Epidemiology of Chronic Diseases in Adolescence in Mediterranean Populations

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Burden of Chronic Diseases In USA , estimated by a number of key measures

- Chronic Diseases account for the majority, of mortality, morbidity and disability
- More than 15% of adult population suffer chronic disabling conditions (arthritis, heart diseases, hypertension, diabetes)
- Direct health care costs account for 75% of total expenditure for health care
- Behaviour that lead to many chronic diseases (smoking, alcohol, unhealthy diet) account for approximately 800.000 deaths)

Clinical Epidemiology Objectives

- **Occurrence of Disease**
- **Population Characteristics**
- **Disease Clinical Manifestation**
- **Modes of Prevention and Management**
- **Characteristics of organism and mode of transmission (Infectious Disease)**

Main Implementation of Epidemiological Data

- **Burden of Disease in the population**
- **Formulation of prevention and management**
- **Evaluation of effectiveness of prevention and management programs**

Type of Clinical Epidemiology

I. Descriptive

II. Causative or Analytic

- Cross Sectional
- Prospective
- Retrospective

III. Experimental

Indices of Quantification of a Disease

- **Incidence Rate:**

The number of times the disease is noted in a population during a period of time

- **Prevalence of Disease:**

Proportion of population affected by a disease in a defined period

Causes of Death and Age in USA in 1993

(Nelson's Textbook Pediatrics 1996)

Rank	Causes	Rate per 100.000
	1-4 yrs All causes	44
1	Injuries	
2	Congenital abnormalities	
3	Malignant Neoplasms	
4	Homicide Legal Internention	
	5-9 yrs All causes	23
1	Injuries	
2	Malignant Neoplasma	
3	Congenital Anomalies	
4	Homicide	
	10-14 yrs All causes	23
	Injuries	
	Malignant Neoplasma	
	Suicide – Homocide	
	Congenital Anomalies	
	15-24 yrs All causes	98
1	Injuries	
2	Homicide	
3	Suicide	
4	Malignant Neoplasm	
5	Aqcuired Immunodeficiency Syndrome (AIDS)	

Factors influencing diversity of prevalence of chronic diseases (CD)

- Lack of definition of chronic disease
- Lack of definition of age period for adolescence
- Enormous number of chronic diseases (mainly rare)

Definition of chronic illnesses

"…Physical illnesses … that determine the planning actions and feelings of a child and his family in a more or less threatening way over the space of several years or a lifetime"

Petermann, Noekker and bode (1987)

Common Chronic Diseases in Childhood and Adolescence

I)Common in both Age groups

1) Genetic Disorders
2) Chronic Infectious Diseases
(Tuberculosis – Chronic Hepatitis B,C e.t.c.)
3) Respiratory Diseases (Asthma)
4) Neurological Disorders (Seizures)
5) Endocrinological (Obesity – Diabetes)
6) Gastrointestinal Diseases (Chron, Coeliac)
7) Malignancies

II)Predominantly in Adolescence

1) Accidents Sequelae
2) Psychosocial Problems
- Substance Abuse (Drugs – Alcohol – Smoking)
- Anorexia Nervosa
- Depression

3) Sexually Transmitted Diseases (AIDS, HBV)

Groups of Genetic Diseases

- Single Gene Disorders
- Chromosomal Abnormalities
- Congenital Malformations
- Diseases with Polygenic Inheritance
- Common Diseases with Significant Genetic Component
- Mitochondrial DNA Disorders
- Somatic Cell Mutations

Total Load of Genetic Diseases (Weatherall, 1991, Modified)

Type of Disease	Frequency/ 1000 population
• Single Gene	
Dominant	1.8-9.5
Recessive	2.2-2.5
X-Linked	0.5-2.0
• Chromosome Abnormalities	6.8-7.0
• Congenital Malformations	(19-22)[+]
• Common Disorders with a significant Genetic Component*	(7-10)
Total (approximate)	(37.3-52.8)

+. Figures in bracket, gross approximation
*. Diabetes, schizophrenia, etc.

Prevalence of Common Chromosome Disorders

Condition	Frequency/ 1000 births
I. Numerical	
• **Sex chromosomes**	
45XX	0.1
47XXX	1.0
47XXY	1.3
47XYY	1.0
• **Autosomal**	
Trisomy 21	1.4
Trisomy 18	0.1
Trisomy 13	0.1
Others	0.2
II. Structural	
Balanced Translocation	2.0
Unbalanced	0.5
Total	7.7

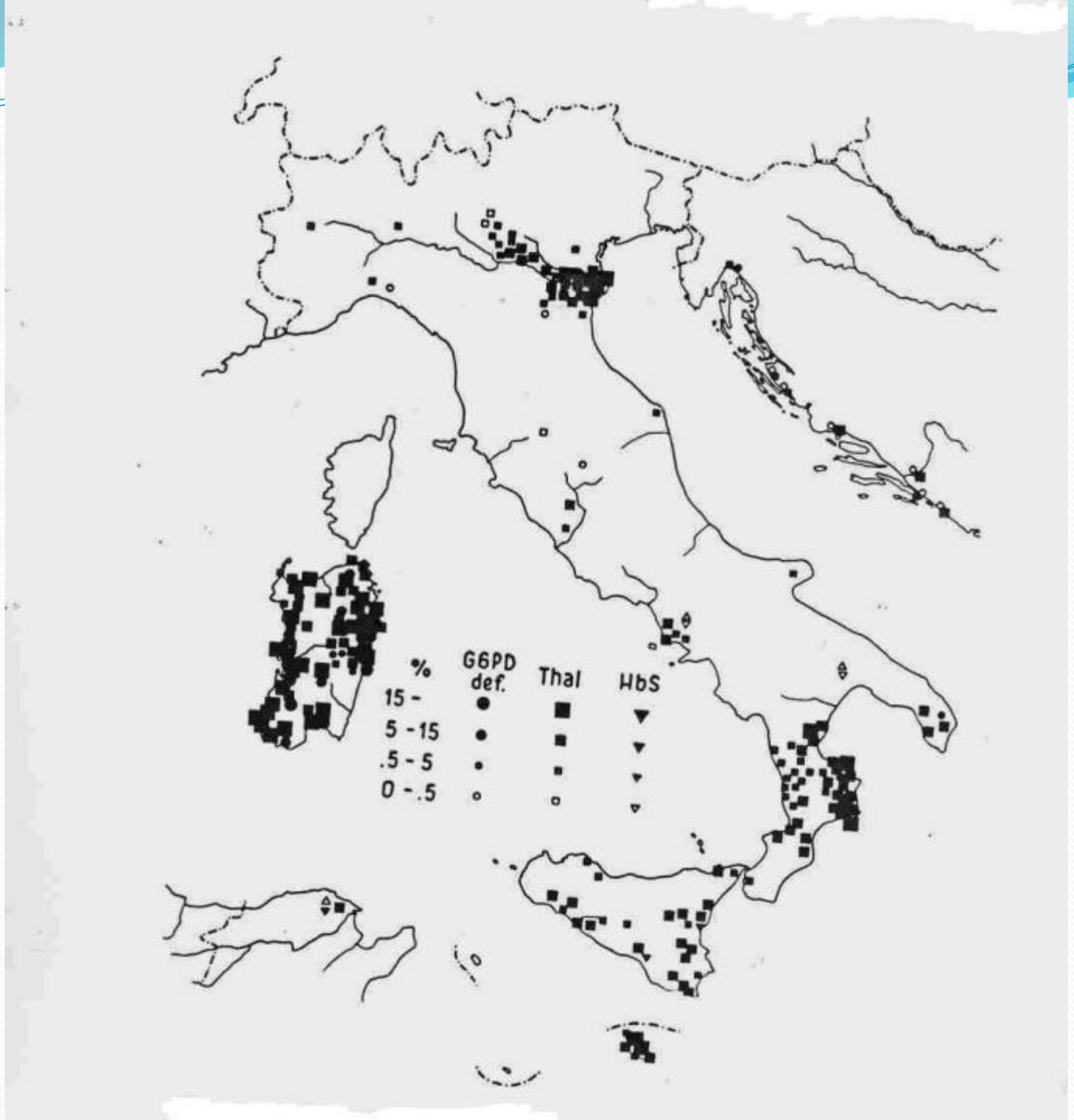
Monogenic Diseases: (McKusick, 1997)

Phenotypes:	
Autosomal Dominant	**3,348**
Autosomal Recessive	**3,928**
X-Linked	**1,371**
Y-Linked	**81**
Total	**9,348**

Remarkable Differences in the Frequency of Genetic Diseases among races.

Disease	Race	Frequency/ 1000 births
• Thalassemia	Mediterranean-Oriental- Northern Europeans	5-15 0.0
• Sickle Cell Anemia	Africans- Northern Europeans	5-15 0.0
• Androgenital Syndrome	Yupik Eskimos North Americans	2.0 0-0.25
• Tay-Sachs	Ashkenazi Jews Sephardi Jews	0.2-0.4 0.001-0.003

Source: Weatherall 1991 (Modified)


%
15 -
5 - 15
.5 - 5
0 - .5
G6PD def.
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HbS

Most common monogenic diseases with chronic clinical manifestations in childhood and adolescence, in Northern Europeans.

Disorder	Frequency/ 1000 births
I. Autosomal Dominant	
• Polycystic Disease of kidney	1.00
• Neurofibromatosis	0.25
• Spherocytosis	0.2
• Early Childhood Deafness	0.1
II. Autosomal Recessive	
• Cystic Fibrosis	0.5
• Phenylketonuria	0.1
• Adrenal Hyperplasia	0.1
• Hemoglobinopathies*	0.1-0.3
III. X-Linked Disorders**	
• Muscular Dystrophy	0.3
• Hemophilia	0.1
• Fragile – X syndrome	0.9 ?

* In immigrants

** per 1000 males

Data on Rare Diseases in EU

- Definition: Based on prevalence;
  U.K. 1 in 50,000; Sweden- Denmark 1 in 10,000
  Other EU member states 1 in 2,000

- Total Number: Estimates 5,000-8,000
- Incidence:
  - ✓ 6-8% of EU population (27-36 million)
  - ✓ At least 80% of genetic origin
  - ✓ 50% affect children and adolescents

European Basic Projects for Rare Diseases

- EUROPLAN; implement national action with EU strategy
- EURORDIS: Build Pan-European community of patients organizations
- ORPHANET: Reference portal of information on RD, orphan drugs and expert services in EU.
- ECORN-CF.eu: Expert advice on cystic fibrosis.
- CARE-NMD: Multidisciplinary care for Muscular Dystrophy.
- RARECARE: Data on European rare cancer.
- EUROCAT: Surveillance of Congenital in Europe.

European activities and projects to improve experts services for RD

I) **National Centres of Expertise**: a) approval of designated regional and national expertise centres. b) Recommendations for national designation of expertise centres .

II) **European Reference Networks (ERNS):**

Organization and function of a number of pilot ERNs, financed by the commission in the context of Community action on RD.

Financed ERNs are: ***Dyscerne**, for dysmorphology ;***ECORN_CF**.eu, cystic fibrosis;***NEUROPED**, rare paediatric neurological diseases; ***NEUROHISTIONET**, histiocytosis; ***TAG**, rare Genodermatoses

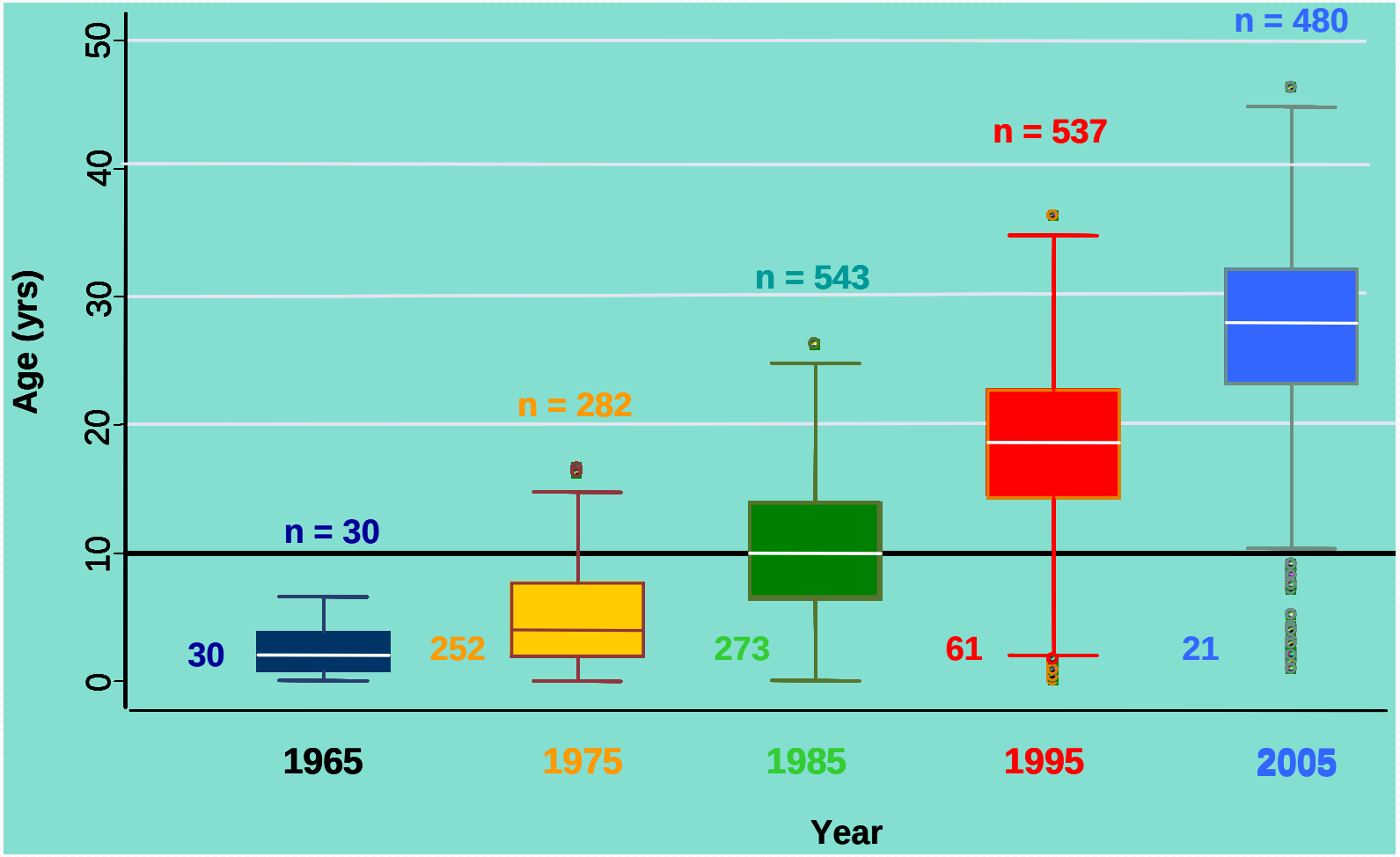
PAAIR**, alpha -1 deficiency ;EPANET** , porphyria ; ***EN-RBD** , bleeding disorders; ***CARE-NMD**, Duchene Muscular Dystrophy;and

***ENERCA**, for rare and congenital anemias.

III) Expert Clinical Laboratories.

Expert clinical laboratories for diagnostic tests (especially for gene identification)

Mean age and annual input of thalassemic patients in the Thalassemia unit of Athens University 1965-2005 (in parenthesis number of patients <10yrs)



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Thanks to my collaborators

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