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*Research Article*

# Community Genetic Services in Iran

**Shirin Atri Barzanjeh,[1] Mozhgan Behshid,[1,2] Mohammad Bagher Hosseini,[3] Maryam Ezari,[1] Mahdieh Taghizadeh,[4] and Saeed Dastgiri[4]**

[1] *School of Nursing and Midwifery, Tabriz University of Medical Sciences, Tabriz 5166615739, Iran*
[2] *Health System Research Committee, Tabriz Health Services Management Research Centre, Tabriz University of Medical Sciences, Tabriz 5166615739, Iran*
[3] *Pediatric Health Research Centre, Tabriz University of Medical Sciences, Tabriz 5166615739, Iran*
[4] *Tabriz Health Services Management Research Centre, School of Medicine, Tabriz University of Medical Sciences, Tabriz 5166615739, Iran*

Correspondence should be addressed to Mozhgan Behshid, mozhganbehshid@hotmail.com

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The aim of the study was to report a description of the primary, secondary, and tertiary level services available for genetic disorders in Iran. For the purpose of this study, essential data were collected from every facility providing community genetic services in Tabriz city of Iran using a prestructured checklist. Technical information was filled in the predesigned forms using diagnostic records of each client/patient. Information was also gathered from community genetic services clients through a face-to-face interview at these facilities to assess the quality of services provided. Primary prevention measures were available in 80 percent of centres in the study population. Diagnostic techniques were fully available in the study area both in public and private sectors. Screening of congenital hypothyroidism and thalassemia has been successfully performed across the country by the Ministry of Health. Other screening programs have also been initiated by the country health authorities for neural tube defects, Down syndrome, and phenylketonuria. The high cost of genetic services at secondary and tertiary levels does not allow many people to get access to these services despite their needs. Governments will therefore need to allocate necessary resources to make the essential genetic services available for everyone needing these in the community.

## 1. Introduction

Prenatal genetic service has a key role in detecting the genetic risks and carrier screening for thehereditary conditions.

Prenatal genetic service has a key role in detecting the genetic risks and carrier screening for certain hereditary conditions such as sickle cell disease, cystic fibrosis, Tay Sachs disease, and alpha/beta thalassemia. According to the previous studies in the region, the genetic disorders and congenital abnormalities can affect individuals in different ages ranging from 2 percent at birth to 65 percent of individuals in their lifetime period. They account for approximately 30 percent of pediatric hospital admissions ranging from mild to very severe conditions [1, 2].

The essential goals of Community Genetic Services (CGS) are to manage the prevention, clinical, and psychosocial demands of individuals suffering from/at risk of hereditary disorders. This can be achieved through timely and precise clinical and laboratory diagnosis, genetic consultation, and primary health care services [3].

As for any disease, prevention methods in genetic disorders are performed at three levels including primary, secondary, and tertiary. Primary prevention is to avoid the occurrence of a disease (i.e., genetic consultation). Secondary prevention methods are to find and treat patients as early as possible (i.e., drug treatments for some heart anomalies). Tertiary prevention includes every effort to reduce the negative effects of the disease through treatment or rehabilitation (i.e., rehabilitation for limb anomalies).

The aim of the study was to report a brief description of the primary, secondary, and tertiary level services available for genetic disorders in Tabriz city of Iran as a sample pattern of CGS for similar regions.

## 2. Methods

This study was carried out in Tabriz city of Iran. The city is one of the three major cities in the northwest of the country. Health houses, primary care centres, public/private clinics and hospitals, and Welfare and Rehabilitation Organization provide CGS in the area covering about 5 millions of population in the northwest of the Iran.

For the purpose of this study, essential data were collected from every facility providing CGS in the area using a pre-structured checklist. Technical information was filled on the predesigned forms using diagnostic records of each client/patient. Information was also gathered from CGS clients through a face-to-face interview at these facilities to assess the quality of services provided.

The study took place between March and June 2010. A total number of 19 public and private hospitals and clinics and 15 heath centres were checked for the purpose of this study. A total number of 140 clients/patients were interviewed to assess the quality of services, costs, and so forth.

Approval for this study was obtained from the Regional Committee of Medical Ethics of Tabriz University of Medical Sciences. Accordingly, an informed consent from the study participants was obtained and data were gathered anonymised at the source of data collection to keep personal information private.

## 3. Results

Some of the current services at primary, secondary, and tertiary levels available for genetic disorders in Iran are listed in Table 1.

Primary prevention measures were available in 80 percent of centres and facilities in the study population. These measures include preconceptional care and folic acid supplementation, pregnancy health care services, maternal and children health care services, vaccination (i.e., rubella, measles, etc.), and various educational programmes for the primary prevention of congenital anomalies and genetic disorders.

Diagnostic techniques including ultrasonography, DNA extract tests (including PCR), serum alpha fetoprotein, non-conjugated estradiol, amniocentesis, chromosomal analysis of amniotic fluid, maternal phenyl alanine test, urine and plasma amino acids analysis fetal echocardiography, MRI scan and CT scan, electroencephalography, chest radiography, electrocardiogram, different type of echocardiography, color ultrasonography, pulse and color Doppler, angiocardiography, pulmonary angiography, liver function tests, IV urography and urethrography, blood gas analysis, chest radiography, radiography by barium swallow, thoracentesis, bronchography, lung function tests, lung biopsy, lung aspiration, bronchoscopy, laryngoscopy, thoracoscopy, radionucleotide scan, fluoroscopy, arteriography, and aortography were all fully available in the study area both in public and private sectors.

Screening of congenital hypothyroidism and thalassemia has been successfully performed across the country by the Ministry of Health [4]. Other screening programs have also been planned/started by the country health authorities for neural tube defects, the Down syndrome, and phenylketonuria.

Selected types of congenital anomalies (i.e., heart defects) were routinely treated by various types of drugs as well as the neurosurgical technique for hydrocephaly and Spina Bifida, shunting for hydrocephaly with or without myelomeningocele, reconstructive surgery for cleft lip, and palate and for patent ductus arteriosus (PDA), ventricular septal defect (VSD), Atrioventricular (AV) canal, tetralogy of fallot, pulmonary valve stenosis and noncyanotic congenital heart diseases, urology corrective surgery for hypospadias, cryptorchidism, undiscerning testis, arterioplasty for coarctation aorta, valvuoplasty and valve replacement for aortic valve stenosis, craniosynostosis surgery, dialysis and kidney transplants for congenital renal diseases, and bone marrow transplantation for different type of bone marrow failure.

## 4. Discussion

In this study, we reported the current status of genetic services for the prevention, treatment, and rehabilitation of congenital anomalies in Iran provided by public and private sectors in the area.

This study showed that primary prevention of congenital anomalies and genetic disorders (i.e., folic acid supplementation before and during pregnancy, etc.) is now well established in the region. Despite some differences, similar findings have been reported in previous studies [5–7] indicating that the activities by the Ministry of Health and private sectors for the prevention of genetic disorders are well understood/accepted in the country. Research reports from the nine European countries, Georgia, Venezuela, and India showed that primary prevention measures have been implemented in these regions too [8–11]. However, the structure of the preventive strategies may vary by the region based on the population demands and financial sources.

Our study indicated that despite availability of diagnostic and treatment facilities for genetic disorders in the area, they are mostly too expensive to be fully accessible for many people in the country as they responded in the face-to-face interview. It is obvious that as more advances in genetic technology occurring, the demands for CGS are growing in the population. However, the high cost of genetic services does not allow many people to get access to these services despite their needs. Governments will therefore need to allocate necessary resources to make the essential genetic services available for everyone needing that in the community. More investigations are needed to assess the cost effectiveness and health economics aspects of this matter.

## Conflict of Interests

The authors declare that they have no competing interests.

Table 1: Community Genetic Services in Iran.

| Services | Congenital abnormalities |
|---|---|
| Alpha fetoprotein | Neural tube defects, nephrosis, chromosomal abnormalities, omphalocele |
| Amniocentesis | Neural tube defects, chromosomal abnormalities |
| Amniotic fluid chromosomal analysis | Trisomy 13, 18, 21 and chromosomal abnormalities |
| Aortography | Congenital heart abnormalities |
| Arteriography | Congenital heart abnormalities |
| Arterioplasty with balloon | Coarctation aorta |
| Blood gas analysis | Congenital respiratory diseases |
| Bone marrow transplantation | Hydroxyurea |
| Bronchography | Congenital respiratory diseases |
| Bronchoscopy | Congenital respiratory diseases |
| Chest radiography | Congenital respiratory diseases |
| Chest radiography | Congenital heart abnormalities |
| Chorionic gonadotropin level | Trisomy 13, 18, 21 |
| Color ultrasonography and ventriculography | Congenital heart abnormalities |
| Cordocentesis services | Hemoglobinopathies, fetal anemia, acid-base disorders, thalassemia |
| Craniosynostosis surgery | Nervous system anomalies |
| Dialysis and kidney transplants | Congenital renal diseases |
| DNA services | Thalassemia, muscular dystrophy, phenylketonuria (PKU), Turner syndrome, cystic fibrosis |
| Drug treatments | Congenital heart abnormalities |
| Echoencephalography procedure | Neural tube defects |
| Electrocardiogram | Congenital heart abnormalities |
| Embryo echocardiography | Congenital heart abnormalities |
| Embryoscopy | Facial and limb abnormalities |
| Exercise testing, congenital heart disease | Congenital heart abnormalities |
| Fetal skin biopsy | Albinism |
| Fetoscopy | Limb abnormalities |
| Fluoroscopy | Congenital heart abnormalities |
| Genital ambiguous surgery | Urogenital malformations |
| Growth monitoring | Congenital anomalies diagnosed after birth |
| Hemoglobin electrophoresis | Sickle cell anemia |
| Herpes and cytomegalovirus tests | Mother and fetus |
| Intrauterine surgery | Diaphragmatic hernia and myelomeningocele |
| Iron levels and hemoglobin electrophoresis | Thalassemia |
| IV urography and urethrography | Obstructive urethral congenital disease |
| Laryngoscopy | Congenital respiratory diseases |
| Liver function tests | Hemochromatosis |
| Lung aspiration | Congenital respiratory diseases |
| Lung biopsy | Congenital respiratory diseases |
| Lung function tests | Congenital respiratory diseases |
| Maternal health care services | Congenital anomalies |
| Maternal phenyl alanine | Fetal microcephaly |
| MRI scan and CT scan | Neural tube defects (hydrocephaly and spina bifida) |
| Neurosurgical techniques | Hydrocephaly and Spina Bifida |

Table 1: Continued.

| Services | Congenital abnormalities |
|---|---|
| NICU and special delivery services | Pregnancies with omphalocele |
| Nonconjugated acetyl test | Trisomy 21 |
| Preconceptional folic acid consumption | Congenital anomalies |
| Pregnancy health care services | Congenital anomalies |
| Public education | Congenital anomalies |
| Pulse and color doppler, angiocardiography | Congenital heart abnormalities |
| Radiography by barium swallow | Congenital respiratory diseases |
| Radio-nucleotide scan | Congenital respiratory diseases |
| Reconstructive surgery | VSD, PDA, AV, tetralogy of fallot, pulmonary valve stenosis, and noncyanotic congenital heart diseases |
| Reconstructive surgery | Cleft lip and palate |
| Screening programme | Congenital hypothyroidism, adrenal hyperplasia |
| Shunting | Myelomeningocele with hydrocephaly |
| Surgical treatment | Neurofibromatosis |
| Thoracentesis | Congenital respiratory diseases |
| Thoracoscopy | Congenital respiratory diseases |
| Treatment by dexamethasone | High risk pregnancy for congenital adrenal hyperplasia |
| Two-dimensional echocardiography | Congenital heart abnormalities |
| Ultrasound diagnostic services | Neural tube defects, heart defects, diaphragmatic hernia |
| Urine and plasma amino acids, serum ammonium | Fetal microcephaly |
| Urology corrective surgery | Hypospadias, cryptorchidism, undescending testis |
| Vaccination | Congenital defects caused by viral infections |
| Valvuloplasty and valve replacement | Aortic valve stenosis |
| Varios clinical tests | Eye defects, thyroid anomalies, speech and hearing defects |

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