



Introduction to the module of Community Genetics

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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.
- Community genetic services can provide couples with the best chance of having a healthy infant.

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.

Definition of Community Genetics

- Community genetics is the art and science of the responsible and realistic application of health and disease-related genetics and genomics knowledge and technologies in human populations and communities to the benefit of individuals therein. Community genetics is multi-, inter- and transdisciplinary and aims to maximize benefits while minimizing the risk of harm, respecting the autonomy of individuals and ensuring equity.

Leo ten kate et al, *J of Community Genetics*, 2010

(<http://www.springerlink.com/content/rh7761342130p643/fulltext.html>)

Activities and areas of interest within the field of community genetics

- *Genetic screening*
- *Genetic literacy/education*
- *Access and quality of genetic services*
- *Genetics in primary care*
- *Genetics in middle and low income countries*
- *Genetics in disadvantaged subpopulations*
- *Registries of congenital and genetic disorders*
- *Genetics in preconception care*
- *Public consultation about genetic issues*
- *Epidemiologic issues*
- *Economic issues*
- *Psychosocial issues*
- *Ethical and legal issues*
- *Policy issues*

*Leo ten kate et al, J of Community Genetics, 2010
(<http://www.springerlink.com/content/rh7761342130p643/fulltext.html>)*

Community Genetics and Clinical Genetics

- Although benefit to the individual is central to both community genetics and clinical genetics, community genetics seeks to locate people within the wider community who may be at increased risk of a genetic problem, but have not yet been identified or helped. Whereas clinical geneticists deal with persons or families with a particular problem or concern who have requested or been referred for a consultation.

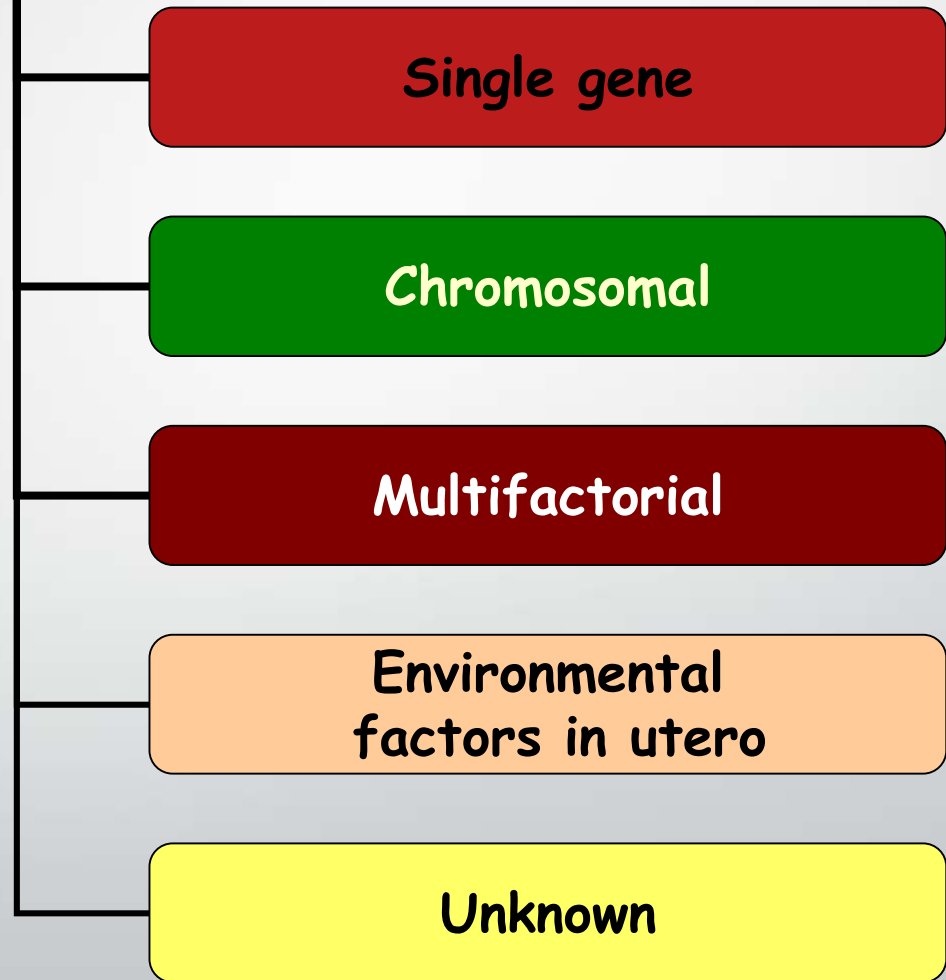
Burden of congenital disorders

- 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

Birth defects (Congenital Disorders)

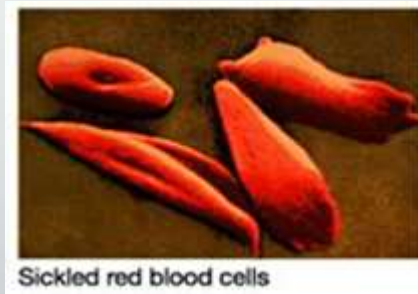
- Congenital disorder or Birth defect includes any morphological (structural), functional and biochemical-molecular defect that may develop in the embryo and fetus from conception until birth, whether detected at birth or later.

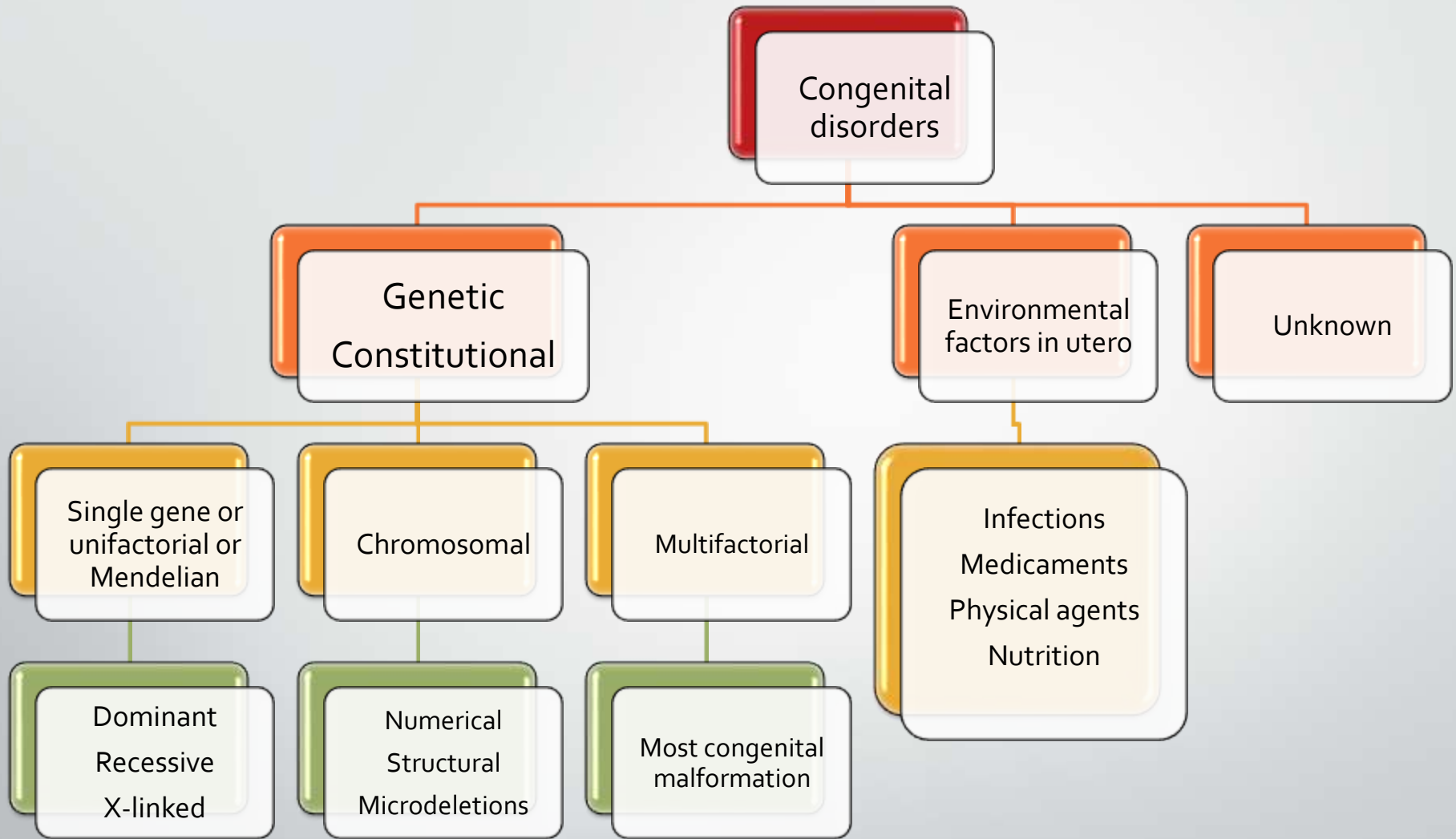
Underlying etiology of Congenital disorders/birth defects



Categories of Congenital Disorders

- Congenital malformations (structural defects) such as cleft lip and palate
- Genetic disease (Chromosome such as Down syndrome or single gene abnormality such as sickle cell anemia)
- Intrauterine infections as Rubella and exposure to teratogenic drugs such as thalidomide

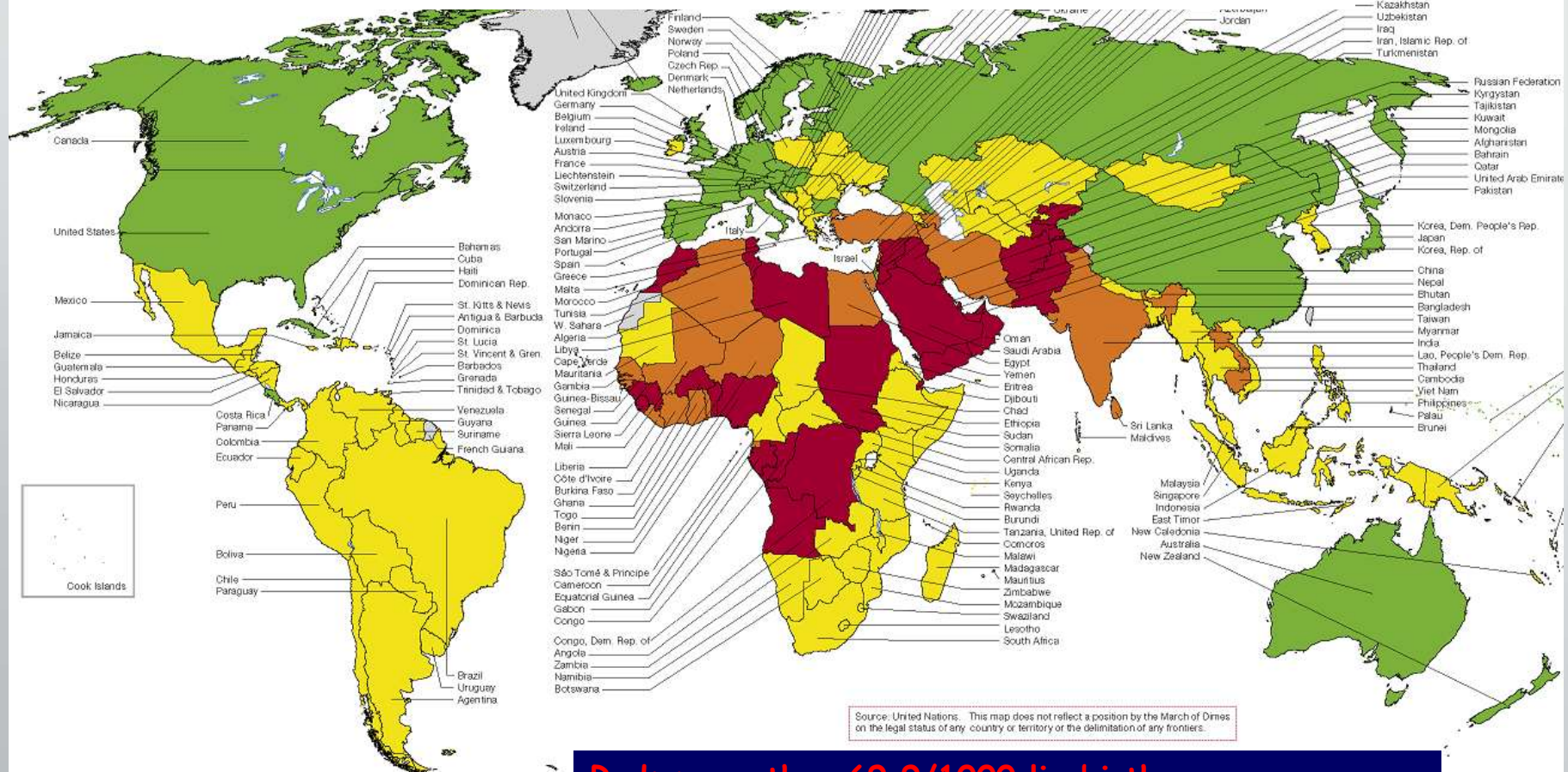




MARCH OF DIMES

GLOBAL REPORT ON BIRTH DEFECTS

THE HIDDEN TOLL OF DYING AND DISABLED CHILDREN



Source: United Nations. This map does not reflect a position by the March of Dimes on the legal status of any country or territory or the delimitation of any frontiers.

Red: more than 69.9/1000 livebirths

Orange 61-69.9

Yellow 52.1-60.9

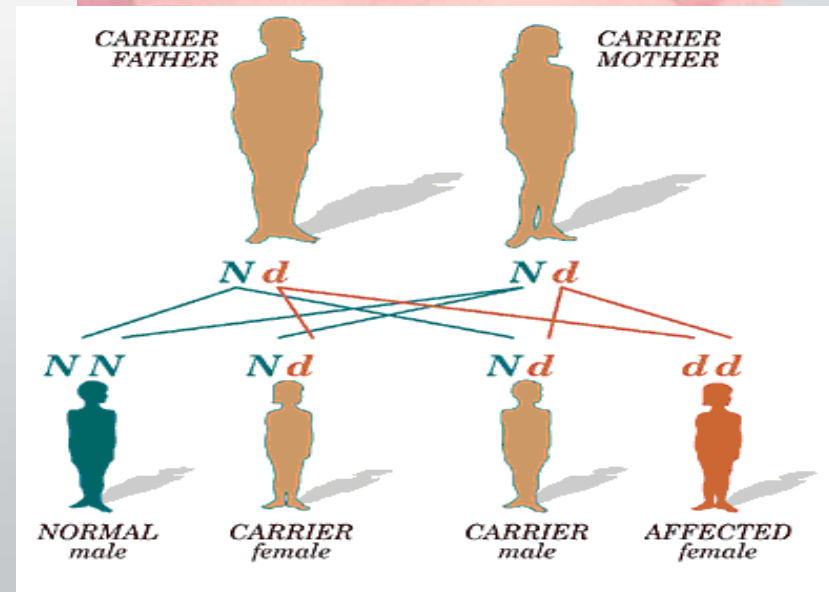
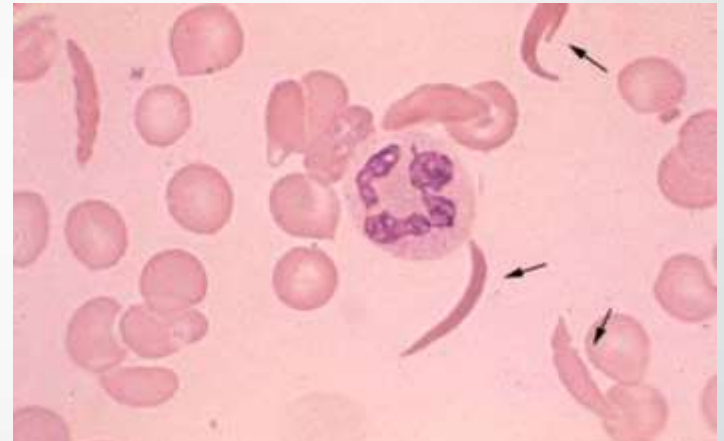
Green less than 52.1

Grey no data

Christianson A, Howson C,
Modell B, 2006

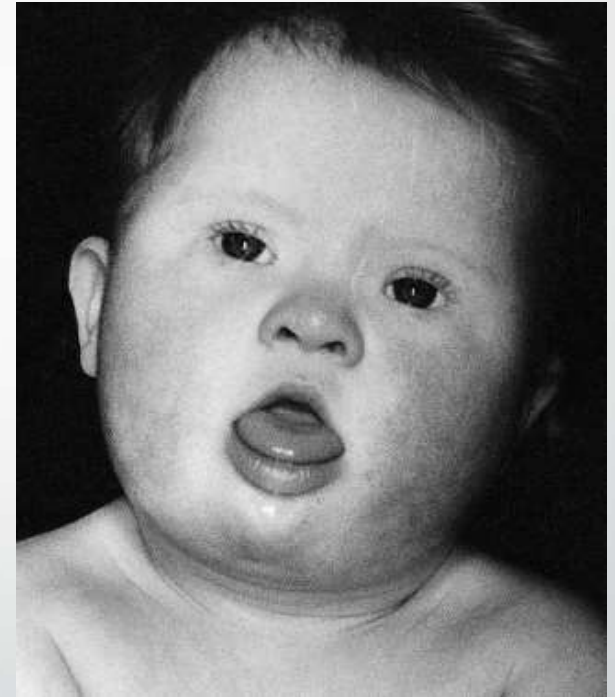
Single gene disorders

- 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.*

Prevention and care of Congenital disorders

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graph TD; A[Prevention and care of Congenital disorders] --> B[Primary]; A --> C[Secondary]; A --> D[Tertiary];
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Primary

Preconception care
Premarital and preconception
screening

Secondary

Prenatal screening
Testing and management

Tertiary

Newborn screening
Management of affected

About 70% of congenital disorders can be preventable

Community based interventions to reduce burden of congenital disorders I

Preconception care

- Giving protection: nutrition, vaccinations, genetic counseling..
- Managing conditions: chronic maternal diseases, infections..
- Avoiding exposures known to be teratogenic or otherwise harmful: medicaments, infectious agents, smoking, alcohol, illicit drugs....

Community based interventions to reduce burden of congenital disorders II

Health care services and national programs

- Genetic counselling and prenatal diagnosis
- Management and rehabilitation of affected
- Food fortification
- Immunizations
- Surveillance and Registries
- Promoting healthy lifestyle

Community based interventions to reduce burden of congenital disorders III

Population screening programs

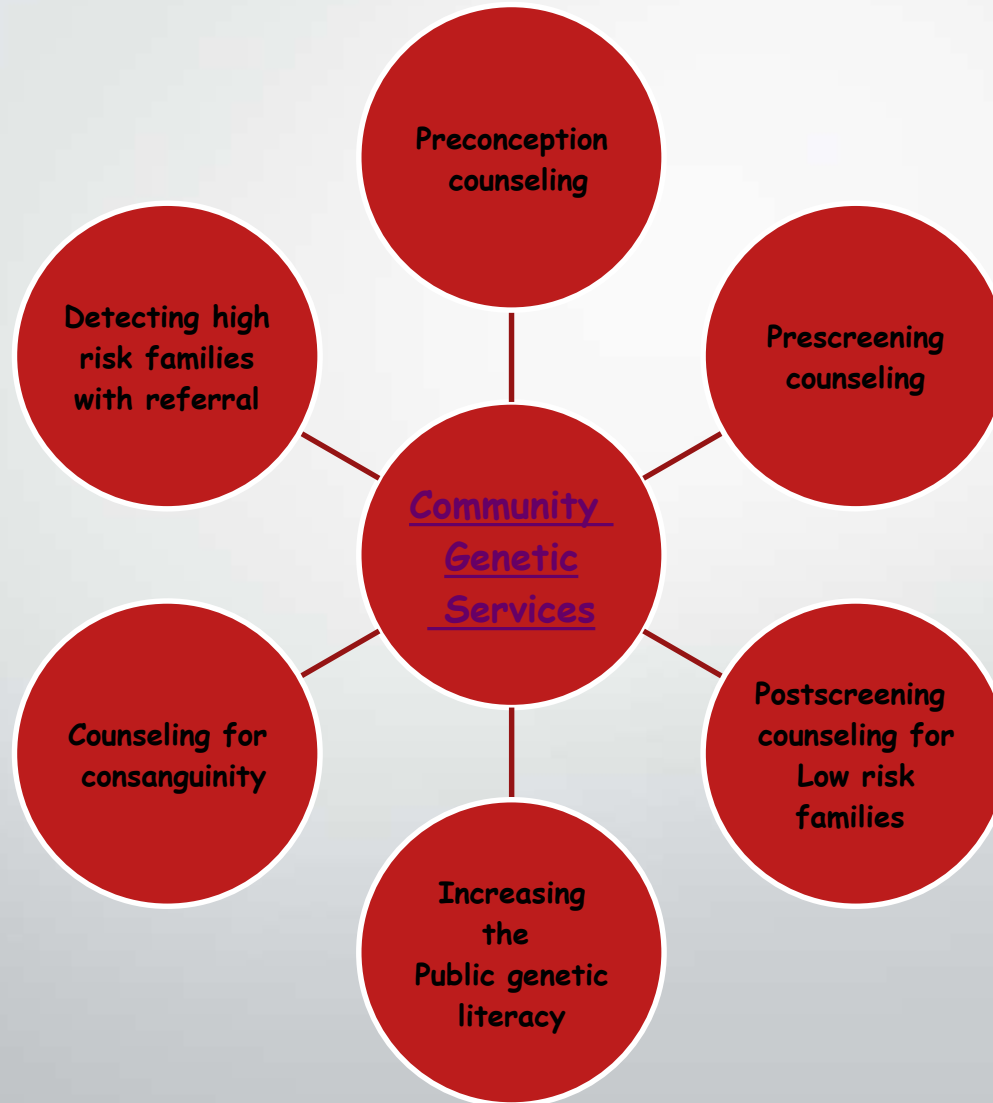
- Premarital and preconception carrier screening
- Prenatal screening
- Newborn screening

Community based interventions to reduce burden of congenital disorders IV

**Training
and
education**

- Of all health care providers
- Of the public
- Of medical and nursing students

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

Principles of Genetic Counseling

- Basic principles, aims, and ethical issues in genetic counseling
- Ethical issues in medical genetics and genetic services
- Consanguinity and counseling

Prevention strategies

- Community genetic services
- Preconception care and counseling
- Prenatal screening and diagnosis
- Prevention of congenital disorders
- Epidemiology, care and prevention of hemoglobinopathies
- Research priorities and writing for publication

Research and publication

- Research priorities in integrating community genetic services in existing health care systems
- Scientific Research writing and publication

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

- Understand and clearly explain the principles of inheritance of diseases.
- 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.
- Offer prescreening counseling, and know how to detect at risk couples or individuals for referral to special centers.
- Offer counseling related to consanguinity in communities with high consanguinity rates.

Upon completion of this module, participants will be able to

- Know the basic ethical principles and techniques of genetic counseling
- Recognize the feasibility and importance of providing preconception care to every woman of childbearing age at any health care encounter.
- Understand and implement the core interventions of preconception care.
- Understand the basis of common congenital disorders in the community and acquire information on available means for their care and prevention
- Access appropriate information materials for patients, families and the general public

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.