SCUOLA INTERNAZIONALE DI SCIENZE PEDIATRICHE

WORKSHOP
"MUSCLE GLYCOGENOSES"

27-28 October 2006
Badia Benedettina della Castagna
Genova-Quarto Italy

DIRECTORS OF THE WORKSHOP

Bruno Claudio, *Genova*
DiMauro Salvatore, *New York, USA*

ORGANIZING COMMITTEE

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INVITED SPEAKERS

Amalfitano Andrea, *East Lansing, USA*
Andreu Antoni L, *Barcelona, Spain*
Angelini, Corrado, *Padova*
Arenas Joaquin, *Madrid, Spain*
Bembi Bruno, *Trieste*
Bruni Stefano, *Modena*
Bruno Claudio, *Genova*
Comi Giacomo P, *Milano*
DiMauro Salvatore, *New York, USA*
DiRocco Maja, *Genova*
Filocamo Mirella, *Genova*
Franceschetti Silvana, *Milano*
Kilimann Manfred, *Uppsala, Sweden*
Martinuzzi Andrea, *Conegliano*
Minassian Berge A, *Toronto, Canada*
Minetti Carlo, *Genova*
Quinlivan Ros, *Oswestry, UK*
Pittis Maria Gabriela, *Trieste*
Raben Nina, *Bethesda, USA*
Reuser Arnold J, *Rotterdam, The Netherland*
Spirito Paolo, *Genova*
Toscano Antonio, *Messina*
Vissing John, *Copenhagen, Denmark*
Vorgerd Matthias, *Bochum, Germany*
Zara Federico, *Genova*

Istituto Giannina Gaslini

Aim of the Workshop:

Biochemical defects in glycogen synthesis, glycogenolysis, and glycolysis have been described since the early 1950s, but although muscle glycogen storage diseases have been studied for decades, new biochemical defects are still being discovered, especially in the glycolytic pathway.
The workshop, addressed to pediatricians, neurologists, 6edical genetics, and researchers in the field of metabolic myopathies, has been organized to provide the latest scientific information on muscle glycogenoses, and it will focus on the most recent advances on genotype-phenotype correlation, diagnostic tests, genetic analysis, and pharmacological and enzyme replacement therapy.
Our goal is to create an appropriate platform to share most recent data in the field, and to promote discussion and scientific interactions.

Directors of the Workshop:

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The Venue:

Pivotal to the success of Gaslini International is the Badia Benedettina complex, comprising the former convent, the historic Villa Quartara, and a large park sloping down toward the sea. This complex (which is still undergoing extensive renovation) currently hosts state-of-the-art training facilities, and is set to ultimately house research laboratories and housing, both for the families of children hospitalized at the Gaslini Institute and physicians and researchers in training at the Institute. Situated less than a kilometer from the Giannina Gaslini Children's Research Hospital, the Badia will form a hub around which a number of strategic initiatives of the will take root and develop.

Preliminary Program

DAY 1 (Friday 27 October 2006 - morning and afternoon)

8.30 Registration of participants
9.00 Opening

➢ **GLYCOGEN AND ITS METABOLISM**

Chairman: C Minetti

9.15 S DiMauro Clinical and pathogenesis of muscle glycogenoses
9.45 J Vissing Diagnostic exercise tests in muscle glycogenoses
Discussion

10.30 *Coffee break*

10.45 Lecture:
S Bruni Pompe disease and other Lisosomal Storage Diseases: state of the art and novel therapeutical perspectives

➢ **GLYCOGENOSIS TYPE II: CLINICAL AND THERAPEUTIC ASPECTS**

Chairman: C Angelini

11.30 **M DiRocco** GSD type II: clinical overview
11.50 **AJ Reuser** Enzyme therapy for Pompe disease: from science to industrial enterprise
12.10 **B Bembi** Enzyme replacement therapy in Glycogenosis type II: the Italian experience
12.30 **N Raben** Search for the therapeutic enzyme: tails of mistargeting and autopaghy in Pompe disease
12.50 **A Amalfitano** Future gene therapy for GSD type II
Discussion

13.30 *Lunch*

➢ **MCARDLE'S DISEASE: GENETIC UPDATE AND TREATMENT**

Chairman: J Arenas

14.30 **AL Andreu** Molecular genetic analysis of McArdle's disease
14.50 **R Quinlivan** Pharmacological and nutritional treatment for McArdle's disease
15.10 **J Vissing** Carbohydrate supplementation in McArdle's disease
15.30 **M Vorgerd** Treatment of glycogenosis type V with creatine and ketogenic diet
15.50 **A Martinuzzi** Pilot trial of ACE inhibitor in McArdle's disease
Discussion

16.30 *Coffee break*

➢ **GENOTYPE-PHENOTYPE CORRELATION**

Chairman: S DiMauro

17.00 **MG Pittis, M Filocamo** Mutation profile of the GAA gene in Italian GSD2 patients
17.20 **GP Comi** Genotype-phenotype correlation in Glycogenosis type III
17.40 **C Bruno** Clinical and genetic heterogeneity of Glycogenosis type IV
18.00 **M Kilimann** Muscle phosphorylase kinase deficiency and pseudo-phosphorylase kinase deficiencies
18.20 **A Toscano** Tarui disease and Distal Glycogenoses: clinical and genetic update
Discussion

DAY 2 (Saturday 28 October 2006 - morning)

- **GLYCOGEN STORAGE DISEASES AND CARDIOMYOPATHY**

Chairman: S DiMauro

9.30	**P Spirito**	Hypertrophic cardiomyopathy and other forms of left ventricular hypertrophy
9.50	**S DiMauro**	Danon disease: clinical, morphological and genetic aspects
10.10	**M Kilimann**	Fatal nonlysosomal cardiac glycogenosis: PRKAG2 gene mutations, genetic heterogeneity, and the issue of secondary glycogen storage diseases
	Discussion	

10.40 *Coffee break*

- **THE POLYGLUCOSAN BODY DISEASES**

Chairman: N Raben

11.00	**S DiMauro, C Bruno**	Adult polyglucosan body disease and branching deficiency
11.20	**S Franceschetti, F Zara**	Clinical and genetic findings in Lafora disease
11.40	**BA Minassian**	Lafora's disease: towards a clinical, pathologic, and molecular synthesis
	Discussion	

12.30 *Concluding remarks*
Final test (for Italian CME credits)

Lunch