ERNDIM National Representative Meeting

May 9, 2008

Basel

Organization of BGT testing in Italy

U. Caruso, Genova

National Representative

2008


EUROPE
Barents Sea
Norwegian Sea
White Sea
ICELAND
SWEDEN
FINLAND
Gulf of Bothnia
NORWAY
ESTONIA
Baltic Sea
LATVIA
RUSSIA
NORTH ATLANTIC OCEAN
North Sea
DENMARK
LITHUANIA
IRELAND
UNITED KINGDOM
NETHERLANDS
GERMANY
POLAND
BELARUS
BELGIUM
LUXEMBURG
CZECH REPUBLIC
UKRAINE
SLOVAKIA
FRANCE
AUSTRIA
HUNGARY
MOLDOVA
SWITZERLAND
SLOVENIA
Bay of Biscay
ITALY
CROATIA
ROMANIA
Sea of Azov
MONACO
BOSNIA-HERZEGOVINA
SERBIA
YUGOSLAVIA
Adriatic Sea
BULGARIA
Black Sea
ANDORRA
FYROM
ALBANIA
PORTUGAL
SPAIN
HELLAS (GREECE)
TURKEY
Mediterranean Sea
Ionian Sea
Aegean Sea
MALTA
MOROCCO
ALGERIA
TUNISIA
CYPRUS
SYRIA

2008

ITALY:

20 Regions

2008

Newborn Screening

Newborn screening in Italy is generally organized on regional basis. Some Regions have more than one Centre (Sicily, Puglia…), some Regions refer to a single Centre (north-east).

This is due mainly to historical reasons. NBS born and grew from local Paediatric Departments (first in Genoa in 1973). Central Health and political Authority demonstrated interest for NBS only in the last years.

NBS for PKU and CH is mandatory, recommended for CF.

Some local Centres perform NBS also for CAH, galactosemia, MSUD, biotinidase def., hypermethioninemias, G6PD def.

Newborn Screening 2006

- Live births: ~ 560000
- Coverage:
  - PKU and CH: 100%
  - CF: 77.6%
- Detection rate:
  - PKU and HPAs 1 / 3652
  - CH 1 / 392
  - CF 1 / 5503

Newborn Screening

Newborn screening in Italy is generally organized on regional basis. Some Regions have more than one Centre (Sicily, Puglia…), some Regions refer to a single Centre (north-east).

This is due mainly to historical reasons. NBS born and grow from local Paediatric Departments (first in Genoa in 1973). Central Health and political Authority demonstrated interest for NBS only in the last years.

NBS for PKU and CH is mandatory, recommended for CF.

Some local Centres perform NBS also for CAH, galactosemia, MSUD, biotinidase def., hypermethioninemias, G6PD def.

Expanded NBS is ruled by regional law in Tuscany from December 2004.

Pilot programs are active in Genoa (full Region coverage) and in Rome (partial coverage) from 2005.

In Padua is performed locally from 2001.

Expanded NBS programs

Active

Planned

P= partial coverage

2008

Expanded NBS 2006 (*)

	screened	AA	OA	ß-OX	Total	Rate (1/X)
Florence	33601		2	2	4	8400
Genoa	12299	1		2	3	4100
Rome	5312			2	2	2656
Padua	4094	-	-	-	-	-
TOTAL	55306	1	2	6	9	5690

(*) Data from SISN Report on Newborn Screening Programs in Italy

2008

Questionnaire:

1. General information:
    - Clinical Department in the same Hospital
    - Confirmatory testing for expanded NBS
    - Training
    - Replacement plan
2. Participation do ERNDIM schemes
3. Metabolite analysis
4. Biochemistry (enzyme activities)
5. Genetic assay
6. Prenatal testing
    - Metabolite
    - Enzyme
    - Genetics

Testing for IMD in Italy

29 Laboratories

24 Hospitals

20 Cities

14 / 20 Regions

Submitted data

Presumptive data

2008

OVER 29 LABORATORIES	N	%
Clinical Department in the same Hospital	25	86.2
Confirmatory testing for expanded NBS	15	51.7
Training programs	13	44.8
Replacement plan	7	24.1

Participation in ERNDIM schemes	N	% *	% total
Aminoacids	12	60	41.4
Organic acids qualitative	8	62	27.6
Organic acids quantitative	2		6.9
SA serum	5		17.2
SA urine	6		20.7
Purines pyrimidines	2	50	6.9
Lisosomal disorders	2	20	6.9
Acylcarnitines	2	29	6.9
Proficiency Testing	6		20.7

* Among the labs doing the assay

2008

Metabolite assay - 1	N	%
Aminoacids	20	69.0
Organic acids qualitative	13	44.8
Organic acids quantitative	7	24.1
Ammonia	14	48.3
Orotic acid	8	27.6
creatinine	14	48.3
Total homocysteine	12	41.4
Succinylacetone	7	24.1
Acylcarnitines	7	24.1
Free carnitine	9	31
Acylglycines	3	10.3
Neurotransmitters	3	10.3

Metabolite assay - 2	N	%
Lisosomal metabolites	10	34.5
VLCFA	6	20.7
Phytanic / pristanic	4	13.8
Plasmalogens	2	6.9
Pipecolic	3	10.3
Bile acids	1	3.4
7-dehydrocholesterol	3	10.3
Mevalonic	5	17.2
Creatine – guanidinoacetate	7	24.1
Syalotransferrins	7	24.1
Purines/ pyrimidines	4	13.8
Pterines	2	6.9
Carbohydrates and metabolites	6	20.7

Biochemistry	N	%
Organic acidurias	2	6.9
Aminoacid disorders	-	-
Creatine defects	2	6.9
GDG	1	3.4
ß-oxidation defects	1	3.4
Carbohydrate metabolism defects	3	10.3
Mitochondrial defects	3	10.3
Lisosomal disorders	10	34.5
Peroxsisomal disorders	-	-
Neurotransmitter disorders	-	-

Genetics	N	%
Organic acidurias	2	6.9
Aminoacid disorders	7	24.1
Creatine defects	2	6.9
GDG	-	-
ß-oxidation defects	4	13.8
Carbohydrate metabolism defects	3	10.3
Mitochondrial defects	4	13.8
Lisosomal disorders	5	17.2
Peroxsisomal disorders	-	-
Neurotransmitter disorders	1	3.4

Prenatal investigations	N
Metabolites	1
Biochemistry	4
Genetics	7

The Scientific Societies

- SISMME – Italian Society for the Study of IMD
- SISN – Italian Society for Neonatal Screening
- SIGU – Italian Society of Human Genetics
- SIBIOC – Italian Society of Clinical Biochemistry

Only the SISMME and the SISN are involved in IMD and NBS respectively.

These two Societies are going to make a unique Society within this year.

The Scientific Societies

- Working groups:
  - Lisosomal disorders - SISMME
  - Mass spectrometry (MSITA) – SISMME/SISN
    www.sismme.it/gismet
  - Creatine (GISMeT-creatina) – SISMME/SISN
    www.sismme.it/gismet
  - Italian Guidelines for Expanded NBS - SISMME/SISN
  (work in progress)

- QA schemes:
  - PKU screening (from 1995) – SISN
    All screening Centres (involve also a few foreign Centres)
  - AA - AC (in 2006) – SISMME/SISN
    All Centres doing the assay
  - PT for expanded NBS (from this year) – SISMME/SISN
    As above (involve also a few foreign Centres)

Prof. Cesare Romano, Pediatrician

passed away on April 23rd, 2008, at the age of 83

2008

•Trained in the renowned school of Pediatrics founded and directed by Giovanni de Toni

•1963: Cesare Romano first describes the syndrome that in part bears his name: the Romano-Ward syndrome.

• Careful and painstaking clinician, he was interested in all facets of pediatrics; nevertheless, he was particularly drawn to two fields of special scientific and socio-medical impact: cystic fibrosis and inborn metabolic diseases (IMD).

• As Director of the Endocrine and Metabolic Disease Center of the University of Genoa Pediatric Clinic, he was a pioneer in IMD and in newborn screening programs in Italy, as well as a fervent advocate of the first Italian regional law (Liguria Region, 1973) on newborn screening.

• 1978: Director of the University Department of Pediatrics and, thereafter, of the Residency Program in Pediatrics.

• Beginning in the 1980s, he regularly conceived, convened and animated yearly national conferences on newborn screening programs in Italy, efforts that lead in 1995 to the establishment of the Italian Society for Newborn Screening (SISN), of which he was a founding member and first President.

•Member of the SSIEM and President in occasion of the 37th Annual Symposium held in Genoa in 1999.

2008

He was enlightening teacher for his students, and an inspiring and charismatic leader for all of his co-workers.

The void that Cesare Romano has left in his pupils, colleagues and peers can only be filled by the memory of his kindness, of his propriety and of his teachings.

The quality of our service today is a testament to his efforts.

2008