# Introduction to the module of Community Genetics

Professor Hanan Hamamy

Geneva University Hospital

Training Course in Sexual and Reproductive Health Research
Geneva 2010

# Reproductive Health and Community Genetics

- The World Health Organization definition of Reproductive Health encompasses the right of men and women to be informed and to have access to safe, effective, affordable and acceptable methods of fertility regulation of their choice, and the right of access to appropriate health care services that will enable women to go safely through pregnancy and childbirth and **to provide couples with the best chance of having a healthy infant**.

- **The goal of community genetic services would be to maximize the chances for having healthy babies**

# What is meant by Community Genetic Services?

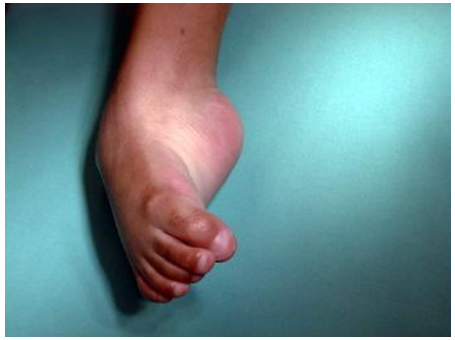
- It means the care and prevention of congenital disorders through community based services
- Provide services with **equity** and **efficiency** to all those in need
- Care and prevention of birth defects is an essential and integral part of maternal and child health programs.

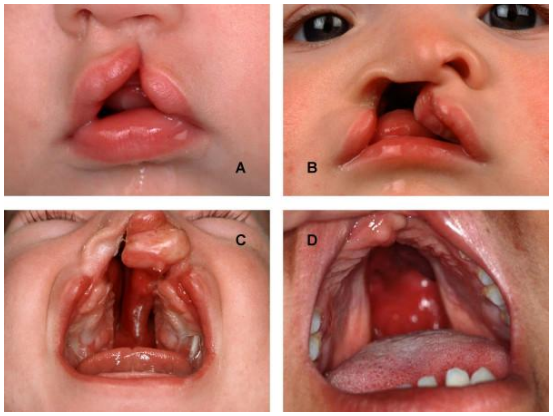
# Birth defects (Congenital Disorders)

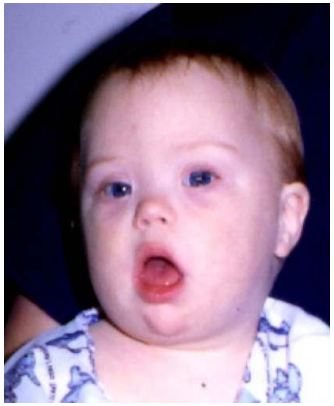
- Congenital disorder or Birth defect includes any morphological (structural), functional and biochemical-molecular defect that may develop in the embryo and fetus from conception until birth, whether detected at birth or later.

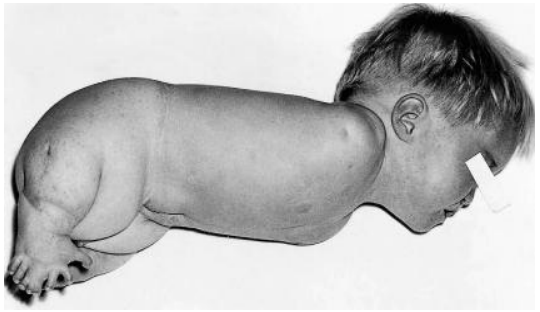
# Categories of Congenital Disorders

- Congenital malformations (structural defects)
- Genetic disease (Chromosome or single gene abnormality)
- Intrauterine infections as toxoplasmosis and exposure to teratogenic drugs







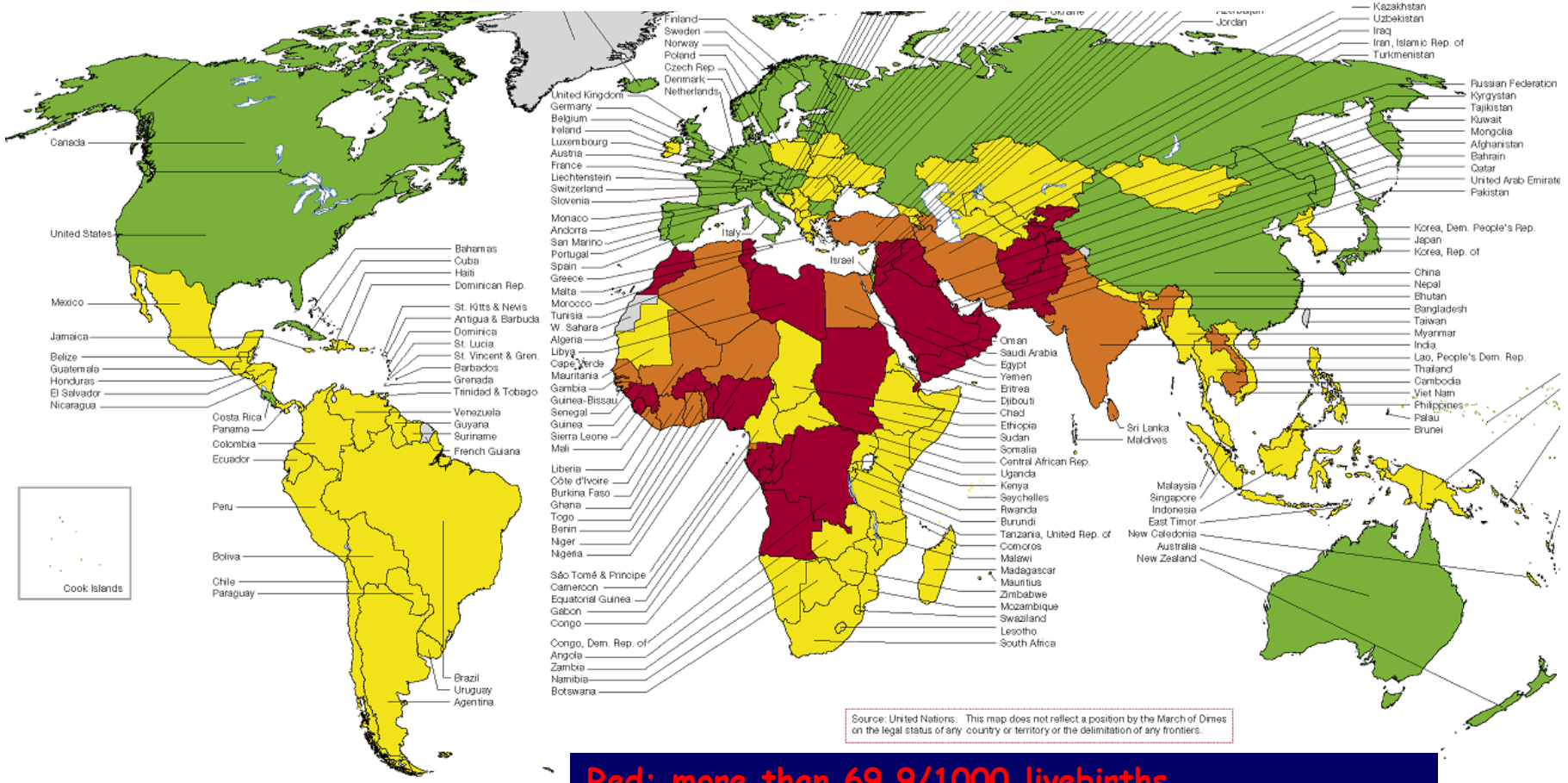
# Burden of congenital disorders

- Every year around 8 million infants are born with a serious birth defect, of which several hundred thousands are caused by teratogens (alcohol, rubella, syphilis, iodine deficiency). 3.3 million die, 3.2 survive with severe disability
- According to experience from industrialized countries, up to 70% of birth defects can be prevented or adequately managed

# MARCH OF DIMES

## GLOBAL REPORT ON BIRTH DEFECTS

THE HIDDEN TOLL OF DYING AND DISABLED CHILDREN




Red: more than 69.9/1000 livebirths

Orange 61-69.9

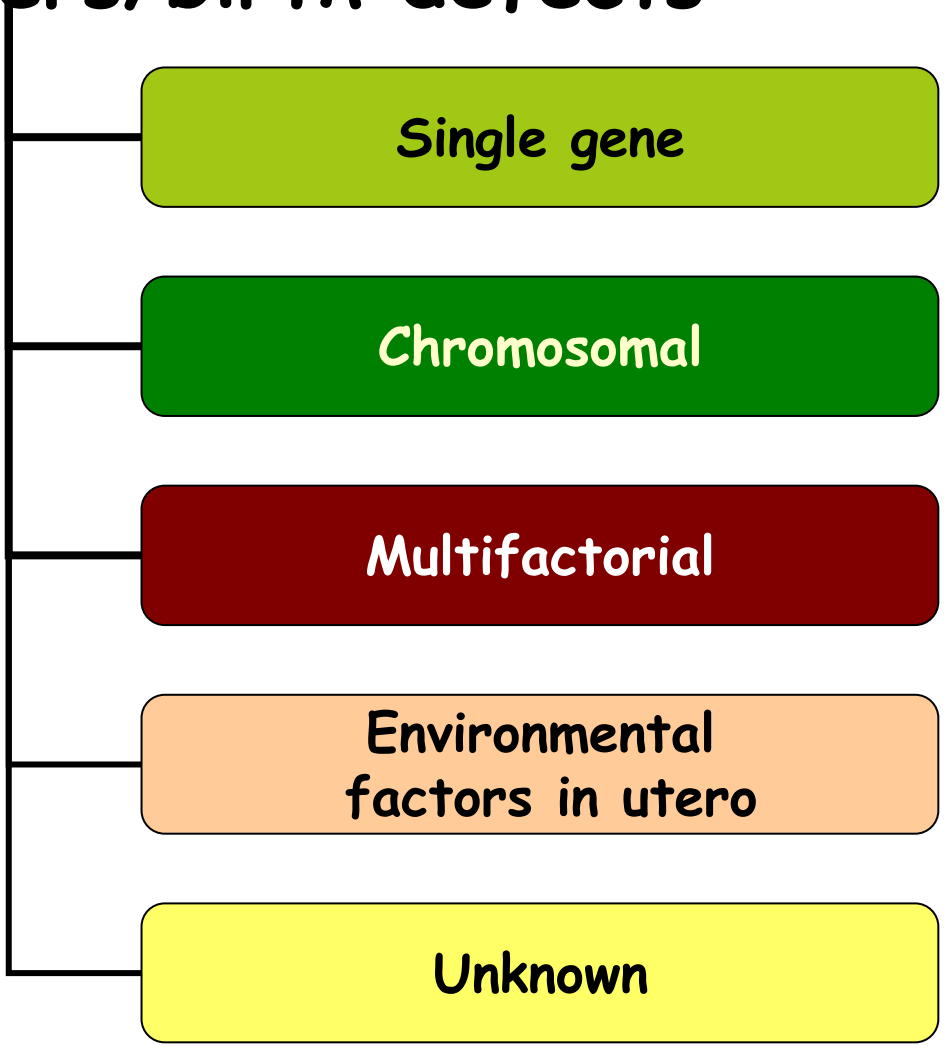
Yellow 52.1-60.9

Green less than 52.1

Grey no data

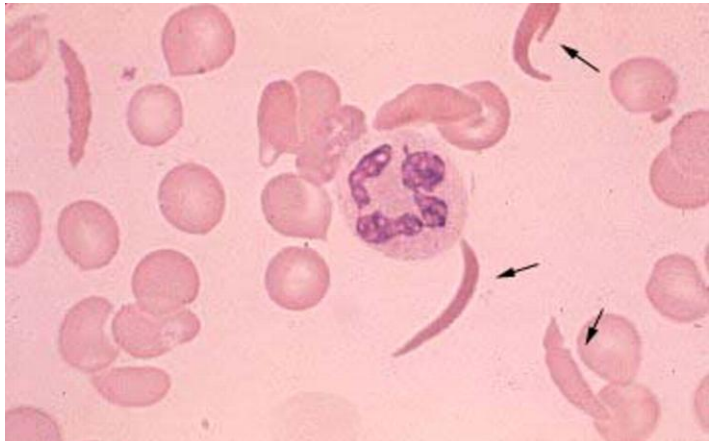
Christianson A,Howson C, Modell B, 2006

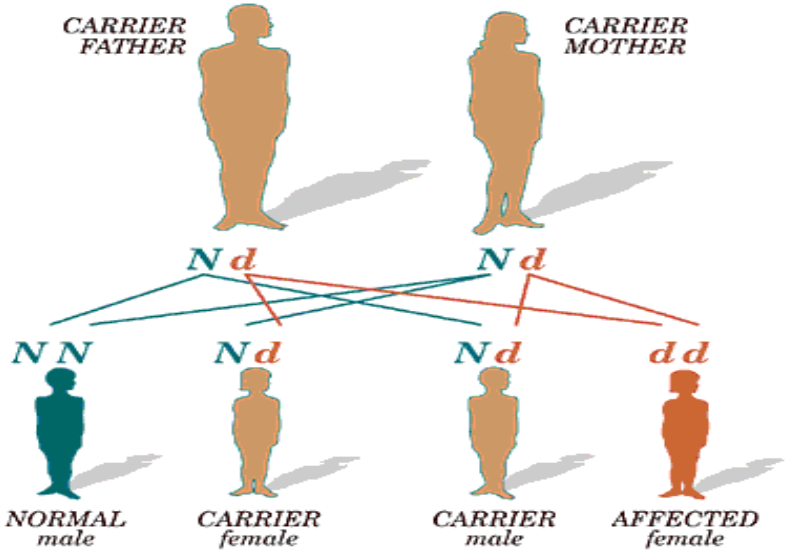
# Underlying etiology of Congenital disorders/birth defects



# Single gene disorders

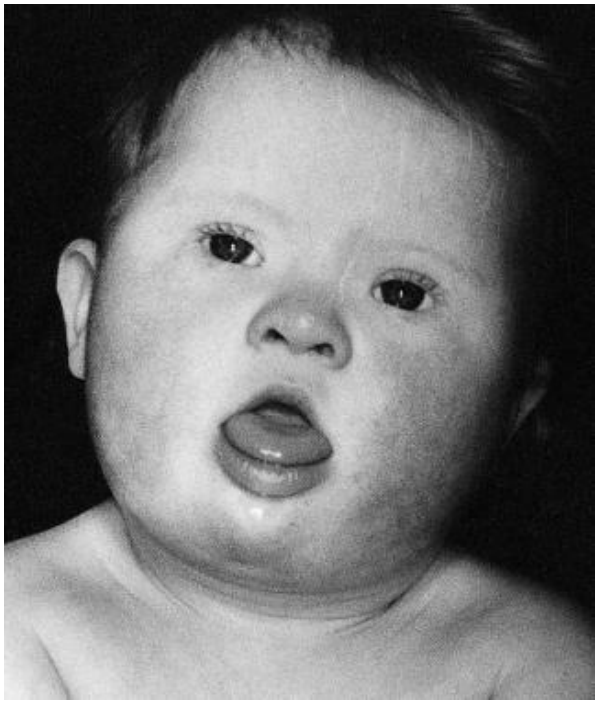
- Single-gene disorders occur in about 1% of in neonates
- There are thousands of different single gene disorders
- **sickle cell anemia is one example**
- **If a child is affected, risk of affection for future pregnancy is 25%**





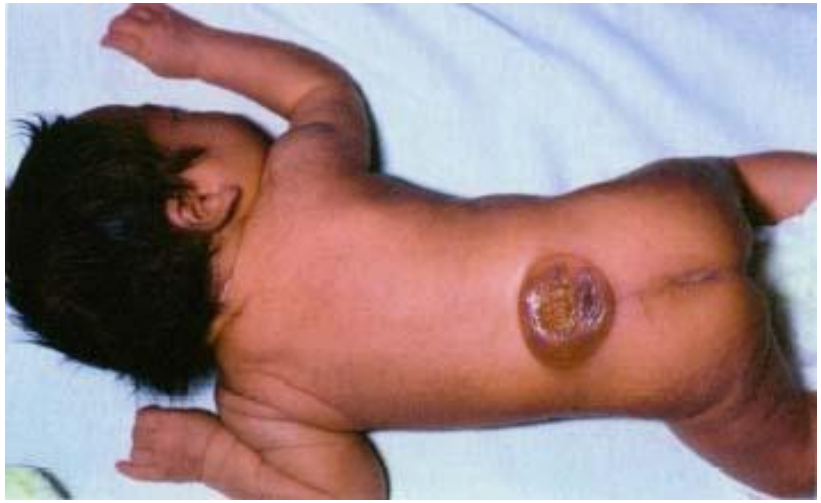
# Chromosomal disorders

- Chromosome abnormalities occur in about 0.5% in neonates
- More than 20,000 different chromosomal abnormalities have been reported
- Down syndrome is the most common (about 1/600 livebirths)
- ***Risk of having a child with Down syndrome increases with advance in maternal age***



# Multifactorial disorders : congenital abnormalities/malformations

- Of all neonates, 2-3% have at least one major congenital abnormality (structural defect).
- Most **congenital abnormalities** such as congenital heart defects, neural tube defects and cleft lip/palate have multifactorial etiology which is an interaction between both genetic and environmental factors to cause the condition
- After the birth of a child with for example spina bifida, the risk of recurrence in a future pregnancy is around 4% and increases with increase in number of affected in the family
- Supplementation by folic acid pre- and postconception can markedly decrease the risk for neural tube defects

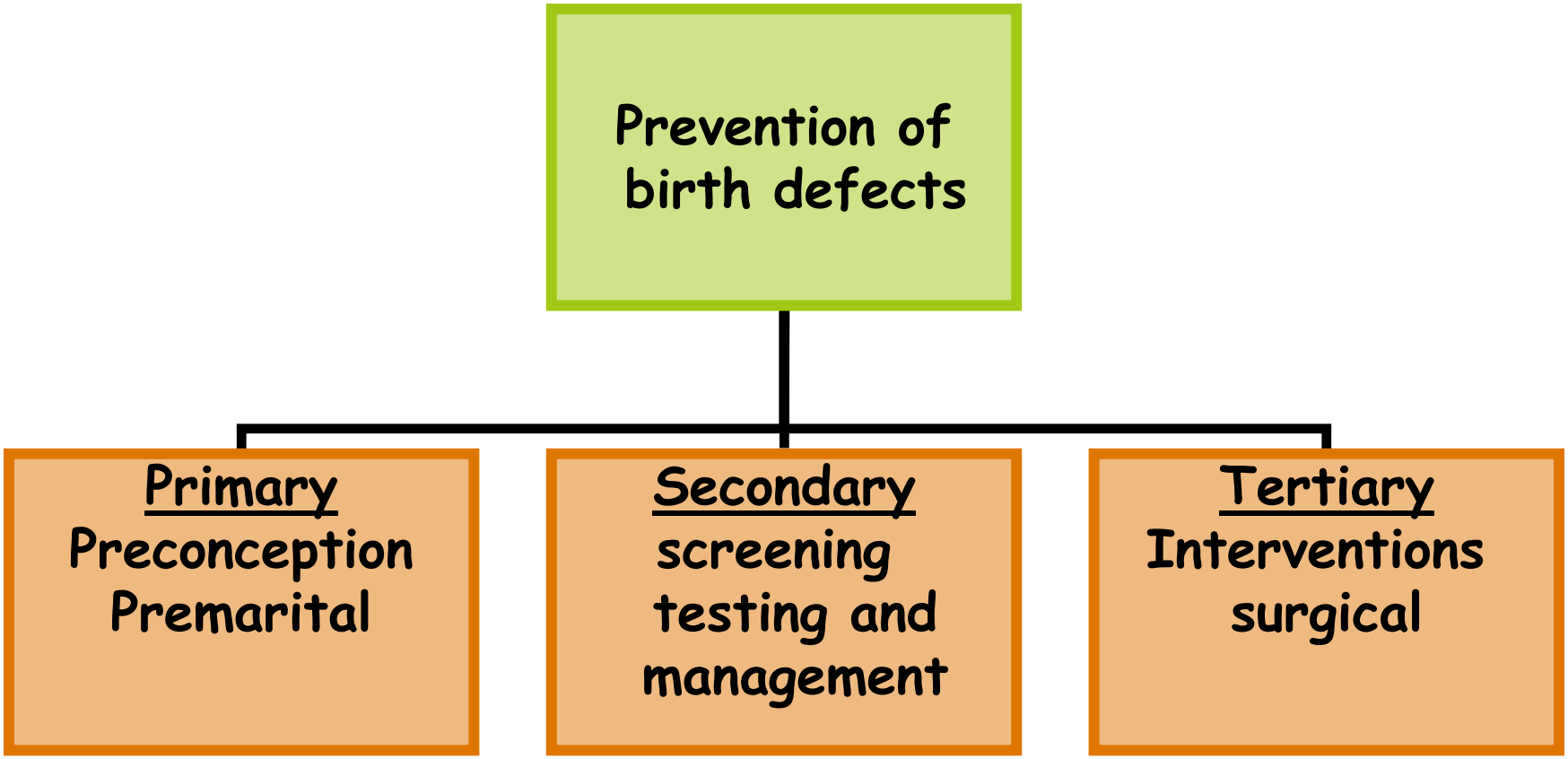


# In utero environmental factors (infections, drugs, chemicals)

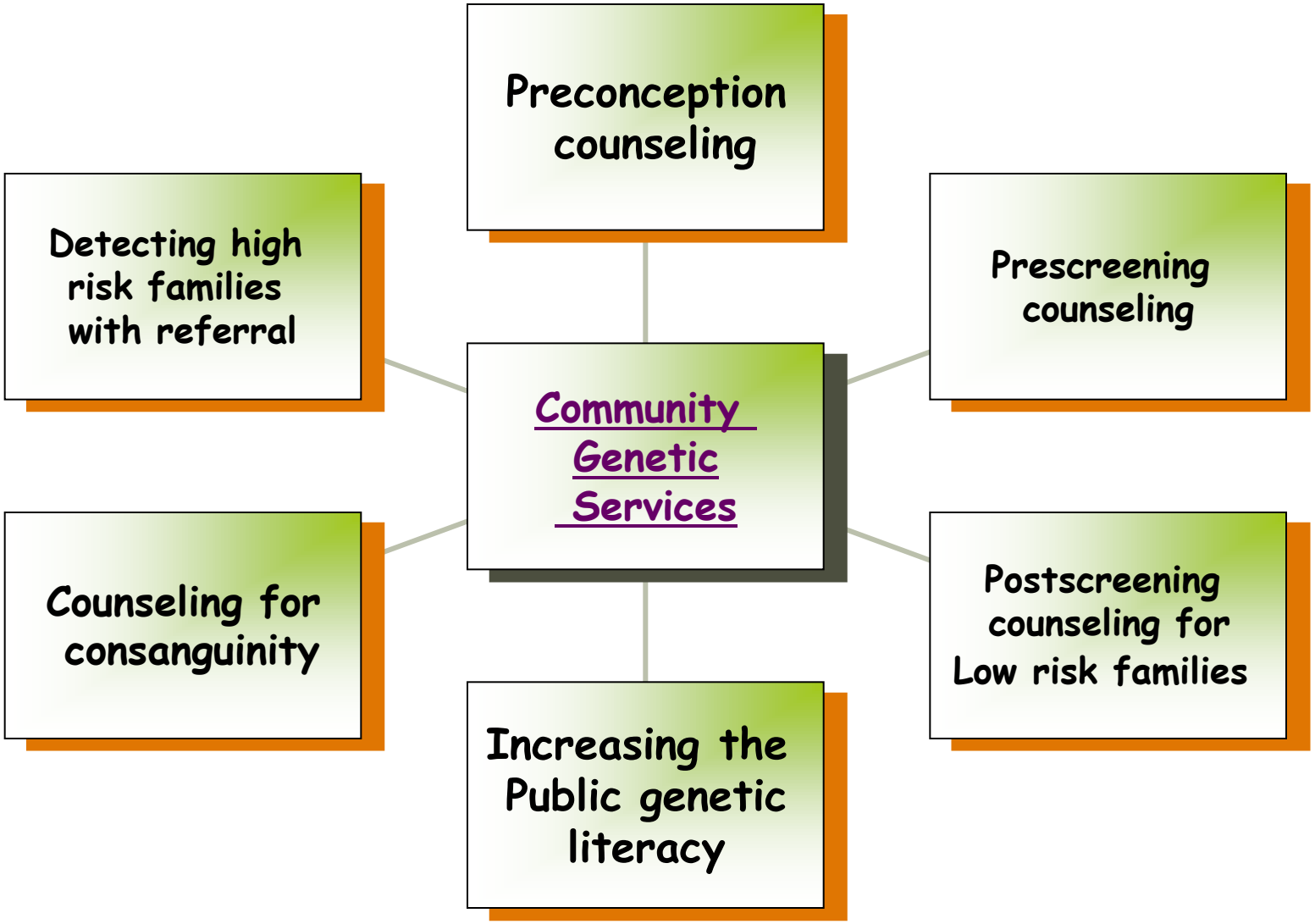
## Example: congenital syphilis

- Pregnant women who are infected with syphilis can transmit the infection to their fetus, causing congenital syphilis, with serious adverse outcomes for the pregnancy in up to 80% of cases.
- An estimated two million pregnancies are affected annually; approximately 25% of these pregnancies end in stillbirth or spontaneous abortion, and in a further 25% the newborn has a low birth weight or serious infection, both of which are associated with an increased risk of perinatal death.
- *A large reduction in congenital syphilis is feasible with relatively simple interventions focused on maternal and newborn care.*

About 70% of birth defects are preventable



# Basic Community Genetic Services that can be offered by primary health care providers



# Preconception counseling to maximize the chances of a couple to have a healthy baby

1. Preconception nutritional supplementation for example proper intake of folic acid can minimize the risks for neural tube and other defects
2. Prevention and treatment of maternal infections (Rubella, TORCH, Syphilis)
3. Diagnosis of maternal Rh status
4. Information on risks of advanced maternal age at conception
5. Avoidance of teratogenic drugs and chemicals
6. Cessation of smoking and alcohol intake
7. Family planning
8. Monitoring of maternal health before and during pregnancy (diabetes, hypertension, epilepsy, hyperthermia)

# Counseling related to population screening programs

- Newborn screening for example for congenital hypothyroidism and phenylketonuria
- Screening for carriers of common autosomal recessive conditions for example beta thalassemia
- Prenatal screening
- Referral of high risk couples to specialised centers

## Identification and referral of individuals and families at increased genetic risk to specialized centers

**Main services provided at a genetic center**

- Genetic counselling to families reporting to the centre
- Laboratory services for the diagnosis of genetic disorders (cytogenetics, molecular genetics, biochemical tests)
- Teaching and Training of health professionals on the principles and practice of medical genetics
- Research

## Counseling related to consanguinity in communities where consanguineous marriages are common and referral when indicated

- Detailed family history should be taken to recognize any congenital disorder
- If a genetic disorder is suspected in the family, refer the couple to a specialized genetic counseling clinic
- If there is no known inherited disorder in the family, first cousin marriages are given a risk for birth defects of double the population risk (for example instead of 2.5%, it becomes 5%)

# Basic content of the community genetics module

# Principles of Human Genetics

- Introduction: Categories of genetic diseases, introduction to human genetics
- Single gene disorders, pedigree construction and risk calculation
- Chromosome disorders and cytogenetic techniques
- Multifactorial inheritance
- Basic concepts in dysmorphology
- Principles of population genetics

# Principles of Genetic Counseling

- Basic principles, aims, and ethical issues in genetic counseling
- Ethical issues in medical genetics and genetic services
- Preconception counseling
- Consanguinity and counseling
- Counseling for chronic non-communicable diseases

# Prevention strategies

- Community genetic services
- Prevention of congenital disorders
- Birth defects registry
- Genetic screening and testing
- Prenatal screening and diagnosis
- Prevention programs for hemoglobinopathies

## Upon completion of this module, participants will be able to:

- **Understand and clearly explain the principles of inheritance of diseases.**
- **Understand the basis of common congenital disorders in the community.**
- **Take a basic genetic family history and construct a pedigree.**
- **Know how to detect families with genetic risks and where and when to refer them.**
- **Know the basic ethical principles and techniques of genetic counseling.**
- **Offer preconception counseling.**

## Upon completion of this module, participants will be able to:

- **Offer prescreening counseling, and knows how to detect at risk couples or individuals for referral to special centers.**
- **Offer counseling related to consanguinity in communities with high consanguinity rates.**
- **Know the basic strategies for the prevention and care of common genetic disorders in the community.**
- **Access appropriate information materials for patients, families and the general public.**

# Assignments

- At the beginning of each week you will be asked to read a number of powerpoint presentations and documents and to answer the questions assigned to that week.

The overall goal of community genetic services would be to maximize the chances for having healthy babies and to achieve a progressive decline in the rates of congenital disorders for the benefit of families in the community.