WHAT IS GENETICS?

- Genetics is the science of genes, heredity and variation in living organisms.
  - The fact that living things inherit traits from their parents has been used since prehistoric times to improve crops/plants and animals through selective breeding.
Normal male karyotype (46, XY)
Normal female karyotype (46, XX)

A – adenine
C – cytosine
T – thymine
G - guanine
Gene with a G to T nucleotide change
MEDICAL GENETICS

- inheritance, diagnosis, treatment of diseases caused by mutations, chromosome abnormalities
  - Newborn Screening
  - Clinical Genetics
    - application of genetics to clinical problems
  - Cytogenetics
  - Biochemical Genetics
  - Molecular Genetics
    - application of molecular biology to diagnosis and treatment of disease

PERSONALIZED MEDICINE

- rapidly advancing field of health care that promises greater precision and effectiveness than traditional medicine because it is informed by each person’s unique clinical, social, genetic, and environmental information to prevent, diagnose, and treat disease
WHAT IS GENETIC COUNSELING?

- The process by which patients or relatives at risk of a hereditary disorder are advised of
  - the consequences of the disorder
  - the probability of developing or transmitting it
  - the ways in which this may be prevented, avoided or ameliorated

Practical Genetic Counselling by Harper, 6th ed

WHAT IS GENETIC COUNSELING?

- It focuses mainly on susceptibility to disease in individuals who are:
  - Suspected of having a heritable disease
  - At risk because of their family history
  - Concerned about the possibility of having an affected child based upon personal or family history, age or ethnicity
WHAT IS GENETIC COUNSELING?

- communication process
- address individual concerns relating to development/ transmission of hereditary disorder
- strong communicative and supportive element
  - Allows those who seek information to reach their own fully informed decisions without undue pressure or stress

WHAT IS THE ROLE OF GENETIC COUNSELING?

- identify families at risk
- interpret information about the disorder
- analyze inheritance patterns and risk of re-occurrence
- review available option/s with the family
- provide genetic information
- make their clients’ best interest their foremost priority
WHO ARE GENETIC COUNSELORS?

- Genetic counselors are health professionals with specialized graduate degrees and experience in the areas of medical genetics and counseling.

- Most enter the field from a variety of disciplines, including biology, genetics, nursing, psychology, public health and social work.

  (NSGC 1983)

GENETIC COUNSELORS

- Assist patients in addressing the scientific and emotional issues that arise
  - Diagnosis of genetic disorder in a newborn
  - Identification of a mutation conferring elevated risk of cancer, etc.

- Help individuals make informed decisions based on their own values and individual circumstances.
GENETIC COUNSELORS

- Must keep pace with new developments in biomedical research
  - Diagnostics
  - Pharmacogenetics

GENETIC COUNSELORS

- Interact with clients and other healthcare professionals in a variety of clinical and non-clinical settings
WHERE DO GENETIC COUNSELORS WORK?

- Majority of genetic counselors work at:
  - University medical centers
  - Private or public hospitals

- Some genetic counselors:
  - Work in laboratories
  - Coordinate research studies
  - Work in private industry
  - Educators and resource people
  - Community

WHAT DO GENETIC COUNSELORS DO?

- Provide information and support to
  - families who have members with birth defects or genetic disorders
  - families who may be at risk for a variety of inherited conditions

- Identify families at risk, investigate the problem, interpret information about the disorder, analyze inheritance patterns and risks of recurrence and review available options with the family.
WHO SHOULD HAVE A GENETICS CONSULTATION?

- Those concerned about a genetic disease
  - Information
  - Coping strategies

- Reasons for referral are often grouped into:
  - Preconception/prenatal
  - Pediatric
  - Adult

COMMON REASONS FOR PRECONCEPTION CONSULTATION

- Mother ≥ 35 years old at delivery
- Abnormal fetal ultrasound results
- Personal/ Family Hx of a known or suspected genetic disorder, birth defect, or chromosomal abnormality
- Exposure to a known or suspected teratogen
- Mother has a medical condition known to affect fetal development
- 2 or more pregnancy losses
- Close biological relationship of parents
- Ethnic predisposition to certain genetic disorders
### COMMON REASONS FOR PEDIATRIC GENETICS CONSULTATION

- Abnormal newborn screening results
- One or more major malformations in any organ system
- Mental retardation or developmental delay
- (+) known or suspected genetic disorder or chromosomal abnormality
- FHx of known or suspected genetic disorder, birth defect, or chromosomal abnormality

### COMMON REASONS FOR ADULT GENETICS CONSULTATION

- Mental retardation
- Personal/ Family Hx of hereditary cancers
- Personal/ Family Hx of a known or suspected genetic condition or chromosomal abnormality
- Development of a degenerative disease
- Risk assessment for pregnancy planning
WHO NEEDS GENETIC COUNSELING?

- Known genetic disorders
  - Ex. Down’s syndrome, Metabolic disorders
- Birth defects
  - Ex. Cleft Lip and Palate, Congenital Heart Disease
- Inherited Cancers
  - Ex. Breast and Bowel Cancers
- Intellectual disability
- Infertility or multiple miscarriages or infant deaths
- Genetic defects occurring frequently in specific populations
  - Ex. Sickle cell Anemia, Thalassemia

RESPONSIBILITIES OF A GENETIC COUNSELOR

- Provide expertise in clinical genetics
- Counsel and communicate with patients on matters of clinical genetics
- Provide genetic counseling services in accordance with professional ethics and values

APPROVED June 2007 by the National Society of Genetic Counselors USA
www.abg.net
PRE-REQUISITES OF GENETIC COUNSELING

- Detailed family history
- Accurate diagnosis
- Understanding the medical aspect of the disorder (etiology, natural history, treatment, prognosis, burden)
- Understanding the inheritance pattern (recurrence risk)
- Understanding the psycho-social impact of the information
- Training / experience in counselling techniques
- Understanding the concepts of health / disease / healthcare in the appropriate cultures

WHAT HAPPENS DURING A GENETIC COUNSELING SESSION?

- Determine why the patient or family is seeking genetic counseling
- Identify what information they wish to get out of the session
- ask for any genetic disorder, multiple miscarriages and early or unexplained death of relative (if any)
- look for medical records including
  - Ultrasound
  - Parental test results
  - Other diagnosis which have been made in the past.
- An accurate pedigree chart is made by seeking information from the patients, their family members, cousins, in –laws, sibling and grandparents to determine the presence of any inherited genetic disorder
WHAT HAPPENS DURING A GENETIC COUNSELLING SESSION?

- A family having problems and difficulty will have to decide what to do next
- The genetic counselor will help the patient/family face and understand the problems
- The family will have to prepare themselves for the challenges they will face
- The counselor explains the way heredity contributes to the disorder risk of re-occurrence
- The counselor will make the family understand the alternatives for dealing with the risk
- The counselor will make them choose a course of action and make the best possible adjustment.

ESTABLISHING THE DIAGNOSIS

- **most crucial step** in any genetic counseling
- if incorrect - totally misleading information could be given with tragic consequences
- reaching diagnosis involves three fundamental steps
  - taking a history
  - examination
  - undertaking appropriate investigations
WHAT IS A PEDIGREE CHART?

- Pedigree is used to help make a diagnosis of genetic disease.
- To determine a person’s risk of developing a genetic disease.
- Contains vital medical information like birth date, age of death, cause of death, health problems and results of genetics test.
- To determine the risk of a relative to develop a genetic disease or have a child with a genetic disease.
- On the basis of information gathered from the first degree relatives i.e. parents & siblings, second degree of relatives i.e. aunts & uncles and third degree relatives i.e. cousins & grand parents, a pedigree chart is made.

SYMBOLS USED IN PEDIGREE CONSTRUCTION

- Male
- Female
- Mating
- Parents & Children 1 boy 1 girl (in order of birth)
- Dizygotic twins
- Monozygotic twins
- Sex unspecified
- Number of children of sex indicated
- Affected individuals
- Heterozygotes for autosomal recessive
- Carrier of sex-linked recessive
- Death
- Abortion of stillbirth sex unspecified
- Proband
- Method of identifying persons in a pedigree
- Here the proband is Child 2 in Generation II
- Consanguinous marriage
COUNSELING

- Session can be so **intense and intimidating** that amount and accuracy of information retained is very disappointing.
- **Letter** summarizing the topics discussed at counseling session is often sent to family.
- **Follow-up home visit or clinic appointment** to clarify any confusing issues.

Ethical Guidelines and Approaches Principles

**Non-directive counselling**

- The counselor is expected to give a **non-directive counselling** which involves the presentation of the facts in an unbiased manner, leaving the entire responsibility of decision with the consultee.
- It is an **individual-centered approach to care by focusing on the thoughts and feelings of each person.**
- **Parental autonomy in abortion choices.** Pre-natal diagnosis must be provided when parents need the information to prepare themselves for the birth of a possibly affected child.
- Practitioners need to **disclose to the consultee the risks and benefits of each procedure.**
IN SHORT, GENETIC COUNSELING IS

- **Determine the facts**
  - Diagnosis, etiology, and inheritance patterns, prognosis, natural history, treatment and re-occurrence of risk

- **Transmitting the information**
  - To those requesting it in a sensitive, culturally appropriate, understandable way

- **Supporting the decision**
  - Supporting the decision making process of the couple

- **Genetic counseling**
  - It is non-directive

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- **Medical genetics in the Philippines is fast expanding**

- **MS Genetic Counseling is a 2-year degree program**
  - health care providers with specialized training in psychosocial counseling of families with risks for genetic disorders

  - work closely with the clinical/medical geneticist who provides the clinical diagnosis and management of a patient

  - work with a team of other health care providers (i.e. social workers, nurses, nutritionists, and other specialized medical professionals) to provide the best care to patients and their families.
### MS in GENETIC COUNSELING

- **2009** - strategic planning bet. local medical geneticists and international genetic professionals to develop the curriculum
- **January 2011** - Board of Regents at the University of the Philippines approved the proposed curriculum
- **June 2011** - training of the Philippines’ first cohort of genetic counseling students commenced

![Group Photo]

### APPLICATION FORMS MAY BE OBTAINED FROM

- NATIONAL GRADUATE OFFICE FOR THE HEALTH SCIENCES, UP Manila
  - Tel. # 526-5850; 523-1495
  - www.ngohs.upm.edu.ph

### FURTHER INQUIRIES MAY BE SENT TO

- DEPT. OF PEDIATRICS, UP-PGH
  - Tel. # 554-8400 loc 2100
- INSTITUTE OF HUMAN GENETICS, NIH
  - Tel. # 307-0780
THANK YOU!