Meeting Report

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Rodney Harris
Manchester

Report of ESHG Satellite Meeting: EU Concerted Action on Genetic Services in Europe (CAGSE)

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European Union (EU) Concerted Actions are for the coordination of already on-going research in member states through scientific and administrative management support, organisation of meetings and workshops, exchange of staff, exchange of materials, centralised data handling and dissemination of results [1]. It is recognised that increasingly heavy demands will be placed upon health care systems by the elderly and those with chronic disorders. Consequently, much hope is placed on there being major progress in prevention of diseases, with the availability of better and less expensive technologies and new methods of care delivery.

It was in this context that the Concerted Action on Genetic Services in Europe (CAGSE) was funded under Biomed 1 as Health Services Research. CAGSE sets out to identify optimum means for delivering better prevention and health care resulting from the Human Genome Project in our multicultural and multilingual Europe. This work was initiated by a working party of the European Society of Human Genetics [2].

Introduction to CAGSE

Rodney Harris (project leader) in welcoming participants[1] to the meeting noted that CAGSE incorporates four main principles:

(1) *Prevention* of genetic disorders requires education, counselling and informed consent.

(2) The specialty of *medical genetics* is the essential *infrastructure* for clinical molecular advances, diagnosis and counselling.

(3) Many *other health care professionals* are involved in medical genetics and require appropriate training and continuing links with genetic centres.

(4) *National characteristics* have a profound influence and include type of health service and its economics, as well as historical, social and ethical factors.

Certification

There are still only four EU countries which officially recognize the specialty of medical genetics, in spite of the rapidly growing need for genetic counselling resulting from the Human Genome Project and increasing public awareness. There was general support for a participant who proposed that an important aim of CAGSE should be to establish formal certification for specialists in medical genetics in the EU and to provide factual data to demonstrate examples of good organisation/practice as well as poor organisation. It was agreed that different models for medical genetic services for individual countries may develop rather than one universal model.

[1] *Present:* Nicole Baumann, France; J.J. Cassiman, Belgium; M. Fellous, France; Rodney Harris, UK; Hilary Harris, UK; Lena Koch, Denmark; G. Lucote, France; Margareta Mikkelsen, Denmark; J.P. Moatti, France; M. Niermeijer, The Netherlands; Irmgard Nippert, Germany; P.C. Patsalis, Cyprus; Judith Rhind, UK; Giovanni Romeo, Italy: S.C. San Roman, Spain; D. Stemmerding, The Netherlands; Alistair Stewart, UK; C. Stoll, France; N. Tommerup, Denmark; Lisbeth Tranebjaerg, Norway. Apologies: J. Houghton, Ireland; Kare Berg, Norway; M. Kettner, Germany; C. Legum, Israel; E. Maher, UK.

Dr. Rodney Harris
Department of Medical Genetics
University of Manchester
St. Mary's Hospital, Hathersage Road
Manchester M13 0JH (UK)

S. Karger AG, Basel
1018–4813/94/
0024–0300$8.00/0

Partners then described the themes of CAGSE, genetic counselling by non-geneticists, the social dimension, medical genetics in relation to health services, laboratory genetics, primary health care, genetic epidemiology and south Europe.

Genetic Counselling by Non-Geneticists

Rodney Harris described the UK Confidential Enquiry into Genetic Counselling (CEGEN) funded by the Department of Health and with the collaboration of the medical royal colleges and many others. CEGEN follows the highly successful precedents of the National Confidential Enquiries into Maternal and Perioperative Deaths which rely on total confidentiality and the cooperation of clinicians who had cared for the patients.

CEGEN looks at the quality of genetic counselling given by non-geneticists – obstetricians, paediatricians, general surgeons and general physicians – with the goal of widening understanding of genetics in medicine and improving clinical standards and services. Of the seven disorders included[2], the Down's syndrome study was almost complete. Remediable faults related to systems of care rather than failings of individual doctors. These included delays in women booking with their general practitioner (GP) and between this and their first antenatal appointment at the hospital. Long delays between referrals resulted in women not being offered screening and prenatal diagnosis for Down's syndrome. Data were shown comparing prenatal diagnosis (PDN) uptake in Caucasians with that of non-Caucasian women as well as social class differences. There were difficulties in establishing why different social classes choose different PND options. One suggestion was the differing cultural views within different classes, as well as factors such as whether the mother was single, married or with a partner. However, there was once again clear evidence that delay in visiting the GP and antenatal clinic were important and potentially remediable deficiencies. Similar studies of inherited cancers and NTDs were well advanced and it was anticipated that published reports would alert doctors, nurses, managers and politicians to the needs of patients with genetic problems and their families.

The meeting agreed that CAGSE would investigate the possible introduction of comparable schemes in other EU countries (see notes on the presentation by J.P. Moatti below). Problems encountered, for example, in ascertainment and gaining cooperation of professionals would be highly instructive of differences in health care systems but also of societies and cultures.

The Social Dimension: Actors, Networks and Entrenchments

Lena Koch began by informing participants about the work carried out to date with Dirk Stemmerding and future plans for CAGSE in this area which had begun with a planning meeting in April 1994 in Twente, The Netherlands.[3]

She agreed with participants that sociological terms needed to be defined if cooperation with medical geneticists was to be fruitful and it would be important to keep the terminology relatively simple, otherwise much of the discussion/research time would be spent debating and agreeing on these definitions, rather than carrying out the research.

Entrenchment was identified as 'a process through which new technical options (in genetics) become viable and established practices in society'. *Actors* included professionals (researchers, clinicians), governments, patient organisations, funding organisations, churches, commercial firms, consumers and, of course, the media. *Need* was defined as 'a professional evaluation of the occurrence of disease and of the technical possibilities of diagnosis and therapy', while *demand* was 'a sociological concept/articulated by different actors, consumers, doctors and authorities', and depends on access to services, information on services and other factors. In society, need does not speak for itself, and demand must be articulated by social actors. *Attunement* is a measure of the quality of entrenchment. Questions which needed to be addressed in this area include 'to what extent does initial attuning take place?' and 'in what ways are relevant actors involved?' *Acceptability* was noted to be a sociological concept which must be articulated by actors in society through assessment studies, ethical committees, government legislation and public debate, and depends on historical, social, political and cultural factors.

The main *methodological* issues were (1) what is the object of research – what particular area should the

[2] The Events studied in CEGEN are: Down's syndrome births to women over 37 years; neural tube defect (NTD) births; cases of multiple endocrine neoplasia II; cases of bowel cancer in patients under 45 years; siblings with cystic fibrosis (CF); births of children with thalassaemia and new cases of haemophilia.

[3] *Present:* R Harris, Irma Nippert, Teresa Marteau, Matteas Kettner, Dirk Stemmerding, Lena Koch.

social aspect of the study address: genetic services, diagnostic practices, genetic technologies? (2) how is a *balance* achieved between analysis of baseline (existing services) and developments possible in the future, and (3) what is the best methodological design to collect this information – questionnaires and/or interviews?

Difficulties in deciding which aspects to study were noted as being (1) selection of persons or institutions – for example, heads of department would have different opinions to their junior colleagues; (2) selection of countries – which countries should be selected and why? and (3) selection of diseases/services.

Medical Genetics, Health Services and Economics

Jean-Paul Moatti outlined the areas which would be covered in CAGSE. These included a review of EU countries' economic policies supported by evidence from the economic literature.

The definition of *optimal criteria* for genetic screening would be calculated by cost effectiveness, cost benefit and decision analysis. The analysis of *determinants of diffusion* (entrenchment) of genetic services as dependent on the characteristics of the health care systems, by attitudes of the medical profession, by institutional and financial incentives, or barriers, and by patients' demand. Patients' attitudes and preferences needed to be gathered, possibly by doing a European comparative study. This could be achieved either by monographs covering each country, by case studies or by surveys of the medical professions. It was stressed that economic analysis should facilitate ethical choices and not the other way round. Economics allows the demonstration of choice, making them transparent. Additionally, when economic evaluations are made, the size of a laboratory and the optimal size of services and limits, for example, in the case of individual laboratory tests, need to be taken into consideration.

Moatti described a survey which he and colleagues had carried out among women at delivery in Marseille in 1990 as an example of the sort of comparative study which could be included in CAGSE and which would be compared with the UK CEGEN described above. The demographic and other features of the sample of 514 women were described. The reasons women above and below 37 years had not had amniocentesis were detailed. These were (1) women refusing amniocentesis; (2) physicians' negative counselling and (3) failure of the physician to offer and the woman to ask for amniocentesis. Cost and benefit studies of screening pregnancies for Down's syndrome, the use of 'willingness to pay' and the acceptability of termination of pregnancy were also described.

Laboratory Genetics in Europe

Niels Tommerup reported on laboratory information collected as part of a Europe-wide study by the ESHG Committee on Genetic Services. This had sought information from senior professionals and from the relevant official in the British Embassy in each country. He concentrated on validating the information for Norway and Denmark whose laboratories were well known to him.

He commented that there were instances where different organisations/authorities had listed the same laboratory under a different name, and information has thus been duplicated. Additionally some laboratories had extended the range of test/investigations and provided additional services which had not been recognised. It was felt that this information was an important first step and that the quality of information would be improved by CAGSE. Suggestions to improve the listings already gathered included the selection of 'regional' experts with much local knowledge able to contact each individual laboratory. Such regions would be limited in size to no more than 10 million population per expert. The information collected would be cross-referenced with other EU programmes which liaise extensively with such laboratories. Special attention would be given to the relation between national health services and private laboratories.

The benefit of collecting such information should be made explicit when asking individual laboratories for their cooperation.

Genetics and Primary Health Care

Martinus Niermeijer summarised the areas for inclusion by CAGSE as (1) *educational* programmes for GPs and genetic nurses; (2) standards relating to the offer and follow-up of presymptomatic testing; (3) *technical* infrastructure availability and (4) *government* policy matters in relation to public interest.

Specific areas within genetics which were considered for development in primary health care included (1) *counselling* for genetic disease in adulthood, cancer, hyperlipidaemias and dementias; (2) the *roles* of different types of doctors and nurses working with clinical geneticists, and (3) *screening* programmes for carriers of CF and maternal biochemical serum screening for NTD and Down's syndrome for all pregnant women.

Niermeijer outlined Dutch government policies and actual developments as examples of problems that genetic health care was facing in the Netherlands which would probably recur in other countries.

Hilary Harris described research funded by the Wolfson Foundation which is introducing family history taking and genetic counselling by GPs using, as a

vehicle, CF carrier screening at the first diagnosis of pregnancy in eight general practices in north-west England. Counselling for CF carrier testing together with a short family history is integrated into the first antenatal booking appointment at an average 8 weeks gestation. Evaluation of the programme at 1 month and at 1 year by questionnaire and structured interview in the patients' homes is carried out independently by two genetically trained nurses. Evaluation focuses on the acceptability of the timing, the person doing the counselling and the location in general practice. The evaluation is also concerned with whether patients felt that they had enough time with the doctor in a busy GP setting and whether women feel that they have been given enough time and information to make an informed choice. The special problems of counselling CF carrier couples with no experience of affected individuals is tackled jointly with the Department of Medical Genetics. The whole programme is seen as a model for many counselling and screening opportunities with the additional advantage of beginning the process of educating the primary health care team.

With the assistance of data collected for the Dutch College of General Practitioners [3], a comparison was made of the features of primary health care in each country which might be relevant to medical genetics. Patients are registered in primary care in seven EU countries offering more opportunities for continuing care and the long-term maintenance of medical records. Seven countries have systems of group practice which are more conducive to the organisation of genetic screening than single-handed practice. Again, in seven countries, the GP clearly acts as the gatekeeper to secondary health care, an important consideration if patients are not to use services inappropriately and are to benefit from effective multidisciplinary intervention. Overall workload factors will contribute to the success of GP involvement: the doctor:patient ratios varied from 1:588 (Belgium) to 1:2,319 (The Netherlands). The clinical implementation of new genetic technologies will require the participation of genetically informed young GPs. The best opportunity to achieve this will be to introduce clinically relevant genetics into the GP postgraduate training programmes that exist in 10 countries (although these programmes are not mandatory in all of them). Only four countries have practice nurses working with the primary health care team.

Genetic Epidemiology with Special Emphasis on Southern Europe

Giovanni Romeo stressed the importance of *certification* in the speciality of medical genetics and the need for training in genetics for general doctors. He demonstrated for Italy the geographical spread of services both public and private indicating the nature and range of services each institution offered for biochemical genetics, cytogenetics and molecular laboratories.

He suggested that Italy was an excellent example for the study by CAGSE of the organisation of the delivery of genetic services in southern Europe, involving six areas. (1) *Analysis of existing censuses* of medical genetics clinics, laboratory services for biochemical genetics, molecular genetics and cytogenetics. (2) *Audit of the validity* of these censuses through (a) questionnaires, (b) comparison with other Concerted Actions and (c) site visits by ad hoc CAGSE subcommittees. (3) *Evaluation of training programmes* in (a) clinical genetics through the European school of Medical Genetics and (b) laboratory genetics services through the courses in Preventive Medicine held yearly in Sestri Levante, Italy. (4) A *regional test case* in the Liguria region of Italy (population: 1.5 million). (5) *Comparative data* for other south European countries (Greece, Spain, Portugal). (6) *Focal questions*: (a) how to train medical and paramedical personnel involved in genetic services in southern Europe; (b) how to certify this training – the need for a European Board of Medical Genetics; (c) how to organise quality control tests for laboratory services at the European level. A workshop was organised on genetic services by the Hellenic Society of Human Genetics during the First Balkan Conference on Human Genetics in Thessaloniki (31 April – 3 September 1994) at which representatives from Bulgaria, Greece, Romania, Turkey and parts of ex-Yugoslavia agreed to contribute data to CAGSE.

Future Plans for CAGSE

During the next 3 years, workshops covering the above areas will be organised by partners and these will be published as monographs, reports in journals and formal reports to Brussels.

References

1 Thiers G: Looking forward to the next Biomedical Health Research Programmes. Eur Union Biomed Health Res 1994;5(1):1.
2 Harris R, Rhind JA: The speciality of clinical genetics: European Society of Human Genetics survey. J Med Genet 1993;30:147–152.
3 Boerma WGW, de Jong FAJM, Mulder PH (eds): Health Care and General Practice across Europe. Netherlands Institute of Primary Health Care, 1993.