Genetic Services in Egypt:

Current Situation and Needs Assessment

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Capacity Building for the Transfer of Genetic Knowledge into Practice and Prevention

Prepared by

Randa Kamal A. Raouf

Professor of Pediatrics,

Institute of Postgraduate Childhood Studies

Ain-Shams University

Cairo, Egypt

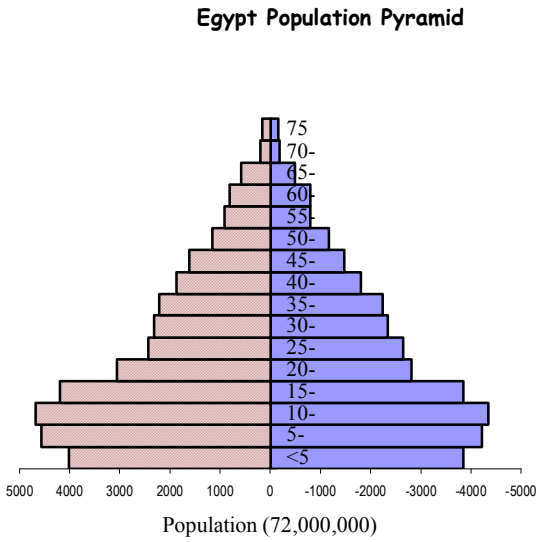
Map of Egypt

● **Genetic counseling clinics**

Health indices:

The Egyptian population is the largest among Arab countries; it has now reached approximately 78.7 millions and is rapidly growing. The total fertility rate is 3.1/woman, the birth rate is 22.9/1000 population and the number of live births per year is approximately 1.8 millions. The death rate is 5.09/1000 population. Consanguineous marriages reached 37.9% in rural areas and 24.1% in urban areas (EDHS, 2006).

Egypt is one of the countries passing through a phase of epidemiological transition and although infectious diseases and malnutrition are not yet fully controlled still there is a great improvement with a reduction of below five years mortality rate from 130/1000 live births in 1985 to 28/1000 live births in 2008. Infants mortality rate was also reduced from 97/1000 live births to 24/1000 live births. Maternal mortality rate was reduced from 174/100.000 live births in 1990 till 55/100.000 in 2008. We have already achieved the millennium development goal for the under five mortality rate and we are so close in achieving it for the maternal mortality (EDHS, 2005,2008 & MOHP 2008).



This progress is due to an improvement in maternal and child health care services provided through the 3960 primary health care (PHC) facilities that belong to the ministry of health and population (MOH&P) and are distributed allover Egypt.

The most relevant PHC preventive programs provided by the ministry of health:

- **Maternal programs:**
  1. The prenatal follow-up program; where regular follow-up of the pregnant women is done by well trained PHC physicians. The service includes thorough physical examination, ultrasonographic imaging of the fetus, laboratory investigations,

health education sessions on the avoidance of teratogens, proper nutrition and appropriate multivitamins prescriptions.

2. The essential obstetric care; all the hospitals and the majority of the PHC centers that belong to the MOH&P have now well prepared delivery rooms and resuscitation rooms. There is a training program for physicians, nurses and birth attendants for conducting safe, normal deliveries.
3. Postnatal care is an important program for the wellbeing of the mother and the newborn. In rural areas, nearly 60% of the deliveries occur at home, an outreach program has been implemented in some governorates where well-trained PHC nurses provide postnatal care services including checking for any postpartum complications, examining the newborns for any detected abnormalities, taking dried blood spot samples for neonatal screening for congenital hypothyroidism and offering health education messages to the mother concerning hygiene, nutrition, breast feeding and care of the newborn. The nurses report all the findings to the responsible PHC physician in the corresponding PHC setting.

- **Child care programs:**

Egypt adopted an organized MCH program in 1927 that was comprehensive by the state-of-art at that time. Since the 1980s, MOH&P adopted a CDD, ARI and EPI programs; which significantly reduced child mortality and morbidity caused by common preventive diseases. This success encouraged the Ministry to continue upgrading child programs and developing new interventions. Some of the current programs are exclusively promotional (e.g. breastfeeding), others are purely preventive (e.g. EPI), some are curative (e.g. ARI), but some combine more than one field (e.g. IMCI combines the 3 fields, and Care of children with special needs works on early detection, prevention, treatment and rehabilitation. Current childcare programs are coordinated at the central level as they fall under PHC undersecretary. At district level, they are under the supervision of one DHO assistant. At the PHC facility level, the same health provider implements all programs. Most of the programs already have their protocols, and others are under way. These current programs are:

1. Monitoring under-5 child growth and development program.
2. Neonatal care program.
3. Promotion and support of breastfeeding.
4. Micronutrient supplementation program.
5. Expanded program of immunization (EPI).
6. Control of diarrhoeal diseases program (CDD).
7. Control of acute respiratory infections program (ARI).
8. Integrated management of childhood illness program (IMCI).
9. National Neonatal Screening Program for Congenital Hypothyroidism (NNS).

Relevant preventive child care programs:

❒ Monitoring under-5 child growth and development program:

The program has been established on a regular basis with the development of *"Child Health Card"* in September 1996 for the purpose of detecting deviation in growth and development, and early detection of ill health &/or disability. It articulates on 3 activities:

- Monitoring weight and height for age to assess protein-energy malnutrition.
- Measuring the blood hemoglobin level to assess anemia.
- Monitoring mobility development, hearing & speech, and social behavior, using Denver II developmental scale.

To ensure highest coverage, the program policy links the periodicity of monitoring children up to age 18 months with immunization visits in addition to the first contact at birth or within 15 days, and the end of each year of age, as well as any visit for medical care. HIO shares in implementing this program. Data generated during each visit, is recorded in the child's health card given to parents at the time of birth notification. The service covers all PHC facilities at national level. No doubt that tying the monitoring occasions with EPI schedule is useful in increasing coverage, the issue to be considered is the quality of the service, given that the day of immunization session is generally a busy day.

The program plans to extend use of the newly developed growth curves up to age 21 years, and expands measures to include head circumference, and body mass. It also plans to increase public awareness about assessing child development, and training providers on the Denver II developmental scale.

❒ Neonatal Care Program:

The program was initiated in 1992 in light of the high proportion of neonatal deaths. It started with 30 hospitals by providing a limited number of incubators. At present, the program is implemented at national level. Two hundred neonatal care units are now established in hospitals that belong to the ministry of health and are classified into 3 categories according to the level of care, and skills and equipment needed; either normal care units, intensive care units and tertiary care units. A defined referral system from each level up is in place. Each level is equipped with the relevant equipment, as well as an ambulance equipped at least with an incubator. Though the program is hospital-based, it is established and coordinated by the PHC sector.

As a result the case fatality rate in neonatal care units dropped from 23.0% in 1995 to 16.4% in 2002. The top cause of death is respiratory distress syndrome particularly among the "very" low birth weight children (<1.5 kg).

The important constraints are:

- Shortage in financial resources to cover the high costs of care, and in "some" governorates HIO does not cover the costs of needed ancillary services e.g. laboratory tests, blood transfusion, and radiography.

- Some units need renovation to control pollution and infection.
- The need to maintain some equipment with limited allocated budget.

❒ Micronutrient Supplementation Program:

As micronutrient deficiencies have an effect on body functions and resistance, MOH&P is implementing a supplementation program.

- Iodine:

The 1992 nutritional survey showed that the iodine deficiency rate surpasses the recommended rate of 5% for different age groups in all governorates. MOHP studied iodization of table salt with the principal producing company, which was provided with the needed equipment with UNICEF support. Production started in the year 1996. Iodized salt is the single salt legally allowed to be commercially marketed since the year 2000. By that time, MOHP generalized the application of iodine detector in the course of the "food control program". Results showed that the proportion of its availability in the market reached 94%. MOHP supported DHS testing iodine content of table salt available at households since 2000. The rate of households using iodized salt appreciably increased from 55.9% in 2000 survey, to 69.2% in 2006 survey. This result implies that non-iodized salt is still marketed or individually got from places producing sea or rocky salts. However, it is believed that its actual use is limited to the freshly prepared foods rather than other use such as pickling. Results of recent research prove that iodine deficiency is the cause for the high incidence of congenital hypothyroidism in some areas in Egypt.

- Vitamin "A"

The 1995 nutritional survey found that the rate of sub-clinical deficiency (plasma retinol <10/100 ml) was 11.9% among children and 10.2 among mothers. In light of these results, MOHP started vitamin "A" national supplementation for children from January 1999, and for mothers one year later. The policy calls for giving 2 doses for children along with vaccination sessions; 100,000 units at 9 months and 200,000 units at 18 months, with at least 4 months apart. Mothers are given a single dose of 200,000 units within 4 weeks of childbirth. Program data show that coverage reached 97% of children and 84% of mothers in 2005. However, the impact of this program is not measured yet.

- Iron:

The issue of iron deficiency was raised in Egypt since a long time, as different surveys found that anemia is highly prevalent among children and pregnant and lactating women; reaching around 30%. The rates vary by social class and residence, higher among the low class and in South Egypt. It was found that anemia is primarily a result of iron-folic acid deficiency. Until present, there is no consensus on the 1st intervention thought of; i.e. bread fortification. However, bread fortification is being piloted at Fayoum Governorate. As a result of the arguments on bread fortification, MOPH initiated a supplementation program in January 1999. The program policy requires giving a weekly dose of iron preparation for children, in addition to iron-folic acid supplementation for pregnant and lactating women. The

program is implemented at all PHC facilities the national level. MOH 2003 data shows that coverage reached 85% of pregnant and lactating women. Unfortunately, child coverage is not reported. On the other hand, general information indicates that child foods fortified with iron and vitamin "A" are available in the market, but only accessible to the minority who can afford its cost.

❒ Expanded Program for Immunization (EPI):

The immunization program is one of the oldest programs in Egypt; initiated in 1891. Egypt was a pioneer country in eradicating smallpox long before initiation of the global campaign for its eradication. BCG vaccination was introduced in 1956, and polio vaccination in 1968. The program was evaluated in 1984, and upgraded to EPI in 1985, with special focus on the cold chain. Then after, two more vaccines were added, hepatitis B in 1992, and MMR in 1999. The program thus addresses 9 diseases, rather than the 6 globally targeted diseases.

The program purposes are:

- Reduction of incidence and specific mortality from targeted diseases.
- Eradication of poliomyelitis.
- Elimination of neonatal tetanus and measles diseases.
- Prevention of mumps and rubella.

The program strategy calls for:

- Immunizing all children against the 9 diseases.
- Organizing national and limited campaigns against polio, tetanus neonatorum, rubella and measles, implemented in collaboration with WHO, UNICEF and Rotary Club.
- Coverage is continuing to be over 90% over the past 15 years.

❒ Integrated Management of Childhood Illness (IMCI) Program:

IMCI is a strategy rather than a program, however Egypt dealt with it as a program in MOHP structure. It is based on both CDD and ARI programs. The strategy is characterized by:

- Being a holistic approach, integrating sick child management of the common diseases into one protocol, requiring full examination of the sick child irrespective of the complaint.
- Combining preventive aspects with case management; such as immunization, breast-feeding, child feeding and other factors influencing child health and development.
- Emphasizing child home care to complement the care provided at health facility during health and illness.

National newborn screening program for congenital hypothyroidism:

- Neonatal screening for congenital hypothyroidism (CH) is an essential and productive preventive public health program. The Egyptian Ministry of Health and Population started to implement the screening program for (CH) in year 2000, and by the end of year 2003 all 27 governorates were covered.

- The aim of this program is to decrease the incidence of mental retardation caused by (CH), and to provide free of charge services for diagnosis and management for the discovered cases in different specialized and public health facilities.

- The Screening method is primary TSH measurement in dried blood spots collected from neonates aged 3-7 days using the ELISA technique.

- The cut-off point for the neonatal TSH is ≥ 15 µu/ml.

- Collecting blood samples took place through 3200 public health facilities, in addition to implementing an outreach program for the underserved population.

- Samples are transferred to 14 central & regional laboratories where measurements of neonatal TSH & other confirmatory tests take place.

Diagnosis, treatment and follow up of cases discovered through the screening program take place in 25 health insurance centers geographically distributed all over Egypt.

- Results: A total of 4,778,549 neonates were screened, out of 6,434,844 live births till the end of 2005.

- The coverage ranged from more than 95% in some rural governorates to 75% in urban governorates.

- The overall incidence of hypothyroid patients was 1/2020 in the year 2005.

- Percentile curves of TSH values for neonates and protocol for management and follow up were developed.

Congenital and genetic disorders in Egypt: current situation and available preventive and intervention services

Magnitude of the problem of genetic and congenital disorders in Egypt

Analysis of the available epidemiological data clearly indicates that hereditary disorders and congenital malformations are rapidly becoming a major public health concern in Egypt.

The prevalence of congenital and genetic disorders (among infants and young children) in Egypt is estimated to range from 2.8% in urban areas in metropolitan governorates to 8.4% in rural areas in Upper Egypt. This relatively high prevalence might be attributed to several factors including:

• The high rate of traditional consanguineous marriages, which increases the frequency of autosomal recessive disorders.

• A relatively high birth rate of infants with chromosomal disorders related to advanced maternal age such as Down syndrome and other trisomies;

• A relatively high birth rate of infants with malformations due to new dominant mutations related to advanced paternal age.

• Large family sizes, which may increase the number of affected children in families with autosomal recessive conditions.

• The lack of public health measures directed at the prevention of congenital and genetically determined disorders..

• Heavy economic, social and health burdens are imposed on the affected family as well as the society. In general, genetic diseases are relatively prevalent among the Arab population, and are a significant cause of morbidity and mortality in this population.

Based on known incidence figures, these are some examples of the genetic impact on health:

- 3-5% of all births result in congenital malformations
- 20-30% of all infant deaths are due to genetic disorders
- 30-50% of post-neonatal deaths are due to congenital malformations
- Down syndrome (1/800 live births and increases with advanced maternal age)
- Old maternal age (risk of chromosome aneuploidy)
- Fragile X syndrome (1/1,000 males and 1/800 female carriers of which 30% will be mentally retarded)
- Duchenne muscular dystrophy (200/million male births)
- 11.1% of pediatric hospital admissions are for children with genetic disorders and 18.5% are children with other congenital malformations .
- 12% of adult hospital admissions are for genetic causes.
- 0.5% of all newborns have a chromosomal abnormality
- 7% of all stillborns have a chromosomal abnormality
- 50% of individuals found to have mental retardation have a genetic basis for their disability
- 15% of all cancers have an inherited susceptibility
- 10% of the chronic diseases (heart, diabetes, arthritis) which occur in the adult populations have a significant genetic component.

The first step to reduce the frequency of congenital malformations/inherited diseases is to choose combined strategies, including efforts focused on health education and health promotion, with an emphasis on the medical consequences of marriages within the family to find the cause of these diseases and identify the genes responsible for the disease. The Second step is to find cures for these disorders and map the demographic prevalence of genetic disorders.

Incidence of congenital malformations among Egyptians ranges from 1,16 to 3,17 % . This is probably due to the high consanguinity rate (20 – 40 %) among Egyptians. Early diagnosis of various genetic disorders and malformations with proper intervention (medical, hormonal, dietary, and interventional by stimulation of motor & cognitive development) will reduce

the burdens of genetic disorders at the individual, familial and community levels.

Today genetic facilities are widespread in developed countries. Genetic counseling and prevention of hereditary, congenital and common diseases of adulthood are available for these populations. In developing countries genetic services are still rare and absent in some countries.

The magnitude of the impact of genetic disorders on all societies is quite significant necessitating their control which can be principally achieved by prevention.

The model for prevention, control and care of genetic diseases incorporates: Services, Education, Teaching, Registry and Research, is entertained as follows:

1. Provision of services that must reach the entire population in a country in order to provide equitable health services to all citizens. This involves establishment of genetic clinics and genetic centers, well-trained and knowledgeable family doctors, nurses and social workers and consultants.
2. Education for the health care personnel, including the family doctors as partners in these services.
3. Teaching to improve knowledge of the medical and non-medical personnel, patients, their families and the general public at large.
4. Preparation of the Registry of genetic diseases in the population.

5. Research to identify the molecular pathogenesis of the genetic diseases, establish more definitive methods for the identification and diagnosis of the genetic defect, establish appropriate management strategies and identify ways and means of prevention and control.

This program incorporates field research, laboratory investigations and clinical trials for management of commonly encountered genetic disorders in our population.

This protocol requires a concerted national effort to determine what environmental, genetic, occupational, nutritional, and behavioral factors cause or contribute to birth defects.

Prevention

To initiate a nationwide intervention program for the control of any health problem, there are two prerequisites. The first is evidence that the magnitude of the problem is significant, and the second is an indication that prevention is both feasible and cost-effective. The health care needs of most Arab populations necessitate that this problem be addressed promptly. Moreover, great advances have been made in our knowledge of genetic disorders, and the principle of equity in health care demands that the gap

between medical progress and health care services should be narrowed whenever possible.

The prevention of genetic disorders could be effective at two levels:

- General population.
- Families and individuals at risk.

Health education programs improve health, identify and reduce disease risks, manage chronic illness, and improve the well-being and self-sufficiency of individuals, families, organizations and communities.

Primary prevention is conducted by genetic counseling prior to marriage or prior to the first pregnancy, and secondary prevention focuses on early genetic screening during the pregnancy.

In Egypt, genetic services cover diagnosis, screening and testing, counseling, education, clinical research, the ongoing management of individuals and families with particular birth defects and genetic disorders, and finally prevention of such diseases.

In order to provide the necessary tools for such prevention programs, genetic research must first characterize the common genetic diseases in the communities, and identify their responsible mutations.

Strategies and feasible approaches

While the overall objective of a national program for the prevention of genetic and congenital disorders in the community, the strategies adopted to achieve this objective should be carefully selected to match the unique demographic, cultural and religious characteristics of the population and should take into consideration the priorities set and the resources available. In all countries, irrespective of the resources available, certain public health measures capable of reducing the burden of genetic and congenital disorders can be feasibly implemented without major resource implications. These primary prevention measures, which should be integrated into primary health care, include the following:

• To reduce genetic disorders related to advanced parental age, such as Down syndrome and autosomal dominant conditions due to new mutations, as part of the family planning services.

• To reduce the occurrence of congenital abnormalities such as neural tube defects, and avoiding the sequelae of micronutrient deficiencies such as mental retardation due to iodine deficiency by promoting healthy nutrition for women.

• To prevent congenital rubella syndrome by immunizing against rubella infection.

• To reduce mortality and chronic handicap due to rhesus haemolytic disease through routine prenatal screening.

• To reduce congenital abnormalities and stillbirths by better control of maternal diabetes prior to and during pregnancy.

• To reduce the risk of miscarriage, congenital abnormality and fetal growth retardation through avoidance of smoking and alcohol intake during pregnancy.

• Avoidance of congenital abnormalities caused by certain infections such as syphilis by prevention, early detection and prompt treatment.

• To reduce the occurrence of hereditary disorders in high-risk families through genetic counseling.

• To provide information on the implications and availability of carrier testing for common disorders such as the haemoglobinopathies and PKU.

Genetic services in Egypt:

Genetic services are health measures implemented to help people with a genetic disadvantage and their families to live and reproduce as normally as possible.

Our Mission:

Genetic Services strive to improve the health of children who have genetic disorders. We try to provide all the Egyptian population with equal access to all of our services. We want to give opportunities for Egyptians to achieve the best progress in their pursuit of wellness.

We are trying to accomplish our mission by coordinating, providing and supporting:

* Direct access to genetic Health Care Services

*Availability of comprehensive and high quality genetic services

*Assessment, Planning and Policy Development

Strategic aims

1. To reduce morbidity and mortality from inherited conditions and birth defects.
2. To improve the quality of life for individuals and families impacted by inherited conditions and birth defects.
3. To empower people to make informed decisions about genetics and health.

Priorities for the provision of genetic services in Egypt:

- Coordination and Coverage of Services:

  People with a genetic condition need coordination of medical, social, and educational services. Medical genetic services are not well understood and are often poorly reimbursed.

- Access to Quality Services:

  There are few formal mechanisms in place to assess the quality of clinical and laboratory genetic services.

- Privacy & Confidentiality of Genetic Information:

  Genetic advances have resulted in increased concern for the privacy and confidentiality of genetic information, as well as the potential for genetic discrimination.

- Raising Awareness of Genetic Issues:

Rapid advancements in the field of genetics have resulted in the need for genetic education among a variety of people.

Clinical genetic services:

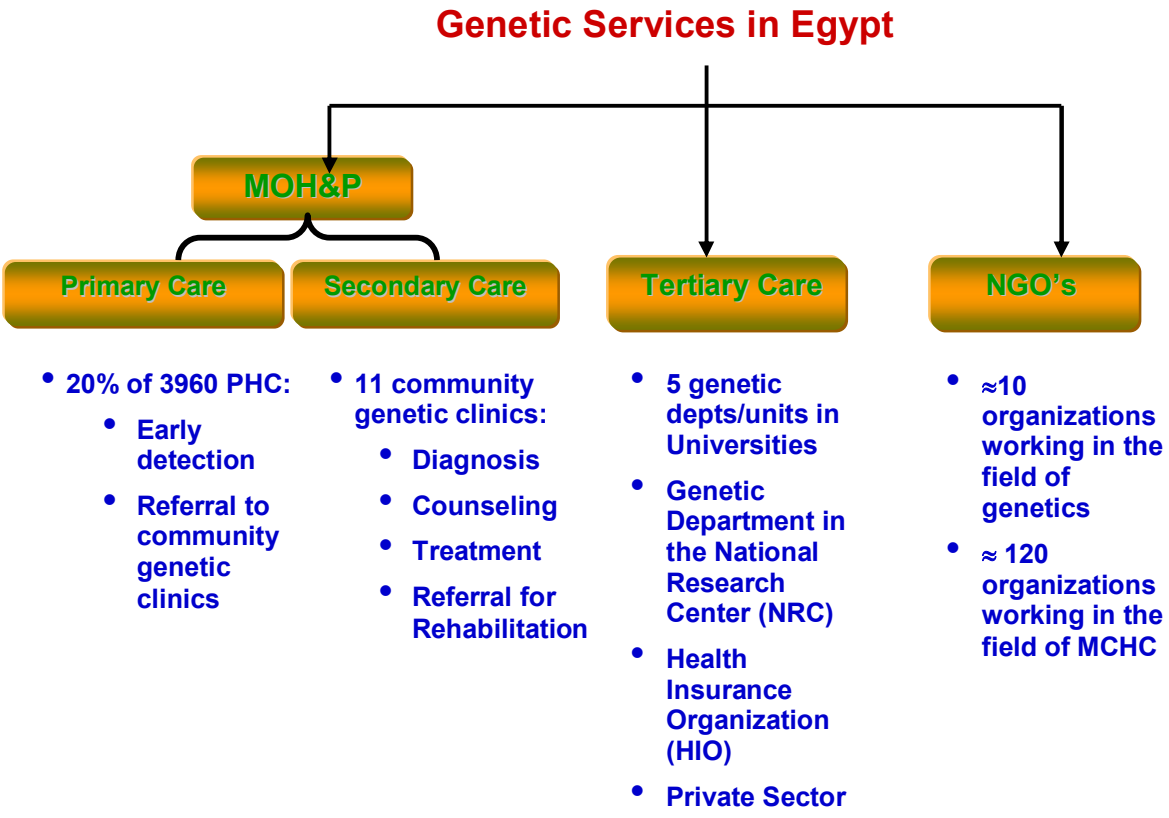
Currently available services in Egypt include:

- Clinical diagnostic services for individuals with birth defects and genetic disorders of childhood
- Diagnostic, predictive and counseling services for individuals at risk of, or affected by, adult onset genetic disorders (such as neurodegenerative disorders).
- Information and counseling for people considering prenatal diagnosis, or following the diagnosis of fetal abnormalities.
- Counseling of individuals, couples and families affected by (or perceived to be at risk of) genetic disorders or birth defects, and referral to appropriate community resources, including support groups.
- Identification of affected individuals and carriers of genetic disorders in extended family members, as appropriate.
- Involvement in the long term management of individuals with rare complex genetic disorders, such as inborn errors of metabolism.

Structure and logistics of genetic services in Egypt:

Genetic services in Egypt are provided through 4 main categories; primary, secondary, tertiary care and the NGO's. Primary and secondary care services are delivered through health facilities that belong to the MOH&P. Only 20% of the 3960 PHC facilities in the

country offer the services of early detection of genetic and congenital disorders. The facilities are mainly located in the catchment areas of the community genetic clinics.



At the secondary care level, 11 genetic counseling clinics distributed in different governorates in Egypt offer the services of diagnosis, counseling, and treatment of genetic disorders as well as referral for rehabilitation through the genetic counseling program.

❒ Genetic counseling program:

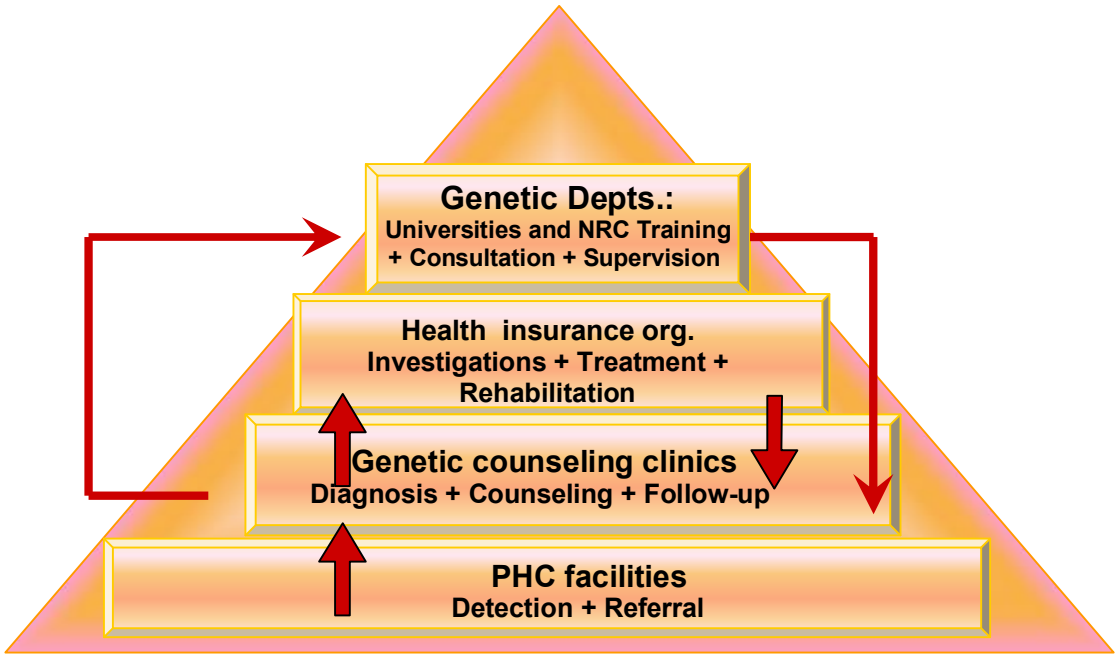
In 2002 a national committee for community genetic services was established. Its main objective was to develop policy guidelines for the prevention and management of genetic disorders. The committee recommended the necessary establishment of a national program for genetic services delivery. At the end of 2002 the minister of health responded to the request of the committee and signed an approval on a proposal presented by the children with special needs department/MOH&P that included an action plan and time frame for the implementation of a genetic counseling program.

The program started 5 years ago with the establishment of only one clinic in Giza governorate. Now there are 11 clinics in different governorates in Egypt.

The main objectives of the program are:

1. To provide genetic diagnostic and counseling services free of charge for the major part of the community who cannot afford to get such services.
2. To determine the prevalence of various genetic disorders among Egyptian children.
3. To raise community awareness regarding genetic diseases and the value of early detection and intervention.

The clinics work through a system of referrals from primary care to secondary care then tertiary care level of services. The early detection of genetic and congenital disorders is done at PHC level, and then cases are referred to the genetic counseling clinicsin the catchment area where services of diagnosis, counseling and follow-up are provided by well trained secondary care physicians. Continuous on-job training for the physicians, supervision, and consultation are provided by genetic University professors. Investigations, treatment and rehabilitation services are provided by the health insurance organization, and then cases are referred again to the clinics for follow-up.

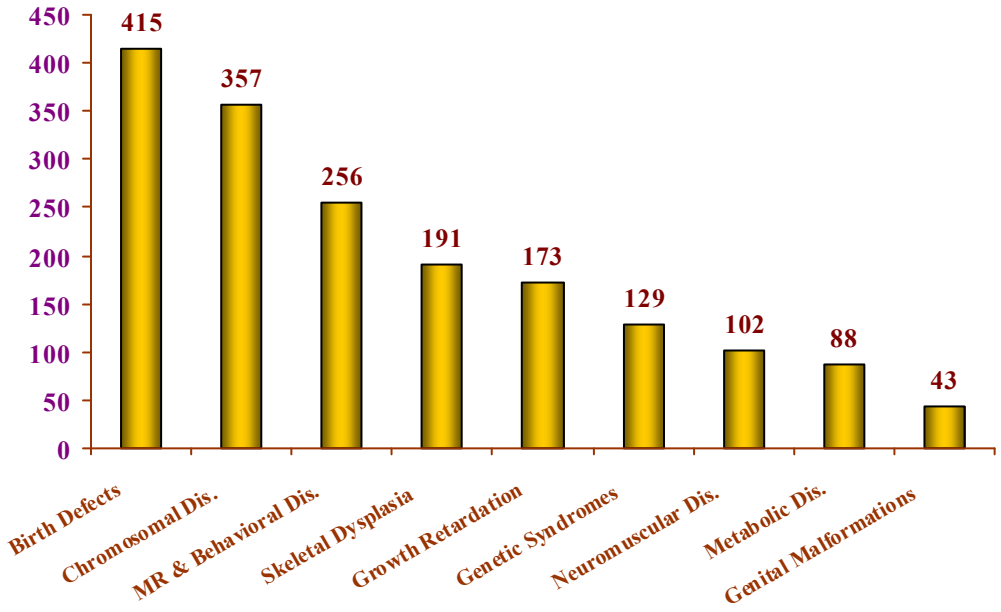


Egyptian module for Genetic Counseling Program

Genetic counseling clinics in Egypt:

Governorates	Locations of genetic counseling clinic
1- Cairo:	1. Nozha Health Center
	2. Moniera General Hospital
	3. Manshiat El-Bakry Hospital
2- Giza:	1. Om El-Atebaa Hospital
3- Alexandria:	1. El-Raml pediatric hospital
	2. Fawzi Moaz Medical Center
	3. El-Qabary Medical Center
4- Port-Saied	1. El-Manakh Medical Center
6- El-Sharquia	1. Menia El-Qamh General Hospital
7- El-Menia	1. Suzan Mubarak Center
8- Assuit	1. El-Walidiah Medical Center

The figure below shows the classification of cases seen in five of the genetic clinics from 2004 to 2006. Children with birth defects were the most commonly seen in the clinics, they constituted 25% of cases coming to the clinics, followed by cases with mental sub-normality and behavioral disorders representing 20% of cases, then chromosomal disorders representing 15% of cases with Down syndrome being the most common disorder. Severe forms of skeletal dysplasia and also growth retardation mainly short stature are often seen in the clinics.



Genetic Counseling Program Classification of Registered Cases (n=1653) in Five Genetic counseling clinics(2004-2006)

How to promote genetic counseling

In order to promote genetic counseling some decisions should be taken

1. Genetic disorders must occupy priority place in public health policy in our country. National or regional programs have to be established taking account of the epidemiological pattern of genetic diseases and the cultural structure of the community.
2. Education of population has to be developed, that will contribute largely to the success of health programs in this field.
3. Genetic counseling clinicsand genetic laboratories are to be established for genetic services provision.
4. Genetic counselors training.
5. Training programs for midwives can be conducted to facilitate the diagnosis of congenital abnormalities and prompt referral of affected neonates to appropriate centers.

Training of Genetic Counselors

Providing genetic counseling needs competence in clinical diagnosis, probability calculation and knowledge in interpreting laboratory diagnostic studies. Psychological, ethical, legal and Socio-cultural issues are also important in genetic counseling practice.

Training includes conferences and practical training, followed all together by physicians' evaluation.

Training curriculum provides the physicians with basic knowledge on:

- inheritance
- clinical expression of genetic diseases
- cytogenetic and molecular basis of genetic disorders
- Statistics for probability calculation.

Practical training will allow physicians to get the required knowledge and skills in the following fields:

- family history and pedigree drawing
- physical examination
- laboratory studies interpretation
- risk calculation
- communication of the risk assessment
- Ethical attitude

The following table shows the number and types of genetic education courses conducted to improve the knowledge and practice of the PHC nurses and physicians and the secondary care physicians who are working in the community genetic clinics.

Several training courses were conducted through the department of children with special needs. In cooperation with the genetic university professors and the national research center (NRC), the department was able to train PHC nurses on how to detect and refer children with birth defects. Seventy six orientation courses and 15 practical courses were conducted to teach and train PHC physicians on the detection of congenital and genetic disorders and referral to the community genetic clinics. This service was provided for children coming to receive PHC services at the PHC facilities.

Concerning physicians working in the community genetic clinics, they received condensed practical training courses. The duration of the course was 2 months. The courses were conducted in 3 main genetic departments in university hospitals and the NRC. The physicians also attended several specialized courses, for example courses on dysmorphology, premarital counseling, genetic lab results interpretation, and prenatal testing and diagnosis.

Continuous on-job training was provided to those physicians by the genetic professors who visit the clinics weekly for supervision and offering genetic consultation for those coming to receive genetic counseling services.

Medical Genetic Education Courses (2002 – 2006)

Target Population	Type of Training Course				No. of Trainee
	Theoretical		Practical		
	No.	Duration	No.	Duration	
1) PHC Nurses	10	3 days	15	3 days	116
2) PHC Physicians	76	2 days	15	3 days	1371
3) SHC Physicians	32	3-5 days	10	2 months	40

Tertiary care is provided through five genetic departments and units in 4 different universities and a genetic department in the national research center. The health insurance organization provides financial coverage for investigations

and treatment as well as rehabilitation services including physiotherapy, speech therapy, early intervention therapy and behavioral modification. The private sector also offers clinical genetic services.

There are 5 renowned genetic departments in university hospitals (Ain-Shams University, Cairo University, Alexandria University, El-Mansoura University and the high institute of research, Alexandria University) in Cairo, Alexandria, Dakahlia governorates. They offer clinical genetic services and they have fully equipped laboratories including molecular lab.

The national research center (NRC):

The department within the Division of Human Genetics and Genome Research at the National Research Center, provides the following functions:

*Assessing pediatric congenital anomalies, including inborn errors of body chemistry and hearing disorders.

*Identifying pediatric screening and genetic resources.

*Informing the public about pediatric screening and genetic services.

*Assisting policy makers in developing pediatric screening and genetic services-related policies.

*Supporting private and public health-care providers.

*Developing programs and information systems.

*Providing clinical consultation and educational activities, especially related to pediatric screening and genetic services.

*Developing and distributing guidelines and educational materials on pediatric screening and genetics services.

*Participates on a statewide, interagency system of prevention/early intervention services to assure that families can meet the needs of their children who are at risk for disabilities from birth through age two.

Support groups/NGOs/Patient organizations:

The Genetic Support groups aim to ensure that people with genetic disorders have appropriate and accurate information and support to enable them to manage the challenges to their health and wellbeing. They facilitate an exchange of information, resources and assistance in order to support a number of existing genetic disorders and assist in the development of new groups.
In Egypt there are approximately 10 NGO's providing services for those who have congenital or genetic disorders and 120 organizations working in the

field of maternal and child health care and the care and prevention of disabilities.

Genetic diagnostic laboratory services:

Laboratory services include a range of genetic techniques to diagnose and assist in managing particular genetic conditions.

Current genetic diagnostic laboratory services in Egypt include:

- Newborn screening - all newborn babies are screened for congenital hypothyroidism.
- DNA diagnostic tests - DNA from individuals is examined to look for mutations in genes that may indicate a specific disorder.
- Metabolic diagnostic tests – pilot testing for screening inborn errors of metabolism in several governorates.
- Cryptogenic diagnostic tests - these are used to identify changes in the number or structure of chromosomes and may help to diagnose a genetic disorder.

In 2003 the MOH&P has established 3 genetic laboratories in the central health laboratory a cytogenetic lab including FISH technique, a biochemical lab and a molecular lab.

There are about 10 private genetic labs. 4 of them offer full genetic lab services.

Relevant community based genetic research:

- **Collaboration project between the children with special needs department/ Ministry of Health and Population and the Clinical Genetics Department of the National Research Center:**

A joint project was conducted between the Ministry of Health and Population and the Clinical Genetics Department of the National Research Center for the detection and estimation of the prevalence of various genetic disorders among Egyptians during the period from 2004 till 2006. the project was conduted to:

1. Evaluate the possible causes of birth defects.
2. Improve the diagnosis and treatment of birth defects.
3. Establish a mechanism for informing the parents of children identified as having birth defects and their physicians about the health resources available to aid such children

The main goal of the project was:

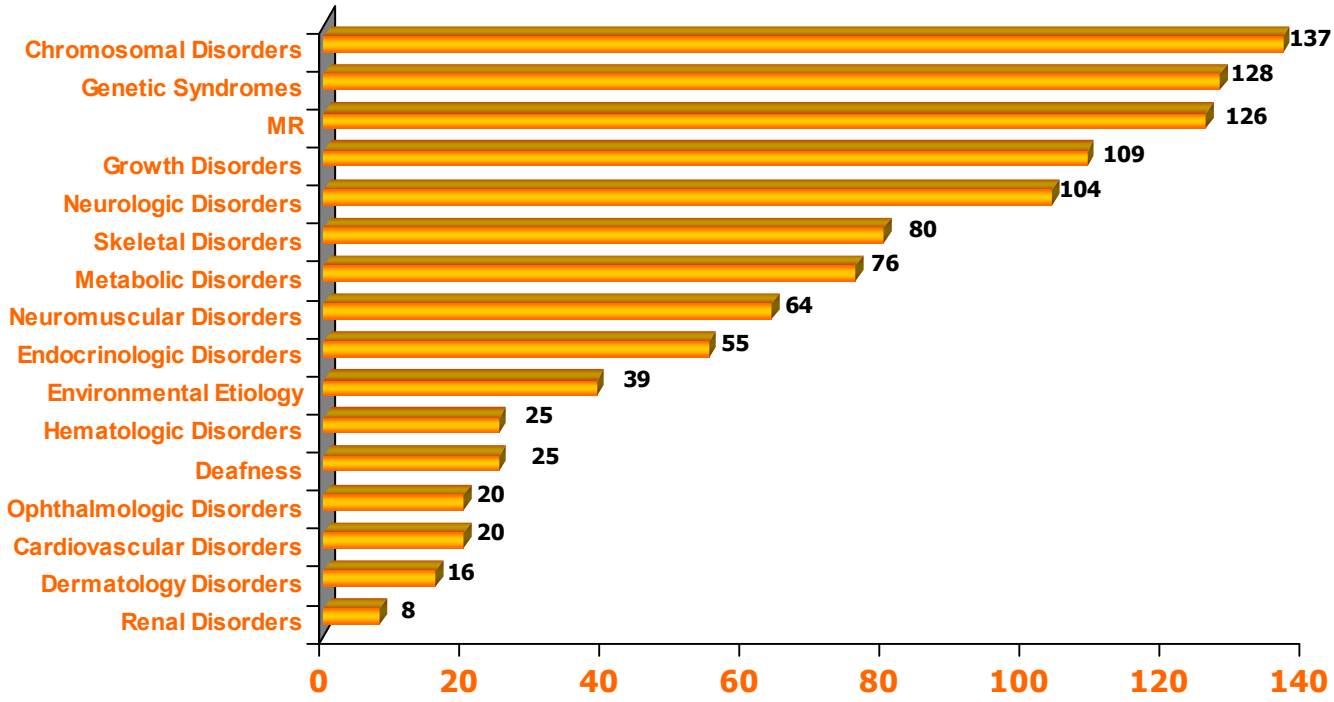
to provide a brief outline of the epidemiology and magnitude of genetic disorders and congenital abnormalities in Egypt and examines feasible preventive strategies and intervention approaches that are appropriate to the country.

Specific objectives:

1. To provide care for patients with a variety of rare genetic disorders.

2. To offer an additional opportunity for training in clinical genetics, dysmorphology and metabolic genetics at the Clinical Genetics Department of the NRC.
3. To discover new research opportunities in the aspects of diagnosis and care for specific genetic diseases.

The project included 1700 cases referred from the outpatient clinics and the neonatal care units of two governmental hospitals (Boulak El-Dakrour & Imbaba hospitals,Omm el atebba hospital) and primary health care centers (El-Giza & El-Talbia PHCCs).



Pattern of Genetic and Congenital Disorders (n=1700) in a rural and a Slum Area in Giza Governorate (Joint Project: MOH&P + NRC, 2004-2006)

Clinical evaluation of patients was performed. In addition, genetic counseling services were provided to patients and their families to assess risk, and give information on preventive measures, and testing options. Disorders that were studied included chromosomal and Mendelian disorders of childhood and/or adult onset, congenital anomalies and/or birth defects, dysmorphic syndromes, multifactorial disorders, and metabolic abnormalities. Standard, medically indicated laboratory or radiological studies were performed to confirm a diagnosis or to help in the management of the patients. In some cases, patients received medical or surgical treatment for their disorder, according to current clinical practice. Some patients and/or family members with genetic disorders have consented on DNA testing.

Each patient was subjected to meticulous clinical examination, pedigree construction, anthropometric measurements, and differential diagnosis. Some patients were referred to other departments and laboratories to perform the

required investigations needed to confirm the final diagnosis, and then they were given the proper genetic counseling.

To reach the proper final diagnosis the Clinical Genetics Department – when needed - referred the patients to other departments and laboratories to do the required specific investigations to confirm the provisional diagnosis.

Patients were referred for:

Cytogenetic analysis using G-banding technique, high resolution banding, induction of chromosomal breakage using DEB, or fluorescence in situ hybridization (FISH) technique .

Biochemical analysis including metabolic screening of plasma & urine for amino acids, estimation of specific enzyme activities, detection of glucosaminoglycans and oligosaccharides in urine

Molecular analysis for detection of different mutations to confirm the clinical diagnosis for some patients with neuromuscular disorders, PKU, ambiguous genitalia and congenital heart defects.

Orodental examination to detect changes that may be a part of the genetic disorder.

Prenatal diagnosis if the female was pregnant and suspected to be carrying a fetus with a genetic disorder

Other investigations according to specific cases.

Through the project the MOH&P and the NRC were able to fulfill the following:

1) *Establish an integrated classification* for the genetic disorders

2) *Offer proper diagnosis and genetic counseling* of the patient and his/her family.

3) Highlight the genetic disorders' burden through *increasing the physicians awareness* of the problem and increase their skills in detecting and diagnosing genetic disorders (e.g. training courses both theoretical and practical, etc).

4) *Attempt to improve the quality of life of patients* with genetic disorders, through regular follow-up of cases and applying the new approaches in management of patients (e.g. dietary regime for patients with metabolic disorders, etc….)

5) *Early detection and treatment of diseases* to reduce mortality and minimize morbidity. This is the basis of successful neonatal screening for inborn errors of metabolism where treatment or modification of lifestyle can modulate disease.

6) *Detect consanguineous relationships* and significant genetic morbidity through screening pedigrees administered by a trained link worker.

- **Screening for birth defects in east Alexandria:**

  Research title: Pattern of birth defects among newborns at east medical locality of Alexandria.

  The objectives of the study were:

  1. To determine the prevalence and pattern of different types of birth defects among newborns.
  2. To highlight the importance of early detection of birth defects.
  3. To obtain reliable information suitable for evaluating the need for a national program for early detection and intervention of birth defects.

Twenty thousands newborns were screened within one year from September 2003 till the end of August 2004. They were consecutively recruited from PHC centers and units during screening and vaccination sessions. The early detection of any congenital or genetic disorders was performed by well-trained PHC nurses. Cases were then referred to one of the genetic counseling clinicsin Alexandria where they were examined by physicians working in those clinics. The newborns were diagnosed, offered suitable intervention therapy and followed by genetic specialized physicians and consultants.

Limitations to the study included:

1. The cost of intervention was considered high.
2. Inability to identify newborn deaths caused by birth defects due to lack of reliable deaths registry.
3. The PHC nurses lacked the required skills for the detection of internal malformations like congenital heart diseases.

The prevalence of birth defects in newborns is 3%.

The pattern of birth defects indicates that congenital malformations of genital organs are the most common defects (164/10.000 live births), followed by malformation and deformation of the musculoskeletal system (79/10,000 live births).

The following table shows the classification of cases detected and diagnosed during the study according to the WHO classification (ICD-10).

Pattern of Birth Defects According to WHO "ICD-10" Classification

ICD-10	Main Group	No.	Prevalence /10,000
Q00-Q07	Congenital Malformations of the Nervous System	15	7.7
Q10-Q18	Congenital Malformations of Eye, Ear, face and Neck	17	8.7
Q20-Q28	Congenital malformations of the circulatory system	6	3.6
Q30-Q34	Congenital malformations of the respiratory system	-	0.0
Q35-37	Cleft lip and cleft palate	9	4.6
Q38-Q45	Other congenital malformations of the digestive system	43	22
Q50-Q56	Congenital malformations of genital organs	321	164.6
Q60-Q64	Congenital malformations of the urinary system	1	0.5
Q65-Q79	Congenital malformations and deformations of the musculoskeletal system	155	79.5
Q80-Q89	Other congenital malformations	80	41.0
Q90-Q99	Chromosomal abnormalities, not elsewhere classified	14	7
Total	Birth defects	661	338

- **Screening for genital anomalies among newborns in Great Cairo (2006):**

This is a collaborative research between the children with special needs department and the national research center.

Screening for genital anomalies, abnormal sexual differentiation and congenital adrenal hyperplasia was performed on 20'000 newborns from PHC units and centers in Cairo, Giza and Qualiobia governorates. the results revealed some important data concerning the incidence of CAH and ambiguous genitalia (not published yet).

- **Pilot newborn screening for metabolic disorders in Cairo, Giza, Qualiobia and Assiut governorates (2007-2009):**

A collaborative research between the children with special needs department and Kasr-El-Einy friends association funded by the EU commission

Ten metabolic disorders are screened using Tandem mass (MS/MS technique) for screening and diagnosis. Filter paper from 25000 newborns utilized in screening for congenital hypothyroidism is used in this study.

- **Pilot newborn screening for metabolic disorders in Alexandria governorate (2008-2009)**:

A collaborative research between the children with special needs department and Alexandria child neurology organization funded by the EU commission. Screening for PKU and galactosemia is performed on 60.000 filter paper used in neonatal screening for congenital hypothyroidism in three main zones in Alexandria.

- **Pilot testing on screening newborns for congenital adrenal hyperplasia in Cairo (2008-2010):**

A collaborative study between the department of pediatric endocrinology, the department of clinical pathology in Abo-El-Reech pediatric hospital/Cairo University and the children with special needs department/MOH&P. The study will be performed on 50.000 newborns in Cairo governorate.

- **Identification of ß-Thalassemia carriers among secondary school students in Great Cairo (2004):**

A feasibility study was performed by the children with special needs department in collaboration with the health insurance organization and the central health laboratories, mainly to assess the possibility of implementing a national program for Thalassemia carrier detection among the Egyptian population and to offer genetic counseling for positive carriers. It was conducted on 5000 secondary school students from public and private schools in Great Cairo. The prevalence of ß-Thalassemia carriers was 1.47%.

- **A national study on the early detection of birth defects and all types of impairments that may lead to disabilities in infants and children below the age of six years (2004/2005):**
This national study was performed to assess the feasibility and validity of a suggested Egyptian tool for early identification of disabilities.

Prevalence rates of disabilities among infants and children below the age of six

Age category	Total examined	No. of disabled	Prevalence rate (%)
1st month	1678	26	1.55
8-12 weeks	1322	39	2.95
9-12 months	1465	57	3.89
18-24 months	1541	161	10.45
2.5-3 years	1065	128	12.02
5-6 years	976	106	10.86
Total	8047	517	6.42

- **Research on neonatal hearing screening in Cairo governorate (2005/2006):**

  This study was conducted to build-up a proper image to the problem of hearing loss in Egypt in an attempt to promote national neonatal hearing screening. Fifteen hundreds and two neonates were recruited and randomly selected from four primary health care (PHC) centers distributed in East Cairo. Primary screening by Echo screen Evoked Otoacoustic Emission was conducted by trained primary heath care physicians and nurses. Complete audiological evaluation to detect the type, degree of hearing loss and risk factors was conducted at audiology clinic Ain Shams University to all neonates who failed secondary screen. Sensorineural hearing loss has been detected in 0.8% of the examined neonates; hence representing a prevalence higher than other international rates for neonatal hearing loss.

- **Screening for autistic disorders among children aged 18 to 24 month in Cairo and Alexandria governorates (2006):**

  The study was conducted by the children with special needs department/MOH&P on 10.500 children from PHC centers and units in Cairo and Alexandria governorates.

Needs assessment: Assessment framework, Gap analysis, goals and strategic planning

The following tables illustrate the current situation, gap and needs in health facilities and health providers involved in genetic services provision and activities of early detection of birth defects and disabilities:

- ❖ At the PHC level the total number of rural and urban facilities (centers and units) are 3512 and 452 respectively, distributed throughout the entire country. Those facilities serve mainly women and children. Only 792 rural and 116 urban facilities have been involved in the early detection of genetic and congenital disorders. Physicians working in those facilities have been well trained on the early detection of birth defects and referral of cases to the genetic counseling clinics in the catchments' areas of those health facilities. So they serve only the areas around the genetic counseling clinics in only 7 governorates of the 29 governorates in the country. What is needed is to cover all the 3512 facilities with services of early detection of genetic and congenital disorders and referral through a well established system of referral to the second level of service at the nearest genetic counseling clinic. A better planning for availability of service in underserved areas is required.

- ❖ Concerning the community genetic clinics, having already 11 clinics in 7 governorates, we need to extend the service to other governorates. A plan should be implemented to increase the number of clinics to 23 to cover heavily populated governorates in Egypt. This is a long term

plan expected to be implemented within 6 years. It is also planned that at least six of the clinics will be located in PHC centers or hospitals in rural areas.

- Available laboratories either molecular, cytogenetic or biochemical laboratories include central labs that belong to the MOH&P, university hospital labs, national research genetic labs, Vaccera labs. and private labs. Most of the labs are mainly present in the big cities like Cairo, Giza, Alexandria and El-Mansoura. There is a lack in genetic labs in the southern part of upper Egypt especially Sohag, Quena, Luxor and Aswan governorates. What is needed is to extend the genetic lab. services to cover those areas as a first step, then focus on the other underserved areas like the Suez Canal governorates, North and South Sinai.

- There are 5 genetic specialized centers and units all belonging to the university hospitals and the national research center. The plan is to establish 4 regional specialized genetic centers that would be a tertiary care level for referral and will provide all genetic health care services. Those centers will belong to the MOH&P but genetic services will be provided by genetic specialized physicians, consultants and genetic university staff.

- To be able to provide the services of early detection of congenital and genetic disorders in all the PHC units and centers. At least one trained physician is required in each facility. The number of PHC physicians who are trained over the last 5 years is 1371. A number of 2593 should be trained over the coming 5 years, one per each PHC facility to be able to detect and refer genetic cases to the secondary care level.

- PHC nurses involved in neonatal screening activities will be trained on early detection of birth defects. A long term implementation plan to train 3134 nurses in five years is required.

- The majority of genetic lab specialists are working in the central labs in Cairo that belongs to the MOH&P, the university hospital labs. the labs. in the NRC, and the lab. in Vaccera. it is mandatory to train other physicians, scientists and pharmacists in each genetic lab specialty. Some of them will be responsible for providing services in the regional genetic labs in the upcoming plan (3 from each lab specialty/regional lab). A short term implementation plan of 3 years is required.

- We still have shortage in the number of professional medical geneticists and genetic counselors. We have only 40 secondary care physicians trained to provide clinical genetic services including counseling to the community. They are working in the community genetic clinics, serving a population of at least 10 million (1:250000 individuals). There are also 110 genetic specialized physicians working in the genetic departments in the university, the NRC, Vaccera and the private sector. At least twice the number of medical geneticists and

counselors is needed in the meantime, but to be able to achieve this goal a minimum period of five years is needed to educate and train them. It is also mandatory to start involving certified nurses in counseling.

- Genetic professors (holding PhD or Doctoral degrees) are working in 5 departments in only 4 universities and in the NRC; their total number is approximately 62. They are mainly serving populations living in big cities including Cairo, Alexandria, Giza, and El-Mansoura. According to the number of individuals seeking genetic counseling at the genetic centers in the university hospitals each counselor serves 30 to 40 individuals per month.
- The number of medical genetic University staffs is considered small in relation to the large population they are serving. There are only 5 fully staffed departments of genetics in 4 universities and a comprehensive devision with 8 specialized genetic units in the national research center. There are only 2 departments in 2 universities (Ain-Shams and Alexandria universities) that offer postgraduate medical genetic certification including a master and a doctoral degree in medical genetics.
- At least six main universities in Egypt (Al Azhar, Suez-canal, Menia, Assiut, Menofia, and Zagazig universities) are required to establish genetic departments with certified staff to be able to serve such a large population. It is estimated that each university will need at least 5 genetic staffs as a start for the early establishment of genetic departments, so the total additional number of university staff needed is 30.

health facilities: Gap analysis and implementation time frame

Health facilities	Current situation (number of facilities)		Gap (number of facilities)		Needs (number of facilities)		Implementation time frame (years)	
	Rural	Urban	Rural	Urban	Rural	Urban	Short term 1-3 y.	Long term 4-7 y.
1. PHC units & Centers	792	116	2720	336	3512	452		✓
2. Community genetic clinics	1	10	6	6	7	16		✓
3. Laboratories								
• Molecular Lab	-	12	-	4	-	16	✓	
• Cytogenetic lab	-	20	-	4	-	24	✓	
• biochemical lab	-	10	-	8	-	18	✓	
4. Genetic Specialized Centers	-	6	-	4	-	9		✓
5. Genetic depts. in universities & NRC	-	5	-	6	-	11		✓

Health providers: Gap analysis & implementation time frame

Health providers	Current situation (number of health providers)	Gap (number of health providers)	Needs (Number of health providers)	Implementation time frame	
				Short term 1-3 years	Long term 4-7 years
1. PHC physicians	1371	2593	3964		✓
2. PHC Nurses	116	3134	3250		✓
3. Lab Specialists (physicians):					
• Biochemical genetics	30	20	50	✓	
• Cytogenetics	68	20	88	✓	
• Molecular genetics	30	12	42	✓	
4. Clinical geneticists physicians*	40 + 110	40 + 110	300		✓
5. Genetic university Professors and consultants	62	30	92		✓

* Clinical geneticists are physicians working in the genetic counseling clinics + physicians working in genetic departments in universities/NRC (graduate students for master or doctoral degrees in genetics and those holding a master degree and doctoral degrees in genetics but not including professors).

Barriers to genetic health services provision

The following barriers were identified through genetic needs assessment process:

- ❖ Difficulties in the integration of a new service along with so many vertical programs that are established within the public health system.
- ❖ Lack of knowledge about how and where to access such services (among both health care providers and the general public)
- ❖ Long distance from residence to the genetic clinics.
- ❖ Unavailability of genetic services in many governorates.
- ❖ Cost of services and lack of insurance coverage or adequate reimbursement
- ❖ Cultural and religious beliefs.
- ❖ Concerns about privacy and confidentiality.

- ❖ Social stigma of using genetic services or having a genetic condition.

Key priority goals identified through needs assessment process:

Goal 1: Promote comprehensive and accessible genetic services for the Egyptian population

Genetic services are available to help individuals and families understand their specific genetic condition, or risk of genetic condition, and ultimately improve health by reducing morbidity and mortality associated with these conditions. Due to advances in genetic technology and increasing clinical applications for more common conditions, the need for genetic services is growing. Comprehensive genetic services include evaluations, testing, counseling, treatment, management, educational activities, family support and follow-up for all members of the population. Assuring quality and accessible genetic services to the public is a fundamental component of the state genetic services plan.

Objective 1

Evaluate and improve the availability and accessibility of genetic services in Egypt

Strategies:

1. Evaluate genetic services delivery in Egypt and identify gaps.
2. Monitor the national Newborn Screening Program in order to identify, evaluate and incorporate, when appropriate, new technologies to facilitate screening and follow-up.
3. Expand the screening laboratories and follow-up clinics capacity to support comprehensive and emerging testing, tracking, and treatment options for genetic conditions.
4. Add other disorders that are prevalent in the country like PKU, galactosemia and congenital adrenal hyperplasia to the screening program.

Objective 2

Support and sustain the national committee for community genetic services to share strategies for providing quality, accessible and comprehensive services to all residents.

Strategies

1. Recruit committee members for regular meetings to Develop mission, goals, structure, desired outcomes and administration of the national community genetic counseling program.
2. Identify existing protocols or develop guidelines describing appropriate genetic services provision on a public and private level.
3. Develop strategies for providing accessible and comprehensive services.
4. Adapt or develop quality indicators for genetic services in Egypt.

Goal 2: Reduce barriers to access to genetic health care services

Advances in genetic technology and genomics have led to the development of primary, secondary and tertiary prevention strategies. However, access to genetic services is not equally available and utilized by all segments of the population, particularly in underserved rural and low-income communities. Understanding and reducing potential barriers to access to genetic services is key in providing equal opportunities for disease prevention and health promotion.

Objective 1

Improve access for low income individuals by addressing economic barriers.

Strategies

1. Raise public and professional awareness of available at no cost genetic services at the community genetic clinics.
2. Link clients to available NGO's and support groups.

Objective 2

Improve access for people living in rural and remote areas by addressing geographic barriers.

Strategies

1. Evaluate genetic services delivery and identify gaps.
2. Examine the need for establishing additional genetics outreach services and mobile teams in underserved areas.
3. Identify and increase funding opportunities for providing genetic services in underserved areas.
4. Promote referral of cases from PHC level in rural and remote areas to the nearest genetic counseling clinic.
5. Develop a strategy to extend genetic clinical services to areas currently without such access.

Objective 3

Improve access for rural community and close communities (Bedouins and Nubians) by addressing cultural barriers.

Strategies

1. Assess cultural barriers to receiving genetic services.
2. Determine methods of providing information to those in need, including raising awareness of existing services and costs of those services.
3. Evaluate current genetics education literature for cultural competence and develop resources where there is a gap in existing materials.
4. Determine methods for addressing fixed cultural beliefs, customs and religions beliefs.

Objective 4

Assess and assure adequate access to genetic health care providers.

Strategies

1. Develop a hot telephone line and online genetic information and consultation services.

2. Promote Partnership with genetic professionals and consultants to investigate opportunities for providing genetic consultation and counseling through the national genetic counseling program referral systems.
3. Assess referral patterns of primary care providers to genetic service providers, including the appropriateness of referrals.
4. Develop and support undergraduate programs and postgraduate medical education programs that will lead to an adequate and diverse genetic provider workforce.
4. Conduct ongoing surveys, or obtain existing data from the country situation survey report, to determine supply and distribution of qualified genetic providers.

Goal 3: Increase genetic awareness

Objective 1:

Increase genetic awareness and knowledge in the general population.

Strategies

1. Identify community leaders in rural and minority communities for the purpose of building relationships and disseminating information about genetics and public health.
2. Identify and develop appropriate health education materials e.g. brochures, posters and leaflets.
3. Develop and coordinate seminars and workshops for community genetic education
4. Identify existing genetic and family history awareness programs/materials and replicate successful models and projects.
5. Identify religious professional leaders for the purpose of building relationships and disseminating information about genetics and public health.
6. Identify “mass media” mechanism for promoting family history.
7. Develop relationships with media contacts.

Objective 2

Increase genetic awareness and knowledge among policymakers, including legislators, local peoples' assembly and other public health leadership.

Strategies

1. Form a coalition of partner organizations.
2. Develop a strategy for approaching health policymakers.
3. Educate and raise awareness of public health policymakers and agency leaders about the importance of integration of genetics into public health services.

Goal 4: Promote integration of genetics into health care delivery systems through education of health professionals and health care providers

It is now becoming increasingly important to provide education and training to health care professionals and the health providers to enhance awareness, build competencies and ensure integration of genetic services in the health care delivery system. Health care providers’ current level of awareness and understanding of genetic conditions and genomics has not kept pace with the

current increase in genetic knowledge. Therefore, by increasing provider awareness and knowledge of genetics, health care professionals may make more informed decisions regarding delivery of genetic services.

Objective 1

Identify the educational needs and assess the current state of genetics knowledge of the public health and health care providers.

Strategies

1. Identify existing genetic competencies for health professionals.
2. Conduct a needs assessment study to assess genetic knowledge and integration of genetics into medical practice among health professionals and the health care providers.

Objective 2

Increase health professionals' genetic knowledge through identification, development and dissemination of educational resources.

Strategies

1. Identify existing genetic educational resources for health care providers.
2. Assess and evaluate the quality of available genetic resources for health care providers and identify gaps in educational materials.
3. Develop regular and ongoing genetic education and updates.
4. Develop genetics curricular materials where there is a gap in available educational resources.

Goal 5: Develop a data base for genetic services, facilities and information

Objective 1

Improve the accuracy and availability of data regarding genetic conditions and services.

Strategies

1. Improve the accuracy and completeness of the data currently collected by the physicians and consultants working in the field of genetic.
2. Develop a network for data collection linking the entire genetic counseling clinics and insure availability of data at the central administrative level.
3. Increase awareness of public health program staff and partners regarding the types and quality of data currently collected by and available through the network.

Needs Assessment Framework

Strategic objectives	What we have accomplished	What we need to establish
1. Evaluate effectiveness, accessibility, and quality of personal and population-based genetic services.	▪ Supervision system for monitoring the quality of genetic services and providing continuous on job training for physicians working in the genetic counseling clinics. ▪ Genetic services are accessible to the population through a referral system from the PHC facilities to the genetic counseling clinics in the catchment areas of those clinics.	▪ Insure sustainability of the supervision system and to expand this system to cover all genetic health facilities. ▪ Put quality standards for genetic services provision and do periodic quality check. ▪ Establish other community genetic clinics to reach the inaccessible communities in rural, remote and slum areas.
2. Diagnose and investigate genetic health problems and health hazards affecting children and families.	▪ Eleven genetic counseling clinics are established in several governorates. Affected children and families have an access to tertiary care services through a well established system of referral from the clinics to the genetic specialized centers and labs. ▪ Three genetic labs are established in the central health laboratories that belongs to the MOH&P.	▪ Establish more genetic counseling clinics to cover the rest of the governorates. ▪ Establish regional tertiary care centers to cover the 4 regions in Egypt. ▪ Establish 4 regional genetic labs. ▪ Provide antenatal genetic diagnostic services.

Strategic objectives	What we have accomplished	What we need to establish
3. Provide information and education to the public and families about genetic health issues.	▪ There are no activities for promoting community awareness and public education about genetic health.	▪ Develop appropriate genetic health education messages for the community ▪ Develop and coordinate seminars and workshops for community genetic education ▪ - Identify existing genetic awareness programs and materials and replicate successful models and projects.
4. Mobilize community partnerships between policymakers, health care providers, and the general public to identify and solve genetic problems.	▪ The Egyptian national childhood and motherhood counseling have established a committee for children with special needs including those who have genetic disorders and their families. The committee consists of policy makers (29 governors, representatives of executive authorities in health, education and social solidarity personnel, NGO's, and community leaders). They make policies, strategies and action plans for all relevant health issues.	▪ Provide support and put in action all the decisions made by this committee. ▪ Health authorities should adopt and enforce strategies and action plans decided by the committee and should provide the necessary resources for implementation.
5. Promote and enforce legal requirements pertaining to genetics that protect and promote child and family health.	▪ Constitution for provision of free of charge PHC services for all mothers and children. ▪ The law # 12 that passed in 1996 for children rights in health, education and social issues. ▪ Recently an amendment occurred to that law in June	▪ Law mandating insurance reimbursement or coverage for genetic services such as genetic counseling, premarital genetic testing, prenatal genetic testing, etc. ▪ Enforcement and empowerment of the health reform policy.

Strategic objectives	What we have accomplished	What we need to establish
	2008 to promote and enhance the rights of children and highlighting the rights of disabled children in all services. ▪ Recently an amendment for the punishment law # 58 was done in June 2008 for enforcement of punishment for child neglect and abuse. ▪ Amendment of the Civil law # 143 in 2008 for age specification of children to be up to 18 years forbidding marriages below this age. ▪ The law mandated premarital examination and testing before any marriage. ▪ A modification in Feb. 1993 of the law for the social health insurance services delivery to ensure expansion of health services to cover all school students. ▪ A ministerial decree in 1997 for providing health insurance services for neonates and children below the age of 6 years. ▪ A ministerial decree in 2000 for providing at no cost neonatal screening for congenital hypothyroidism for all neonates.	▪ Getting an agreement from the peoples' assembly on the new health insurance law that will provide coverage for all Egyptians (including the disabled), for all age groups and occupations.

Strategic objectives	What we have accomplished	What we need to establish
6. Assess and monitor health status of children and families affected by genetic conditions and other special health needs, and respond to identified problems.	▪ Some limited existing genetics health data from the genetic counseling clinics are collected on a yearly basis. ▪ Data for the national neonatal screening program are regularly collected. ▪ Data from genetic related research and projects are collected.	▪ Establish a data base system for collection and analysis of data. ▪ Establish a birth defect registry. ▪ Establish a network for collection of information concerning genetic and congenital conditions, available services and facilities at all levels of referral in the genetic counseling program.
7. Assess and assure genetic knowledge and practice of genetic health care providers	▪ Continuous ongoing clinical genetic training to the physicians working in the genetic counseling clinics is provided by genetic professors and consultants during supervision visits. ▪ Training courses for PHC physicians on early detection of genetic and congenital disorders. ▪ Training courses for PHC nurses on early detection of genetic and congenital disorders.	▪ Develop a training curriculum in genetics for PHC physicians and nurses and secondary care physicians. ▪ Conduct regular orientation and training courses for the PHC physicians and nurses and secondary care physicians
8. Support research and projects to gain new insights and innovative solutions to genetic health problems	▪ Many national community-based research and projects in the field of genetics were implemented (see above).	▪ National community based studies to determine the prevalence of various genetic conditions among the Egyptian population. ▪ National studies for carrier detection of certain prevalent genetic recessive disorders like Thalassemia, sickle cell anemia, G6PD… ▪ Implement feasibility studies for genetic services provision especially services like antenatal screening and testing for genetic conditions.

❖ Future Scenarios (Egypt in 10 years)

1. Demography:

- Population number will increase to 91 millions
- No significant change in population distribution
- Millennium goals → success

2. Description of health care system

- Complete coverage with health reform system
- Health insurance coverage for the majority of the population including the disabled
- Health insurance coverage for all genetic health care services
- Expansion of community genetic clinics to all governorates
- Expansion and improvement of the National newborn screening program
- Increase genetic services utilization due to improved awareness

3. National Policy:

- Health policy makers will consider genetic disorders as public health priority.

References:

1. **Abd-Al-Salam E. (1994):** Hereditary disorders. A report presented to the Regional Consultation on Community Genetic Services, World Health Organization Regional Office for the Eastern Mediterranean, Alexandria, 17-21 April, (available on request from the Non-Communicable Diseases Unit, World Health Organization Regional Office for the Eastern Mediterranean, Alexandria, Egypt).

2. **Abel E, Horner SD, Tyler D, Innerarity SA (2005):** The impact of genetic information on policy and clinical practice. Policy Polit Nurs Pract 6: 5-14.

3. **Alex Faulkner, Nicola Mills, David Bainton, Kate Baxter, Paul Kinnersley, Tim J Peters, and Deborah Sharp (2003):** A systematic review of the effect of primary care-based service innovations on quality and patterns of referral to specialist secondary care. Br J Gen Pract., 53(496): 878–884.

4. **Bittles A. Consanguinity in the Eastern Mediterranean. (1994):** A paper submitted to the World Health Organization Regional Consultation on Community Genetic Services, (available on request from the Non-Communicable Diseases Unit, World Health Organization Regional Office for the Eastern Mediterranean, Alexandria, Egypt).

5. **Christianson A. and Modell B.(2004):** Medical genetics in developing countries. Annu. Rev. Genomics Hum. Genet. 2004. 5:219–65

6. **Czeizel AE. (1993):** Prevention of congenital abnormalities by periconceptional multivitamin supplementation. British medical journal, 306:1645-8.

7. **El-Zanaty F and Way A (2006):** Egyptian demographic health statistics. MOH&P, Egyptian national population council; Cairo, Egypt.

8. **Glyn Elwyn, Adrian Edwards, Rachel Iredale, Peter Davies, and Jonathon Gray (2005):** Identifying future models for delivering genetic services: a nominal group study in primary care. BMC Fam Pract.; 6: 14.

9. **Hafez M et al. (1985):** Autosomal recessive genes causing mental deficiency in Egyptian children. Egyptian journal of genetics and cytology, 14:259-64.

10. **ICD-10-CM (2003):** International classification of diseases, 10th version, clinical modification . World Web site: http:// www. cdc.gov/nchs/icd9.htm.

11. **JA Knottnerus (2003):** Community genetics and community medicine. Family Practice Vol. 20, No. 5, 601-606.

12. **Jon Emery, Eila Watsona, Peter Rose and Anne Andermanna (1999):** A systematic review of the literature exploring the role of primary care in genetic services. Family Practice Vol. 16, No. 4, 426-445.

13. **Meyer BF (2005):** Strategies for the prevention of hereditary diseases in a highly consanguineous population. Ann Hum Biol 32: 174-179.

14. **Ozand PT, Odaib AA, Sakati N, Al-Hellani AM (2005):** Recently available techniques applicable to genetic problems in the Middle East. Community Genet 8 : 44-47.

15. **Suther S, Goodson P.(2003):** Barriers to the provision of genetic services by primary care physicians: a systematic review of the literature. Genet Med.;5(2):70-6.

16. **Teebi A, Farag T. (1996):** Genetic disorders among Arab populations. Oxford monographs on medical genetics No.30. Oxford, Oxford University Press.

17. **Teebi AS, Teebi SA (2005):** Genetic diversity among the Arabs. Community Genet 8 : 21-26.

18. **Temtamy SA, Meguid N, Mazen I, Ismail SR, Kassem NS, Bassouni R (1998):** A genetic / epidemiological study of malformations at birth in Egypt. Eastern Medit Health J. 4: 252-25.