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Identifying Regional Priorities in the Area of Human Genetics in SEAR

*Report of an Intercountry Consultation
Bangkok, Thailand, 23-25 September 2003*

WHO Project: NEP NCD 001



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1. INTRODUCTION

An intercountry consultation was held at Bangkok, Thailand from 23 to 25 September 2003 to identify regional priorities in the area of human genetics. The meeting was organized as a follow-up of the recommendations made by the 26th Session of the WHO South-East Asia Advisory Committee on Health Research held at Thimphu, Bhutan in April 2001 and aimed at initiating a human genetics programme in the South-East Asia Region.

The inaugural address of Dr Uton Muchtar Rafei, Regional Director, SEARO was read out by Dr Somchai Peerapakorn Ag. WR Thailand. The Regional Director stated that the countries of the SEA Region having entered epidemiological transition, were suffering from the double burden of communicable and noncommunicable diseases. Although the burden of infectious diseases was still large, cardiovascular diseases, cancer, mental illness, diabetes mellitus and common genetic diseases were increasingly emerging as a major health challenge. The progress in human genetics that occurred in the developed countries had little impact in the developing countries where the genetic services are negligible. In most countries of the Region these services were at an early stage of development and in many had yet to be established. There were many barriers including lack of expertise and general awareness. Genetic services were incorrectly perceived as expensive, concerned with rare diseases, and considered mainly diagnostic rather than preventive and therapeutic.

2. OBJECTIVES

The following were the objectives of the Consultation:

- (1) To identify regional priorities in the area of human genetics in the SEA Region;
- (2) To map centres of excellence in the area of identified priorities in human genetics in the Member Countries of the SEA Region;
- (3) To discuss a draft format for standardized profile of national centres of excellence in the area of identified priorities in human genetics in the Region, and
- (4) To develop a framework for networking of centres of excellence in the Region in the area of human genetics.

3. WHO GLOBAL AND REGIONAL HUMAN GENETICS PROGRAMME

Dr V Boulyjenkov, MNC/HGN HQ stated that the reduction in the mortality and morbidity caused by communicable diseases and malnutrition resulted in recognition of congenital and hereditary genetic diseases as a major health burden. Due to recent progress in molecular genetics, it was evident that inherited predisposition was also important in a number of common diseases of later life, such as coronary heart disease, hypertension, diabetes mellitus, and some rheumatic, oncological and mental illnesses. He introduced the global human genetics programme (HGN) of WHO. The programme was mapping genetic approaches to the prevention and control of diseases and provided expertise and technical assistance to countries in their efforts to establish and manage national programmes. Genetic services could be delivered effectively to the community only if information and screening were integrated into primary health care. He informed that thalassaemia control programmes were being developed in a number of European countries, as well as in Brazil, China, India, Myanmar, Pakistan, Thailand, Tunisia and Turkey and sickle cell disease (SCD) programmes had been established in Cuba, Guadeloupe and Jamaica.

He referred to the "WHO Guidelines on Ethical Issues in Medical Genetics", the primary purpose of which is to assist policy-makers, officials, practitioners and other health workers in ensuring that genetic information and services are introduced into the broad medical practice in ethically acceptable ways. The broadening role of human genetics in research and health systems development raises an increasing need to examine the concomitant ethical, legal and social issues (ELSI), especially surrounding intellectual property rights, gene patenting, genetic testing and screening, pharmacogenetics, DNA banking and database development.

Dr J Leowski, NCS SEARO reviewed regional activities in human genetics. He said that the Resolution RC48.R3 on Prevention, Control and Treatment of Thalassaemia adopted by the Regional Committee in 1995 gave an important boost to this area as it took cognizance of the magnitude of this problem in some countries. Since then India, Indonesia, Sri Lanka and Thailand strengthened national capacity to control thalassaemia and a comprehensive national thalassaemia programme was established in Maldives with WHO support.

He referred to the two scientific debates on (i) Regional Perspectives in Human Genetics and (ii) Health Research on Prevention and Control of Thalassaemia held during the 26th and 28th Sessions of the SEA Advisory Committee on Health Research in 2001 and 2003 respectively. The WHO Advisory Committee on Health Research report on "Genomics and World Health" published in 2002 addressed a number of key issues related to utilization of genome technology in reducing inequalities in health, developing health care innovations and improving prevention, diagnosis and management of diseases. The regional input to this global document was provided through a consultation organized by the Thai Health Research Forum. He concluded that genetic diseases were not rare, some of them were of public health importance, and introduction of simple, effective and affordable approaches was the need of the hour. Countries should have equal access to knowledge of recent technologies. Thalassaemia control could be taken up as a model programme in Member Countries to start with. Mapping centres of excellence, assessing the availability of genetic services in the Region and addressing the ethical, legal and social implications of the genetic services and research should receive high priority in the SEA Region.

Dr Vasantha Muthuswamy from Indian Council of Medical Research, India provided an overview of the prevalent situation in the SEAR countries. Genetic disorders, which remained largely unrecognized and underestimated, imposed a heavy burden on the population. This burden was further compounded by a complete lack of awareness at all levels, absence of screening programmes and improper record keeping. In most of the countries of the Region genetic services were almost non-existent in government hospitals and only confined to a few centres in the private sector. She said that none of the countries of the SEA Region had any newborn screening programmes for genetic diseases. Clinical genetics was not part of the curriculum in medical colleges and most physicians had poor knowledge of the latest developments. The training of other health professional groups including nurses and technicians was also poor and the expertise was confined to few medical geneticists concentrated in academic institutions and/or in private practice. Little attention has also been given to educate society.

The results of the human genome project led to remarkable advances in developed countries, but had almost no impact in the developing countries. With a rise in the burden of noncommunicable diseases including genetic disorders, there was a need to develop genetic services such as screening, prenatal diagnosis, carrier detection, and genetic counselling. Haemoglobinopathies constituted a major public health problem in South-East Asia.

She informed the participants that a few countries like India, Indonesia and Thailand were working on models for the control of haemoglobinopathies, and suggested that common genetic disorders such as neural tube defects, Down syndrome and haemoglobinopathies could provide the point of entry for introducing genetic services in the Member Countries. Screening for inborn errors of metabolism could also be introduced initially for common genetic diseases like phenylketonuria and congenital hypothyroidism. Carrier detection, prenatal diagnosis and genetic counselling services offered to the families and individuals at risk by adequately trained counsellors could also go a long way in addressing the problems imposed by the genetic disorders. She further emphasized the need for capacity building, establishing networks and fostering collaboration at regional and global levels.

Dr Sawat Ramaboot, CHP SEARO referred to the major advances in early diagnosis and screening technologies accomplished during the past decade. He stressed the importance of WHO's role in supporting countries in developing comprehensive strategies for the control of genetic disorders with an emphasis on prevention. Primary prevention of nongenetic diseases starts after birth, while primary prevention of genetic diseases requires intervention much earlier, even before marriage.

There were numerous genetic diseases and countries had to prioritize which should be given attention first. Criteria for prioritization may include prevalence of disease, availability of reliable and simple technologies for diagnosis, screening, and acceptable prenatal diagnosis. Implementation of the programme should be delivered through the existing health systems. The reproductive health programme could be considered as the most appropriate entry point for the control of genetic diseases. Implementation could not be successful if carried out by a single professional group or an institute. There was a need for collaboration between several sectors backed by governmental commitment.

4. SELECTED REGIONAL PRIORITIES IN HUMAN GENETICS

4.1. Inborn Errors of Metabolism, Mental Retardation and Malformations

Dr Shubha R Phadke, Sanjay Gandhi Post Graduate Institute of Medical Sciences, Lucknow, India said that inborn errors of metabolism, mental retardation and congenital malformations constituted a major group of genetic

disorders requiring diagnosis, management and genetic counselling. Over the last few decades, a few genetic centres had come up in the SEA Region, providing clinical, laboratory and prenatal diagnostic services. Strengthening these centres and establishing genetic units in medical colleges was essential for bringing up the level and availability of clinical genetic services. She stressed the need to establish or improve clinical genetics services including laboratory diagnosis, neonatal screening, screening of pregnant women for Down syndrome, training on targeted ultrasonography for prenatal detection of malformations, short-term training of obstetricians and paediatricians in counselling, identification of common genetic disorders and timely referral to genetic centres, incorporation of medical genetics in undergraduate and postgraduate courses and networking of genetic centres providing different types of diagnostic services.

4.2. Control of Thalassaemia

Dr Wanchai Wanachiwanawin, Division of Haematology, Mahidol University, Thailand discussed the measures to be taken in order to control thalassaemia as a model for developing clinical genetic services. He apprised the participants about the current situation in the Region and the future projections. He said that the number of estimated thalassaemia patients in Thailand was in the range of 523 750. He described the pathophysiology of thalassaemia and other haemoglobinopathies, carrier frequencies of thalassaemia of which β -thalassaemia had a frequency of about 3-9%, and clinical symptomatology of haemoglobin disorders. He emphasized the need for developing the control programme because of the magnitude of the problem, changing demography, increased demand for blood transfusions and other resources and the limited accessibility of curative services.

Dr S Fucharoen, Mahidol University, Nakornpatham, Thailand discussed the epidemiology of thalassaemia in Thailand and the results of the molecular studies identifying various mutations. He said that a national control programme of thalassaemia was necessary to provide the best treatment to patients and prevent births of new cases. The programme on the prevention and control of thalassaemia in Thailand started in 1994 involving the Ministry of Public Health, Mahidol University and the Thalassaemia Foundation. The Thailand Thalassaemia Network (TTN) strives towards developing and strengthening research plan, human and economic development plan and coordinates national registration, service delivery and research. In 1997, a pilot programme for screening of pregnant women carried out in 17 provinces

helped in decreasing the number of new births by 10% and in 1998, the plan was extended to the whole kingdom. The thalassaemia programme was integrated into the existing health care system in 2000 in all provinces.

4.3. Ethical Issues in Genomics - SEAR Perspective

Dr A Wibowo, EIP/RPC SEARO informed that rapid advancements in medical technology and epidemiological transition helped in bringing genetic and hereditary diseases into focus. This had important ethical, legal and social implications. Human genetics had the power to predict the health of individuals and the potential to alter a diversity of populations. Informed consent was a must for conducting genetic testing/screening. At the same time, confidentiality of results was to be maintained, as the predictive power of medical genetics might be misused to discriminate against or stigmatize individuals. On the other hand, the access to treatment of genetic disorders may be limited only to those who could afford to pay, and thus would widen health inequalities. On the issue of medical termination of pregnancies affected by serious genetic disorders, Dr Adik Wibowo said that Member Countries should make their own decisions whether the practices were compatible with their laws, culture, religion, and traditions. Genetic information could also be misused for sex selection. India had strong regulations to prevent this practice. A situation analysis on ELSI in human genetics/genomics in some SEAR countries was needed.

4.4. Teaching and Training in Human Genomics and Genetics

Dr Shubha Phadke, pointed out that advancements in genetics had changed the scenario of medicine. To keep up with these advances, it was essential to improve the facilities for teaching and training. Various types of educational programmes were needed to develop necessary manpower. These included training in clinical genetics for medical practitioners, training in clinical and laboratory genetics for medical geneticists and short-term training courses in clinical genetics for paediatricians, obstetricians and other clinicians dealing with common genetic problems. Technical training programmes for technicians in molecular genetics, cytogenetics and biochemical genetics was also needed. Counselling training for genetic service providers was a must. Nurses, psychologists and medical staff with other background could be trained in genetic counselling. The recent developments in genetics should be incorporated in the undergraduate and postgraduate medical curriculum.

5. COUNTRY PRESENTATIONS ON NATIONAL PRIORITIES IN THE AREA OF HUMAN GENETICS AND AVAILABILITY OF CLINICAL GENETIC SERVICES

5.1. Bangladesh

Although there was no reliable epidemiological data on prevalence of genetic diseases in Bangladesh, it was observed that the common diseases included the hereditary haemoglobinopathies like thalassaemia and haemoglobin E disorder. Rough estimates suggested that there were about 3.6 million carriers of thalassaemia and more than 2000 thalassaemic children might be born every year. The priorities identified included screening for common genetic disorders like thalassaemia, Down syndrome and haemophilia, provision for genetic counselling services, prenatal diagnosis, and creating public awareness to prevent marriage of carriers. The importance of setting up genetics/molecular lab in government medical colleges, manpower development and training, introduction of advance courses on medical genetics need to be stressed. Treatment of thalassaemics was available at government hospitals and the Thalassaemia Foundation of Bangladesh showed keen interest in supporting the cause. Genetic counseling was provided to individual families by the clinicians.

5.2 Bhutan

The Royal Government of Bhutan and the Ministry of Health were fully cognizant of the increasing global and regional interest in the area of genetics and genomics. While recognizing the need to invest time and efforts in understanding the potential of such services, Bhutan was currently not in a position to initiate such services due to varying priorities and resource limitations. However, it was important to initiate a review and appraisal of needs related to genetic services and promote a better understanding among policy-makers and health professionals. Specialized genetic services were not available at present and patients requiring attention were referred to centres outside the country, mainly to India and Thailand. Bhutan looked forward to assistance from more developed countries of the Region with the support of WHO to initiate activities in this area.

5.3 India

India is witnessing an accelerating shift towards noncommunicable diseases. Congenital malformations and genetic disorders were the third commonest cause of mortality in neonates in cities. Estimates for genetic disorders showed that 495 000 infants with congenital malformations, 390 000 with G6PD deficiency, 21 400 with Down syndrome, 9 000 with β -thalassaemia, 5 200 with sickle cell disease and 9 760 with amino acid disorders were born each year. The various factors influencing the high prevalence of these disorders included consanguineous marriages, high birth rate, improved diagnostic facilities, poor governmental support facilities, and lack of expertise in genetic counselling. A task force on human genetics had been set up at the Indian Council of Medical Research and the Department of Biotechnology.

A nation wide network of genetic centres capable of providing diagnosis, counselling and antenatal diagnosis was the need of the hour. Similarly, training of laboratory personnel to strengthen expertise in cytogenetics and molecular techniques was required for areas such as inborn errors of metabolism and metabolic disorders. Establishment of a major national programme for the control of thalassaemia and haemoglobinopathies as well as neural tube defects (NTD) was urgently needed. Teaching and training of genetics was a major initiative in recent years, both in clinical genetics as well as genetic counselling. An ICMR multicentric study on genetic disorders in the year 1996 identified repeated abortions, chromosomal disorders and mental retardation to be the most common reasons to seek genetic counselling. The common single gene disorders observed in this study included thalassaemia, achondroplasia, haemophilia and G6PD. About 35-40 centres offered cytogenetic services, while only few clinics had advanced cytogenetic, biochemical, prenatal diagnostic services and counselling. A much larger number of genetic clinics were needed in the country to reach out to 200 medical colleges, 520 district hospitals, university genetics departments, private and charitable hospitals. Development of trained manpower also needed attention.

5.4 Indonesia

The human genetic programme in Indonesia started with formal university teaching and clinical chromosome analysis services introduced in the early 1960s. The fundamental research that led to generation of the early evidence for the molecular basis of the thalassaemias was conducted. The main feature

of the current human genetic activities was rooted on the vast human genetic diversity of the populations of the Indonesian archipelago. A nation-wide network of human geneticists initiated at the Eijkman Institute were involved in the study of population genetic structure and the association of genetic diversity to the distribution of single gene disorders such as thalassaemias and haemoglobinopathies, SEA ovalocytosis, and G-6-PD deficiency. Research on susceptibility to infectious diseases such as malaria and tuberculosis, complex polygenic disorders such as diabetes mellitus, obesity and preeclampsia, and drug metabolizing enzymes was carried out. Some fundamental research activities in molecular pathobiology investigating phenotypic expression of disease genes were also carried out. Harappan Kita hospital at Jakarta, University Diponegoro and Telogorejo Hospital at Semarang, IUC Biotechnology at Bandung and Eijkman Institute at Jakarta were some of the centres providing cytogenetic diagnostic services. However, only some of these centres provided genetic counselling. Studies on thalassaemia indicated that the carrier frequency ranged from <1 to 35 and was mostly due to 29 common mutations. It was necessary to establish a national thalassaemia control programme in Indonesia.

5.5 Maldives

Identified national priorities in human genetics were thalassaemia and other haemoglobinopathies, G6PD deficiency, congenital malformations, inherited disorders and lifestyle-related diseases with genetic background. Maldives was keen to develop a public health policy for promoting expertise in genetics, assess burden of genetic diseases in the country and collaborate with regional centres in harnessing expertise in clinical genetics. Constraints in healthcare delivery system included fragmentation of the population and difficult accessibility, low resources and lack of trained professionals.

It was important that services be provided and awareness increased regarding thalassaemia. Genetic counselling services needed to be made available to all carriers, couples with an affected child and those seeking prenatal diagnosis. The National Thalassaemia Centre (NTC) operated by the government, primarily concentrated on the care of thalassaemics across the nation. Thalassaemia was present in the formal curriculum of primary, secondary schools and tertiary training institutes for teachers and health care workers. Pre-marital counselling and screening for thalassaemia was a legal requirement. Prenatal diagnosis (PND) and medical termination of pregnancy (MTP) were legalized in 1999.

5.6 Nepal

Although there was no programme on genetics in Nepal, the country is realizing the need for integration of human genetics into the primary health care services by involving the primary health care doctors, paramedics and specialists other than geneticists. Information, education and training had to be provided to the communities as well as health professionals. It was also important to provide affordable and acceptable genetic tests and genetic counselling. At present, no clinical genetic services were available in the country. A retrospective analysis of haemoglobin electrophoresis carried out at a teaching hospital showed the presence of thalassaemia, sickle cell disease and abnormal haemoglobins in a few cases. The Nepal Haemophilia Society had about 200 members. The Government of Nepal supported introduction of need-based new technology in the country and established a National Forensic Science Laboratory. The laboratory was equipped with the basic facilities including equipment for serological and biological tests and had competent manpower. The laboratory carried out DNA profiling for preparing a database of the Nepalese population and genetic disease diagnosis. Gene therapy and pharmacogenomics were also considered as potential future areas.

5.7 Sri Lanka

The only place in Sri Lanka that provided a comprehensive clinical genetic service was the Human Genetics Unit of the Faculty of Medicine in the University of Colombo established in 1983. The centre provided genetic diagnosis and counselling along with a Genetic Awareness Programme (GAP) to sensitize and educate the public on preventive and management aspects of genetic disorders and birth defects. The centre offered genetic consultation, chromosomal analysis and cytogenetic tests. It carried out blood cultures, bone marrow cultures, foetal pathology assessment and preconception counselling for birth defects.

It also took the initiative to conduct undergraduate and postgraduate teaching programmes and was also pursuing active research in genetics. Information was available on counselling, appointments, education, teaching and training at the website – www.infolanka.com/org/genetics. There is a need for a comprehensive genetic unit that may provide diagnosis, screening and counselling for all genetic disorders.

5.8 Thailand

Although the magnitude of the problem is considerable, the epidemiological data on incidence/prevalence of genetic diseases is lacking. The community-based genetic services available at present included prenatal carrier screening and diagnosis of thalassaemia and newborn metabolic screening for congenital hypothyroidism and phenylketonuria. The Ministry of Public Health provided the regional laboratory services and clinical genetic services were mostly available in the medical school hospitals. The national priorities in Thailand included improving the availability of quality genetic services, provision of genetic education to health professionals, general public and schools. Resource allocation was a major area requiring greater attention.

Thalassaemia carrier screening and prenatal diagnostic facilities were available at antenatal clinics. New born metabolic screening facilities were also available for congenital hypothyroidism, PKU, CAH and inborn errors of metabolism. Patient as well as family-based genetic services were also in place. The Thai Medical Genetics Association also provided laboratory services including cytogenetic, molecular and biochemical services; prenatal screening; ultrasound; α -feto protein, and hCG facilities. There were three hospitals in the country offering clinical genetic services.

6. GROUP WORK

The group discussions on the priority ethical, legal and social, as well as research issues in the field of human genetics resulted in identification of the following areas for special consideration:

6.1. Ethical, Legal and Social Issues

- (1) Privacy and confidentiality issues in relation to patients, their families, health insurers, employers, legal systems;
- (2) Gender issues such as gender bias, sex identification, X-linked disorders;
- (3) Autonomy including informed consent;
- (4) Genetic testing and pretest counselling;
- (5) Legal issues in relation to prenatal diagnosis and selective abortion;
- (6) Social issues such as education on medical ethics, role of mass media and insurance companies, and
- (7) Consanguinity and its implications.

6.2 Research Priorities

- (1) Surveys on prevalence and availability/accessibility of genetic services for four major groups of genetic disorders: (a) β -thalassaemia, G6PD deficiency and other haemoglobinopathies; (b) Down syndrome and other chromosomal abnormalities; (c) congenital malformations; and (d) muscular dystrophies;
- (2) Community-based studies on attitude/ belief/knowledge on genetic diseases;
- (3) Clinical studies with a focus on developing simple screening/ diagnostic tests, diagnosis, new treatment modalities, and identification of environmental hazards;
- (4) ELSI related research;
- (5) Research on specific chromosomal disorders and on inborn errors of metabolisms;
- (6) Identification of new and specific mutations using molecular biology tools, and
- (7) Resource mobilization for research including twinning and funding from national and international agencies.

7. COLLECTION OF INFORMATION AND NETWORKING

The content of the draft proformas for collection of information on (i) Country Profile on Availability of Clinical Genetic Services, and (ii) Standardized Profile of Centres of Excellence in Human Genetics developed by the Regional Office prior to the workshop were presented, discussed in detail and finalized during the working group and subsequent plenary session. It was agreed to distribute the format for Country Profile on Availability of Clinical Genetic Services questionnaires to Member Countries. The selected genetic institutions represented in the Consultations agreed to fill the Standardized Profile of Centres of Excellence in Human Genetics and contribute to strengthening regional collaboration through networking. The major areas for networking identified by the participants included developing comprehensive models on medical genetic services linked to primary health care systems, developing capabilities to address ethical issues of genetics and promoting educational and training programmes.

8. CONCLUSIONS

After detailed deliberations the following conclusions were reached:

- (1) Human genetic disorders are assuming a prominent role in the health care services due to the observed transition in the disease pattern in the Region.
- (2) There is a lack of good epidemiological data on genetic disorders from the countries.
- (3) Human resources are limited in the SEA Region in the area of human genetics.
- (4) Public education is a core component for the success of the genetics programme.
- (5) Ethical, social and legal issues of genetics and genomics are emerging in a big way.
- (6) Thalassaemia has emerged as the commonest genetic disorder of public health importance in the SEA Region. It can be taken as the entry point for control of genetic disorders in the Region.
- (7) WHO has a pivotal role to play in promoting genetic services development, research and training in the Member Countries.
- (8) The proforma on (i) Standardized Profile of Centres of Excellence in Human Genetics and (ii) Availability of Clinical Genetic Services in the Member Countries, finalized by the consultation are useful instruments for collecting information required for planning of regional genetic programme.

9. RECOMMENDATIONS

The participants agreed to impress upon the concerned authorities to initiate appropriate action in the priority areas of human genetics recognized during the deliberations of the consultation and develop ways and means to achieve the same. In particular:

Member Countries should:

- (1) Utilize opportunity created by advances in genetics and genomics in addressing priority health issues.
- (2) Assess importance of genetic disorders as a cause of ill health.

- (3) Promote generation of public awareness about common genetic disorders and the potential for their effective prevention and management.
- (4) Strengthen collaboration and partnership with national and international NGOs in prevention and control of genetic disorders.
- (5) Conduct situational analysis on the availability of clinical genetic services and create an inventory through appropriate databases.
- (6) Identify and support national centres of excellence in the area of human genetics.
- (7) Promote capacity building for genetic research and for prevention and control of common genetic disorders.
- (8) Assess and address ethical, legal and social issues of genetics and genomics.

WHO should:

- (1) Develop and update guidelines on management of common genetic disorders.
- (2) Promote collaboration and networking of institutions in the area of human genetics and genomics while identifying centres of excellence.
- (3) Facilitate transfer of knowledge and technology in the area of human genetics and genomics.
- (4) Provide technical support in developing and strengthening prevention and control programmes for genetic disorders of public health importance.
- (5) Conduct regional assessment of availability of genetic services in the SEA Region.
- (6) Support research in areas identified during the consultation viz. thalassaemia, Down syndrome and other chromosomal abnormalities, congenital malformations, and muscular dystrophies.
- (7) Promote development of a quality control programme for biochemical, cytogenetic, hematological and molecular tests.
- (8) Facilitate human resource development in the area of genetics through existing programmes such as fellowships and new innovative approaches.

Annex 1

PROGRAMME

Tuesday, 23 September 2003

- 0830-0900 hrs Registration
- 0900-0945 hrs Inaugural Session:
- 1000-1230 hrs Plenary Session:
- Global Human Genetics Programme - *Dr V Boulyjenkov, MNC/HGN, WHO HQ*
 - Human Genetics Programme in SEAR – *Dr J Leowski, NCS, SEARO*
 - Review of Regional Priorities in Human Genetics in SEAR – *Dr V Muthuswamy, Indian Council of Medical Research, New Delhi, India*
 - Genetic Diseases and Primary Prevention – *Dr S Ramaboot, CHP, SEARO*
 - Selected Regional Priorities in Human Genetics: Inborn Errors of Metabolism, Mental Retardation and Congenital Malformation – *Dr Shubha Phadke, SGPGI, Lucknow, India*
- 1300-1500 hrs Plenary Session:
- Control of Thalassaemia: A Model of Clinical Genetics Services – *Dr W Wanachiwanawin, Siriraj Hospital, Mahidol University, Thailand*
 - Strategy for the Prevention and Control of Thalassaemia in Thailand – *Dr S Fucharoen, Thalassaemia Research Centre, Mahidol University, Thailand*
 - Ethical Issues in Genomics – SEAR Perspective – *Dr A Wibowo, EIP/RPC, SEARO*
 - Teaching and Training in Human Genomics and Genetics – *Dr Shubha Phadke, SGPGI, Lucknow, India*
- 1515-1700 hrs Plenary Session:
- Country Presentations on National Priorities in the Area of Human Genetics

Wednesday, 24 September 2003

- 0830-1030 hrs Plenary Session:
Country Presentations on Availability of Clinical Genetic Services
- 1045-1230 hrs Group Work on:
- Format for Standardized Profile of National Centres of Excellence (Moderators: *Dr J Leowski and Dr Vasantha Muthuswamy*)
 - Priority Ethical Issues in the Area of Human Genetics in SEAR (Moderators: *Dr A Wibowo and Dr S Fucharoen*)
- 1330-1500 hrs Plenary Session:
Reports of Group Work
- 1515-1700 hrs Group Work on:
- Format for Situational Analysis on Availability of Clinical Genetic Services (Moderators: *Dr J Leowski and Dr Vasantha Muthuswamy*)
 - Research Priorities in the Area of Human Genetics in SEAR (Moderators: *Dr S Ramaboot and Dr W Wanachiwanawin*)

Thursday, 25 September 2003

- 0830-1030 hrs Plenary Session:
Reports of Group Work
Finalizing Formats for Standardized Profile of National Centres of Excellence and for Situational Analysis on Availability of Clinical Genetic Services
- 1045-1230 hrs Plenary Session:
Framework for Regional Network of Centres of Excellence in the Area of Human Genetics
- 1330-1500 hrs Conclusions and Recommendations
- 1500-1600 hrs Closing Session

Annex 2

LIST OF PARTICIPANTS

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