

**HUMAN GENETICS POLICY GUIDELINES
FOR THE MANAGEMENT AND PREVENTION
OF GENETIC DISORDERS, BIRTH DEFECTS
AND DISABILITIES**

INDEX

	Page
PREFACE	1
TERMINOLOGY USED IN THIS DOCUMENT	4
EXECUTIVE SUMMARY	6
1. SITUATIONAL ANALYSIS	9
1.1 CURRENT POSITION	9
1.2 HISTORICAL OVERVIEW	9
1.3 LAY ORGANISATIONS	10
1.4 STAFFING	10
2. GOALS AND OBJECTIVES	11
3. PRIORITY MEDICAL GENETIC SERVICES	12
3.1 SERVICES PRIOR TO CONCEPTION	12
3.2 SERVICES DURING PREGNANCY	12
3.3 SERVICES AT BIRTH	13
3.4 SERVICES IN INFANCY AND CHILDHOOD	13
3.5 SERVICES IN ADOLESCENCE AND ADULTHOOD	13
4. STRATEGIES FOR PREVENTION OF GENETIC DISORDERS AND BIRTH DEFECTS	14
4.1 GENETIC COUNSELLING	14
4.2 PREVENTION STRATEGIES	15
4.3 PRE-CONCEPTION METHODS OF PREVENTION	18
4.4 ANTE-NATAL METHODS OF PREVENTION	18
4.5 POST-NATAL DIAGNOSIS	19
4.6 POPULATION SCREENING	19
5. DELIVERY OF GENETIC SERVICES	21
5.1 HOME	21
5.2 COMMUNITY LEVEL CARE	21
5.3 DISTRICT SUB- STRUCTURE OF DEPARTMENT OF HEALTH ...	22
5.4 REGIONAL LEVEL	23
5.5 PROVINCIAL DEPARTMENT OF HEALTH	24
5.6 NATIONAL DEPARTMENT OF HEALTH	25

6.	EDUCATION AND TRAINING	26
6.1	MEDICAL SCHOOLS	26
6.2	GENETIC COUNSELLORS	28
6.3	NURSES	29
6.4	MEDICAL SCIENTISTS AND TECHNOLOGISTS	32
6.5	HUMAN GENETICS IN SCHOOL EDUCATION	33
7.	LABORATORY SERVICES	34
7.1	NATIONAL REFERENCE LABORATORY (NRL)	34
7.2	CLINICAL DIAGNOSTIC LABORATORY SERVICES	38
7.3	RESEARCH AND DEVELOPMENT, TEACHING AND ACADEMIA .	43
7.4	FUNDING	44
8.	EVALUATING A HUMAN GENETICS PROGRAMME	45
8.1	DATA COLLECTION	45
8.2	MONITORING OF BIRTH DEFECTS IN THE IMMEDIATE POST BIRTH PERIOD	46
8.3	OBJECTIVES AND INDICATORS FOR HUMAN GENETICS	48
9.	ETHICAL GUIDELINES	49
9.1	GENERAL ETHICAL GUIDELINES IN MEDICAL GENETICS	49
9.2	ETHICAL PRINCIPLES FOR GENETIC PROFESSIONALS	50
10.	OUTREACH CLINICS/HUB AND SPOKE MODEL	52
11.	FINANCIAL IMPLICATIONS	53
11.1	CURRENT COSTS	53
11.2	PLANNED COSTS	56
12.	SUMMARY AND CONCLUSION	59
13.	REFERENCES	60
14.	CONTRIBUTORS	62
APPENDIX	64

**ANNEXURE - REPORT OF THE SUBCOMMITTEE ON GENETIC
LABORATORY SERVICES**

HUMAN GENETICS POLICY DOCUMENT FOR THE MANAGEMENT AND PREVENTION OF GENETIC DISORDERS, BIRTH DEFECTS AND DISABILITIES

PREFACE

Health and welfare is a basic right and this includes the individual with a genetic disorder or birth defect. Section 27 of the South African Constitution guarantees everyone the right of access to health and welfare services. Section 27 part 2 reads *“The State must take reasonable legislative and other measures within its available resources, to achieve progressive realisation of each of these rights”*.

Free health care for all children under the age of six years and for pregnant women was made available under the Reconstruction and Development Programme (1994). Prevention and promotive health programmes for mental health and care of the disabled (the blind, deaf, and the physically and mentally disabled) contribute to meeting basic health care needs. As a signatory to the United Nations Convention on the Rights of the Child (CRC), South Africa is required to provide health and welfare services for the disabled child. Article 23 of the CRC specifically deals with the physically or mentally disabled child. Section 4 of this Article reads *“States and parties shall promote, in the spirit of international co-operation, the exchange of appropriate information in the field of **preventive health care and of medical, psychological and functional treatment of disabled children** including dissemination of, and access to, information concerning rehabilitation, education and vocational services, with the aim of enabling state parties to improve their capabilities and skills and to widen their experience in these areas. In this regard, particular account shall be taken of the needs of **developing countries**”*.

In December 1997, the white paper on an Integrated National Disability Strategy was launched from the Deputy President’s Office. It provides guidelines to government departments on how to include disability related programmes in their functions. The recommendations made in this document for the Department of Health relate to *“**prevention, health care and rehabilitation of people with disabilities**”*.

Genetic services are essential to all levels of medical care including pre-marital counselling, family planning, pre-conceptual and pre-natal care, and in paediatric,

adolescent and adult medicine. Genetic services can be fully integrated into Primary Health Care (PHC), as advocated by the World Health Organization (WHO), where the core principles are concerned with equity, efficacy, effectiveness, community participation, and improving the health and well being of populations.

Within the National Department of Health, genetic services are integrated into Maternal, Child and Women's Health services (MCWH). Genetic disorders and birth defects impact at every level of service delivery (see Section 3). Inter-disciplinary integration of services is therefore essential to foster appropriate client appraisal and treatment.

The general objective of genetics services is to **“help people with a genetic disadvantage to live and reproduce as normally and responsibly as possible”** (WHO, 1985).

Birth defects include inherited genetic disorders and non-genetic (congenital or structural) abnormalities. They range from minor physical problems (postminimus polydactyly) to very serious, chronic and life-threatening, disabling conditions (Fetal Alcohol syndrome) . Some birth defects are visible at birth (e.g. neural tube defects, albinism and Down syndrome) while others have late onset and manifest later in life (e.g. Huntington disease, breast cancer, hypertension, diabetes mellitus). Serious birth defects have enormous emotional, psycho-social and financial implications for the family, and are important causes of ill health and human suffering. Some birth defects can be treated, but management is usually costly and often only partially successful. Other abnormalities, however, may be wholly preventable by dietary means, pre-natal diagnosis, selective termination of pregnancy, or even by early diagnosis and treatment.

The incidence of birth defects in developing countries is not well documented. It has been estimated that the cumulative incidence of severe (significant) birth defects and genetic disorders reaches eight percent by the age of five years. If all genetic-related disorders of late onset, such as hypertension, diabetes, certain cancers and psychoses are included, it is estimated that 60 – 65% of the population will suffer from such a condition during their lifetime. The estimated number of births for 1996 in South Africa is 1 312 040 (Statistics South Africa) and it can thus be determined that approximately 150 000 of children born annually are affected by a significant birth defect or genetic disorder by the age of five years. These anomalies, serious by definition, result in disorders that persist throughout life and culminate in premature mortality or affected persons not reaching their full potential. **The cost to society and the State in burden of disease is estimated at several billion annually (see Section 11).**

To deliver a comprehensive genetic service equitably to all South Africans, a considerable revision of clinical and laboratory staffing will be required. The United Kingdom report on “Clinical Genetics Services into the 21st Century” (1996) recommends that two full-time clinical geneticists are required per 1 000 000 of the population. Using these criteria, about 80 clinical geneticists would be required in South Africa. However, there are currently only four full-time, practising, registered clinical geneticists in the country. Furthermore, according to the British criteria, each geneticist should be supported by four genetic counsellors / trained nursing staff registered with the Health Professionals Council of South Africa (HPCSA). South Africa currently has fewer than 20 genetic counsellors, whereas 320 are ideally required. Each clinical team (one clinician and four counsellors) should be supported by at least five laboratory scientists for the provision of cytogenetic, molecular and biochemical investigations. This means that 400 medical scientists and technologists are needed, but currently such State-employed persons countrywide number fewer than 50. These are supplemented by approximately 50 university-employed or research-funded individuals. The current national situation with regard to genetic staffing is critical and requires urgent attention.

These Human Genetics Policy Guidelines are designed to facilitate the integration of genetic services into the comprehensive Primary Health Care package, focussing on patient care and primary prevention. Medical genetic services are not “stand-alone” programmes and must not be viewed as such. The collaboration of many role players is essential for a comprehensive genetic programme to succeed, and the saving in terms of human suffering and costs to the State will be profound.

TERMINOLOGY USED IN THIS DOCUMENT

Birth defects include any abnormality of body structure or function present at birth, some of which may only be recognisable or diagnosable later in life. Severe birth defects persist as a disability throughout life, reducing an individual's longevity and potential for full societal participation.

A **gene** is a section of DNA (deoxyribonucleic acid) that carries the instructions for making an enzyme or other protein. A gene is a specific hereditary unit and occupies a particular position on a chromosome. Each gene has a specific effect upon the phenotype (the physical, biochemical and physiological characteristics) of an individual. Genes may undergo mutations, which alter the effect they have on a person's phenotype.

Genetic counselling is a communication process which aims to assist individuals or families with a genetic disorder or birth defect to understand the medical implications, diagnosis, prognosis and management of the disorder; the inheritance pattern and risk of recurrence; the options available to deal with the risk and the choices and actions required; and how to make the best possible daily life adjustment to the disorder.

Genetic counsellors / genetic nurse counsellors are, appropriately-trained health professionals registered with the Health Professional Council of South Africa (HPCSA), who provide information and psychosocial support to individuals or families who have, or who are at risk for, a genetic disorder or birth defect. These counsellors identify such individuals and families, investigate the clinical problems, interpret medical information to the clients, analyse inheritance patterns and risks of recurrence, review available options with the clients, and educate the community about genetic disorders.

A **genetic disorder** is a pathological condition due to a mutation in one or more genes. Such a gene has mutated (changed) so as to increase the risk of or cause a genetic disorder.

A **genetically-trained nurse** is a registered nurse with basic and post-basic training, whose major function is to deliver a genetic service at a Primary Health Care level.

Genetic services are services forming part of integrated comprehensive health care, aimed to assist individuals with a genetic disadvantage to live and reproduce as normally and responsibly as possible. The components include clinical diagnostic

services, counselling, laboratory support, prevention strategies, public awareness campaigns in collaborations with NPOs, and training. All medical genetic services should be comprehensive.

A **medical geneticist** is a registered medical practitioner with an appropriate specialist qualification and registration with the HPCSA in the sub-speciality of Medical Genetics.

Medical Genetics is a speciality that aims to diagnose, manage and prevent genetic disorders and birth defects.

A **mutation** is a change in the structure of DNA. Such changes can be passed on to the next generation. A few mutations are beneficial, but others can cause genetic disorders.

A **teratogen** is a physical or chemical agent that can cause damage to the embryo or fetus, resulting in a birth defect. Teratogens include drugs (e.g. alcohol, warfarin), maternal infections (e.g. rubella, cytomegalovirus, herpes), maternal illness (e.g. diabetes mellitus) and environmental chemicals (e.g. lead, methyl mercury).

EXECUTIVE SUMMARY

The care of people (and their families) with a genetic disorder or birth defect includes both management and prevention.

MANAGEMENT

Good management consists of establishing the diagnosis using appropriate investigations, and providing the individual or family with genetic counselling, psycho-social support, and the best possible medical treatment.

PREVENTION

Primary level prevention of common conditions such as Down syndrome (particularly in women over 35 years of age), neural tube defects (by means of peri-conceptual folic acid) and fetal alcohol syndrome (by discouraging the drinking of alcohol during pregnancy), is emphasised. Management at Primary Health Care level includes prevention of disease complications, e.g. eye and skin care for persons with albinism, mental stimulation for children with Down syndrome, and provision of psycho-social support to affected individuals and their families.

Secondary level prevention includes voluntary pre-natal diagnosis and selective termination of pregnancy for genetic disorders and birth defects, e.g. anencephaly, Trisomy 18, Trisomy 13 and inoperable fetal cardiac defects.

Tertiary level prevention involves correction of genetic disorders or birth defects in order to restore normal function, e.g. repair of cleft-lip/palate, orthopaedic management of club feet, and cardiac surgery for children with Down syndrome.

PRIORITY CONDITIONS

- Down syndrome
- Neural tube defects
- Fetal alcohol syndrome
- Albinism
- Cleft-lip and palate
- Talipes equinovarus (club feet)
- Congenital infections, e.g. rubella and cytomegalovirus

Genetic deafness, blindness, physical handicap and mental retardation
Other common disorders include haemophilia, cystic fibrosis, thalassaemia, Fragile X syndrome, Duchenne muscular dystrophy and genetic cancers.

PEOPLE BENEFITING FROM A COMPREHENSIVE GENETIC SERVICE

Affected individuals or those with a family history of a genetic disorder or birth defect

Couples with recurrent miscarriages or stillbirths

Pregnant women exposed to alcohol, teratogenic medications, recreational drugs and chemicals

Women over the age of 35 planning a pregnancy

Women identified as high risk through antenatal tests

Couples in a consanguineous marriage

Ethnic groups with a high risk for certain disorders (e.g. Albinism amongst South African Blacks and Tay-Sachs disease in Ashkenazi Jews)

TRAINING AND EDUCATION NEEDS

Under-graduate human genetic teaching of medical and nursing students

Post-graduate training of medical geneticists

Development of a distance-learning medical genetic education programme for nursing and paramedical staff

Training and supervision of the Primary Health Care professional

Training for a registerable, post-basic nursing diploma in medical genetics / counselling

Training for a registerable Masters degree in genetic counselling

Training of medical scientists and technologists in the fields of cytogenetics and molecular and biochemical genetics

For all of the above, continuing professional development (CPD) is required to ensure the maintenance of knowledge and standards.

LABORATORY SERVICES FOR HUMAN GENETICS

Due to the complexity of genetic disorders and the laboratory tests thereof, close collaboration is required between medical geneticists, counsellors and scientists to deliver a well-planned, comprehensive service. In line with the goals of the future National Health Laboratory Service (NHLS) and due to the intricate nature of genetic testing and service delivery, it is suggested that:-

- (1) cytogenetic laboratory tests and fluorescent *in-situ* hybridisation (FISH) investigations only be undertaken in academic health centres and in laboratories accredited by a national standards body;
- (2) molecular genetic investigations be undertaken in designated laboratories familiar with the tests envisaged and accredited by a national standards body;
- (3) biochemical testing for genetic disorders be undertaken by designated laboratories with relevant expertise and accredited by a national standards body;
- (4) medical genetic counselling become a compulsory component of any organisation offering genetic investigation, and that counsellors be registered with HPCSA; and
- (5) the costs of investigations for indigent clients be met by the National Sub-directorate Human Genetics, who will be allocated an appropriate budget by the Department of Health for these purposes.

MONITORING AND EVALUATION

Notification of genetic disorders and birth defects in the immediate post-natal period, as well as later in life, should be collated at a National level. Information should be collected at regional level by nurse co-ordinators, and forwarded to both the Provincial Health Information Services and the National Sub-directorate Human Genetics.

ETHICAL GUIDELINES

The use of genetic information has major ethical implications for affected individuals and those related to them. Ethical guidelines adapted from the WHO document “Hereditary Disease Programme 1995” and other relevant publications, have been included in the document. Genetic counselling should be non-directive, supportive, responsive to patients’ needs, and should respect the choices of clients and families. Information conveyed to the patient and/or the family will be conducted in a language that is easily understood, enabling them to make informed, independent decisions. Informed consent is always to be obtained prior to investigation or treatment.

1. SITUATIONAL ANALYSIS

1.1 CURRENT POSITION

The Sub-directorate Human Genetics forms part of the Maternal, Child, Women's Health and Nutrition (MCWH) Cluster of the National Department of Health (DOH). Genetic Services are also integrated with MCWH services at Provincial level. The Universities of Cape Town, the Witwatersrand and Pretoria, with their Departments of Human Genetics and medical, paramedical and laboratory staff, provide a major share of the genetic services for South Africa. The Universities of Stellenbosch and the Free State, comprising divisions of medical genetics within their medical schools, also undertake mainly-provincial genetic services. The University of Natal Medical School is in the process of initiating a medical genetic service. MEDUNSA supplies a regional cytogenetic service.

Approximately ten provincial "genetic nurses" (previously employed by the DOH Sub-directorate Human Genetics) sited in major cities and towns, also help provide a genetic service. Additionally, limited numbers of nurses have received some training in medical genetics, and assist in service provision. The background, development and limitations of the present service have been described by Jenkins (1990).

1.2 HISTORICAL OVERVIEW

The first academic department of Human Genetics was established in South Africa in 1972 and others followed in 1975 and 1989; units or divisions are found in two other medical schools. The National DOH created a Genetic Services division in the early 1970s and, with a relatively small budget, set up a network of genetic nurses, totalling 16 posts to serve the whole country. The DOH trained these health professionals by means of annual one-week in-service training courses and offered short courses (lasting one, three and five days) in medical genetics to nurses throughout the country. Prior to 1994, nurses from the "Homelands" were similarly trained and some were employed as part-time genetic nurses by the regional authorities. An attempt was made by the DOH to co-ordinate genetic services countrywide, beginning in the late 1980s. Funding was provided to bring major stakeholders together once a year but no DOH posts were created for this purpose. By and large, the Human Genetic departments within medical schools provided the major share of the

genetic service, funded to a greater or lesser extent by the National Department of Genetic Services, provincial authorities and research funding. They operated largely in the cities in which they were situated.

The DOH initially concentrated on screening institutions for the mentally handicapped, physically handicapped, deaf and blind. It funded laboratory investigations, produced leaflets on common genetic disorders for mass distribution and offered some genetic counselling which was provided by its nursing staff. A few nurses were attached for a while to academic departments. Some academic departments undertook outreach clinics to rural areas (the DOH contributed to the cost) but genetic services were not available to large areas of the country. Academic departments also provided graduate training for BSc(Hons), MSc and PhD degrees, as well as lectures to medical students, nurses and the general public.

1.3 LAY ORGANISATIONS

The South African Inherited Disorders Association (SAIDA) and other Not-for-Profit Organisations (NPOs) provide information and support for affected families; operate on community participation principles; and provide valuable input into genetics services while attempting to improve their accessibility.

1.4 STAFFING

The staff complement presently available for genetic services is inadequate to provide a comprehensive service to the whole South African population, estimated at 40 million people. Outreach services and training are provided from Universities in Cape Town, Bloemfontein, Pretoria and Johannesburg to other urban and rural areas. However, these are too few because of the limited number of staff available. An additional 70 medical geneticists, supported by 300 genetic counsellors / genetic nurse counsellors and 300 laboratory scientists / technologists (cytogeneticists and molecular geneticists), would be required, as judged by First-World recommendations. The current services are presently of world-class standard in some urban areas, but they are neither equitable nor accessible to most of the remaining population.

2. GOALS AND OBJECTIVES

To reduce the burden of genetic disorders and birth defects to the individual, the family and society in general

To empower individuals with genetic disorders and birth defects, and their families, to live and reproduce as normally and responsibly as possible

To create awareness of the psychosocial and fiscal impact of genetic disorders and birth defects

- 2.1** To provide a national, PHC -based, comprehensive, medical genetic service for the diagnosis and care (management and prevention) of genetic disorders and birth defects in South Africa.
- 2.2** To integrate medical genetic services into primary, secondary and tertiary levels of care as part of the comprehensive health care system with an appropriate referral network.
- 2.3** To develop a medical genetic component of health services through capacity building, re-orientation of health professionals, training of PHC workers particularly midwives, nurses and others concerned with Maternal, Child and Women's Health.
- 2.4** To establish the infrastructure and technology with which to deliver medical genetic services effectively and equitably to the community at large.
- 2.5** To develop appropriate public education and community awareness for health promotion strategies in the field of medical genetics.
- 2.6** To co-ordinate medical genetic services at inter-provincial and national levels.
- 2.7** To establish a national monitoring and evaluation system for genetic disorders and birth defects.
- 2.8** To establish inter-sectoral collaborations with relevant stakeholders such as the Departments of Education, Labour, Welfare (Social Development), the private sector, NPOs and CBOs.

3. PRIORITY MEDICAL GENETIC SERVICES

3.1 SERVICES PRIOR TO CONCEPTION

Target population: Women of reproductive age and individuals and families at high risk for genetic disorders and birth defects

- Creation of awareness of risks associated with advanced maternal age
- Ascertainment of genetic risk based on a family history
- Discouragement of exposure to teratogens, e.g. alcohol, tobacco, rubella
- Encourage folic acid supplementation to reduce the risk of neural tube defects
- Creation of awareness of medical conditions that predispose to birth defects, e.g. epilepsy, diabetes mellitus
- Identification of women who have had multiple miscarriages

3.2 SERVICES DURING PREGNANCY

Services during pregnancy should include genetic counselling, pre-natal diagnosis and the option of termination of pregnancy.

Target population: Women of reproductive age and individuals and families at high risk for genetic disorders and birth defects

- Regular ante-natal care
- Early identification of pregnant women of advanced maternal age
- Ascertainment of genetic risk based on a family history
- Identification of women who have been exposed to teratogens
- Continuation of folic acid supplementation
- Identification of high-risk pregnancies, e.g. diabetic mothers
- Offering of appropriate diagnostic tests

3.3 SERVICES AT BIRTH

Target population: Parents and their newborns, and individuals who have undergone selective termination of pregnancy

- Physical examination of the neonate for genetic disorders and birth defects
- Physical examination and investigation of the stillborn baby
- Parental referral for genetic counselling
- Patient referral for medical care

3.4 SERVICES IN INFANCY AND CHILDHOOD

Many genetic disorders and birth defects are not recognisable at birth.

Target population: Children and their families

- Encouragement of attendance at well-baby clinics
- Monitoring of growth and development
- Identification of late-onset genetic disorders and birth defects
- Referral for appropriate care and genetic counselling

3.5 SERVICES IN ADOLESCENCE AND ADULTHOOD

Sixty-five percent of the population will suffer from a genetic-related disorder or birth defect in their lifetime.

Target population: All adolescents and adults

- Ascertainment of genetic risk based on family history
- Screening of high-risk populations, e.g. Afrikaner population for hypercholesterolemia
- Referral for appropriate care and genetic counselling

4. STRATEGIES FOR PREVENTION OF GENETIC DISORDERS AND BIRTH DEFECTS

4.1 GENETIC COUNSELLING

4.1.1 Definition

Genetic counselling is defined as a communication process which deals with human problems associated with the occurrence of a genetic disorder or birth defect in a family.

The process involves the following:-

- comprehension of medical facts, diagnosis, prognosis and management;
- appreciation of the genetics of the disorder and risk of recurrence in relatives;
- understanding the options for dealing with the risk of recurrence;
- choice of an appropriate course of action in view of the risks and family goals; and
- making of the best possible adjustment to the disorder within the family.

Genetic Counselling includes the provision of information necessary for rational decision-making regarding prenatal testing, termination of pregnancy and the prevention of genetic disorders and birth defects, as well as referring the family for appropriate services and care.

4.1.2 Individuals who benefit

Individuals and families who may benefit from genetic counselling include:-

- persons affected by, or families with a family history of a genetic disorder or birth defect;
- persons affected by, or families with a history of, a physical, intellectual, hearing or visual disability, or other condition which may have a genetic basis;

persons with a history of multiple miscarriages, stillbirths or early infant deaths, or with primary infertility;
women aged 35 years and over who are planning a pregnancy or who are pregnant; women identified as high risk through ante-natal screening (e.g. maternal serum screening and ultrasound);
pregnant women exposed to teratogens, which include alcohol, certain medications, infectious agents (eg. rubella, syphilis), recreational drugs, and certain chemicals;
persons in a consanguineous relationship;
persons in ethnic groups or geographic areas with a high risk for specific genetic disorders or birth defects; and
persons with medical disorders which adversely affect pregnancy, e.g. diabetes mellitus.

4.2 PREVENTION STRATEGIES

4.2.1 Primary Prevention

Primary prevention aims at preventing first occurrences of specific genetic disorders and birth defects. Conditions most amenable to primary prevention are birth defects, caused by environmental factors that can be removed or neutralised, and non-hereditary genetic disorders such as chromosomal abnormalities, in which the association with advanced maternal age offers a basis for prevention.

Strategies for primary prevention include:-

- public efforts to improve health, nutrition, education and self reliance, particularly of women;
- avoidance of unintended pregnancies, and proper birth spacing through access to contraception and other methods of family planning;
- improved access to, and quality of, pre-natal care and genetic counselling;
- improved quality of birth care;
- control of possible occupational risks;
- peri-conceptual supplementation of folic acid to women of reproductive age to reduce the risk of neural tube defects and perhaps other congenital defects;
- encouraging women to procreate at the ideal reproductive ages

(20 – 35 years) to reduce the risk of chromosomal abnormalities; and avoidance of exposure to teratogens (e.g. alcohol, recreational drugs, and certain chemicals and infectious agents) during pregnancy.

4.2.2 Secondary prevention

Secondary prevention aims at the pre-natal identification of women at increased risk for having a child with a genetic disorder or birth defect. Further it offers the option of termination of pregnancy.

Strategies for secondary prevention include:-

- identification of pregnant women at risk;
- identification of pregnant women aged 35 years or more;
- identification of pregnant women exposed to teratogens, e.g. alcohol, recreational drugs, infections (e.g. rubella, syphilis), medicines (e.g. phenytoin) or chemicals;
- ultrasound evaluation for all pregnant women for accurate gestational aging in order to detect fetal defects;
- amniocentesis or chorion villus biopsy in appropriately-selected pregnancies, e.g. previous chromosomal abnormality; and
- termination of pregnancy for serious genetic disorders or birth defects.

4.2.3 Tertiary prevention

Tertiary prevention of genetic disorders and birth defects aims at averting deterioration, complications, disability and dependency of the patient and the family.

Strategies for tertiary prevention include:-

- anticipatory guidance (e.g. prevention of obesity in Down syndrome);
- proper intervention to avert complications (e.g. laminectomy to alleviate spinal cord compression in achondroplasia);
- rehabilitation of disabilities (e.g. speech therapy, hearing aids in hypoacusia, physical therapy in neuromuscular diseases); and

pyscho-social support of affected individuals and their families.
(This is an essential, albeit commonly-neglected component of care relating to genetic disorders and birth defects.)

4.2.4 Prevention strategies for priority conditions

4.2.4.1 Down syndrome

Educate women regarding their increasing risk as they advance in age

Identify pregnant women who are at increased risk because of their family history, age (35 years or older), or maternal serum screening results

Offer pre-natal genetic counselling, investigation (ultrasound or amniocentesis), diagnosis, and voluntary selective termination of affected pregnancies

Offer diagnosis of Down syndrome in newborns, infants and children

Refer appropriate individuals for secondary and tertiary preventative measures

Refer affected individuals for early stimulation programmes and rehabilitation

Provide anticipatory guidance and genetic counselling

4.2.4.2 Neural Tube Defects

Encourage all women of reproductive age to have a healthy, balanced diet

Encourage folic acid supplementation

Identify at-risk women by family history, and offer peri-conceptional folic acid supplementation

Offer pre-natal, mid-trimester maternal serum alpha-fetoprotein screening, ultrasound evaluation, counselling, and voluntary selective termination of pregnancy to all at-risk women

Offer neo-natal detection, and appropriate referral for secondary and tertiary care

4.2.4.3 Fetal Alcohol syndrome

Educate all women regarding the deleterious effects of alcohol on the fetus

Educate all women to avoid alcohol throughout pregnancy

Offer early detection of FAS, with appropriate referral of affected individuals and their parents for counselling and care

4.2.4.4 Albinism

Offer genetic counselling for couples with a previous affected child or family history

Offer early clinical detection, and appropriate referral of affected individuals for proper management

4.3 PRE-CONCEPTION METHODS OF PREVENTION

At-risk couples need to be identified and offered genetic counselling and information on prevention

Specific maternal medical conditions in pregnancy, e.g. maternal diabetes mellitus, epilepsy (anti-convulsants) and certain cardiac problems (warfarin)

Target all women of reproductive age with the following messages of awareness:-

advanced maternal age increases the risk of chromosomal abnormalities, so aim to complete reproduction by the age of 35 years

be aware of any history of genetic disorders or birth defects in your family
alcohol, smoking and substance abuse can damage the fetus, so avoid these during pregnancy

get immunised against rubella, which can cause birth defects

take a folic acid supplement to prevent neural tube and other defects

eat a healthy, balanced diet

4.4 ANTE-NATAL METHODS OF PREVENTION

For these services to be optimally delivered, pregnant women must present for ante-natal care as early as possible (ideally before 18 weeks gestation) in order to maximise options available to them.

The following pre-natal diagnosis services, where appropriate, can be offered:

Detection of at-risk women, e.g. those of advanced maternal age

Ultrasound scans to detect congenital defects

Maternal serum screening to detect certain chromosome disorders and neural tube defects. Accurate gestational age is important for the interpretation of these tests

Amniocentesis or chorionic villus sampling to detect genetic and congenital disorders

Voluntary selective termination of pregnancy for serious genetic disorders and birth defects

4.5 POST-NATAL DIAGNOSIS

Early diagnosis (including pre-natal diagnosis) should be encouraged so that, where necessary, early treatment can be initiated to prevent morbidity associated with certain genetic disorders and birth defects.

Clinical screening of newborns for treatable genetic disorders and birth defects, and referral of affected individuals for appropriate health care, should be routinely offered.

Parents of affected children should be referred for genetic counselling to help them understand the risks relating to future pregnancies, if appropriate.

4.6 POPULATION SCREENING

Specific at-risk populations can be screened for particular genetic disorders and birth defects, according to provincial needs. Examples include screening of the Afrikaner population for familial hypercholesterolaemia; individuals of European descent for cystic fibrosis; individuals of Greek and Asian extraction for thalassaemia; and those of Ashkenazi Jewish descent for Tay-Sachs disease. However the psycho-social disadvantages must be weighed up against the advantages of such screening. Population screening can result in the prevention of births of affected individuals as well as prevention of genetic disease in at-risk individuals.

Population screening methods include:-

total at-risk population screening to detect healthy gene carriers and individuals who may benefit from health education, e.g. familial hypercholesterolaemia;

cascade screening, whereby only at-risk consenting relatives of affected individuals are screened; and

screening of newborns for treatable genetic disorders such as phenylketonuria (PKU).

5. DELIVERY OF GENETIC SERVICES

5.1 HOME

Involves the health of the whole family. It is supported by health promotion via the various media, health care workers undertaking home visits, or outreach programmes.

Involves the individual child or woman herself, husband/partner, the parents, other family members and caregivers, teachers and other community members

Supported by

Health projects of community-based organisations and extensions of the health services in:

- maintenance of good health and fostering of healthy lifestyles
- early recognition and management of minor ailments, problems and life-threatening issues
- prevention and promotion
- health promotion through the media and health care worker
- support for special needs (*Such as disability*)
- tracing of births, deaths and high risk groups
- providing information and support to enable communities to access appropriate care for each of these conditions and problems
- participation in community-based and social mobilisation health-related activities

5.2 COMMUNITY LEVEL CARE

One to two persons per facility able to counsel, treat, manage, and refer on most common conditions. This person would have other duties as well. Needs regular genetic training with regular updates from genetic co-ordinator.

Facility: Community Health Centre

Delivery of integrated comprehensive Maternal, Child and Women's Health including a genetic service for most common conditions (*with daily extended access and attention to priority genetic problems*)

Emergency care

Referral

Screening and early detection of disability

Health promotion and prevention

Integration with community-based health facilities

Support for visiting specialists, clinical geneticists and other academic experts

Community and NGO liaison.

Functions of genetically trained health worker:

Delivery of an integrated MCWH service, including genetic services

Management of the most common genetic conditions and birth defects (see Section 4)

Data collection of most common disorders (listed in Section 8)

Organisation and delivery of outreach services

Inter-sectoral collaboration (e.g. networking with environmental health officer or teacher)

Community and NGO liaison (including support groups)

Community Health Committee/Clinic Committee

Facility: Clinic

Delivery of integrated genetic services

(with daily extended access to all components of health care and including promotion, prevention and management with referral of minor problems and ailments)

Emergency care

Health surveillance and data collection

Facilitate community outreach and integration

Liaison with community health committees

5.3 DISTRICT SUB- STRUCTURE OF DEPARTMENT OF HEALTH

One "Genetic nurse" (not necessarily "dedicated nurse") identified per district appointed to co-ordinate district services and data collection.

This nurse has other duties and is responsible for identifying common genetic disorders, investigating such individuals appropriately, document and counsel them and notify to SPN in region (co-ordinator). Has two week course and receives annual in-service training. In future this nurse will have completed the MGEP course, with practical training.

Functions of District Co-ordinator:

Co-ordination of genetic and health services for district
Data collection
Collaboration and liaison (e.g. School principal, Rural Foundation).

Facility: District Hospital

24 hour service
Emergencies
Deliveries
Specialist consultations and management of referrals
Chronic diseases (control and management)
Diagnostic and therapeutic support services (where available)
Secondary care
(second level of referral at level 2 hospitals)
Co-ordination of delivery of care
(with other levels in the province and district)
Specialist support
(for health care delivery at district level and for in-service training)
Diagnostic basic biochemical tests
Referred cytogenetic services
Amniocentesis
Basic ultrasounds then refer to tertiary service
Basic radiology.

5.4 REGIONAL LEVEL

One Regional Co-ordinator for each region (e.g. MCWH manager). One SPN in each region with a post basic diploma in medical genetics (6 month's training) responsible for co-ordination of genetic services, training of community nursing sisters at district level and collecting data relating to genetic conditions and birth defects in their region.

Functions of Regional Co-ordinator:

- Co-ordination of district services
- Co-ordination of training (e.g. MGEP co-ordinator)
- Monitoring and evaluation (including data collection and surveillance)
- Inter-sectoral collaboration (e.g. environmental health, mental health)
- Support groups.

Facility: Regional Hospital

- 24 hour service and specialist support (basic and specialist)
- Emergencies
- Referral system
- Diagnostic and therapeutic support service (e.g. radiology, basic biochemistry, cytogenetics, amniocentesis and basic ultrasound).

5.5 PROVINCIAL DEPARTMENT OF HEALTH

Genetic services fall under Maternal, Child and Women's Health Services. Need a genetics co-ordinator at provincial level (CPN or NSM, a genetic counsellor or "genetics nurse" with at least 6 month advanced training).

Functions of Provincial Co-ordinator

- Provincial policy
- Monitoring and evaluation (including data collection/surveillance)
- Research
- Training
- Health promotion and media liaison
- Collaboration with other Departments (e.g. Education) and sub-directorates (e.g. mental health)
- Co-ordination of genetic services within province
- Provincial genetic advisory group.

Facility: Tertiary Hospital

Tertiary care (at hospitals in a given province, or in adjacent provinces for those provinces which have no tertiary care facility).

- Medical geneticist/Developmental neurologist
- Sub-specialist referral and support (e.g. neuro-developmental paediatrician, obstetrician, cardiologist)
- Co-ordination of care (between tertiary level and other levels in the province)

Medical specialist support services

Provision of diagnostic and therapeutic support services including diagnostic laboratories (e.g. MRI, DNA tests)

Research

Referral to social and Welfare Support Services.

5.6 NATIONAL DEPARTMENT OF HEALTH

Sub-directorate: Human Genetics forms part of the Cluster: Maternal, Child, Women's Health and Nutrition of the National Department of Health.

Functions:

National MCWH Policy document

Training: Standardisation of curriculae for courses within Provinces, Genetic curriculae in basic nursing training (Universities, Technikons, Colleges)

Data collection/co-ordination (National Genetic Information System)

National advisory group

National awareness campaign/health promotion

Publications

6. EDUCATION AND TRAINING

In order to implement a human genetics programme for the management and prevention of genetic disorders and birth defects in South Africa, information, awareness, specific education and training of medical, nursing and paramedical staff, and the general public will be a prerequisite.

6.1 MEDICAL SCHOOLS

6.1.1 Current situation

6.1.1.1 Training of medical undergraduates in Human Genetics

Formal teaching of medical genetics in undergraduate medical education is undertaken in the five faculties of medicine that have Departments of Human Genetics. Currently, a review of undergraduate training in medical genetics in South Africa is being undertaken by the Southern African Society of Human Genetics (SASHG).

6.1.1.2 Training of medical specialists in Human Genetics

Postgraduate training in human genetics of medical specialists is presently not formally a part of any medical speciality. However, in the different faculties of Medicine, postgraduate teaching of human genetics is undertaken on an *ad hoc* basis to various medical specialists, according to the needs of the specific discipline and availability of human resources to undertake such teaching.

6.1.1.3 Sub-specialty training in Medical Genetics

Post-specialist training of doctors in medical genetics is potentially available at the universities of Cape Town, Stellenbosch, Free State, Witwatersrand and Pretoria. This training, if undertaken by a paediatrician, physician or

obstetrician / gynaecologist registered with the HPCSA, is recognisable by registration with the HPCSA as a medical geneticist. Given the country's circumstances, and bearing in mind the practices of other countries, consideration should be given to broadening the base for sub-speciality training in medical genetics to include other medical specialities.

There is currently one training post available in South Africa. The minimum requirements for sub-specialist registration in Medical Genetics are:-

- (i) specialist registration with the HPCSA in paediatrics, internal medicine or obstetrics / gynaecology; and
- (ii) a minimum of two years sub-speciality training in an academic medical genetic unit accredited by the HPCSA.

6.1.2 Recommended situation

6.1.2.1 Training of medical undergraduates in Human Genetics

Standardisation of a curriculum for all undergraduate medical genetic education in South Africa should be undertaken and recommendations made to the HPCSA.

6.1.2.2 Training of medical specialists in Human Genetics

The present situation in which medical genetic training is not formally included in the curriculae of medical specialists in training, is an issue that requires formal investigation by appropriate bodies, including the SASHG, the HPCSA and the College of Medicine of South Africa. Recommendations should be made to ensure formal medical genetic education is a part of all medical speciality training in South Africa.

6.1.2.3 Sub-specialty training in Medical Genetics

It is urgently recommended that training posts for medical genetics be created in South Africa. Concurrently, posts for qualified medical geneticists need to be established throughout the country.

6.2 GENETIC COUNSELLORS

6.2.1 Current situation

The minimum requirements for registration as a genetic counsellor are:

- (i) a laboratory-based MSc in human genetics, registered with the HPCSA, plus two years post-MSc training in genetic counselling at an academic medical genetics department accredited with the HPCSA; or
- (ii) an MSc in medical genetic counselling, registered with the HPCSA, plus one year post-MSc training at an academic medical genetics department accredited with the HPCSA.

Nursing staff qualified with a BSocSc (Nursing) will be eligible to obtain registration as genetic counsellors by following career paths that include BSc(Hons) in human genetics, MSc in Medical Genetic Counselling, plus one year post-MSc training at an academic medical department accredited with the HPCSA.

6.2.2 Recommendation

The current situation with regard to training is satisfactory. It is recommended that posts for genetic counsellors are urgently made available on a country-wide basis.

6.3 NURSES

In order to ensure the successful implementation of a national medical genetics programme it is recognised that the training of community-based nursing staff is of the highest priority, and that the common genetic disorders and birth defects recognised as part of the National Primary Health Services are appropriately managed.

6.3.1 Undergraduate nursing education

6.3.1.1 Current situation

Medical genetics forms a part of the present nursing curriculum. The teaching of this component of the curriculum, however, varies according to the facilities and staffing available for its teaching in the various colleges.

6.3.1.2 Recommendation

Consideration should be given to assessment of the present medical genetic component of the nursing curriculum, as well as the facilities and staffing available to teach medical genetics throughout the country. This should be done with a view to ensuring the appropriateness and standardisation of content and teaching of this curriculum.

6.3.2 Training of post-basic clinical nurse practitioners

6.3.2.1 Current situation

Post-basic clinic nurse practitioners currently receive no training in medical genetics.

6.3.2.2 Recommendation

A medical genetic component, with an emphasis on the recognition and management for common genetic disorders and birth defects, should be included in the training of post-basic clinical nurse practitioners.

6.3.3 Genetic training for nursing staff

6.3.3.1 Current situation

Currently, post-basic genetic training for nursing staff comprises one-, four- and five-day courses in medical genetics, followed by yearly in-service training programmes. The five-day courses and the in-service training are organised and presented under the auspices of the Human Genetics Sub-directorate, National Department of Health. The one- and four day courses are organised by provincial Departments of Health.

6.3.3.2 Recommendation

Consideration should be given to the above being replaced by the following education programme:

- (i) **One-day courses** would involve broad-based, PHC-orientated day programmes offered by provincial Departments of Health. The contents of the course would be standardised, and lectures would be given by genetically-trained nursing staff (GTNS), genetic counsellors and medical practitioners trained in medical genetics.
- (ii) The **Genetic Education Manual (GEM)** course would be a distance learning, self-administered education programme similar to the present Perinatal Education Programme (PEP) which has enjoyed wide success in South Africa. It would be offered for nursing staff,

particularly those involved in maternal, child and women's health, who wish to undertake further training in medical genetics.

- (iii) A **two-week medical genetics course** would be a practically-orientated course in medical genetics offered to nursing staff who have completed the above GEM course. It is intended that the curriculum be standardised by the Human Genetics Sub-directorate of the Department of Health, but the course itself would be offered by provincial Departments of Health. Topics to be covered would include the diagnosis and management of common genetic disorders and birth defects; laboratory procedures; principles and practice of genetic counselling; psychosocial support; and the collection and collation of statistics. This course should provide some degree of flexibility, to cater for the special training needs of provinces, geographical areas and/or communities. Subsequently, an examination covering the contents of both this and the GEM course would need to be passed for qualification as a genetically-trained nursing sister (GTNS).

- (iv) Yearly **in-service training programmes** should be made available in each province to ensure that GTNS maintain their level of education, and to keep abreast of current developments. This could be part of Continuing Professional Development (CPD).

6.3.4 Genetic Nurse Counsellor (GNC)

6.3.4.1 Current situation

Although there are several nursing staff who have extensive experience in genetic counselling, there are no formally-designated genetic nurse counsellor posts.

6.3.4.2 Recommendation

It is proposed that a career path be developed for GTNS to undergo further training, which would result in their recognition and registration as Genetic Nurse Counsellors (GNC), with equivalent status to that of a Genetic Counsellor. It is proposed that the minimum requirements for qualification as a GNC be:

- (i) a GTNS qualification, with a 12-month nursing diploma in medical genetics (this diploma has still to be negotiated with the relevant nursing authorities and colleges); and
- (ii) one year post-diploma experience and in-service training including part-time attachment to an accredited academic medical genetics department.

6.4. MEDICAL SCIENTISTS AND TECHNOLOGISTS

6.4.1 Current situation

Training for medical scientists and medical technologists is available at most of the medical faculties in South Africa. However the availability of posts is very limited, and many of the qualified individuals leave the country or change profession. It is recognised that there is an urgent need to ensure the availability of training posts for medical scientists and medical technologists in medical genetics. This is necessary to maintain and develop medical genetic laboratory services country-wide, which are essential for the future equitable development of medical services in South Africa. It is noted with concern that present government funding for such posts is inadequate, and that there is a disturbing loss of trained scientists from government/ academic laboratories and the country at the present time.

6.4.2 Recommendation

It is recommended that more funding be made available for both the training and creation of medical scientist and technologist posts.

6.5 HUMAN GENETICS IN SCHOOL EDUCATION

The introduction of appropriate medical genetic topics into the biology curriculum of secondary education is considered to have a high priority. In this manner adolescents can be “conscientised” on matters related to genetic health and disease, including their rights regarding genetic care. This should be seen as a part of present life skills programmes now being developed and undertaken in secondary schools.

7. LABORATORY SERVICES

South Africa has the greatest capability in the field of medical genetics in Sub-Saharan Africa. The incorporation of medical genetic laboratories into the new National Health Laboratory Services (NHLS) will provide an opportunity for improving access to medical genetic services in the country.

The recommendations of the Ad Hoc Committee on Genetic Laboratory services of how medical genetics should be incorporated into the NHLS are outlined below. The complete report is included as an annexure.

PROPOSAL FOR THE INTEGRATION OF MEDICAL GENETIC LABORATORIES INTO THE NHLS

This will be discussed under the headings given to the subcommittee of

- 1) National Reference Laboratory
- 2) Clinical Diagnostic Laboratory Services
- 3) Research Development, Teaching, Training and Academia

7.1 NATIONAL REFERENCE LABORATORY (NRL)

For Medical Genetics services to function efficiently, a NRL is necessary, but that it should function as a national advisory body for medical genetics laboratory services. It would be directly responsible to the NHLS Board, funded by that Board, and be responsible for nationally organisable tasks. It would undertake the requisite research and development and then place proposals for policy before the NHLS Board (Figure 2). Because the NHLS Board falls under the direction of the National Ministry of Health, the Medical Genetics Advisory Board could be utilised for similar functions related to medical genetics for the National Department of Health.

Medical Genetic Advisory Board (MGAB)

Proposed composition:

- 4 people in Medical Genetics (Clinical and laboratory based)
- Deputy Director: Human Genetics, National Department of Health
- One person qualified in Law and/or Ethics

The chairperson could either be nominated by the NHLS Board, or elected by the members of the MGAB and would report to the NHLS board. The MGAB would meet on a 3 to 4 monthly basis, and may co-opt national, and if necessary international, expertise to assist in the performance of its task.

Secretariat

A secretariat based at the SAIMR, Johannesburg would be employed to administer and co-ordinate the functions of the MGAB.

Functions of the MGAB

The MGAB would have responsibility for the research and development of proposals for policy with respect to:

- i) the development of standards for laboratories involved in the provision of service in medical genetics.
- ii) the development of a system of laboratory quality control and proficiency testing.
- iii) quality assurance and laboratory licensing which would take place outside the auspices of this body.

The policies set by the NHLS board, including quality control, proficiency testing, quality assurance and licensing should apply to all laboratories (public and private) that perform genetic testing in the country. In addition, patients having genetic tests should be provided with genetic counselling. As a general rule for each genetic test, at least one counselling session should occur. Test results often have implications for other family members and therefore issues of confidentiality also need to be carefully addressed. The provision of laboratory services must therefore be linked with the clinical/counselling genetic services thus enabling the patient to make informed reproductive health and management choices.

The principles for the provision of medical genetic services are similar to those of HIV. Preventive measures may be taken, there is a need for pre and post test counselling, the calculation of risk, and the management of psychosocial aspects.

- iv) The determination of NHLS research and service priorities for genetically determined conditions and birth defects. To undertake this, it would be necessary for the MGAB, through The Secretariat, to maintain a surveillance system.

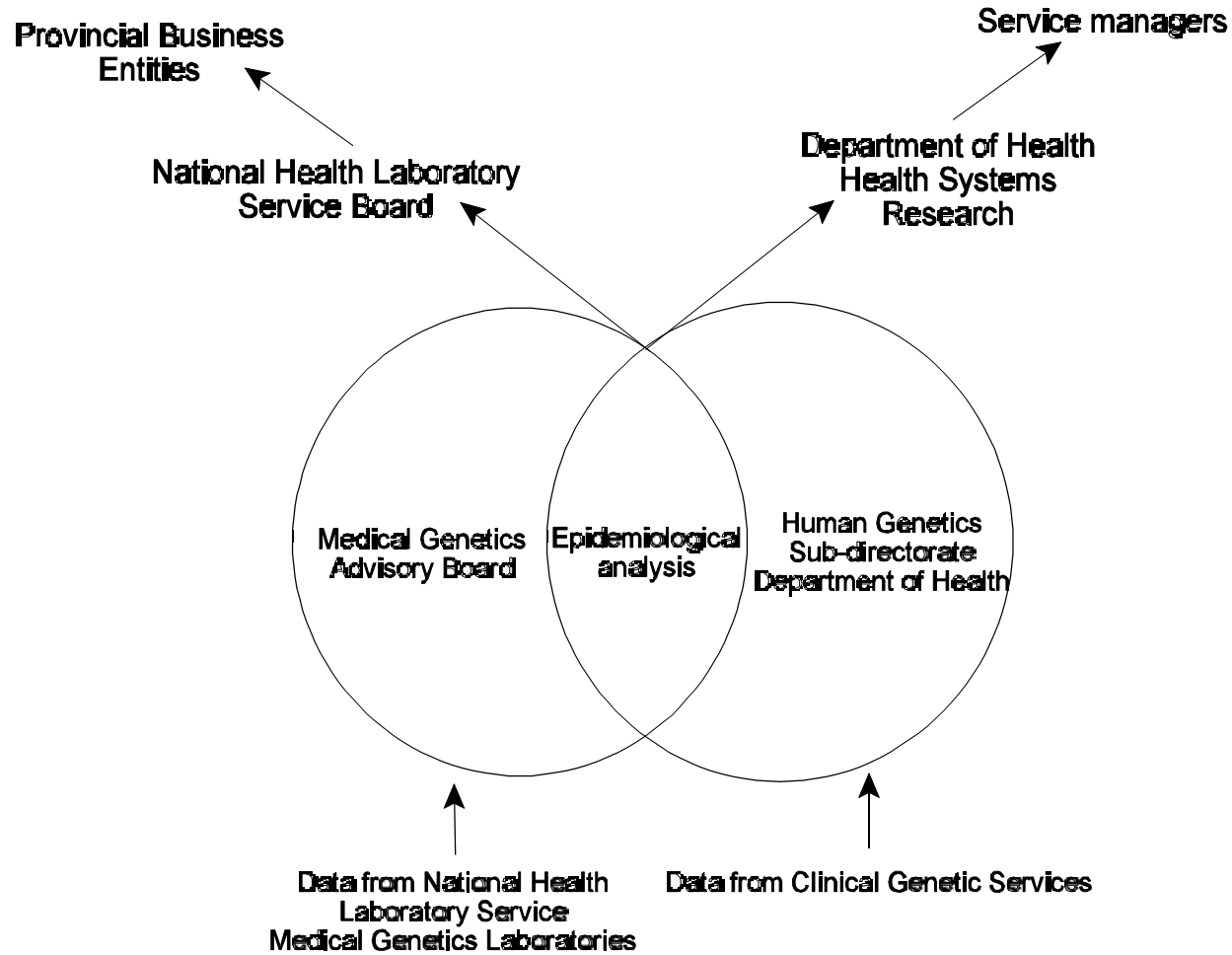
A centrally regulated genetics laboratory service with support from the national health system will allow access to (i) data on patients requesting genetic tests, (ii) clinical data on patients for whom no laboratory test may be offered. The information can be processed in a unit where expertise on the analysis of genetic conditions would be required. The processed information can then be forwarded to the Health Systems Research Unit in the Department of Health and to Service managers in the various provinces; as well as to the NHLS board and the PBEs (Figure 1).

*The raw data collected from the laboratory and the clinics needs to be handled so that it would **not** be possible to identify individual patients (including patients with rare conditions) and therefore these data cannot be made part of the routine reporting process.*

- v) make recommendations to the NHLS board on the allocation of research funds. It would also be appropriate for the MGAB to maintain a system of review of research protocols, for NHLS priorities, using national and international experts.
- vi) address ethical and administrative issues in medical genetic services (laboratory and clinical) as the need arises. This would include issues such as pre-implantation diagnosis, cloning, and the release of genetic information to third parties (eg insurance companies and employers).
- vii) develop policy proposals for the establishment of genetic laboratory services in under served areas where and when appropriate e.g. KwaZulu-Natal, Eastern Cape and the Northern Province.
- viii) co-ordination of medical genetic laboratory service to minimise unnecessary duplication of genetic tests for rare conditions. This would include the collaboration with overseas laboratories for those rare tests that are not done in South Africa.
- ix) setting fee structures for laboratory services
- x) other tasks as directed by the NHLS board.

FIGURE 1:

**MANAGEMENT OF DATA GENERATED FROM
MEDICAL GENETIC SERVICES**



LEGEND: Currently the Human Genetics sub-directorate of the Department of Health collects (opportunistic) data from the clinical genetics services. Requests for laboratory tests would allow for clinical data on patients to be accessible to the NHLS. However, due to patient confidentiality issues, the data would have to be processed and analysed within an epidemiological unit prior to dissemination for service and research prioritisation and health planning.

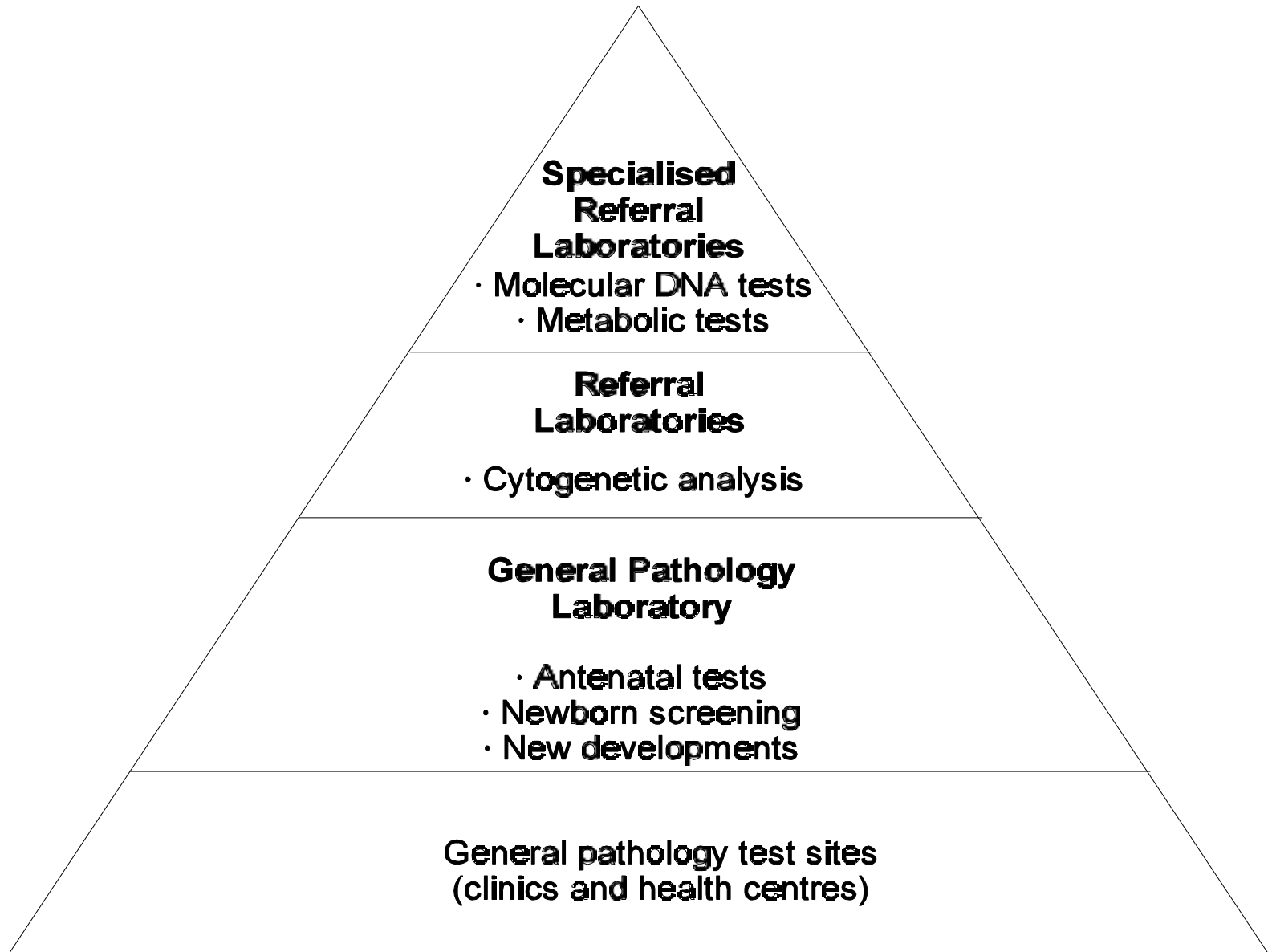
The MGAB should be seen as a body that bears responsibility for policy development, with respect to the regulation, auditing and accounting of medical genetic laboratory services. This body will therefore require an appropriate constitution and mandate.

7.2 CLINICAL DIAGNOSTIC LABORATORY SERVICES

Presently almost all diagnostic laboratory tests in the field of medical genetics (molecular DNA testing, metabolic tests, and cytogenetic tests) are undertaken in academic centres. The continued maintenance of these services in the proposed NHLS would thus fall under the auspices of the Provincial Business Entities (PBE). As the majority of these tests are specialised it would be prudent to suggest that a national grid of “referral laboratories” is developed to undertake this testing as discussed below (Figure 2).

FIGURE 2:

**MEDICAL GENETIC SERVICES WITHIN THE
PROVINCIAL BUSINESS ENTITIES**



7.2.1 SPECIALISED REFERRAL LABORATORIES

Molecular DNA Testing

Several laboratories presently provide molecular DNA diagnostic services for an array of conditions. To date informal agreements between these laboratories has, to some extent, ensured that each test offered is only undertaken at one laboratory unless the prevalence of the condition or other reasons dictate otherwise. The MGAB would oversee the coordination of this policy. As noted above there are exceptions to this “rule” and the future involvement of such tests in the field of common multifactorial diseases will require the timeous development of appropriate policy.

The positioning, rationalisation and regulation of molecular DNA technology for three special applications requires consideration.

Paternity Testing

With the new laws presently in place for child maintenance and support the need for paternity testing has increased. Presently this is undertaken by several laboratories in academic departments, some blood transfusion services, and in private practice. This includes three separate laboratories in Cape Town. This situation requires further investigation with a view to rationalisation and regulation.

Forensic Molecular DNA Testing

There is currently a proposal that the South African Police Services Forensic Science Laboratory develops a “DNA criminal intelligence database”. There are many legal and ethical issues involved in the use of DNA molecular technology for forensic purposes. The control of the use of this technology by the SAPS raises further ethical and legal considerations. It is therefore suggested that serious consideration be given to placing the laboratory services involved in this work under the NHLS and thus subject to the regulations developed for all medical genetic laboratory services.

Pre-implantation Genetic Diagnosis

This is a field of endeavour which is developing in South Africa, particularly in private practice. Its regulation is considered to be a matter of urgency.

Metabolic Disease Testing

Presently there is only one laboratory, in the Department of Biochemistry, University of Potchefstroom, that specialises in the laboratory diagnosis of metabolic disorders. There are however other laboratories which do have capability in the field, providing a limited service. The inclusion of the services of the University of Potchefstroom laboratory into the NHLS is desirable, and the need for a second such facility may need to be investigated in the future.

7.2.2 REFERRAL LABORATORIES

Cytogenetic Testing

Cytogenetic testing forms the backbone of current medical genetic laboratory services and is used for prenatal diagnosis of birth defects, postnatal diagnosis of birth defects, and in the diagnosis and management of malignancies, particularly haematological cancers. There are cytogenetic laboratories in six academic centres, the Blood Transfusion Service in Durban and a provincial laboratory in Potchefstroom. Laboratories are also functioning in private practice. Two laboratories are situated in the Western Cape (Universities of Cape Town and Stellenbosch) and three in Gauteng (SAIMR / University of Witwatersrand, MEDUNSA, University of Pretoria) and one at the University of the Free State. For the present these laboratories should take responsibility for providing services to their particular PBE.

To ensure other provinces have access to cytogenetic laboratories, it is suggested that the Eastern Cape and Northern Cape (south half centred on Springbok) are aligned to the Western Cape PBE; the Free State PBE covers the Northern Cape (northern half centred on Kimberley) and the Gauteng PBE provides services for Mpumalanga, the North West and the Northern Provinces.

7.2.3 GENERAL PATHOLOGY LABORATORY

Maternal Serum Screening

The development of antenatal care services may lead to the future

possibility of maternal serum screening for alpha fetoprotein or possibly the Triple Test to screen for Down syndrome. Such testing would occur in a general chemical pathology laboratory. However issues related to the ethics and administration of such testing, including counselling, would bear consideration by the MGAB as in essence they are tests for birth defects.

Neonatal Screening

Inherited errors of metabolism do not presently constitute a known significant public health problem in South Africa. However with the advancement of transitional epidemiology this situation will change and the necessity for neonatal screening will evolve. Like maternal serum screening this may be undertaken in a general chemical pathology laboratory but raises ethical and administrative issues that fall within the ambit of medical genetics.

New Developments

It can be anticipated that testing for certain genetically determined disorders will become simpler and less expensive in the future. Again, although the testing may be undertaken in a general pathology laboratory, issues related to their appropriate usage will bear consideration by medical geneticists.

7.2.4 SPECIAL CONSIDERATIONS

University of Natal

The present situation in KwaZulu-Natal requires special consideration. The University of Natal Medical Faculty has, on a research basis developed some molecular DNA testing capacity. A collective will has developed there to establish a medical genetics capability. This coincides with the building of a quaternary care Academic Hospital with accommodation and facilities potentially available for this development. This development should encompass all aspects of medical genetics, including cyto and molecular genetic laboratories and a clinical genetics service. The placement of the present Natal Blood Transfusion Cytogenetic Laboratory within this complex should be a matter for investigation.

University of the Transkei

In collaboration with the Department of Medical Biochemistry at the University of Cape Town the University of Transkei has initiated a molecular DNA research laboratory. At present this has very limited capability. However, given this Medical Faculty's placement in a community of approximately 4 million people, which it services, the possibility of establishing a cytogenetic laboratory and developing the molecular DNA laboratory should be encouraged.

Cytogenetic Laboratory - North West Province

The cytogenetic laboratory situated at Potchefstroom is considered by the committee to be non-viable and has the added disadvantage of not being associated with a clinical genetics service. It is currently handling fewer than 100 samples per year with a staff complement of one cytogeneticist, and a part-time laboratory assistant.

7.3 RESEARCH AND DEVELOPMENT, TEACHING AND ACADEMIA

Presently almost all activities in medical genetics in South Africa, including service, research and development, and teaching and training, occur in Academia. Some work, mainly connected with laboratory services, occurs in private practice. The continued undertaking of research and development, and teaching and training, for the foreseeable future, will depend on the academic institutions, and is therefore dependent upon the relationship that will be developed and maintained between them and the proposed NHLS. To date the nature of this relationship has not been determined, and falls outside the ambit of this report. Given the disparity in the way that different medical genetic departments are structured and financed it must be presumed that this future relationship would have to be negotiated with the individual departments, which will be guided in these negotiations by the policies of their parent University.

The exact nature of this future relationship was a concern of many during the consultations of the subcommittee. The manner in which medical genetic departments, units and laboratories are integrated into the NHLS can be expected to have significant consequences for their structure, functioning, staffing and financing. It will thus determine their ability to undertake research, development, teaching and training. It was not possible to discuss specifics related to these issues without having details of the proposed relationship between the NHLS and academic laboratories. Given the precarious situation in medical genetics in South Africa, all of these aspects of academic endeavour

will be essential to ensure the continued viability of medical genetics and will need to be addressed as part of the future negotiations to integrate the academic departments into the NHLS.

7.4 FUNDING

Funding of academic laboratories within the NHLS to undertake service work is an issue that still has to be determined. However this funding would have to be supplied by the PBEs within which the laboratory resided. In addition to the funds necessary to perform the task at hand it is suggested that consideration be given to the allocation of a proportion of the funds generated to bench research and special developments in the laboratories and some form of departmental incentive scheme.

Currently funds for genetic laboratory tests are in the Human Genetics Sub-directorate budget. It is recommended that these funds are kept in this Sub-directorate until such time as genetic services are developed in all the provinces, and all provinces are able to allocate a portion of their budget to pay for genetic tests. The present situation is that because the provision of genetic services is not seen as a priority, it is difficult in some provinces for clinicians to request genetic tests for their patients.

8. EVALUATING A HUMAN GENETICS PROGRAMME

The short term goal is to provide basic comprehensive genetic services for the diagnosis, management, counselling, and prevention of birth defects, inherited disorders and disabilities to parents, affected individuals, and their families.

Goal of Collecting Data

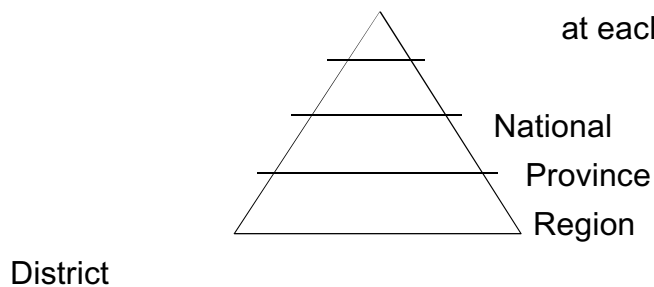
- know the incidence and prevalence of genetic conditions
- determine priorities for intervention
- effective planning
- set objectives
- evaluate and provide feedback

8.1 DATA COLLECTION

Maintain a national database, to be able to get information about changes and trends; time related (seasonal), geographical, based on ethnic differences.

Establish a standardised, user friendly, single genetic notification form which fits in with the Health Information Systems at provincial and national levels.

A pyramid system for the type of detail required at each level is suggested.



Form needs to be simple. Defects should be left in terms of plain language description where it is not possible to make a diagnosis.

Clinical guidelines are needed for proper diagnosis.

Training is required for accurate coding.

Efficiency of data collection is important

Monitor proportion of genetically trained health care workers in the districts,

and monitor that they are appropriately utilised.

8.2 MONITORING OF BIRTH DEFECTS IN THE IMMEDIATE POST BIRTH PERIOD

Basic information: Name, age, sex, gestational age, gravity, parity (include still borns at term), hospital/clinic name, self reported ethnicity.

Conditions to report: identifiable (or measurable) within 24 hours after birth.

Criteria

live births

in newborn period up to a week

first contact with clinic/hospital in the immediate post birth period (within 7 days).

Monitor the incidence of selected birth defects namely:

Neural tube defects

Down syndrome

Albinism

Microcephaly

Isolated Cleft lip and palate

Isolated Hydrocephalus

Other congenital defects (describe)

**INFORMATION TO BE CONTAINED IN A FORM FOR RECORDING OF BIRTH
DEFECTS IN THE IMMEDIATE POST BIRTH PERIOD**

Name

Place of birth

Date of Birth

Hospital name/clinic name

Self reported ethnicity

Sex

Gravity

Parity (includes still borns at term)

Gestational age

CONDITIONS TO BE MONITORED

Neural Tube Defects

Down Syndrome

Albinism

Microcephaly (2 standard deviations below mean of head circumference measurement)

Isolated Cleft lip and Palate

Isolated Hydrocephalus

Other congenital defects (describe/specify)

Stillbirths should be omitted from the reporting because there is poor diagnosis of genetic conditions.

Born before arrival's should also be excluded because of the possibility of counting them more than once.

8.3 OBJECTIVES AND INDICATORS FOR HUMAN GENETICS

PROBLEM STATEMENT: Most congenital / genetic disorders in South Africa are either unrecognised or inappropriately managed. Integration of genetic services into the comprehensive PHC service will ensure the long term decrease in birth defects, inherited disorders and disabilities.

GOAL	OBJECTIVES	INDICATORS
<p>To provide a basic comprehensive genetic service for all individuals to enable them to live and reproduce as normally as possible</p>	<p>Reduce the morbidity of individuals with, or at risk of congenital / genetic disorders. Priority conditions include Down syndrome, neural tube defects, and albinism</p> <p>Provide policy guidelines for the appropriate management (including diagnosis, treatment, counselling and referral) of individuals and families with congenital / genetic disorders</p> <p>Promote awareness of congenital / genetic issues through educational programmes</p>	<p>Proportion of individuals with, or at risk of congenital / genetic disorders</p> <p>Proportion of districts with genetically trained staff and the proportion of districts rendering a basic genetic service as part of the comprehensive PHC service</p> <p>Number of genetic educational programmes per district</p> <p>Number of individuals reached by genetic educational programmes</p>

9. ETHICAL GUIDELINES

9.1 GENERAL ETHICAL GUIDELINES IN MEDICAL GENETICS

[Modified from the WHO document; Hereditary Disease Programme, 1995]

- 9.1.1 Genetic services in a nation should be available equally to everyone regardless of ability to pay and should be provided first to those whose need is greatest.
- 9.1.2 Genetic counselling should be as non-directive as possible
- 9.1.3 All genetics services, including screening, counselling, and testing, should be voluntary, with the exception of screening newborns for conditions for which early and available treatment would benefit the newborn.
- 9.1.4 All clinically relevant information that may affect the health of an individual or fetus should be disclosed.
- 9.1.5 Confidentiality of genetic information should be maintained except when there is a high risk of serious harm to family members at genetic risk and the information could be used to avert this harm.
- 9.1.6 Individual privacy should be protected from institutional third parties, such as employers, insurers, schools, commercial entities, and government agencies.
- 9.1.7 Pre-natal diagnosis should be performed only for reasons relevant to the health of the fetus and only to detect genetic conditions or fetal malformations.
- 9.1.8 Choices relevant to genetic services, including choices about counselling, screening, testing, contraception, assisted procreation, and termination of pregnancy following pre-natal diagnosis, should be available on a voluntary basis and should be respected.

- 9.1.9 Adoptive children or children conceived from donor gametes should be treated equally with biological children under the guidelines.

Duties to family members. *In genetics the true patient is a family with a shared genetic heritage. Family members have a moral obligation to share genetic information with each other. If children are intended, individuals should share information with their partners.*

9.2 ETHICAL PRINCIPLES FOR GENETIC PROFESSIONALS

(Taken from Baumiller *et al*, 1996)

Professionals interact with patients; diagnose, manage and counsel.

Responsibility of the genetic professional to patients and family -

- 1) Serve patients and their families with equity and with respect for each person's feelings, beliefs, ethno-cultural traditions, and social circumstances
- 2) Provide counselling that is non directive, supportive, responsive to the patient's requests, and should respect the choices of patients and families
- 3) Convey information sensitively to the patients, and in a language they understand, so they can make independent decisions and give informed consent
- 4) Refer when indicated or requested to other experts for specialised services or to confirm diagnosis
- 5) Honour the confidentiality of information, shared in the relationship with patients and their families
- 6) Urge patients and families to share genetic information, with relatives at risk, pointing out the possible need for this, early in the relationship

- 7) Inform persons who participate as subjects of research that they may refuse testing, or refuse to receive information, and they may withdraw from research programmes, at any stage without change in care

- 8) Inform patients of possible conflicts of interest, or possible commercial and other uses of their biological materials, direction of storage, and confidentiality. [Cannot use a biological specimen for something that was not initially specified. Patient has the right to demand that you only do the test that the specimen was originally intended for and nothing else. An information sheet with these details has been drawn up by the Southern African Society of Human Genetics].

10. OUTREACH CLINICS/HUB AND SPOKE MODEL

The service needs to be expanded and it is suggested that it follows the “hub and spoke” model which has been very successful in other countries. The Provincial Health Department in each province constitutes the “hub”; with academic centres providing the support and expertise; and the “spokes” extend out through district based services to the peripheral and rural hospitals and clinics, thereby facilitating referrals and access. The laboratory services need to be at the hub and should be controlled on a regional and national level to avoid duplication and maintain quality control. The genetic counselling service can be organised at different levels on a decentralised basis. There should be vertical support for the integrated service provision at district level. This will give the advantage of both equality and economy, and maintain high standards.

The financing of the outreach clinics cannot be left to the academic institutions who have responsibilities for teaching and research as well as service, but should be taken up by the Departments of Health at provincial level. The professional staff employed should be the administrative responsibility of their national and province but supervision and quality control monitoring can be the responsibility of the National, Provincial office and academic centre.

11. FINANCIAL IMPLICATIONS

11.1 CURRENT COSTS:

The cost to society of individuals with significant genetic disorders and birth defects includes

- medical care for the condition and its complications
- special education for some conditions
- loss of income for the care giver
- loss of tax revenue that could have been generated by the individual with a genetic disorder / birth defect.

The medical care currently provided for individuals with genetic disorders and birth defects is not optimal.

Table 1 lists some of the medical procedures required to manage Down syndrome, Neural Tube defects and albinism.

Using **Down syndrome** as an example, each individual requires on average R15,000.00 - R20,000.00 / year for basic medical care. An additional once off cost of R50,000.00 for a heart operation is needed.

In 1996, there were 1,312,040 births in South Africa. Given the incidence of 1 in 500 for Down syndrome, it can be estimated that at least 2,624 children with Down syndrome were born in 1996.

The cost of care for children with Down syndrome vary depending on the type of treatment. Table 2 indicates the possibilities for two different methods of caring for children with Down Syndrome till age 10; method A = R71,000.00 and method B is R124,000.00 per patient.

Cost of medical care (for 1st 10 years)

S Method A

$$R71,000.00 \times 2624 = R186,304,000.00$$

S Method B

$$R124,000.00 \times 2624 = R325,376,000.00$$

TABLE1: SUMMARY OF THE SUB-DIRECTORATE: HUMAN GENETICS' NATIONAL GUIDELINES FOR THE MANAGEMENT, TREATMENT AND COUNSELLING OF THE MOST COMMON GENETIC DISORDERS, BIRTH DEFECTS AND DISABILITIES

DISORDER	INCIDENCE	POP GROUP	TREATMENT	COST
DOWN SYNDROME	1 in 500	All	Physiotherapy (First two yrs) + Speech therapy (two weekly for 5-6 yrs) + Occupational therapy (two weekly for 8 yrs) or Holistic home programme (Weekly for 10 yrs) <i>60% of cases have heart problems</i> Heart sonars (at least 4 sonars before the operation) Operation (once in lifetime) <i>Other</i> Medication (for recurrent infections) Grommets (every 2 yrs) Blood tests (check for hormonal disturbances)	R4,000/child/year R2,000/child/year R2,000/child/year R4,000/child/year R35,000 - R50,000/child/year R2,000/child/year R1,000/child/year R1,000/child/year
NEURAL TUBE DEFECTS	3 per 1000	All	VP Shunt (once off) Surgery to close lesion (once off) Calipers (every 2 yrs) Antibiotics (daily) Incontinence garments (daily)	
ALBINISM	1 in 4000	All	Visual assessment (every 2yrs) Glasses (every 2 yrs) Sunscreen (daily) Lip protection (daily) Moisturizer (daily)	
FETAL ALCOHOL SYNDROME				

**TABLE 2: GENETIC DISORDERS - COSTS OF TREATMENT
(RAND 000s @ 1999 PRICES)**

Down Syndrome

Method A	Year 1	Year 2	Year 3	Year 4	Year 5	Year 6	Year 7	Year 8	Year 9	Year 10	TOTAL
Physiotherapy	4	4									8
Speech Therapy	2	2	2	2	2	2					12
Occup. Therapy	2	2	2	2	2	2	2	2			16
Medication	2	2	2	2	2	2	2	2	2	2	20
Grommets		1		1		1		1		1	5
Blood Tests	1	1	1	1	1	1	1	1	1	1	10
TOTAL											71

Method B	Year 1	Year 2	Year 3	Year 4	Year 5	Year 6	Year 7	Year 8	Year 9	Year 10	TOTAL
Holistic Programme	4	4	4	4	4	4	4	4	4	4	40
Heart Sonars				4							4
Operation				45							45
Medication	2	2	2	2	2	2	2	2	2	2	20
Grommets		1		1		1		1		1	5
Blood Tests	1	1	1	1	1	1	1	1	1	1	10
TOTAL											124

These figures are only for the care of children with Down syndrome. This does not include the cost of caring for these individuals when they have other complications such as Leukemia or Alzheimers.

Using Neural tube defects as an example:

The incidence of neural tube defects in urban areas is 1 in 1000 and it ranges from 4 in 1000 to 6 in 1000 in rural areas. Using an average incidence of 3 in 1000 it can be estimated that at least 3936 children were born with neural tube defects in 1996.

If the lifetime cost of caring for an individual with NTDs is conservatively estimated to be R30,000.00, the cost of providing health care for each cohort will be as follows:

$$\begin{aligned}\text{Lifetime cost} &= \text{R}30,000.00 \\ &\quad \text{R}30,000.00 \times 3936 \\ &= \text{R}117,000,000.00\end{aligned}$$

Approximately 150,000 of the children born in 1996 will have a significant genetic disorder or birth defect by the age of five years and it is therefore expected the costs for the care of children with genetic conditions will be substantial.

11.2 PLANNED COSTS:

11.2.1 Health Promotion

Inclusion of genetic message within other health promotion messages. If planned together with other MCWH programmes extra provincial costs should be minimal

National has a budget for preparing some educational materials to assist with campaigns

Collaboration / partnership with NGOs is currently being established - one of the planned shared responsibilities is that of campaigns.

11.2.2 Initial costs for Training

(i) Genetically trained nurses

- The use of donor funds to run short courses initially will be explored
- Future courses should be run within provinces as part of other training offered by Human Resource Development.

(ii) *Clinical geneticists*

This requires specialists to spend two years in an approved facility under supervision. Provinces need to agree on releasing Paediatricians / Obstetricians for training or to make arrangements for outreach clinics from established Human Genetics Departments/ Divisions to provide ongoing training within the Provinces.

(iii) *Genetic counsellors / genetic nurse counsellors*

6 months post basic training (nurses)

2 years (genetic counsellors)

(iv) *Laboratory personnel*

3 - 4 years training / person.

11.2.3 Implications for Education and Training if the UK Report on Clinical Genetics Services into the 21st Century is adhered to

The funding requirement for the professional staff and their immediate 'tools of trade' can be estimated for each province. The estimates are based on a team - 1 Clinical Geneticist, 4 Counsellor / Nurses and 5 Laboratory Scientists for every 500 000 people - and the population forecasts for each province for April 2000.

Province	Forecast Population April 2000 (Millions)	Number of Teams Required	Cost per Team R000s	TOTAL COST R000s
Eastern Cape	6.75	13	1686	21,918
Free State	2.82	5	1686	8,430
Gauteng	7.93	16	1686	26,976
KwaZulu-Natal	9.06	18	1686	30,348
Mpumalanga	3.06	6	1686	10,116
Northern Cape	0.88	2	1686	3,372
Northern Province	5.45	11	1686	18,546
North West	3.62	7	1686	11,802
Western Cape	4.23	8	1686	13,488
TOTAL	43.80	86		144,996

Note

1. Forecast populations use Statistics South Africa Formulae for calculating changes from the October 1996 Census.
2. The Cost per Team estimate is made up of:

Unit Cost		Rand
263956	1 x Clinical Geneticist (Level 13)	263,956
116185	4 x Counsellor/Nurse (Level 8)	464,740
147502	5 x Lab. Scientists (Level 9)	737,510
	Related equipment/stationery/expenses	<u>219,931 *</u>
		1,686,137

Unit Cost = Last Notch (July 1999) + 30% for Pension etc.

* = 15% of employment costs.

The further funding issues about the scale of training and the infrastructure for diagnosis, counselling, treatment and management are - for this report - assumed to be already available and do not, therefore, require separate calculation. That assumption is reasonable if it is considered that the necessary qualifications are offered already and there is already access to diagnosis and treatment, albeit confined to academic hospitals.

12. SUMMARY AND CONCLUSION

The concept of regional centres each serving 2-5 million people is of value in providing a genetics service headed by a skilled clinical genetics team. However the peripherally based services need to be developed. Future plans should allow for the family-based nature of clinical genetics, the frequent crossing of provincial boundaries, the recognition that genetic services are a part of many facets of health care, that people should have choices opened up for them, and that new developments need to be incorporated (such as counselling for breast cancer). South Africa has an excellent but limited genetic service at present; an excellent and accessible service can be provided in the future, if the policy outlined in this document is adopted.

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APPENDIX

ABBREVIATIONS

BSc	: Bachelor of Science
CBO	: Community Based Organisation
CPN	: Chief Professional Nurse
CSS	: Central Statistical services
DOH	: National Department of Health
FISH	: Fluorescent In Situ Hybridisation
GEM	: Genetic Education Manual
GNC	: Genetic Nurse Councillor
GTNS	: Genetic Trained Nurse
ICBDMS	: International Clearinghouse for Birth Defects Monitoring Systems
HPCSA	: Health Professional Council of South Africa
MCWH	: Maternal, Child and Women's Health
MGEP	: Medical Genetics Educational Programme
MNS	: Medical Natural Scientist
MSc	: Master of Science
NGO	: Non Governmental Organisation
NPO	: Not for profit organisation (includes NGOs and CBOs)
NSM	: Nursing Service Manager
NTD	: Neural Tube Defects
PEP	: Perinatal Educational Programme
PHC	: Primary Health Care
PU/CHE	: Potchefstroom University
SA	: South Africa
SAIDA	: Southern African Inherited Disorders Association
SAIMR	: South African Institute for Medical Research
SASHG	: Southern African Society of Human Genetics
SPN	: Senior Professional Nurse
TBH	: Tygerberg Hospital
TOP	: Act on the Choice on Termination of Pregnancy
UCT	: University of Cape Town
UK	: United Kingdom
UOFS	: University of Orange Free State
UP	: University of Pretoria
US	: University of Stellenbosch
WHO	: World Health Organisation