Community Genetics

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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***

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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.

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Strategies for the introduction of Community Genetic Services

Why do Genetic services have a low priority as health programs in most developing countries

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- other competing priorities, chronic diseases (diabetes, CVD, cancer)

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- Inadequacy of data on magnitude and burden of genetic disorders
- Insufficient numbers of trained health professionals in the area of medical genetics
- other competing priorities
- Cultural, social and religious limitations

What is meant by Community Genetic Services

- It means genetic services targeted to the population with the goal of **reducing the burden** imposed by congenital disorders on individuals in the population

- Involve programs for the **prevention and care** of congenital disorders specifically at the **primary health care** level.

- Provide services with equity and efficiency to all those in need

What is meant by specialized genetic services

- Means genetic services targeted to individuals and families who request them with the goal of enabling people with a genetic disadvantage to live and reproduce as normally as possible and to make informed and voluntary choices on reproductive matters
- Provided by specialists in secondary and tertiary centers

Goals of community genetic Services

Care and Prevention of Congenital Disorders

Primary
Preconception counseling
Premarital screening

Secondary
Prenatal screening and testing

Tertiary
Newborn screening
Management of affected children

Prerequisites for the organization of community genetic services programs

- Political commitment
- Collection of epidemiological data
- Situation analysis of existing health care programs
- Financial resources
- Formation of a national committee

Is this the right time to introduce Community Genetic services in developing countries?

- Declining infant mortality from infectious diseases

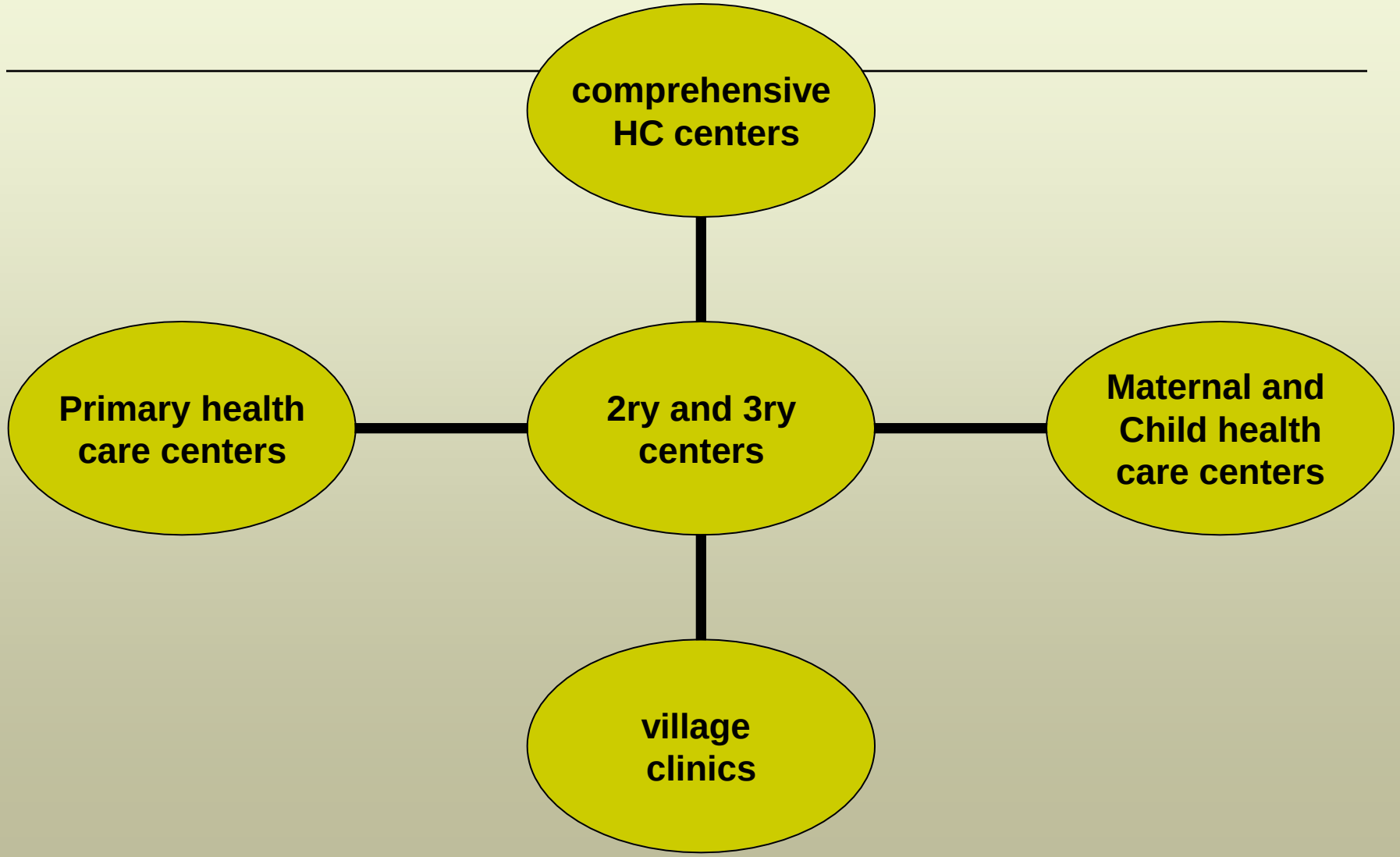
Relation of infant mortality to impact of birth defects

- The lower the infant mortality rate, the higher the contribution of congenital disorders to infant mortality
- Around 20/1000 of all livebirths have a major birth defect, of whom around 40% (8/1000) die in infancy, and 40% live with a severe disability.
- When infant mortality rate is around 20/1000, about **40% of all infant deaths are related to congenital disorders**

Is this the right time to introduce Community Genetic services in developing countries?

- Declining infant mortality from infectious diseases
- Good coverage of reproductive primary health care programs

The health care network in developing countries



Is this the right time to introduce Community Genetic services in developing countries?

- Declining infant mortality from infectious diseases
- Good coverage of reproductive primary health care programs
- Care and Prevention is feasible and cost-effective

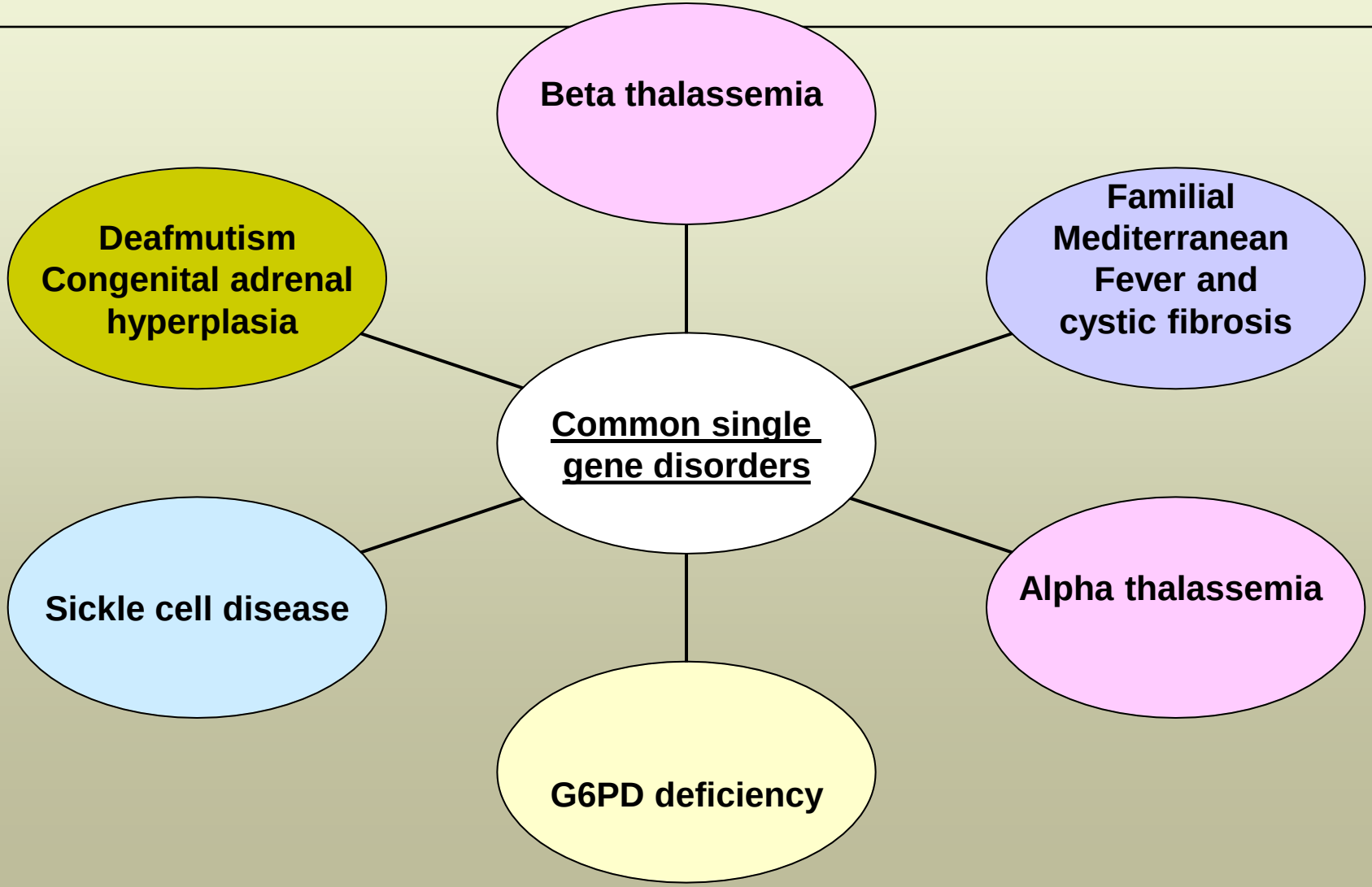
Examples of congenital disorders with feasible community services care and prevention

- Congenital malformations such as neural tube defects can be reduced by periconceptional folic acid supplementation
- Early management of congenital hypothyroidism detected by newborn screening can prevent mental retardation
- Early management of phenylketonuria detected by newborn screening can prevent mental retardation

Common single gene disorders that can be targeted by genetic services

- Hemoglobinopathies (thalassemias and sickle cell anemia): feasible care and prevention community programs are documented
- Early detection of cystic fibrosis, congenital adrenal hyperplasia, hereditary deafmutism and G6PD deficiency helps in proper management

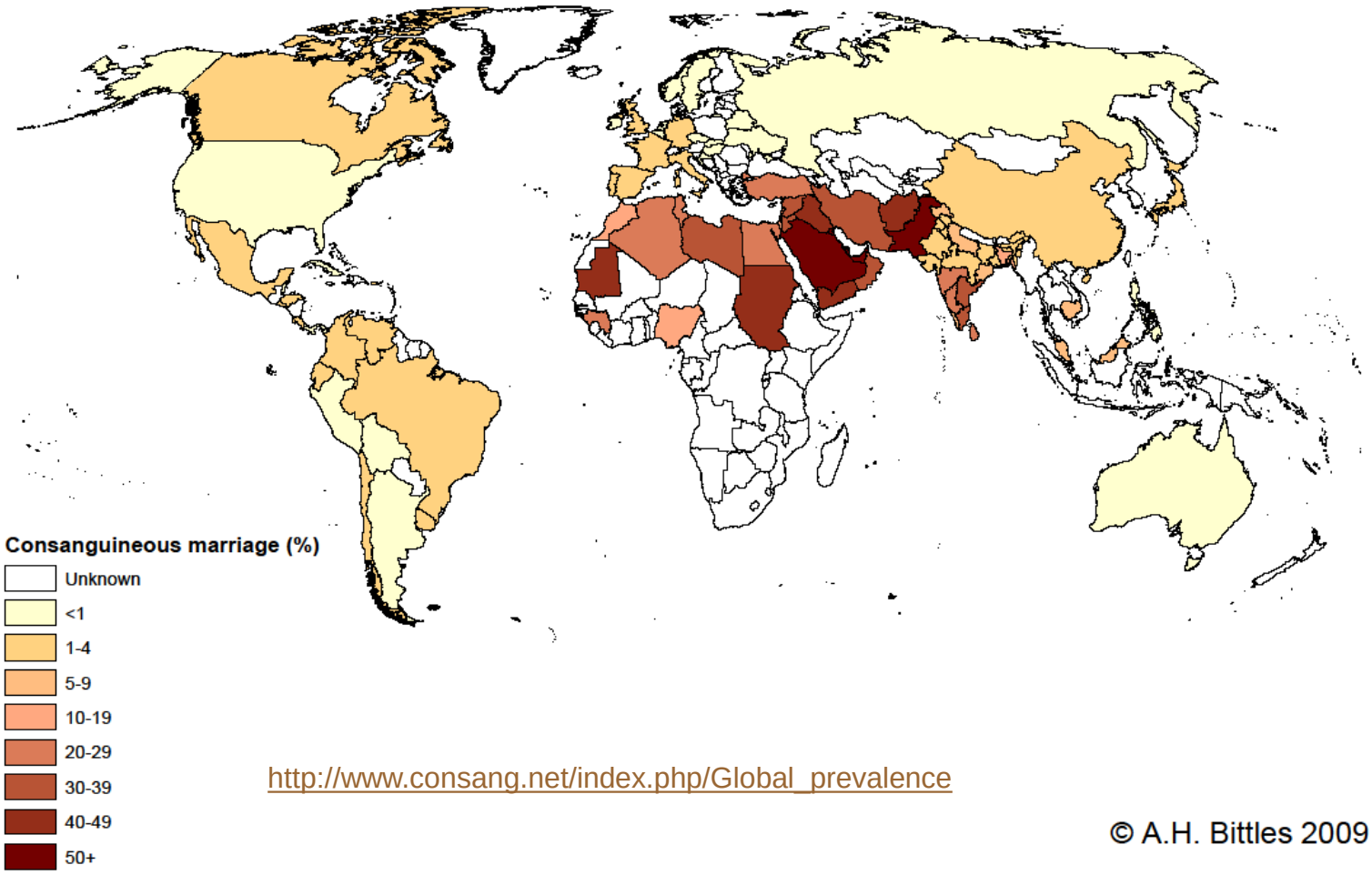
Examples of relatively common single gene disoders



Is this the right time to introduce Community Genetic services in developing countries?

- Declining infant mortality from infectious diseases
- Good coverage of reproductive primary health care programs
- Care and Prevention is feasible and cost-effective
- Proper counseling for Consanguinity

Global prevalence of Consanguinity



Reproductive health among consanguineous and non-consanguineous couples

- Earlier parental age at marriage
- Younger maternal age at first livebirth
- Higher number of infants born to consanguineous parents
- Same or lower rates of abortion
- Higher rates of postnatal mortality in offspring
- Higher rates of congenital malformations in offspring
  Generally speaking, frequency of congenital malformations among newborns of first cousin unions is double the frequency among the general population (around 5%).
- Higher risk of having offspring with autosomal disorder if the gene segregates in the family

Premarital advice regarding consanguineous marriages

- If there is a suspected autosomal recessive disorder or other genetic disorder, refer family for professional genetic counseling
- If there is no known inherited disorder in the family, then first cousin marriages are given a risk for birth defects of about double the population risk. If general population rate for birth defects is 2.5%, it is estimated to be around 5% in first cousin couples.

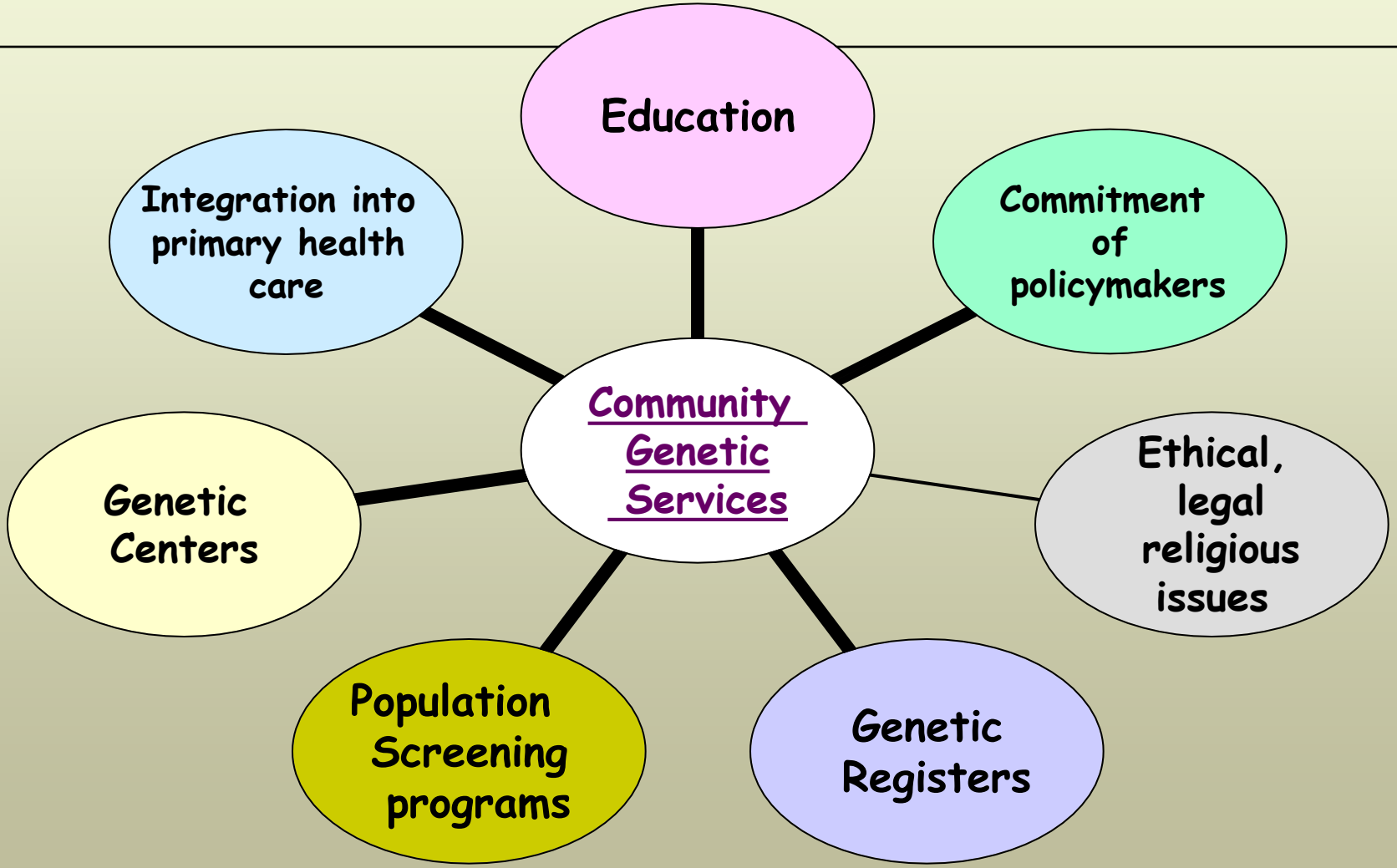
Is this the right time to introduce Community Genetic services in developing countries?

- Declining infant mortality from infectious diseases
- Good coverage of reproductive primary health care programs
- Care and Prevention is feasible and cost-effective
- Proper counseling for Consanguinity
- The public and the health professionals are demanding more genetic services

Strategies for the introduction of community genetic services

- Periconceptional counselling to maximize the chance of having a healthy baby
- Premarital screening for common recessive disorders
- Prenatal screening and counselling
- Newborn screening for treatable congenital disorders
- Initiation of registries for congenital disorders

Pillars for community Genetic Services (Prevention and Care)



Education and training in the context of community genetic services

- Medical education
- Training of health care providers
- Population education

Training of the health sector

Health workers need to be educated in basic scientific principles of genetics and in the ethics and practice of genetic counselling

- Delivering periconceptional counselling
- Delivering prescreening counseling
- When and where to refer families to genetic centers
- Implications of consanguinity on health and the society

Education of the public

Education is an integral part of any community health program. Organized information, education and communication need to be addressed to the population in general through different channels that include

- school curricula
- Population education

1- School curricula

school curricula could include :

- Basic principles of Human genetics
- Information on common congenital disorders in the community and on available screening programs

2- Population Education

- Information acquired through trained primary health care workers
- Health pamphlets addressing methods for the care and prevention of congenital disorders
- Messages through the media. These should be scientific and properly formulated ,since inaccurate messages could do more harm than good and would eventually interfere with the successful implementation of the program

Medical and Nursing Education

- Basic clinically oriented course on the principles & practice of Medical genetics
- Integrated education on applied genetics into the clinical specialties
- Training on the incorporation of community genetic services into the primary health care system

Integration of community genetic services into primary health care systems

- Preconception counseling (nutrition, maternal infections, Rh status, parental age, maternal disease, teratogenic drugs and chemicals, Rubella prevention, cessation of smoking, family planning)
- Monitoring of maternal health before and during pregnancy (diabetes, hypertension, epilepsy, detection and treatment of maternal thrombophilia)
- Examination of all neonates
- Caring for affected children

Main Services Offered by a Medical Genetics Centre

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

Examples of population screening programs for the care and prevention of congenital disorders

- Antenatal screening for Down syndrome, neural tube defects and major malformations
- Newborn screening for hypothyroidism, phenylketonuria, G6PD deficiency
- Premarital screening for beta thalassemia

Genetic registries

- What conditions should be initially registered
- Training personnel
- Defining responsibilities
- Recruiting experts for planning and implementation of the project
- Training physicians and nurses on examination of newborns to detect congenital malformations

Ethical, legal and Religious issues

Generally the main ethical standards that need to be addressed involve the following issues:

- Genetic services should reach all those in need
- Education of the public
- Services should not contradict cultural and religious beliefs
- Definition of guidelines for therapeutic abortion
- Definition of guidelines for premarital counseling
- Definition of guidelines for counseling and media messages

Conclusions

- This is the time to introduce community genetic services in developing countries
- Services can be integrated into primary health care programs
- Education of health care providers is a priority
- Guidelines for prenatal screening, testing and selective abortion of an affected fetus are needed
- In some countries premarital screening programs can be introduced
- Introduction of newborn screening
- Initiation of genetic registries