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 geneticists, 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 pharmaceutical 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.

Programme

Friday October 27, 2006

8.00 Registration of participants
8.30 Opening

GLYCOGEN AND ITS METABOLISM
Chairman: Carlo Minetti

8.45 Salvatore DiMauro
Clinical and pathogenesis of muscle glycogenoses
9.15 Paul Plotz
Clinical mimicry: dissecting phenocopies
9.45 John Vissing
Diagnostic exercise tests in muscle glycogenoses

Discussion

10.30 Break

Lecture
Chairman: Bruno Bembi

10.45 Stefano Bruni
Lysosomal Storage Diseases: state of the art and novel therapeutic perspectives

GLYCOGENOSIS TYPE II: CLINICAL AND THERAPEUTIC ASPECTS
Chairman: Corrado Angelini

11.30 Maja DiRocco
GSD type II: clinical overview
11.50 Arnold J Reuser
Enzyme therapy for Pompe disease: from science to industrial enterprise
12.10 Bruno Bembi
Enzyme replacement therapy in Glycogenosis type II: the Italian experience
12.30 Nina Raben
Search for the therapeutic enzyme: tails of mistargeting and autophagy in Pompe disease
12.50 Andrea Amalfitano
Future gene therapy for GSD type II

Discussion

13.30 Lunch

MARCHE’S DISEASE: GENETIC UPDATE AND TREATMENT
Chairman: Joaquín Arenas

14.30 Antoni L Andreu
Molecular genetic analysis of McArdle’s disease
14.50 Ros Quinlivan
Pharmacological and nutritional treatment for McArdle’s disease
15.10 John Vissing
Carbohydrate supplementation in McArdle’s disease
15.30 Matthias Vorgerd
Treatment of glycogenosis type V with creatine and ketogenic diet
15.50 Andrea Martinuzzi
Pilot trial of ACE inhibitor in McArdle’s disease

Discussion

16.30 Break

GENOTYPE-PHENOTYPE CORRELATION
Chairman: Salvatore DiMauro

17.00 Maria Gabriela Pittis/Mirella Filocamo
Mutation profile of the GAA gene in Italian GSDII patients

17.20 Giacomo P Comi
Genotype-phenotype correlation in GSD type III
17.40 Claudio Bruno
GSD type IV: clinical and genetic heterogeneity
18.00 Manfred Kilimann
Muscle phosphorylase kinase deficiency and pseudo-phosphorylase kinase deficiencies
18.20 Antonio Toscano
Tarui disease and Distal Glycogenoses: clinical and genetic update

Discussion

Saturday October 28, 2006

GLYCOGEN STORAGE DISEASES AND CARDIOMYOPATHY
Chairman: Carlo Minetti

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

Discussion

10.45 Break

THE POLYGLUCOSAN BODY DISEASES
Chairman: Nina Raben

11.00 Salvatore DiMauro
Adult polyglucosan body disease and branching deficiency
11.20 Berge A Minassian
Lafora disease: are we there yet?
11.40 Silvana Franceschetti/Federico Zara
Genotype-phenotype correlation in Lafora disease

Discussion

12.30 Concluding remarks
Final test (for Italian CME credits)
13.00 Lunch
Informations

Registration
The workshop is open to medical doctors, biologists, neurologists, pediatricians, and medical genetists. 50 applicants will be admitted.

Deadline for registration: September 12, 2006
Fill in the attached form and return it to the organizing secretariat.

Registration fee: € 205,00
The registration includes scientific programme, book of abstracts, coffee breaks and lunches, and attendance certificate.

Official language: English

Venue
Villa Quartara, Badia Benedettina della Castagna
Via Antica Romana della Castagna, Genova-Quarto.

How to reach the venue
Plane: from the International Airport Cristoforo Colombo, take shuttle bus n. 100 (runs every 30’) to Genova Brignole railway station, where you can take bus n. 17 and get off in Corso Europa, in front of Via della Castagna.
Train: get off at Genova Brignole railway station, take bus n. 17 and get off in Corso Europa, in front of Via della Castagna.
Car: motorway exit Genova-Nervi, take Corso Europa toward the City center, turn right in Via Schiaffino, pass the Hotel AC and the ESSO gas station on the right, and after 50 mt. turn right into a narrow road, Via della Castagna, go straight 80 mt., enter the gate on the right leaving the church on the left and follow signs for parking.

Accommodation
Please make your arrangement well in advance.
Special rates have been arranged at the following Hotels, located near the Badia:
Hotel AC**** Genova-Quarto +39-010-3071180
(located at a walking distance from the Badia)
Hotel Iris*** Genova-Quarto +39-010-3773914
Hotel Astor**** Genova-Nervi +39-010-329011

Italian CME credits
Italian CME credits for medical doctors (pediatrics, neurology, clinical pathology) and biologists: 11
Dr. Bruni's lecture is not included in the CME programme.

Directors of the Workshop
Bruno Claudio, Genova
DiMauro Salvatore, New York

Invited Speakers
Amalfitano Andrea, East Lansing, USA
Andreu Antoni L, Barcelona, Spain
Angelini Corrado, Padova
Arenas Joaquín, 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
Pittis Maria Gabriela, Trieste
Plotz Paul, Bethesda, USA
Quinlivan Ros, Oswestry, UK
Raben Nina, Bethesda, USA
Reuser Arnold J, Rotterdam, The Netherlands
Spirito Paolo, Genova
Toscano Antonio, Messina
Visking John, Copenhagen, Denmark
Vorgerd Matthias, Bochum, Germany
Zara Federico, Genova

Scientific Organizing Committee
Bruno Claudio, Minetti Carlo, Zara Federico
U.O. Malattie Muscolari e Neurodegenerative
Istituto Giannina Gaslini - Largo G. Gaslini, 5
16147 Genova - Italy
Tel.: +39-010-5636.603/758; Fax: +39-010-3538265
email: patologiamuscolare@ospedale-gaslini.ge.it

Organizing Secretariat
Maria Caterina Cogorno
Scuola Internazionale di Scienze Pediatriche
Istituto Giannina Gaslini - Largo G. Gaslini, 5
16147 Genova - Italy
Tel.: +39-010-5636.805/554; Fax: +39-010-3776590
e-mail: caterinacogorno@ospedale-gaslini.ge.it
web-site: www.sispge.com

WORKSHOP
“MUSCLE GLYCOGENOSES”

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

Directors:
Claudio Bruno
Salvatore DiMauro
Please fill in and return to the Organizing Secretariat by September 12, 2006
Tel.: 010/5638.805/554, Fax: 010/3776590, e-mail: caterinacogorno@ospedale-gaslini.ge.it

Title of the event: Muscle Glycogenoses
ID of the event: 571
Venue: Villa Quartara, Badia della Castagna, Genova
Date: October 27-28, 2006

Personal data:
NAME ……………………………………. FAMILY NAME…………………………………………………………
Private address: ………………………………………………………………………………………………
ZIP code……………………Town…………………………………Country……………………………………
Tel…………………………………… Mobile………………………… Fax…………/………………
e-mail…………………………………………………………………………………………………………
Institute…………………………………………………………………………………………………………
Work address: …………………………………………………………………………………………………
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For Italian registrants only: For purposes of Continuing Medical Education accreditation, please provide the following information:
Data di nascita: ___________________________________________ Luogo di nascita: ___________________________________________
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Disciplina: ___________________________________________

Invoice/Receipt (Please specify to whom the invoice/receipt of your payment should be made out)
The invoice should be made out to …………………………………………………………………………………
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Registration fee
The registration fee of euros 205,00 must be paid within 5 days after the confirmation of acceptance by the Organizing Secretariat

Payment can be made by:
A) Credit card: American Express ✌ - Eurocard ✌ - Maestro ✌ - Mastercard ✌ - Visa ✌
Card N° ______________________________ expiration date __________/___________
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Amount authorised __________________ Signature __________________ Date __________

B) Bank transfer to: Istituto Giannina Gaslini, Account no. 4632/90
Banca CaRiGe SpA, Agenzia 58, sportello Gaslini. - Cod. ABI 6175 - CAB 1583
Please specify on the transfer “Registration for (the name of the event)” on behalf of (registrant’s name).
Please send (preferably by fax) copy of the transfer once it has been ordered
All related banking costs are at the registrant’s expenses

C) Check payable to Istituto Giannina Gaslini and sent to the Organizing Secretariat with this form duly signed
* The registration fee can be reimbursed only with a 5 days notice before the beginning of the event.

Privacy policy; (Art.13, Italian law 196/2003)
SISP will make use of the data provided exclusively for the purpose of the present event including the Italian Continuing Medical Education programme procedures. They will be forwarded to the Italian CME National Commission, stored in the SISP database and used to keep you informed on relevant SISP forthcoming events and in case of your future attendance at SISP events. Please tick the box if you wish we erase your data. ☐
The Istituto G. Gaslini, L.go G. Gaslini 5, 16147 Genova is the holder of the data.

Date…………………… Signature………………………………………………