FOURTH INTERNATIONAL WORKSHOP ON GENETICS, MEDICINE AND HISTORY. GOTHENBURG, 11-12 JUNE, 2010

This two day workshop took as its principal theme:

The Early History of Human Molecular Genetics

41 people attended the workshop, which was held as a satellite meeting of the European Society of Human Genetics. Invited speakers included both historians and scientists, and came from across Europe and from North America, travel costs being greatly helped by grants from Wellcome Trust (087826) and from ESHG.

The principal reason for choosing the theme was the recognition that, while human molecular genetics is a relatively recent area by comparison with other aspects of human and medical genetics, or classical molecular biology, it has had an immense impact during this short time of around 25 years. Also rapid changes in laboratory technology and the predominant use of email, the Web and other electronic techniques make it especially vulnerable to loss of the essential primary sources.

It was recognised that a small workshop of this type could only be a beginning in the process of historical documentation and archiving, but it was felt that a combination of scientists from the field with historians and archivists could make a helpful start and identify some of the key issues.

The speakers, programme and abstracts are attached as part of this report; here the details of individual talks is not given, but some of the main topics covered and discussions arising are mentioned. The entire workshop was recorded and will be sent as a disc when available; also it is planned that a book, edited by the principal organiser, Christos Yapijakis, will be produced.

In the first session some of the beginnings of human molecular genetics were identified. Christos Yapijakis (Athens), in his opening introductory talk, showed that molecular concepts of inheritance were already being proposed in Hellenistic and Roman times. Soraya de Chadarevian (UCLA) showed how early work on haemoglobin and its structure, by Max Perutz, Hermann Lehmann and others in the 1960s, provided a foundation for the subsequent research that allowed the specific analysis of human genes. Correspondingly, Jan Witkowski (Banbury Center, Cold Spring Harbor), illustrated the impact of the Cold Spring Harbor Symposia in developing both the ideas and technologies that would underpin the future human molecular genetics. The value of the extensive documentation and archiving of all aspects sets an example to others.

In the second session, Tom Maniatis (New York), himself one of the key players in the development of human molecular genetics, described the principal discoveries in terms of advances in technology, such as the construction of DNA libraries, and DNA hybridisation and amplification. This was balanced by the presentation of Andrew Read (Manchester), who showed how the new research techniques and findings were first introduced into medical genetics services for important inherited disorders.

The next talks (Judith Friedman, Max Planck Institute, Berlin; Patrick Lestienne, Bordeaux), illustrated how the application of molecular approaches had resolved two important 'problem' areas in human genetics, that of genetic anticipation, with the apparent deterioration across generations explained by DNA instability, and the analysis of the mitochondrial genome and its maternally inherited disorders.

The history of the Human Genome Project was the focus of the next talk, by Ludmila Pollock (Cold Spring Harbor Library and Archives), who described an exciting international initiative (involving Wellcome Trust) to archive digitally as many documents relating to the Project as possible.

The day finished with a general discussion, on which were the priority targets for ensuring the preservation of the history of human molecular genetics. Liz Shaw, one of three Wellcome Trust staff at the workshop, described the Trust's current initiative involving cataloguing and digitisation of genetics records; the importance of interviews was also emphasised, and the problems associated with archiving of electronic records and correspondence discussed. The discussions continued over an excellent dinner!

Day two of the workshop began with two presentations from Mediterranean countries (Dimitris Loukopulos, Athens and Constantinos Deltas, Cyprus) on the applications of molecular techniques to carrier testing and prenatal diagnosis of haemoglobin disorders, showing the profound impact on the frequency of the disorder and the social acceptance of the approach in these populations. Describing the extensive restrictions placed on genetic applications as a consequence of the Nazi abuses, Heike Peterman (Nuremberg) placed these developments in a very different perspective. Peter Harper (Cardiff) then discussed the particular lessons to be learned from Huntington's disease both in terms of understanding its molecular basis and in molecular applications.

Returning to the theme of the Human Genome Project and its predecessors, Sue Povey (London) described the series of Human Gene Mapping Workshops held between 1973 and 1990, which set the stage for the Human Genome Project; everyone agreed that this initiative was important to archive and document historically. Likewise the account by Mary-Claire King (Seattle) of the research in the laboratory of Allan Wilson on human evolutionary

genetics, largely based on the new molecular techniques, illustrated another area of human molecular genetics with major impact.

In the afternoon, Bengt-Olle Bengtsson (Lund, Sweden), showed a film that he had edited, from the 1948 8th International Genetics Congress in Sweden, giving valuable images of many important geneticists involved and showing the importance of the prolonged interactions allowed by such congresses in those years. This film will soon be available on the Web.

The final presentation came from Walter Bodmer (Oxford) on the history of cancer genetics, showing how molecular approaches allowed both the isolation of key underlying genes and also the detection of those at high risk and the prevention of death by early intervention.

Looking ahead, an offer was made by Heike Peterman (Germany) to organise a further workshop in two years which would particularly involve German historians who have worked on the special issues of genetics in Germany as affected by the period of the Third Reich. This would seem to be a theme that will be of wide international interest also to geneticists and to social scientists, and it is hoped that planning such a workshop will begin soon. The ESHG Board has indicated it would support such a meeting as a satellite of the 2012 ESHG conference in Nürnberg. Subject to negotiation, the ESHG would probably provide administrative support and facilities, but would not fund the total cost of the meeting.

Meanwhile the general conclusion is that the present workshop has been both valuable and enjoyable, and that it has helped to put the subject of human molecular genetics firmly on the agenda for detailed historical studies and in particular for measures that will ensure the preservation of the abundant primary material that currently exists but is still in danger of loss.

The workshop organisers would again like to express their thanks to Wellcome Trust and to all others who helped in the planning and organisation of the workshop.