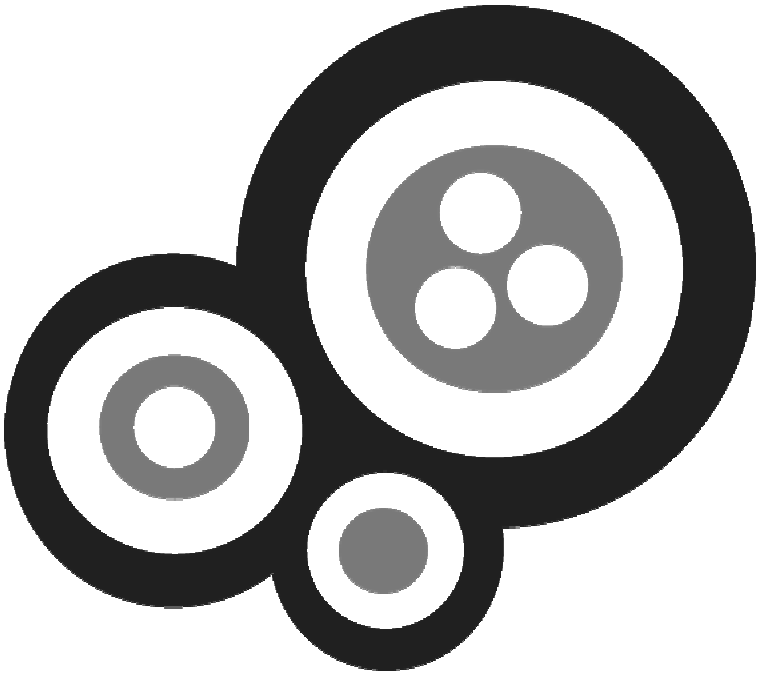

BREAKOUT 5

ROLE OF NGOS AND NETWORKS IN PREVENTION OF BIRTH DEFECTS AND GENETIC CONDITIONS

Moderators:
Ysbrand Poortman – The Netherlands
Rumen Stefanov – Bulgaria

PRECONCEPTION HEALTH CARE DELIVERY IN MIDDLE-INCOME COUNTRIES: STRATEGIES TO INTEGRATE ALONG THE CONTINUUM OF CARE

Mary-Elizabeth Reeve

Global Perinatal Health Education, March of Dimes Foundation, USA

Optimizing preconception health will improve maternal and neonatal health by reducing the risk of poor birth outcomes, including birth defects and preterm birth. In support of its goal to reduce birth defects and preterm birth worldwide, March of Dimes Global Programs is partnering with organizations in a number of middle-income countries to carry out projects aimed at improving both preconception and perinatal health. This presentation will provide a brief overview of preconception health activities within current Global Programs initiatives and explore ways in which they could be adapted for implementation within the countries of Central and Eastern Europe. Global Programs activities demonstrate how preconception health care can be integrated within the broader framework of maternal and neonatal healthcare services. Strategies include improving awareness of the importance of preconception care among patients and their families by creating and disseminating health messages to patients, their families and the community; integrating preconception health care into professional education and training through curricula in medical genetics and in preconception health; integrating preconception health care into clinical services by providing for a preconception wellness visit for couples, and by strengthening medical genetics services. These strategies position preconception health care as an important link in the continuum of care. Extending the maternal, newborn and child continuum to include preconception health supports a life-course perspective on care.

CONTRIBUTION OF THE BULGARIAN INFORMATION CENTRE FOR RARE DISEASES TOWARDS PREVENTION AND TREATMENT

Rumen Stefanov

Information Centre for Rare Diseases and Orphan Drug, Bulgaria

The information centres for rare diseases and orphan drugs have a significant as promoters of awareness, research and policy at national level. The Information Centre for Rare Diseases and Orphan Drugs (ICRDOD) in Bulgaria is the first Eastern European free educational and information service, providing personalized replies to requests from patients, families and medical professionals. The Centre highlights the importance of working simultaneously in 6 main directions – information, education, awareness, support, networking and lobbying. In a result, currently a National health plan for rare diseases is in the process of review and approval in Bulgaria. The specific activities of the Centre towards prevention and treatment are presented as a successful model for integral rare disease approach that can be adapted and applied also in other countries.

THE ROLE OF PATIENT ORGANISATIONS REGARDING NATIONAL PRECONCEPTION POLICY: THE DUTCH EXAMPLE

Elsbeth van Vliet-Lachotzki

Community Genetics VSOP, the Dutch Genetic Alliance, Vredehofstraat, The Netherlands

VSOP, the Dutch Genetic Alliance, is an umbrella organisation of approximately sixty national, disease-linked, parent and patient organisations, concerned with genetic and/or congenital disorders. The VSOP has been active in preconception care for over 30 years, by influencing policy, stimulating research and the development of preconception information. Some examples of these activities are: The first national government sponsored mass media folic acid campaign in 1995, the study preconception counselling in general practice and the publication: ``Preconception care: a good beginning``, by the Health Council of the Netherlands.

During these years parents and patients have often concluded that relevant information on preventive and reproductive choices was not available, or was brought to their attention too late. Addressing risk factors before conception could be the most effective strategy to improve the outcome of the pregnancy. For couples with genetic conditions it offers more reproductive choices.

Many parent and patient organisations are pioneers in the field of preconception. With their personal experience and involvement they aim to safeguard others for the consequences of information being either insufficient or too late.

Patient organisations have the unique position to emphasize the importance of informed choice and preventive options, and simultaneously point to social and economic solidarity for people with hereditary and congenital disorders. Nevertheless, it remains important to realize that hereditary and congenital disorders can not always be prevented. Within such a sensitive ethical field vigilance endures. This can only be guaranteed when the further implementation of preconception policy will fully comprise a patients perspective.

Anyone who realizes how much lifelong care, in every sense, can be prevented through preconception care, will ask him or herself why we would wait any longer. Do all the discussions counterbalance the responsibility for the consequences of doing nothing?

THE CONTRIBUTION OF THE CENTRAL & EAST EUROPEAN GENETIC NETWORK (CEEGN) TOWARDS PREVENTION AND TREATMENT

Susan Szendro, Selena Freisens, Ysbrand Poortman, Teresa Matulka,
Rumen Stefanov, Barbara Czeizel, Grzegorz Wegrzyn,
on behalf of the CEEGN members

Research and Prevention for Families Foundation, Budapest, Hungary

The Central and Eastern European Genetic Network (CEEGN) is a young, patient-driven network organization, consisting of patients, parents, individuals, doctors and scientists (Medico Scientific Committee) from Central and Eastern European countries (CEE). CEEGN functions in line with the European Genetic Alliances' Network (EGAN) and the International Genetic Alliance (IGA).

CEEGN has the main focus on promoting research and development of medicines in countries of Central and Eastern Europe, and on encouraging CEE patients involvement and active participation in research into the causes and cures of genetic diseases. CEEGN streamlines its activities towards early detection and accurate diagnosis, genetic counseling, and ultimately disease prevention in CEE countries.

CEEGN is currently the only network organization from Central and Eastern European Region, with focus on the common needs of CEE patients with genetic disease and their families, related to the backward position of CEE countries. Through supporting the local genetic patient organizations in the countries of CEE, CEEGN ensures that the information from EU has reached various people from those countries who are interested and active in the genetic field.

CEEGN takes part in the EU projects "EuroGenGuide" and "Eurogentest", where it aims to become the main channel for distribution of the outcomes to the patients and scientists in Central and East European countries. CEEGN aims to partner in various major EU/DG projects in 2008 and beyond such as "Roadmap to treatment" and "Patients participation in clinical research". Through these activities, the CEEGN members are able to communicate their opinions and experiences on issues of common interest, and ensure that their country and voice of their people with genetic disease will be heard in the EU.

CEEGN intends to collaborate and partner in different projects of mutual interest with other organizations with similar goals. Finally, CEEGN will refrain from working in the fields where other organizations are already active and will avoid duplication of efforts.

EUROGENGUIDE: PATIENT LED EDUCATION AND DEVELOPMENT FOR GENETIC TESTING IN RESEARCH AND MEDICINE

Alex McKeown

Genetic Interest Group, UK

Others affiliated to EuroGenGuide: VSOP, Policlinico Milan, University of Muenster, WANDA, CEEGN, Alzheimer Europe, Heart EU, Fighting Blindness Ireland, European Federation of Biotechnology, Health Coalition Initiative, Patient Centred Solutions.

EuroGenGuide is a EC funded project aiming to provide information about genetic testing and research to patients, the public and health professionals. The information will take the form of a „manual" with two halves. One half will contain information for patients or people considering taking part in research, and the other half will contain information for health professionals, such that they can provide the right advice and information to their patients.

There is a disparity in Europe between the relatively widespread availability of genetic tests and therapies resulting from research, and the uneven distribution of information and access to it for those affected by genetic disorders. EuroGenGuide aims to reach into areas where clinicians and patients know little about genetics and what help is available to them in taking advantage of new technologies, and help to resolve this disparity. Both halves of EuroGenGuide will dovetail into a comprehensive document and website for use by all, and which will help to generate a robust model of informed consent for those making decisions based on their options in respect of genetic testing or research.

The project is relevant to the conference, as one of the aims of EuroGenGuide is to ensure that those incapable of making autonomous decisions of their own due to a genetic condition, receive care that is in their best interests, and this depends on their carers being able to access relevant information. In the case of chronic conditions such as Parkinson's or Alzheimer's, carers will face difficult decisions about care when help may be available. Similarly, the very young and indeed, unborn, are unable to make rational choices and therefore require others to act in their interests. Whether to carry out predictive or diagnostic tests and make difficult decisions that might follow from a positive result, is a situation in which parents will need access to all the relevant information. Given these facts, EuroGenGuide wishes to attend the conference to build a network of contacts for dissemination of the guide in eastern Europe, and for feedback in the interests of improving upon the material as the project progresses.

CAPACITY BUILDING FOR THE TRANSFER OF GENETIC/GENOMIC KNOWLEDGE INTO PRACTICE AND PREVENTION: THE CAPABILITY INTERNATIONAL COLLABORATIVE NETWORK

Irmgard Nippert

World Alliance of Organizations for the Prevention and Treatment of Genetic and Congenital Disorders, Universitaetsklinikum Muenster, Muenster, Germany

The number of genetic tests is growing each year and increasing knowledge about gene-disease associations will lead to new opportunities to apply genetic/genomic knowledge in practice and prevention. Before genetic tests are introduced into general practice the benefits of their use must be evaluated. Worldwide, health care systems are facing the same challenges: 1) The need to develop an evidence-based evaluation process for genetic tests or other applications of genomic knowledge in transition from research into practice. 2) The need for capacity building to enable health care systems to make effective use of genetic/genomic applications with proven clinical utility.

CAPABILITY (http://www.capabilitynet.eu) is a 3-year model project developed by the European Network of Excellence: Genetic Testing in Europe – Network for test development, harmonization, validation and standardization of services (EuroGentest) (http://www.eurogentest.org), the World Alliance of Organizations for the prevention and treatment of genetic and congenital disorders and by leading experts from: Argentina, Egypt and South Africa, the latter being currently engaged in major development projects to integrate genetic services in primary care and prevention in their countries.

CAPABILITY will:

- develop an analytic framework for evidence-based genetic test evaluation including the domains: efficacy (evidence of utility in controlled settings) and effectiveness (evidence of utility in real settings),
- identify priorities for capacity building by a systematic needs assessment survey and
- validate the project's approach by means of a demonstration project.

CAPABILITY's overall objectives are to contribute to the efforts to establish and sustain a worldwide harmonisation process for quality standards for the integration of genetic test/genomic knowledge applications into practice and prevention and to serve as a model project for successful, sustainable collaboration between EU research centres and centres from non-EU middle and low income countries.