

Organization and Provision of Genetic Services

Victor B. Penchaszadeh, MD
Genetics and Public Health
Mailman School of Public Health
Columbia University
April 15, 2003

Genetic Services

Objectives

“Help people with a genetic disadvantage to live and reproduce as normal as possible” (WHO)

Types of services

- A. Public health (community) genetics programs, with focus in population
- B. Clinical genetic services, with focus in individuals and families

Public Health (Community) Genetics Programs

Objectives and concept:

- Reduce the impact of genetic disorders and birth defects in the population through primary, secondary and tertiary prevention, respecting the dignity, autonomy and reproductive rights of people.
- They are public policies of health systems which offer voluntary services to the eligible population
- Their goal is not “eradication” of genetic disorders
- Objectives are different from eugenics

***Public Health (Community) Genetics
Programs***

Strategy I. Primary prevention of genetic disorders and birth defects .

Instruments and programs:

- Control of environmental factors that interact with genetic susceptibilities: chemical and radiation hazards in environment, foods, workplace, etc
- Public education in genetics
- Family planning and optimization of reproductive age
- Maternal nutrition
- Prenatal care
- Prevention of exposures to mutagens y teratogens

***Public Health (Community) Genetics
Programs***

Strategy II. Prevention based on reproductive options.

Instruments and programs:

- Identification of preconceptional and prenatal genetic risk factors:
 - carrier screening for recessive condition
 - screening pregnancies for fetal anomalies
- Prenatal diagnosis of genetic conditions

***Public Health (Community) Genetics
Programs***

Strategy III. Secondary Prevention.

Early (presymptomatic) detection and medical intervention to reduce clinical manifestations

Examples:

- Newborn screening followed by medical interventions
- Monitoring child growth and development
- Predictive genetic testing followed by medical interventions

Public Health (Community) Genetics Programs

Strategy IV. Tertiary Prevention.

Rehabilitation of disabilities

Examples:

- Child development centers
- Rehabilitation of motor and sensorial deficits
- Rehabilitation of mental retardation

Clinical Genetic Services

- Emphasis in medical and psychosocial needs of individuals affected or at increased genetic risk, and their families
- Emphasis in clinical and laboratory diagnosis, genetic counseling, follow-up and treatment
- Prevention of recurrences through genetic counseling and reproduction options

Needs of patients

- Concern about potential genetic risks to offspring (family history, exposures, genetic disorder)
- Current pregnancy suspected with fetal anomaly
- Child with suspected genetic condition
- Increased risk to develop a genetic condition in the future

Clinical Genetic Services Strategies and Instruments

- Assessment of risks to offspring: family history, medical information, etc
- Prenatal diagnosis to confirm a fetal condition
- Early and accurate diagnosis in symptomatic patients of any age
- Coordination of long-term multidisciplinary management of the patient
- Anticipatory guidance of natural history
- Genetic counseling and psychosocial support

Clinical Genetic Evaluation

- Family history
- Determining inheritance pattern
- Clinical genetic diagnosis, syndrome identification
- Laboratory confirmation:
 - chromosome analysis
 - biochemical tests
 - DNA tests

Genetic Testing

- Genetic testing cannot occur in a vacuum.
- Requires prior education and counseling, and informed consent (it is voluntary)
- It should be part of comprehensive genetic services, which include clinical genetics evaluation and genetic counseling
- Genetic counseling should be provided when informing results
- Follow up

Genetic counseling I

Communication process between a consultand and a professional with the necessary knowledge and skills, with the aims of:

- Understand the role of genetic factors in the causation of a condition
- Comprehend the notion of genetic risk and the probability of occurrence or recurrence of a genetic condition in the family

Genetic Counseling II

- Directiveness and non-directiveness in genetic counseling
- Empower consultands to make their own reproductive and medical decisions to deal with the genetic risk
- Support of patient's decisions
- Genetic counseling and primary prevention
- Genetic counseling and eugenics

Components of Genetic Counseling I

- Eliciting and interpreting individual and family medical, developmental and reproductive histories
- Determining mode of inheritance and risk of occurrence and recurrence of genetic conditions and birth defects
- Explaining the etiology, natural history, diagnosis and management of these conditions
- Interpreting and explaining the results of genetic tests and other diagnostic studies

Components of Genetic Counseling II

- Identify emotional, social, educational and cultural issues
- Evaluating the patient's/family's responses to the condition or risk of occurrence
- Providing patient-centered counseling and anticipatory guidance
- Promoting informed decision-making about testing, management, reproduction and communication with family members

Components of Genetic Counseling III

- Identifying and using community resources that provide medical, educational, financial, and psychosocial support and advocacy
- Providing written documentation of medical, genetic, and counseling information for patients and health professionals

Types of Genetic Services

- General Genetic Clinics
- Metabolic Clinics
- Single Disease Clinics
- Prenatal Genetics Clinics

General Genetic Clinics

Services to individuals affected with, or with a family history positive for:

- Known or suspected genetic disorders
- Congenital anomalies/birth defects
- Mental retardation, developmental disorders
- Consanguinity or ethnicity associated with increased risk for specific disorders

Metabolic Clinics

- Known or suspected inborn errors of metabolism
- Family history of a metabolic disorder

Single Disease Clinics

- Blood disorders (e.g. sickle cell disease)
- Pulmonary diseases (e.g. cystic fibrosis)
- Neurological disorders: e.g. muscular dystrophies, ataxia, Huntington disease, etc
- Specific birth defect requiring multidisciplinary approaches to management (e.g. spina bifida, craniofacial)
- Cancer

Prenatal Genetics Clinics

Risk of unfavorable pregnancy outcome or abnormal prenatal screening results

- Risks associated with maternal age
- couples with previous affected child
- couples with positive family history
- Couples with reproductive loss
- Risks associated with maternal illnesses, medications, exposures or infections
- Pregnancies with abnormal screening results or fetal ultrasound

Genetic Health Care Professionals

- Clinical geneticists
- Genetic counselors
- Genetic nurses
- Oncology nurses
- Perinatologists
- Other specialties
- Laboratory geneticists:
 - cytogeneticists
 - biochemical geneticists
 - molecular geneticists

Responsibilities of Genetic Professionals

- Obtaining and interpreting complex family information
- Providing detailed explanations of genetic testing
- Diagnosing children and adults
- Prenatal diagnosis
- Interpreting complicated genetic test results
- Genetic counseling
- Identifying, counseling and testing individuals and families at higher risk
- Screening entire groups or populations

Certification of Genetic Specialists

- Board of Medical Genetics (doctoral professionals):
 - clinical genetics
 - cytogenetics
 - biochemical genetics
 - molecular genetics
- Board of Genetic Counseling (master professionals)

Characteristics of genetic services

- Multidisciplinary and comprehensive
- Linked to related services: reproductive health and family planning, prenatal care, pediatrics, relevant subspecialties, etc
- Regionalized services by levels of complexity of care
- Access to information databases: syndrome identification, genetic testing, etc
- Links with parent/patient organizations

Comprehensive Tertiary Genetic Centers

- Genetic specialists and counselors
- Close links with:
 - fetal medicine
 - cancer centers
 - other tertiary services
- Laboratory Services
- Research & Education

Tertiary Genetic Centers Special Functions

- Access to and overview of the latest science
- Clinical diagnosis of rare genetic syndromes
- Laboratory (DNA and chromosomal) diagnosis
- Interpretation and integration of complex genetic information
- Genetic counseling
- Dealing with extended families long term

Integration of Genetic Centers with Secondary and Primary Care in the Community

- Outreach to secondary and primary services
 - genetic counselors as first line of consult
 - facilitate referrals of families at higher risk
 - education and liaison of between primary care team and tertiary care center
- Supervision and professional education
- Quality assurance

Issues in the Provision of Genetic Services

- Novel discipline
- Lack of genetic literacy among health professionals
- Deficient coordination and regionalization of services, leading to duplications
- Lack of insurance coverage for some services (i.e. counseling and cognitive services in general)

Special Issues in the Provision of Genetic Services to Ethnic-Cultural Minorities I

- Different native cultures
- Immigration experiences
- Dramatic life experiences and social risks that may dwarf genetic risks in magnitude and relevance

Special Issues in the Provision of Genetic Services to Ethnic-Cultural Minorities II

- World view different from the Western European perspective, particularly on the value of biomedicine and the meaning and acceptability of disability
- Poverty and deficient education
- Financial barriers
- Language barriers

Cultural Factors Affecting Genetic Services to Minorities

- Beliefs about causation of familial diseases and birth defects
- Importance of family life and integrity
- Respect for authority of the elderly
- Use of folk medicines
- Shame and stigmatization
- Expectation for directiveness in counseling

Systemic Barriers in the Delivery of Genetic Services I

- Unfriendly design and organization of services delivery
- Lack of awareness and respect for diverse cultures by genetic providers
- Condescending approach to traditional beliefs and practices
- Stereotypes of patients from ethnic minorities

Systemic Barriers in the Delivery of Genetic Services II

- **Lack of health personnel of the same ethnicity and language**
- **Lack of awareness by the provider of own cultural values**
- **Language, class and cultural asymmetry between providers and patients**
- **Prejudice and social discrimination**
- **Dogmatic reliance of genetic professionals on the Western biomedical model**

Strategies for improving access I:

- Address deficiencies comprehensively with health care in general.
- Implement universal and equitable access to health care.
- Eradicate institutional practices that reinforce discrimination.
- Increase the number of genetic professionals from underserved communities.

Strategies for improving access

II:

- **Provide accessible culturally and linguistically appropriate services**
- **Stimulate community participation**
- **Incorporate the extended family**
- Promote links with parent/patient organizations

Strategies for improving access

III:

- Improve genetic knowledge among health professionals.
- Include meaningful, non-stereotyping cultural sensitivity training for health professionals.
- Public education in medical genetics.
