Introducing Community Genetic Services in Developing Countries

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Why is this topic important to discuss now???

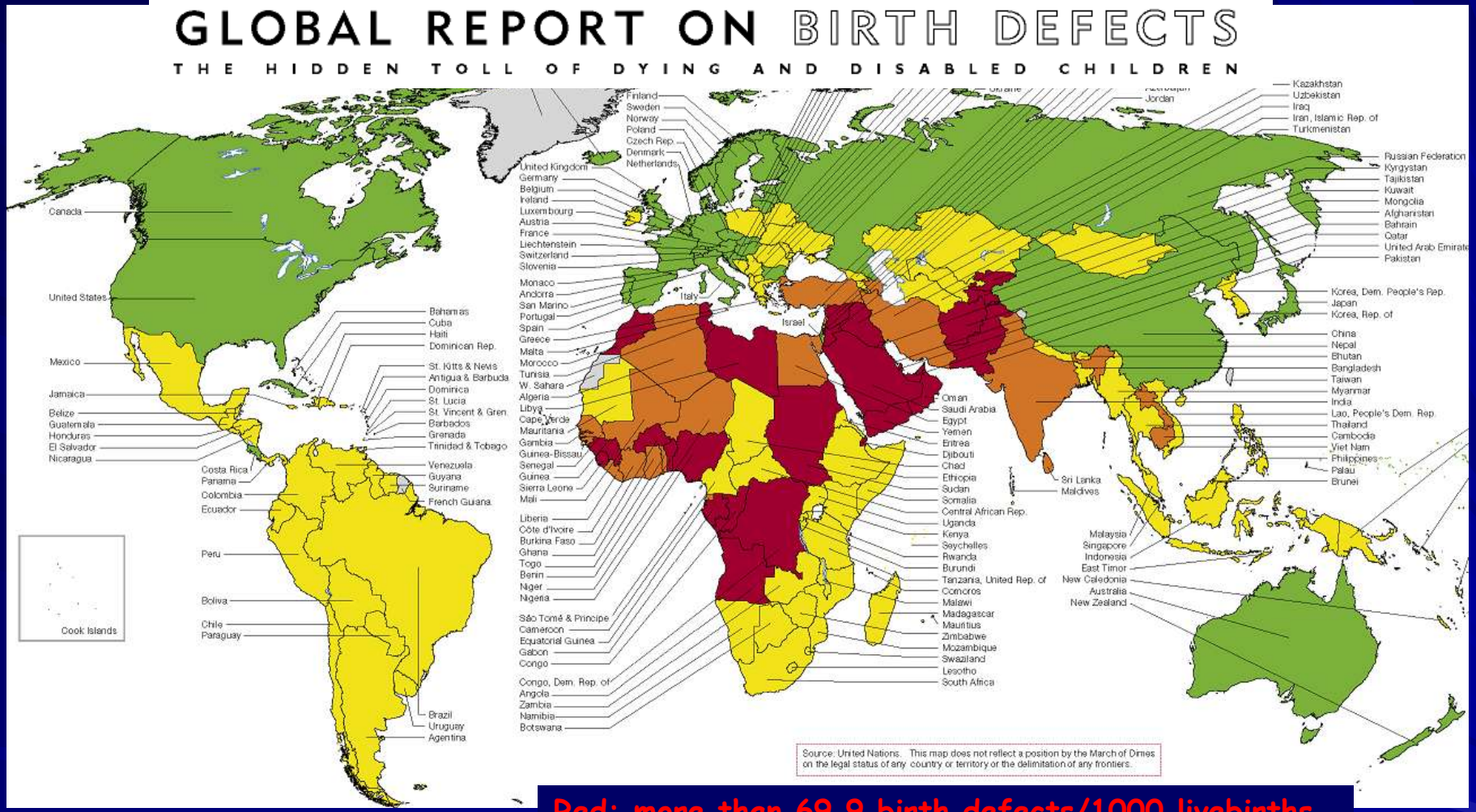
- The rates of genetic and congenital disorders are estimated to be equal or higher than the rates in high income countries
- Ongoing declining morbidity and mortality from infectious diseases, and declining infant mortality rates
- The majority of genetic and congenital disorders can be prevented
- All health care providers have a responsibility in reducing the burden of these disorders on a community and on a family basis

Global rates of birth defects

MARCH OF DIMES

GLOBAL REPORT ON BIRTH DEFECTS

THE HIDDEN TOLL OF DYING AND DISABLED CHILDREN



Christianson et al,2006

Red: more than 69.9 birth defects/1000 livebirths

Orange 61-69.9

Yellow 52.1-60.9

Green less than 52.1

Grey no data

Global rates of birth defects

- Every year around 8 million infants are born with a serious birth defect of genetic or partially genetic origin.
- of which 3.3 million die, 3.2 survive with severe disability

- several hundred thousands are caused by teratogens (alcohol, Rubella, syphilis, iodine deficiency).

- According to experience from industrialized countries, up to 70% of birth defects can be prevented or adequately managed

MOD report: Birth defects defined as structural or functional abnormalities

Impact of genetic diseases

- Of all neonates, about 2-3% have at least one major congenital abnormality, at least 50% of which are caused exclusively or partially by genetic factors.
- Chromosome abnormalities occur in about 0.5% of neonates
- Single-gene disorders occur in about 1% of neonates

Causes of congenital abnormalities (birth defects, present at birth)

Purely genetic (15%)

- Chromosomal abnormalities
- Single gene disorders

Partially Genetic (30%)

- Multifactorial etiology

Environmental (teratogens) (5%)

- Drugs and chemicals
- Infections
- Maternal illness
- Physical agents including hyperthermia

Unknown (50%)

The 5 most serious and prevalent birth defects that constitute about 25% of all disorders are:

- Hemoglobin disorders (thalassemia and sickle cell anemia)
- Down syndrome
- Neural tube defects
- Congenital heart defects
- G6PD deficiency

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Comparison of birth defects rates between UK as a high income country and Jordan as a developing country

country	Children born with birth defects annually	2001 annual births	Rate per 1000 livebirths					
			CVS	NTD	Hbpathies	Down syndrome	G6PD deficiency	Total (all)
Jordan	12,388	169,000	7.9	3.3	0.8	2.1	0.6	73.3
UK	28,602	653,000	7.9	1.7	0.3	1.8	0.1	43.8

CVS: Cardiovascular defects
NTD: Neural tube defects
Hbpathies : hemoglobinopathies

Why are the rates of Birth Defects higher in some developing countries?

Higher rates of autosomal recessive disorders

For example, a high frequency of haemoglobinopathies

- Thalassaemia is common. In most of the Mediterranean area, the Middle East and Asia, 2–18% of the population consists of healthy carriers.
- Carrier couples have a 1 in 4 chance in every pregnancy of having an affected child
- Affected birth prevalence ranges from 0.25 to 9 in 1,000

World Health Organization. Guidelines for Control of Haemoglobin Disorders WHO/HDP/HB/GL/94.1 (WHO, Geneva, 1994).

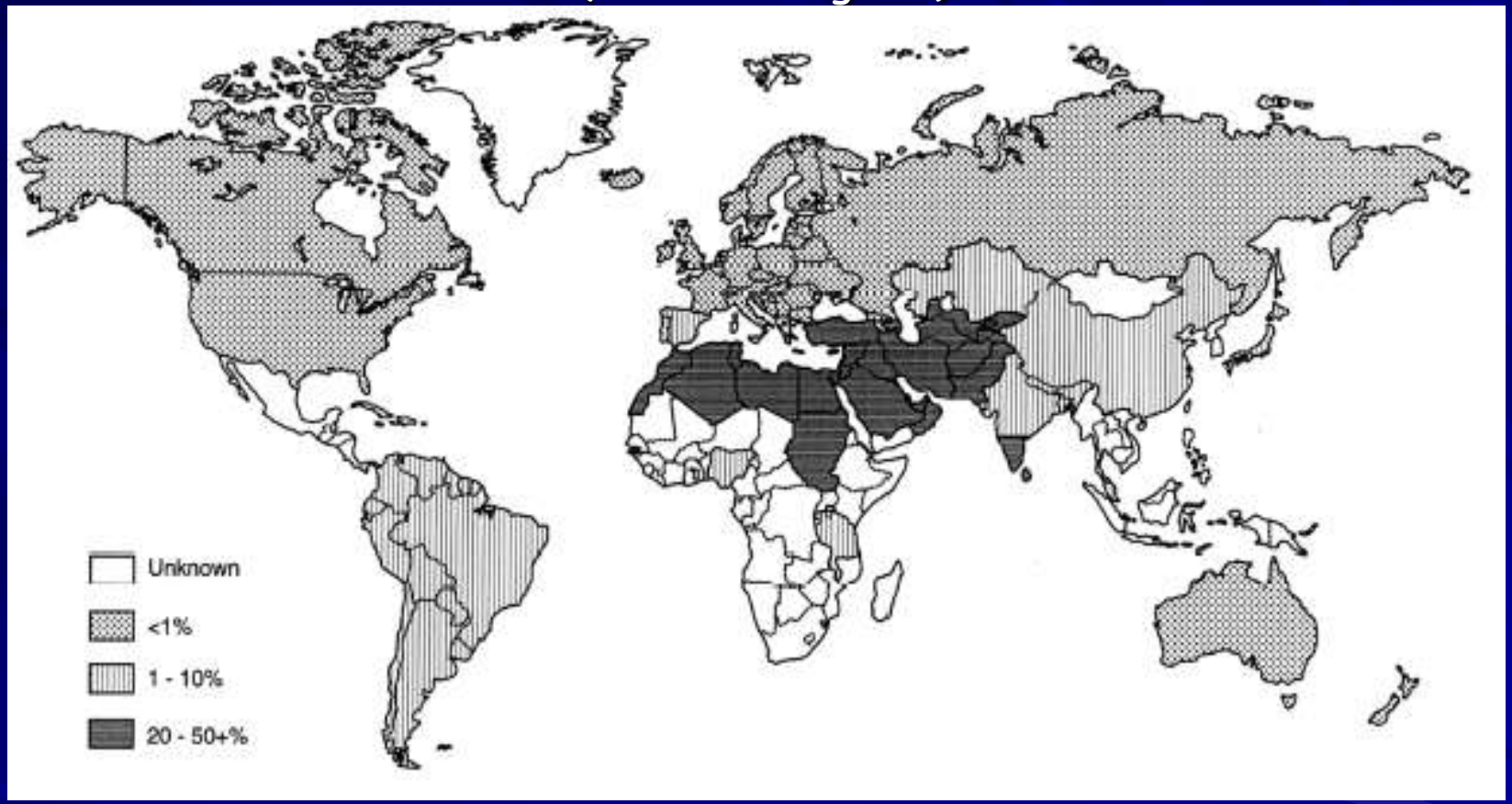
Example of Beta-thalassemia in Jordan

- The carrier prevalence rate of beta thalassemia in Jordan is around 4%. (3-6%)
- The birth incidence for beta thalassemia is about 1 in 2500 livebirths
- The registered number of beta thalassemia patients in the Kingdom is around 1200
- It is estimated that without a control program, 80-90 new cases of beta thalasemia will be born annually

Higher rates of rare autosomal recessive disorders is linked to high consanguinity rates and high fertility rates in some developing countries

Global prevalence of consanguinity

(www.consang.net)



- Less than 1%: North America, Europe, Russia, Australia
- 1-10%: South America, North India,Japan
- **20-50+%: Arab countries, Turkey, Iran, Pakistan,South India.**

Why are rates of chromosomal disorders higher in some developing countries?

- Chromosomal trisomies such as Down syndrome occur more frequently in older than in younger mothers.
- Ratio of pregnant women of advanced maternal age among all pregnant women is higher in many developing countries.
- Prenatal diagnosis for advanced maternal age is not widespread

Why are the rates of congenital malformations higher in some developing countries?

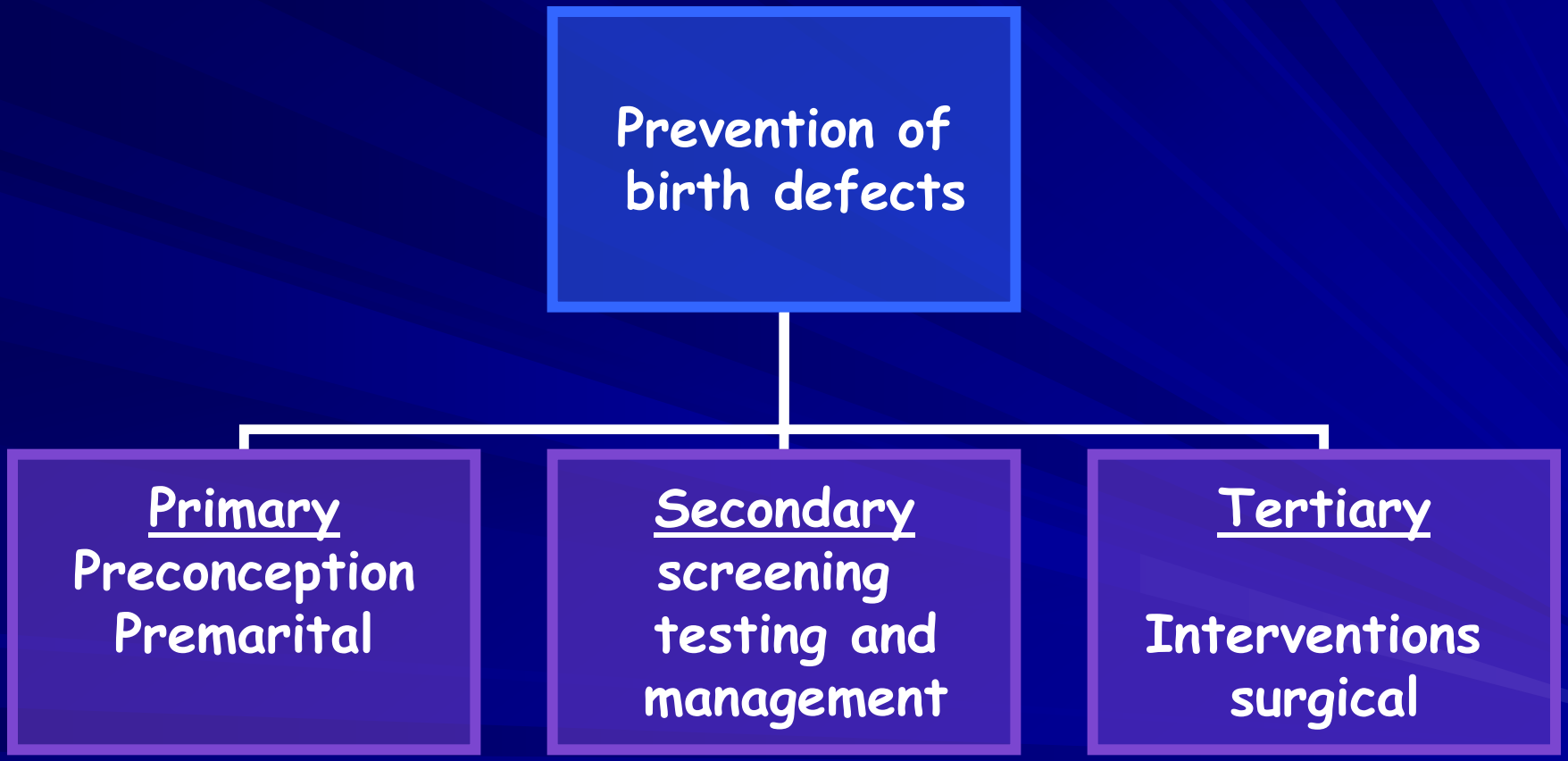
- Low educational levels, low socioeconomic status leading to poor maternal nutrition, more maternal infections, lack of environmental protection
- Main teratogens are: congenital infections, maternal illness(diabetes and hyperthermia) and drugs

Relation of infant mortality to proportion of birth defects

- The lower the infant mortality rate, the higher the contribution of genetic and congenital disorders to infant mortality.

- For example ,with a 22/1000 infant death rate, it is estimated that about **40% of all infant deaths are related to genetic and congenital anomalies.**

About 70% of birth defects are preventable

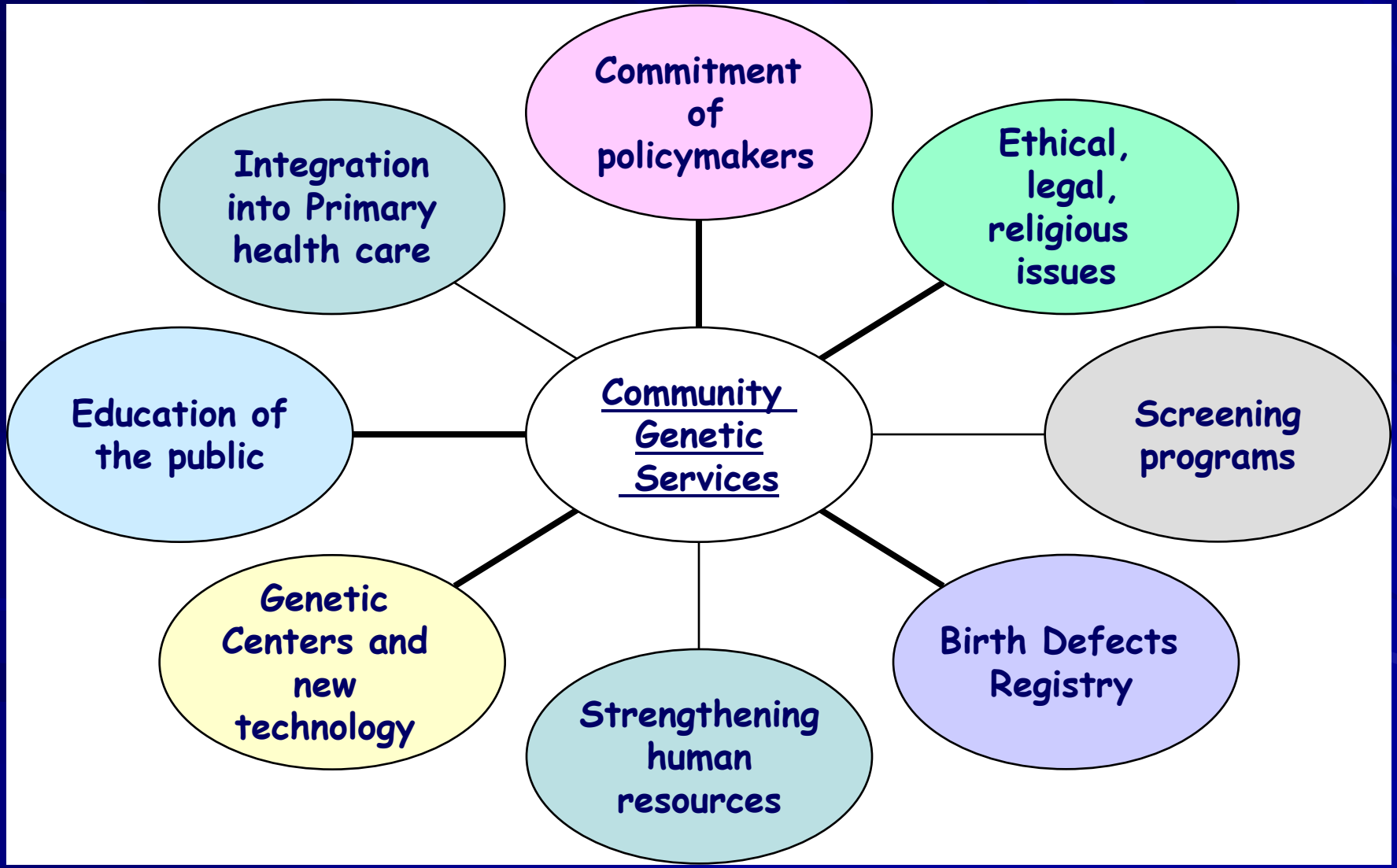


Strategies for the introduction of community genetic services

Basic concepts of the strategy

- Community Genetic Programs should not be vertical programs separate from other health care programs
- Community Genetic programs should be integrated into the already existing national primary health care and reproductive programs

Basic pillars for the introduction of community genetic services



Prerequisites for the organization of community genetic services program

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

Education, training and strengthening of human resources

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

Integration of community genetic services into the primary health care system

- Integration of basic public health approaches into the existing primary care and reproductive health clinics is the most appropriate, sustainable and cost-effective approach.
- Although some additional training and resources will be required, the potential benefits are considerable.

Example of basic public health approaches: Preconception counseling

1. Periconceptional nutritional supplementation
2. Prevention and treatment of maternal infections (Rubella, TORCH, Syphilis)
3. Rh status
4. Advice on maternal age at conception
5. Avoidance of teratogenic drugs and chemicals
6. Cessation of smoking
7. Family planning
8. Monitoring of maternal health before and during pregnancy (diabetes, hypertension, epilepsy).

Screening programs

- Newborn screening for hypothyroidism, phenylketonuria, G6PD deficiency, other metabolic disorders
- Premarital screening for beta thalassemia, other hemoglobinopathies and common autosomal recessive conditions in the population
- Antenatal screening for Down syndrome, neural tube defects and major malformations

Ethical, legal, religious and cultural issues

Some ethical standards that need to be addressed

- Genetic services should reach all those in need
- Services should not contradict cultural and religious beliefs
- Definition of guidelines for prenatal genetic diagnosis
- Definition of guidelines for selective therapeutic abortion of an affected fetus
- Definition of guidelines for premarital screening programs
- Definition of guidelines for newborn screening programs
- Definition of guidelines for premarital counseling
- Definition of guidelines for media messages

National Birth defects Registry

It is important to obtain data on current rates of birth defects and the monitor future rates after implementation of community genetic services

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

Genetics Centers and introduction of new technology

- The establishment of a national centre for the control of genetic and congenital disorders aims at introducing new technology that shapes the diagnostic and therapeutic facilities of present day genetics
- The national genetic center is also responsible for the dissemination of information and for advising on community genetic services.
- Research in the field of genetics in each country is essential to define local issues, to indicate priorities and to solve emerging problems

Conclusions

- Prevention of a large part of birth defects on a community and family basis is feasible
- Community genetic services can feasible be incorporated into the primary health care system of any developing country
- The overall goal of community genetic services would be to achieve a progressive decline in the rates of genetic and congenital disorders for the benefit of families in the community.