Organization and Provision of Genetic Services

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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 and 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


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

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**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

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**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 if 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.