medical genetics was beginning to be lost, with founders in the field dying, records destroyed and nothing tangible being done to improve the situation. I myself had visited several prominent historians of science and medicine in Britain, only to find an almost complete lack of interest in human and medical genetics; most indeed did not know what the field was, some considering it to be synonymous with eugenics, leaving me somewhat discouraged.

Perhaps, though, this had some positive effects. A number of us had discussions at the 2002 European Society of Human Genetics (ESHG) meeting in Strasbourg where we agreed that an initiative was needed urgently, and that we as geneticists would have to undertake it, or at least get it off the ground ourselves, co-opting interested historians, archivists and others along the way. Email addresses were exchanged, a list compiled, and by the end of 2002 the network was in existence, albeit in a primitive form.

If ESHG was the birthplace of GenMedHist, it was also the focus for its first specific event – a part day workshop at the ESHG annual conference in Birmingham (UK), reported in Newsletter 3; this consisted of both a discussion on what the network should aim to do, and also three presentations, including one from Maj Hulten on the human chromosome number and a notable talk from John Edwards on '200 years of genetics in Birmingham'. Characteristically John managed to get his only two slides in the wrong order, and ran out of time before he had reached the second century!

The Birmingham workshop also saw the launch of the GenMedHist website (www.genmedhist.org), designed by our Cardiff web expert Jeff Alderman (I must thank him and the many other Cardiff University staff whose time I have unashamedly infringed on over the years, though in fairness they have said that they found the work much more interesting than what they were officially meant to be doing!). By late 2003 we also had some modest funding, with Wellcome Trust having supported our workshop and given a one-year small grant to establish the website.

So by 2004 the main activities of our network had already become clear and, having started as essentially a 'rescue operation' for preserving the history of human genetics, it steadily evolved into something more comprehensive and ongoing. Most of the main aims, as given in the original Newsletter 1, have each grown into full-size individual projects, with their progress recorded in specific sections of the website and in successive newsletters.

To mention a few of these, the alternate yearly workshops, now organised by Heike Petermann (Muenster), have been a particular success in bringing together historians and geneticists, with workshop 6 scheduled for Glasgow in June 2015. It has been a great help to have these linked to the ESHG conferences, with much of the organisational work now done by its executive officer Jerome del Picchia and his colleagues.

Full reports on all the workshops are already on the website, but I cannot resist one anecdote from the memorable 2005 workshop held in Mendel's Abbey, Brno, where I even had the privilege during a pre-conference planning visit of sleeping in the Abbey itself! During the convivial (to say the least) dinner held in the wine cellar beneath the Abbey, my Cardiff colleague Flo Ticehurst diligently photographed the proceedings (Figure 2), placing the photos on the GenMedHist website, from which they then found themselves on Google Images, to the consternation of some members!

Figure 2: The workshop dinner in the wine cellar beneath Mendel's Abbey in Brno (one of the more discreet photographs).

Recorded interviews

One of the most urgent gaps to fill in documenting the history of human genetics was the absence of all but a very few recorded interviews with older workers in the field. I have written about my own attempts at these in previous newsletters, but at the beginning I did not realise what an important part of our history this aspect would become. As I started by interviewing the oldest members it is inevitable that many should have since died; I very much doubt if these would have had their memories recorded otherwise. The lack of interviews is still especially prominent in America, where the early oral history programme seems to have come to a halt.

My own series has now grown to 100 and there are plans for ESHG to take the interviews forward to cover the next generation of workers, with specific interviewers for each European country. I now intend to conclude my series and to concentrate on analysing and writing about the abundant material contained in the interviews; I hope that others will do so also, since the interviews are almost all on the GenMedHist website and are open to all who wish to use the information that they contain.

As this article is about 'beginnings', I may perhaps be allowed an anecdote about the very first interview in my series, with Paul Polani in late November 2003. Although when I visited him it was a bitterly cold day and he was almost 90, he insisted on meeting me at the train station near his home, and after more than three hours of discussion was just as lively as at the beginning. We concluded by a meal at his favourite Italian restaurant in Guildford, where the staff clearly regarded him as close to being a God. After a very considerable quantity of wine, he then drove me back to the station (without mishap!) just in time for me to catch the late-night train home.

Listening to the audio files of these interviews (all are archived on computer) provides a vivid experience, especially if one has known the interviewee personally, and I regret that so far it has not been possible to place at least a clip of these alongside the transcript text on the website. For a few of the early interviews, though, I was able to do this in the form of a CD accompanying my 2006 book, *First years of Human Chromosomes*, and these clips are now on the GenMedHist website.

The Human Genetics Historical Library is another aspect that has become a fully fledged project and is now standing firmly on its own feet, thanks to the efforts of Karen Pierce and the support of Cardiff University's Special Collections and Archives (SCOLAR) under Peter Keelan. But it began very much as a

response to an emergency, when I was phoned by an Oxford colleague who told me that the entire departmental library was going to be abandoned, apart from a few volumes in current use. Fortunately I was able to drive over at once and found several hundred books spread out on tables. It was clear at once that many of these were of historical interest; the Department had its origins in Alan Stevenson's Medical Research Council Clinical and Population Genetics Unit in the 1960s. It was equally clear that any form of sorting would be a lengthy process so, co-opting some of the more stalwart members of the Department, I loaded the entire collection into my old but capacious Ford Mondeo estate car (figure 3), whose previous owner had used it for collecting wholesale grocery supplies, and drove very slowly back to Cardiff, hoping that the springs of the car would not give way.

Figure 3: Transporting the original volumes of the Human Genetics Historical Library.

Since then the collection has grown to well over 3000 volumes, in part due to the donation of several further large collections, but also from many small donations. The provenance of all books is recorded and thanks to funding from Wellcome Trust the collection has been catalogued in detail on the GenMedHist and Cardiff University libraries websites. Karen Pierce and I have written about the Historical Library in detail (Pierce and Harper, 2010), but a follow-up article is due and is planned. It is also encouraging to learn that Joerg Schmidtke and colleagues have now developed a comparable collection for Germany, especially since in our own collection books in languages other than English are underrepresented.

When the Human Genetics Historical Library started, the question arose whether we should include genetics journals, and we had several offers of long runs of volumes dating back many years. Reluctantly we decided against their inclusion and I am in no doubt that we were right, since over the past decade most journals have digitised their back issues, and their content is now both preserved and available to all, even though their handsome leather bindings may have been lost. We made a similar decision about reprint collections; here historians and archivists seem to be divided about their value but a comprehensive attempt to collect them would have been beyond our capability. What we do have an abundance of in the collection is the so-called 'grey material'- reports, conference proceedings and other items that have not been formally published, but yet contain much valuable information that illustrates the development of human and medical genetics.

The last area that I should like to mention in this note is the one that was directly responsible for the formation of the Network – the identification and preservation of the personal scientific records and correspondence of early workers in human genetics. In America this has been carried out extensively for many years, notably by the American Philosophical Society library, which has a superb collection of the records of early American geneticists, but little had been done in Britain, at least for workers in human and medical genetics. Here the first step was taken before I was myself involved, when John Edwards and Sue Povey were told that the records of James Renwick, a close friend and former colleague of both, who had died some years before, were in immediate danger of being destroyed. They resourcefully rented a van – a large one was needed – and transported the entire set of file cabinets back to the Galton Laboratory, at that time based in Wolfson House, London, where they sat in the basement for several more years untouched. It was around 2003 that I became aware of their existence and their extent; Renwick had played a key role in the early development of human gene mapping and the records provided a wealth of detail on this, as well as extensive correspondence.

At this time I had become aware of the work of the UK National Cataloguing Unit for the Archives of Contemporary Scientists (NCUACS), based at University of Bath, whose senior archivist, Tim Powell, confirmed the value of the Renwick archive and was able to catalogue it in detail. This was soon followed by the records of Malcolm Ferguson-Smith, which had grown in his home to the extent of endangering the safety of visiting grandchildren! Both catalogued collections are now housed in the University of Glasgow Archives and have recently been digitised as part of Wellcome Trust's major initiative on the history of modern genetics.

Sadly the NCUACS was closed not long after this, but senior staff were able to transfer to Cardiff University, where they completed the cataloguing of a series of further records of human geneticists, including a number of the later Galton Laboratory workers, so that a close to complete archive now exists for this key centre in the development of human genetics, spanning over a century from Galton himself up to the year 2000.

Looking back on the history and development of GenMedHist, I am amazed by what it has managed to achieve given the modest financial support underpinning it (mostly from Wellcome Trust). If I had known how it would grow, and the work involved, I would probably not have dared to become involved in the first place – but equally if I had waited until help and funding arrived, most of the Network's achievements would never have happened. Now, with able colleagues increasingly responsible for the various activities, I think that not only has the greater part of the early history of human genetics (at least in the UK), been saved from destruction, but that we have relatively secure foundations for ensuring that the process continues, as well as a large and growing body of written and oral material, which is already beginning to be used by historians and others analysing how human and medical genetics has developed and has influenced science and medicine as a whole during the past 60 years.

Peter S. Harper
Institute of Medical Genetics, Cardiff University School of Medicine, Cardiff, UK

Reference: Harper PS, Pierce (2010). The Human Genetics Historical Library: an international resource for geneticists and historians. *Clin. Genet.* 77, 214-220.

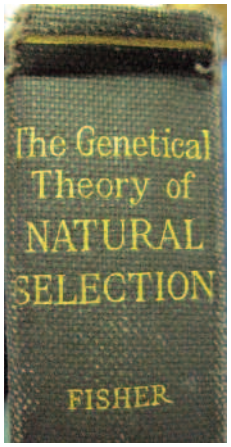
Human Genetics Historical Library Update 2014

ESHG funding for cataloguing

In April this year we were very pleased to receive funding from the European Society of Human Genetics for the cataloguing of 150 books. This will include the 73 published theses from Finland, donated by the Helsinki centre, and another series of books given by Walter Bodmer.

New donations

At the beginning of June we received a new donation from Peter Farndon, CBE (Previously Director, NHS National Genetics and Genomics Education Centre and Consultant Clinical Geneticist, West Midlands) Professor of Clinical Genetics, University of Birmingham. There were 47 items including a selection of textbooks, and course material from the Open University which dated from the 1970s and 1980s.



Over the summer the Cardiff University Library Research Reserve also donated 60 items they had weeded from their collection. The donation included Sewall Wright's four volume work *Evolution and the genetics of populations* (1968-1978), and the first edition of R. A. Fisher's *The genetical theory of natural selection (1930).*

Donations of books not already in the collection and other materials are always welcome. Please contact: Peter Harper (HarperPS@cf.ac.uk) or Karen Pierce (PierceKF@Cardiff.ac.uk)

RLUK conference

At the end of October I was pleased to present a paper about the Human Genetics Historical Library at the Research Libraries UK (RLUK) conference 'Discovering collections, discovering communities' held at the iconic Library of Birmingham. The overarching theme for the conference was 'forging collection based collaboration between archives, museums and academia' and was an ideal place to talk about the success of the HGHL. In my paper I emphasised the idea that we were embracing the concept of 'preservation for the future'. I discussed the unique focus on human and medical genetics in a library collection that came about due to the inspiration of Prof Harper and the MedGenHist network, and the collaboration with library staff at Cardiff University. The panel I spoke within also had a paper from Jenny Shaw of the Wellcome Trust talking about 'Collecting genomics: working collaboratively to document modern science'; her paper was about archives, whilst mine was from a library perspective, and these two approaches complemented each other well.

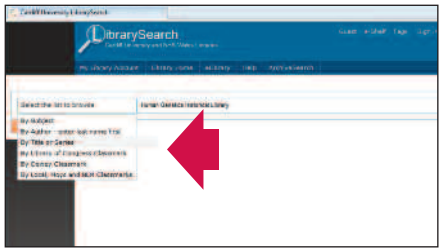
http://www.rluk.ac.uk/events/discovering-collections-discovering-communities/
Presentations are due to be made publically available in early December.

Karen F. Pierce, HGHL cataloguer (Cardiff University)

Accessing the Human Genetics Historical Library Catalogue

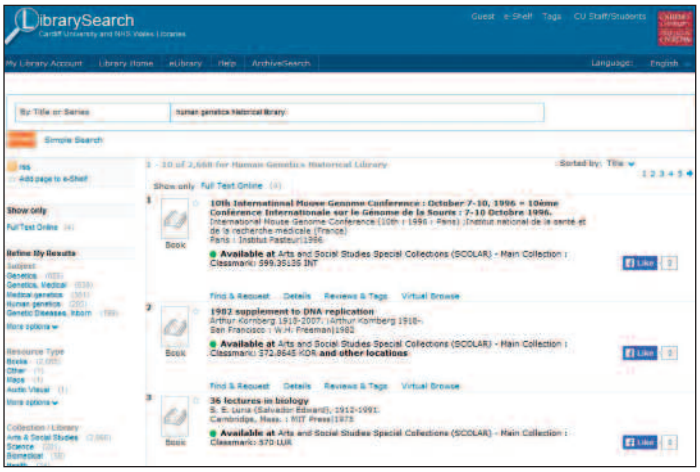
To find out what items are held in the HGHL one can search Cardiff University's library catalogue Library Search (http://librarysearch.cf.ac.uk/primo_library/libweb/action/search.do?vid=CARDIFF_V1)

To accurately retrieve all items in the collection you will need to search using the Browse Library Catalogue facility:



Click on 'Select the list to browse' and choose 'By Title or Series' and then enter **Human Genetics Historical Library** in the search box, and click the Browse button.

This will retrieve a list of titles of which Human Genetics Historical Library will be at the top. If you click on this title entry you will be taken to the list of results with this title.



Multiple editions of a book with the same title will be grouped together under one entry.

All provenance information is recorded on the catalogue records, so if you look in the Details tab of each individual record you will be able to see who donated the item and notes on any marginalia or extra material laid in.

The majority of catalogued items are held within SCOLAR, the special collections unit of Cardiff University library service, and are available to view (reference only) during opening hours, Monday – Friday (9am-5pm). Prof. Harper's personal collection is not currently held in the library, but can be accessed with advance notice. Contact details are available online: http://www.cardiff.ac.uk/insrv/libraries/scolar/news/index.html

Karen F. Pierce (HGHL cataloguer)

Human Genetics Lecture Notes - Call for Material

Most people working in the field of human and medical genetics have been involved in teaching undergraduate or postgraduate students, whether on specific genetics courses, or as part of biology, medicine or other topics. Professor Anthony Edwards, after looking out his own lecture notes for third-year genetics undergraduates at Cambridge from 1969, has written to say that he feels these may be of historical interest; he also wonders how many courses there were at this early time and how many other people have kept their original notes.

To follow up on this suggestion, *GenMedHist* would like to invite anyone with relevant material (from any country) to contact the Editor or Peter Harper. As well as hand-written or typed notes, electronic documents or slide series would also be of interest. If the material has already been included in any existing personal archive it would be helpful to know this.

Do not send any actual material yet, since thought and discussion will be needed before deciding how best to preserve it. This is a preliminary note to gain an idea of how extensive such notes might be; we shall report further in the next *Bulletin*.

Judith Friedman (jfriedma@uvic.ca)
Peter Harper (HarperPS@cf.ac.uk)

Historical Library, Institute of Human Genetics, Hannover Medical School, Germany

The Institute of Human Genetics of Hannover Medical School, Germany, is making its historical library open to the public. It contains books and journals of historical interest, mainly in German, and has an emphasis on ethical, legal, and social issues. It also contains a small collection of lay media "grey literature" on these topics. The historical library is a reference library. Visits can be arranged on request by contacting Dr. Joerg Schmidke (schmidtke.joerg@mh-hannover.de). Catalogues are available at http://www.mh-hannover.de/28073.html.

Prof. Dr. med. Joerg Schmidtke, Institut fuer Humangenetik, Medizinische Hochschule Hannover, Germany

Documenting The Development Of Medical Genetics In Czechoslovakia After 1945

Medical genetics was established in Czechoslovakia as an independent scientific field shortly after the WWII. The first full professor of medical genetics at the Faculty of Medicine of the Charles University in Prague, Bohumil Sekla (1901–1987), was appointed in 1946. Until early 1950s, medical genetics in Czechoslovakia continued in the tradition of reformist eugenic efforts of the second half of the 1930s. After the 1948 Communist takeover, however, classical genetics was gradually suppressed and forcefully replaced by Lysenkoism and Michurinian biology.

Professor Edward Babak in 1922 at the Mendel Centenary. Image used with the permission of the Moravian Museum Brno, Department of the History of Genetics (Mendelianum).

This development significantly affected also on the situation in medicine. This state of affairs remained largely unchanged until early 1960s, when the reception of the then latest discoveries (the discovery of the DNA, of the number of human chromosomes, etc.) made it obsolete. During the same period, medical and clinical genetics started developing in Czechoslovakia. A number of institutions, mostly related with paediatrician care, were created which focused on genetic counselling and prenatal care (Prague, Brno), on basic research in the area of immunogenetics (Prague), and, somewhat later, also cytogenetics (Prague), which was originally covered by endocrinology. Faculties of medicine in Prague and Brno and the Czechoslovak Academy of Sciences played a key role in this process.

Dr. Lionel Penrose in Brno 1968. Image used with the kind permission of Professor Laxova

The main aim of the history project is to map the development of medical genetics by recording authentic testimonies of colleagues who took part in the process through interviews. These can cover the generations reaching recently their 70s and 80s. In this area of oral history, where several dozen interviews are planned, the project is much indebted to the initiative of Sir Peter Harper, Professor Emeritus of the Cardiff University, whose support the authors hereby wish to acknowledge with gratitude.

An independent documentary collection then gathers some written, both published and unpublished, materials and images which would otherwise be irretrievably lost. The collection covers also the earlier history starting with the so-called rediscovery of Mendel's principles in 1900–01. This collection currently includes approximately 600 items including offprints and some rare issues. Their list will be accessible online through pages of the AMG (at (http://www.slg.cz) in the second half of 2015.

The material thus collected should serve two main purposes. Firstly, it should serve as a starting point for a study of the turbulent history of medical genetics in Bohemia, Moravia, and Slovakia in the twentieth century. Secondly, it should be accessible to researchers, Czech and foreign, for comparison with the development of medical genetics elsewhere, especially in CEE countries. Another important part of the project is the preparation of a monograph and edition of interviews concerning the history of medical genetics in Bohemia and Moravia.

The project is to be implemented by the Centre for the History of Sciences of the Academy of Sciences of the Czech Republic and the Society for Medical Genetics of the Czech Medical Association of Jan Evangelista Purkynje (hereinafter SMG).

For further questions, contact Michal Simunek PhD. (simunekm@centrum.cz) or Professor MUDr. Milan Macek Jr., DrSc. (milan.macek.jr@lfmotol.cuni.cz).

Michal V. Šimůnek (Prague)

A New Exhibition Presenting The History Of Genetics In Brno

The original memorial to Gregor Johann Mendel (the 'Mendelianum') was created in 1964 as part of the Moravian Museum in Brno on the occasion of an upcoming international celebration of one hundred years since the presentation of Mendel's discovery. These celebrations, accompanied by the Mendel Memorial Symposium, took place in Brno on August 4–7, 1965 under the auspices of the International Union of Biological Sciences (IUBS), the Czechoslovak Commission for UNESCO, the International Atomic Energy Agency (IAEA), the Council for International Organizations of Medical Sciences (CIOMS), and the Czechoslovak Academy of Sciences (ČSAV).

The original Mendelianum was located on the first floor of the Episcopal Courtyard in the centre of Brno but already ahead of the symposium, it was moved to St. Thomas Augustinian Abbey where Mendel worked and carried out his experiments. And there the Mendelianum remained until December 31, 2001, when after 36 years of successful activity it had to leave due to restitution of the Church property.

The Moravian Museum, the second largest and oldest museum in the territory of the current Czech Republic, was founded by the Moravian–Silesian Society for the Promotion of Agriculture, Natural Sciences, and Local History, which Mendel, too, had joined in 1854. The Moravian Museum has always been highly interested in preserving and promoting Mendel's scientific legacy, both through the now defunct Science Academy for Moravia and Silesia – Economic Society (so-called *Ackerbaugesellschaft*), and through its Society for Natural Sciences (*Naturforschender Verein*). Mendel actively participated in the administration and organisation of most sections of this Society and was thus directly involved in the early development of the Moravian Museum. All meetings of the *Ackerbaugesellschaft* took place in the Bishop's Courtyard, in the historical meeting and lecture hall of the Moravian Museum.

Figure 1. Meeting and Lecture Hall

For these reasons the Moravian Museum in June 2010 applied to the Ministry of Education of the Czech Republic for support of a project of a new visitor centre called 'Mendelianum – An Attractive World of Genetics'. The application was presented within the framework of the Ministry of Education's call for drawing on the EU Structural Funds as part of the Operational Programme 'Research and Development for Innovation'. The project application met all evaluation criteria and project evaluators recommended that the project be financed in 2012–2014.

This is why the new Mendelianum, which presents the world of genetics in an attractive and fresh way, is situated in these historical and highly authentic premises. It provides facilities needed for the popularisation of science, in particular biology, genetics, and their history. The visitor centre offers three basic units which focus on different groups of visitors: 1) From Gene to Function; 2) From Theory to Practice, which focuses on implementation of new research and active participation in laboratory work, and 3) Mendel's laboratory, which presents the history of genetics from Mendel to the most up-to-date developments on the level of Nobel Prizes. This section

includes also historical exhibits. The creation of several alternative tours encourages visitors to employ their own creativity. They can experience science and research 'hands-on', be it through the 3D models of DNA replication and gene expression, by visiting Mendel's laboratory, or by seeing what a modern laboratory of molecular genetics looks like. The various parts of the exhibition help visitors appreciate the importance of genetics in everyday life. They can also gain a deeper understanding of their own biological structure and life rhythms. The new 3D models of DNA replication and gene expression are the largest in the Czech Republic and possibly in Europe, since together with the connected model of translation and of a ribosome they are over 8 meters (26ft) high.

Figure 2. 3D DNA Model

The inauguration of the new exhibition takes place on January 1, 2015.

Images reproduced with the permission of the Moravian Museum Brno, Department of the History of Genetics (Mendelianum)

JIŘÍ SEKERÁK (BRNO), MICHAL V. ŠIMŮNEK (PRAGUE)

Invitation and Call For Papers – Mendel Forum 2015

Mendel Forum 2015

Mendelianum of the Moravian Museum in co-operation with the Institute of Animal Physiology and Genetics, Academy of Sciences of the Czech Republic, Mendel University in Brno and other institutions are pleased to invite you to spend A WEEKEND WITH J. G. MENDEL in the city of Brno.

JOIN US for the weekend March 6 – March 8, 2015 to celebrate 150 years since the publication of Mendel's discovery in Brno and 50 years since the opening of the Mendelianum of the Moravian Museum.

BOTH, passive and active participants are welcome, registration is free of charge.

A unique complex of Centrum Mendelianum based in authentic premises of Mendel´s Scientific Society including Mendel´s Scientific Centre, Mendel´s Visitor Centre and Mendel´s Interactive School will be inaugurated on March 8. See the information attached!

See you in Brno!

www.mendel-brno.cz
MF2015@email.cz
www.mendelianum.cz

1st Announcement, Call for Abstracts

March 6 – 8, 2015 Centrum Mendelianum, Muzejni 1, Brno – city centre, Czech Republic

Registration:

Free of charge, limited number of participants
Deadline for abstract submission: Dec 31, 2014
www.mendel-brno.cz
MF2015@email.cz

Topics:

Mendel as a scientist and multifaceted person
Mendel´s Scientific Society – a great variety of ideas in a small space
Mendel´s Scientific Collegium – Brno and the world
Mendel´s Discovery in modern scientific context
Interpretations of Mendel´s scientific work
Multidisciplinary applications of Mendel´s ideas
Miscellaneous

150 years since the publication of Mendel's discovery in Brno
50 years since the opening of the Mendelianum of the Moravian Museum
Opening of Centrum Mendelianum in the authentic rooms of Mendel´s scientific society in Brno

Abstracts:

Deadline: Dec 31, 2014
Submission: Via the abstract gate at www.mendel-brno.cz, Alternatively, the abstract can be sent to MF2015@email.cz
Guidelines: The abstract must be in English, include title, authors, affiliation and abstract body up to 500 words. Presenting author must be indicated and address for correspondence provided. Accepted presentations will be confirmed by Jan 15, 2015. Abstracts will be published in peer-reviewed journal Folia Mendeliana.

Preliminary Programme - Overview

Friday, March 6, 2015 - Dr. I. Kubistova, IAPG AS CR, v.v.i. - Mendel´s Plants (Talk and new exhibition viewing)
Get together

Saturday, March 7, 2015 - AM - Dr. J. Sekerak, Mendelianum – Johann Gregor Mendel (Talk and new exhibition viewing) Mendel´s Discovery in Context of Science (Talk)
Talks to be announced (selected from submitted abstracts)
Saturday, March 7, 2015 - PM - Dr. A. Matalova, Mendelianum – Mendel´s Scientific Society and Collegium Mendel´s Brno
Talks to be announced (selected from submitted abstracts)
Sunday, March 8, 2015 - AM - Prof. E. Matalova, IAPG AS CR, v.v.i. – Centrum Mendelianum (Concept, guided tour and official opening)
Sunday, March 8, 2015 - PM - Prof. J. Klein, USA – Solitude of a Humble Genius (Introduction of a new expert book about JGM, discussion with the author)

Images reproduced with the permission of the Moravian Museum Brno, Department of the History of Genetics (Mendelianum).

American Association of the History of Medicine Annual Meeting in Chicago

For the Eighty-Seventh Annual Meeting of the American Association of the History of Medicine, historians and physicians met in Chicago from May 8th to 11th 2014. On Thursday afternoon, participants had the opportunity to tour the archives of the American Medical Association; Thursday night featured a showing of "Digital Short Films in the History of Medicine."

A number of talks addressed the history of genetics. In a panel on the Metrics of Race Across Four Continents, Kristin Roebuck, Columbia University, brought attention to eugenic research on konketsuji, "mixed-race" children in post war Japan. Born to a Japanese mother and a foreign father, often a US soldier, these children caused great concern in a US-occupied Japan that feared for its national independence and identity. Segregated in special homes, these children were easily available research objects for numerous examinations that demonstrated their supposed inferiority and degeneration. The konketsuji, Roebuck showed, thus served as a negative image of racial contamination against which Japanese purity, free from foreign influences, could be constructed.

On Saturday afternoon, the panel on Screening and Surveillance brought together the histories of population control, prevention, disease and disability. In his talk on fragile X Syndrome, Andrew Hogan, University of Virginia, pointed out continuities in the genetic prevention of intellectual disability. By the 1950s, improving techniques of chromosome research allowed a relocation of congenital intellectual disability. This, Hogan argues, introduced an age of targeted prevention, as prenatal diagnosis aimed at the level of the individual at-risk pregnancy. Hogan showed how geneticists and physicians delineated fragile X Syndrome from clinical signs and chromosomal research, turning the chromosome into a marker of disease under surveillance. Marion Schmidt, Johns Hopkins University showed how in the first decades of the 20th century, teachers for the deaf adopted eugenics as a means of progressive school reform, yet rejected rigid genetic determinism. Distancing their students from the feebleminded, teachers propagated an image of the responsible deaf citizen who would voluntarily give up procreation if necessary. Rather than coercive eugenics, Schmidt argued, genetic deafness research thus stands for the development of reproductive self-restraint through the internalization of eugenic values, intertwined with contemporary notions of citizenship, productivity and disability. Devon Stillwell, Johns Hopkins University, took the

audience back to the early decades of genetic research of and counseling for breast cancer in the US. Cancer, she pointed out, had long been a concern of heredity research, from the Eugenic Record Office trait files to Sheldon Reed's mid-century research on breast cancer. Stilwell delineated the emergence of "cancer risk families," paying particular attention to the emotional implications of being at risk that were negotiated between (potential) patients and genetic counselors. Adopting the paradigm of cancer prevention through early diagnosis, Stilwell argued, genetic counselors created a state of pre-disease that required constant risk management under professional surveillance.

Taken together, the talks at AAHM 2014 presented a history of genetics that is geographically diverse and strongly pushing the field into the second half of the 20th century.

Marion Schmidt (Johns Hopkins University)

Messenger RNA: From Discovery to Synthesis and Regulation in Bacteria and Eukaryotes http://library.cshl.edu/Meetings/mRNA/

A unique meeting, entitled, "Messenger RNA: From Discovery to Synthesis and Regulation in Bacteria and Eukaryotes," was held August 9-11, 2014, at Cold Spring Harbor Laboratory (CSHL). The meeting began at 7:00 pm on Saturday, August 9th, and concluded after lunch on Monday, August 11th. This meeting combined a historical perspective with updates on exciting developments and research directions of the current day. Speakers included pioneers in the field of mRNA from around the world.

The organizers were: James Darnell, The Rockefeller University; Adrian Krainer, CSHL; and Mila Pollock, CSHL.

The audience included scientists, postdoctoral fellows, graduate students, historians, science writers, journalists, and members of the public who were interested in a topic. Among 140 attendees we had four Nobelists, 18 postdocs and 27 graduate students. It was an excellent opportunity for all participants to learn from and share with the top people in this dynamic field, and with one another.

A unique aspect of the meeting was that scientists, each of whom had made a great contribution to the field, presented their historic work from today's perspective, and then discussed it with the audience. It was incredible to have in one auditorium multiple generations of pioneers in the field.

An introductory presentation by James D. Watson, entitled, "From the Evidence of Volkin-Astrachan RNA to the Jan-July 1960 Discovery at Harvard of T2 and E. coli Messenger RNA," kicked off the meeting. A keynote speaker Sydney Brenner covered the topic: "Messenger RNA: The Idea and Experiment. 1960." This was followed by seven presentation sessions spread over three days (see http://library.cshl.edu/hosted-meetings/history-of-messenger-rna).

The meeting was part of a new series of CSHL/Genentech Center Conferences on the History of Molecular Biology and Biotechnology. These conferences aim to explore important themes of discovery in the biological sciences, bringing together scientists who made seminal discoveries with others whose interests include: the current status of the field, the historical progress of the field, and/or the application of the techniques and approaches in biotechnology and medicine. The first meeting in this series was on the History of Biotechnology (http://library.cshl.edu/Meetings/HistBiotech/), followed by one on the History of Restriction Enzymes (see http://library.cshl.edu/Meetings/restriction-enzymes/). A meeting in September 2014 covered the History of Plasmids (see http://meetings.cshl.edu/meetings/2014/biohist14_plasmids.shtml); and a meeting in 2015 will cover the History of Sequencing.

Major support for this meeting was provided by: CSHL, New England Biolabs, and The Rockefeller University. Additional support was provided by: Lexogen and Life Sciences Foundation. The generous support of our sponsors enabled us to cover the costs of attendance for talented junior investigators, post-docs, historians and students.

Mila Pollock (Cold Spring Harbor Laboratory Library)

"A Lucky Survivor": The Arno Motulsky Papers, American Philosophical Society, Philadelphia USA.

Arno Guenther Motulsky, born in East Prussia in 1923, is a lucky survivor. In a true-life tale of peril and privation, on a journey that took him from Germany to Belgium to France and then Spain, he narrowly escaped lethal anti-Semitism in a Europe falling under Nazi domination. The 17-year-old Motulsky, who had obtained a valid US visa, finally made it to the United States in 1941. Ten days after his departure, he turned 18, the age the Nazi-friendly Spanish government denied people transit. Motulsky was reunited with his father in Chicago; his mother, sister, and brother survived their own harrowing experiences in occupied Europe, and the whole family was reunited in the US after the war. At the age of 18, Motulsky found asylum and could start on the path that would lead him to becoming one of the leading geneticists in America.

Figure 1. Drawing of Arno Motulsky in 1941.

This remarkable tale is documented in the Motulsky Papers at the Library of the American Philosophical Society by among other items interviews, biographical sketches, a brief incomplete diary (in German), a German passport, and a letter (in French) to the commandant of the of the camps in which he was held.

In Chicago, Motulsky took a high school equivalency test and also found work in a virology lab at Michael Reese Hospital. A scholarship allowed him to enroll in premedical courses at Central YMCA College (later Roosevelt University). In an English class he met Gretel Stern, whom he married in 1945. Motulsky was accepted at the Medical School at the University of Illinois in Chicago, but shortly afterward he joined the army and was assigned to the specialized training program, then sent to Yale to finish premedical courses. Returning to Chicago, he graduated from medical school in 1947 and began his internship and residency at Michael Reese, where under the guidance of Karl Singer, his research focused on hematology. Singer and Motulsky came close to demonstrating through experiments with rabbits that the hemoglobin in sickle cells anemia erythrocytes was difference from normal red cells, which was later demonstrated electrophoretically by Harvey Itano and others in Linus Pauling's lab.

After another stint of Army service, during which among other duties saw him assigned to William Crosby's hematology lab at Walter Reed Graduate School, in 1953 Motulsky became an instructor in hematology at the University of Washington, working with Clem Finch, who emphasized the importance of asking the right scientific questions and writing clear scientific papers. Dr. Motulsky began giving lectures in medical genetics in order to acquaint students with the field. Urged to set up a division of genetics, Motulsky visited labs and was influenced particularly by James Neel (whose papers, recently cataloged, are also at the APS). In 1957 the genetics division was established at the University of Washington. Following the establishment of the department, Motulsky spent an academic year at the Galton Laboratory in England with Lionel Penrose that greatly benefited Motulsky and the new division. Dr. Motulsky has spent the rest of his career associated with the department he established.

In the papers at the APS, the documentation of Motulsky's activities in the 1950's and 1960's unfortunately appears to be rather lacking. All the papers have been partially processed, divided into 14 series and reboxed. The dates range from the 1930's through the 2000's, but the bulk is from the 1970's through the 2000's. The papers thus mostly cover Motulsky's later career, when he still did research but also was very active as an editor, board member, teacher, and consultant.

The correspondence series is still arranged chronologically, but when fully processed to be arranged alphabetically, and has letters from a number of important scientists: L. L. Cavalli-Sforza, Victor McKusick, Ernest Beutler, and Friedrich Vogel.

The conferences and meetings series has material from a variety of meetings. There are meetings represented that one could anticipate: the National Academy of Sciences, the American Society of Human Genetics. Other meetings represented in the papers include, for instance, many folders, dating from 1975-1997, on the March of Dimes' birth defects conferences. (In 1996 the March of Dimes Birth Defects Foundation presented Motulsky with the Colonel Harland Sanders Award for Lifetime Achievement in Genetics.) In 1985 Dr. Motulsky attended a meeting of CANDLES—Children of Auschwitz Nazi Deadly Lab Experiment Survivors—founded by twins who survived Dr. Joseph Mengele's medical experiments. At the meeting evidence was heard by a distinguished panel about the ghastly experiments performed by the Angel of Death. Motulsky testified as an expert in medical genetics.

The organizations and committee series documents the many advisory roles that Motulsky has played. Among the extensive files in the series are those on the National Academy of Sciences' Institute of Medicine. Motulsky served on the IOM Council from 1980-82. He also was chair of the Committee on Assessing Genetics Risks, and wrote the preface and contributed to its report, *Assessing Genetic Risks: Implications for Health and Social Policy* (1994). Another extensive file, ranging in dates from 1976-2001, is on the American Society of Human Genetics; Motulsky served as president 1977-78. Also, Dr. Motulsky served from 1979-83 on the President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research and contributed to the reports of the Commission. Among the international committees with significant material is the Radiation Effects Research Foundation, for which Motulsky served as scientific councilor from 1983-97, and since 1969 he has served on the World Health Organization's Expert Advisory Panel on Human Genetics; the collection contains files about both committees. The significant reports produced by the committees on which Motulsky served often concern the intersection of science, ethics, and public policy.

As a leading geneticist of the last half of the twentieth century, Motulsky played an advisory role in the early formation of biotechnology firms. He served on the advisory boards of the firms as they were being established: Genescreen, Mercator, and Amgen. The Amgen files are fairly extensive and look to serve as a good source of information about its formation.

Motulsky has held a number of significant editorships, such as *Human Genetics* (1969-97) and the monograph series *Progress in Medical Genetics* (1974-2000). The papers have files on these and many of the nine editorships he has held and twenty-six editorial boards on which he has served. The papers also have material on the separate monographs edited by Dr. Motulsky, to which he was an important contributor. Among these books are *The Genetic Basis of Common Diseases*, and *Human Genetics: Problems and Approaches*, first published in 1979, now in a third edition and translated into Italian, Japanese, Chinese, and Russian.

Many other aspects to Motulsky's career could be noted and the record of his work explored in the papers: his genetic study of hyperlipidemia that led to the discovery of a new disease (familial combined hyperlipidemia) and made possible the Nobel Prize-winning work of Joseph Goldstein; his determining that a genetic polymorphism affects red color perception; his writings about genetic counseling; his research into diseases of Ashkenazi Jews. Perhaps the most ground-breaking is represented by a paper that appeared in JAMA in 1957. Writing at the request of the AMA subcommittee on blood dyscrasias of the Committee on Research, in three pages he first delineated what Friedrich Vogel in 1959 termed pharmacogentics. At the conclusion of his report, Motulsky writes:

Genetically conditioned drug reactions not only are of practical significance but may be considered pertinent models for demonstrating the interaction of heredity and environment in the pathogenesis of disease. In [the conditions cited in the report] it can be shown clearly how hereditary, gene-controlled enzymatic factors determine why, with identical exposure [to a drug], certain individuals become "sick," whereas others are not affected. It is becoming increasingly probable that many of our common diseases depend upon genetic-susceptibility determinants of this type.[1]

What can be seen now as the common sense (based on the experimental evidence) behind Motulsky's report was at first more or less ignored. But as further research confirmed the evidence, pharmacogentics grew into a discipline and also spawned sister disciplines (which also received Motulsky's attention) such as ecogenetics, nutrigenetics, and pharmacogenomics.

The research series is not large but contains a number of folders about Motulsky's research into glucose-6-phosphate dehydrogenase (G6PD) deficiency. A lack of the G6PD enzyme can cause hemolytic anemia. Motulsky went to the Congo in 1959 to study the deficiencies, which has the genetic benefit of granting the carrier resistance to malaria. The work in Africa resulted in a number of papers, but it was the saved blood samples that by happenstance proved more historically significant. A sample of a patient from Leopoldville, now Kinshasa, was in the mid-1980's shown to have HIV-AIDS antibodies present, the earliest known evidence of the HIV in a human sample. The patient's name is not known, but is designated in the research as L70. Motulsky very carefully followed scientific protocol and did not identify patients by name in his research data, but some of the data sheets themselves are present.

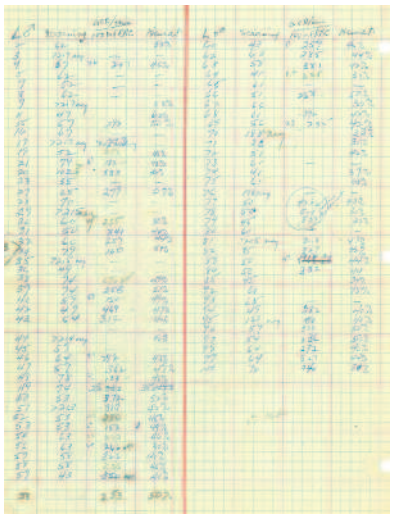


Figure 2. Data sheet from Arno Motulsky's research in the Congo, 1959.

A final note should be made about material that Motulsky saved that proved its worth years later. Decades ago, Margaret Lasker[2] researched the inborn metabolic disorder pentosuria, which affects almost exclusively individuals of Ashkenazi Jewish ancestry and is characterized by high levels of the pentose sugar l-xylulose in blood and urine and deficiency of the enzyme l-xylulose reductase. The condition is autosomal-recessive and completely clinically benign. Lasker entrusted Motulsky with her records. In 2010 Motulsky and his colleague Mary Claire King contacted the APS and asked for the records back. It was an unusual request, which normally would have been fulfilled by sending copies of the material; however, in this case, since the records were not of Motulsky's work, they were sent back to the University of Washington. Dr. Motulsky and colleagues, by following up with the individuals and their families through Lasker's work, were able to identify two mutations in genes that caused the condition.[3]

Figure 3. Arno Motulsky ca. 2000.

The collection at the APS awaits more comprehensive examination, and the preliminary arrangement will likely be revised. Dr. Motulsky will also be sending the APS more of his papers. But going through those now in the Library, two things became clear to me. First, Dr. Motulsky throughout his career has not forsaken clinical medicine, valuing his contact with patients as much as any other aspect of his work. Second, many of Dr. Motulsky's professional interests show his continuing concern about medical ethics and, in a broad, humanistic sense, a concern about the moral life of humankind. "While there are many bad things in this world," said Dr. Motulsky, "there is much good in human beings. We should try to bring out the best in people."[4] As a physician, teacher, researcher, and active professional, Dr. Arno Motulsky has done just that.[5]

Images used courtesy of Arno Motulsky Papers, American Philosophical Society.

Charles Greifenstien, Associate Librarian & Curator of Manuscripts, American Philosophical Society.

[1] Arno Motulsky, "Drug Reactions, Enzymes, and Biochemical Genetics, JAMA, Vol. 165, Oct. 19, 1975, 837.
[2] No relation to the founders of the Lasker Award, Albert and Mary Woodard Lasker.
[3] You can see the results of their work at http://www.pnas.org/content/108/45/18313.full. Dr. King, who among many achievements identified breast cancer genes, demonstrated that humans and chimpanzees are 99% genetically identical, and used genetic sequencing to identify victims of human rights abuses, won the Lasker Award in 2014. Both Dr. King and Dr. Motulsky are members of the American Philosophical Society.
[4] Profile of Dr. Motulsky. "Holding Out Hope in a Cruel World: Geneticist Arno Motulsky Recalls Wartime Europe," UW Medicine, vol. 25, no. 2, Fall 2002, 12.
[5] Information for this piece came from the article in n.4 , and biographical and other files in the Motulsky Papers.

In Memoriam - André Boué (1925-2012), Joëlle Boué (1926-2014)

André and Joëlle Boué as a couple will leave a luminous mark in the history of French genetics. Their careers were from beginning to end original, inseparable and complementary.

Figure 1: André and Joëlle Boué in their laboratory at Longchamp.

They were pioneers in the study of the aetiology of spontaneous abortions. Endowed with the capacity to avoid all ethical conflicts, they developed and then put into practice prenatal diagnosis in France, at the Château de Longchamp, which was the home of the Centre International de l'Enfance (the International Centre of Childhood). Founded in 1949, this centre was run by a group of 6 members, including Robert Debré, and was a prestigious and undisputed authority in the medical community of the time.

When they first met during the 1950s, Joëlle was hoping for a career in surgery, while André was an anaesthetist in resuscitation medicine. But the couple left for Iran, where a surgeon friend had asked for André's help in creating a service for cardiovascular surgery. In Tehran they were welcomed with open arms by the director of the Pasteur Institute, Marcel Baltazard, who put a laboratory at their disposal. They stayed there for nine years, during which time they first set up a transfusion centre, to respond to the needs of the surgical service, but also to study the great diversity of blood groups in the population, which included Shiites, Sunnis, Kurds, Zoroastrians, Russians ... . They next created a virology laboratory for the epidemics of plague, typhus, smallpox, rabies, which were rampant in the region, with cell cultures, initially from monkey kidney, then human kidney (obtained from the numerous stillbirths in neighbouring maternity units).

In 1959 they returned to France, where Professor Debré offered André Boué a post as research leader at CNRS (Centre National de Recherche Scientifique) to direct a virology laboratory. André first worked on the epidemiology of polio virus and on the Sabin vaccine. Joëlle completed her training in cell culture by learning cytogenetic techniques at Hôpital Trousseau as part of the service of Professor Turpin.

At this time the external and endogenous causes of spontaneous abortions were far from being understood and made an excellent subject for research. To achieve this they went together to the Wistar Institute in Philadelphia (Pennsylvania, USA), to carry out training in the laboratory of L Hayflick who had developed long-term culture of diploid cells for making vaccines. During the course of their stay two events occurred that would have a decisive effect on the direction of their work. These were, first, an epidemic of rubella that provided them with the opportunity to explore the pathogenic effect of the virus on embryonic tissues; and secondly the work of David Carr (London, Ontario, Canada) which showed the presence of chromosomal anomalies in spontaneous abortions.

On returning to Longchamp they began a long and important study on several thousand early spontaneous abortions, establishing their frequency and the types of chromosome rearrangement that had a lethal effect on embryonic development, at l'unité INSERM 73 which became Unité de Biologie Prénatale. Around 1970 it had been shown that it was possible, by culturing amniocytes, to establish the karyotype of the future child. It was thus a logical part of her own work for Joëlle Boué to achieve the first diagnostic prenatal diagnoses in France. The decision to terminate pregnancy in the case of severe and incurable abnormality contravened the law of 31st July 1920, which forbade all termination of pregnancy (under the Vichy regime a woman was condemned to death for having undertaken abortions). The first

prenatal diagnosis was achieved in collaboration with Kurt Hirschhorn, then at the Galton Laboratory, London (UK), in 1971, with the approval of Professor Robert Debré. Sadly, the same aneusomy (from which the older brother, very malformed, had died previously), was found again, and the first termination of pregnancy for a chromosomal anomaly was carried out.

In 1975, in Stockholm, they participated in the first European workshop on prenatal diagnosis, organised by EMRC (European Medical Research Council). Joëlle then provided genetic consultations for numerous couples from all over France, whose history justified prenatal diagnosis, until cytogenetics laboratories in other university centres had the capability (technical and ethical) to carry out prenatal diagnosis themselves. In 1980 André became Professor of Medicine (the specialty 'Genetics' still did not exist). He broadened his researches on serological markers in maternal blood and other fields of fetal pathology, and also worked on the immunology of hybridomas and monoclonal antibodies.

For her part, Joëlle extended her activities to the prenatal diagnosis of metabolic illnesses, in collaboration with J-C Dreyfus of institute Cochin, Paris, and of Mendelian disorders, autosomal or X-linked, especially fragile X syndrome in collaboration with J-L Mandel in Strasbourg. In 1984 they both received the Prix de la Fondation Allianz (the Foundation of the Institut de France).

At the end of his career, for a period of 17 years, up to the year 2000, André Boué served as a member of the Comité Consultatif National d'Ethique (CCNE). It was he who drafted most of the 'recommendations' issued by the CCNE on diagnostic and predictive genetic tests, on explanatory information for patients and on confidentiality, as well as on the constitution of cell banks. These recommendations, while not mandatory, are playing a major role in the development of bioethics laws.

Throughout their professional life they succeeded – Joëlle with her courtesy and André with his friendliness – in establishing scientific and personal links with numerous geneticists from Europe and from Canada, who they often welcomed in their pleasant Parisian apartment.

Figure 2: Joëlle Boué (centre) surrounded on the left by Margaretha Mikkelsen (1923-2004) (Glostrup, Denmark), Ted Galjaard (Rotterdam, Netherlands), and on the right by Marie Ferguson-Smith (Glasgow, UK) and Eva Sachs (Rotterdam, Netherlands).

Simone Gilgenkrantz
Professeur émérite de génétique humaine, Faculté de médecine, Université de Lorraine, France

Translated by Peter S. Harper
Institute of Medical Genetics, Cardiff University School of Medicine, Cardiff, UK

Recent Publications

The following recent publications may be of interest to *Bulletin* readers:

Huijnen, Pim, Fons Laan, Maarten de Rijke and Toine Pieters, "A Digital Humanities Approach to the History of Science. Eugenics revisited in hidden debates by means of semantic text mining," in: A. Nadamoto et.al. (eds.), *Social Informatics. SocInfo 2013 International Workshops, QMC and HISTOINFORMATICS Kyoto, Japan, November 25, 2013*. (Springer: Berlin and Heidelberg, 2014).

Rushton, Alan R. "Invited Editorial: Diagnosing the Dead-The Retrospective Analysis of Genetic Diseases." *Journal of the Royal College of Physicians of Edinburgh* 43 (2013): 11-14.

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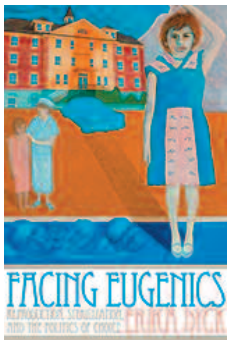
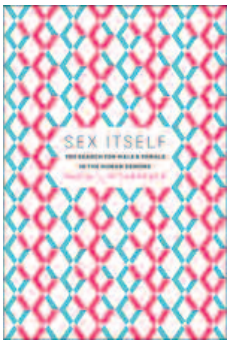
On the bookshelf

The following recently published books may be of interest to readers of the *Bulletin*:



Dyck, Erika. (2013). Facing Eugenics: Reproduction, Sterilization, and the Politics of Choice. University of Toronto Press.



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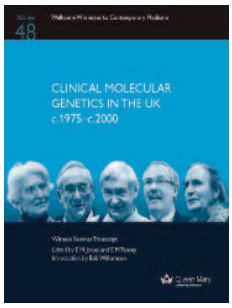

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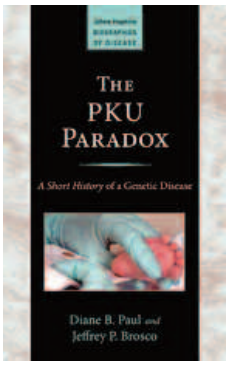

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