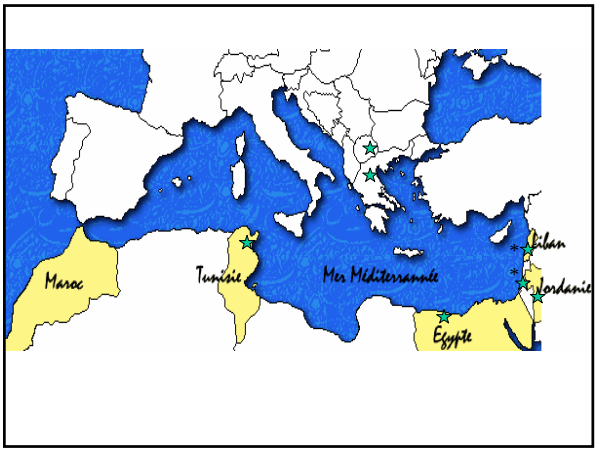
Presentation from TUNISIA

Pr. Habiba Bouhamed Chaabouni.

Department of Congenital and Hereditary Disorders

Laboratory Human Genetics Faculty of Medecine Tunis

Tunisia



TUNISIA * BASIC DATA

- Area 162 155 Km2
- Population 9 947 000 (Est.2004)
- Annual Growth of Population 1.08% (2002)
- Birth Rate 17p.1000 (2001)
- Life Expectancy at Birth 71.1 yr (2001)
- Literacy (10 yr. & up) 68.8% (2003)
- GNP per capita 6 450 PPA US$ (2001)

TUNISIA * BASIC DATA

- Population origin: Arabs, Berbers , African, Turks , Caucasian, Jewish
- Family size 2.8 children
- CONSANGUINITY 32%
- First cousins marriages 23%

Tunisia's Health System Dominated by the Public Sector

- Health services infrastructure : (2003)
  - * 2 beds per 1,000 inhabitants;
  - * 88% of these beds are in public facilities.
  - * PHC / 2028 centers
  - * 1PHC center for every 4 700 inhabitants.
- Facilities for training health professionals in all categories
  - *4 schools of medicine, 1 pharmacy, 1 dentistry,
  - *19 nursing.

DATA FROM MINISTRY OF HEALTH IN TUNISIA, Dr H. Achouri

Tunisia's Health System Dominated by the Public Sector

- Human Resources (2003)
  - * Pysicians in public practice represented 53% of all physicians practicing in Tunisia [7900].
  - * Health Professionals in the public sector > 95% [29 500].
- Financing : 5,6% GDP (2003)
  - * 51% Public Expenditures
  - * 49% Private Expenditures

DATA FROM MINISTRY OF HEALTH IN TUNISIA, Dr H. Achouri

- **There is no national programme of Newborn screening**
- **There is no national programme of genetic diseases prevention**
- **There is national programme of premarital counseling and oriented screening**

THERE IS

- **Good level of medical facilities including genetic facilities.**
- **Diagnosis of common metabolic disorders and family screening starting from propositus.**

- **Hypothyroidism** ~ 40 new cases/year
- **Congenital adrenal hyperplasia** ~ 30 new cases/year
- **Phenylketonuria** ~ 20 new cases/year
- **Hemoglobinopaties**
- **Trisomy 21** ~ 80 new cases/year +30 PND (60% population)

Are the most appropriate conditions to start screening for first

Barriers newborn screening in the country.

- **Financial barriers**
- **Logistic & organisationnal barriers**
  - **Infrastructure , technical platform**
  - **Post-partum period for mothers in public and private hospitals does not exceed 24 hours**
  - **Connection system from hospital to labs.**
  - **Treatment after establishing diagnosis**

Which would be of the greatest assistance to the development or expansion of a newborn screening program in the country?

Logistic improvement

- **Training for health professionals**
- **Organisation of laboratories**
- **Networking**

GENETIC CONDITIONS IN THE COUNTRY.

- **Chromosomal abnormalities**
- **Mental retardation**
- **Hemoglobinopathies : thalassemia, sickle cell disease**
- **Metabolic disorders**
- **Congenital anomalies**
- **Deafness**
- **Spinal muscular amyotrophy & neuromuscular disorders**
- **Cataracts/glaucoma**
- **Genodermatosis**

Once newborn infants and children are identified with a genetic condition?

- Genetic investigation
- Registered in handicap database (children)
- Care management / pediatrician and other specialist
- Therapy possible : investigated and treated
- If absence of treatment , very poor care

What resources are available for these families once these newborns are identified?

- Medical Ressources Genetic counselling,
- Financial Ressources depends upon the medical insurance
- Social Ressources Patients and or Medical association

What about Registries and Databases

- No birth defects registry
- Database : medical departments, neonatal, genetic, pediatric

Research relating to newborn screening

Pilot studies : hypothyroidism, PKU

Newborn screening after propositus diagnosis

CONCLUSION

- High Consanguinity rate,
  High Incidence of AR disorders

Need of genetic diseases prevention programme

Neonatal screening

Prenatal screening