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

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

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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, medical 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.

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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  A Reuser  Enzyme therapy for Pompe disease: from science to industrial enterprise
  11.50  B Bembi  Enzyme replacement therapy in Glycogenosis type II: the Italian experience
  12.10  N Raben  Search for the therapeutic enzyme: tails of mistargeting and autophagy 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  A 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 glycogenesis: 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*