Muscle Glycogenosis: from Diagnosis to Treatment

In-person Meeting, Paris, 13-15 April 2023

Overview

Muscle glycogenosis (GSDs) is an important group of potentially treatable inherited muscle disorders affecting children and adults.
Major advances have been achieved in the diagnosis of these disorders over the past years thanks to improvements in biochemical and molecular techniques.
Better understanding of the phenotypes and clinical course of the various glycogenosis has led to improved multidisciplinary approach to care, follow-up and improved outcomes.
Depending on the aetiologies, various therapeutic approaches are available such as specific diets, enzyme replacement therapy, exercise, and novel drugs or gene therapy are in the pipeline.

Target Audience

This course is aimed at paediatricians, neurologists, neuromuscular specialists, internists, cardiologists, geneticists, biochemists, and other clinicians and scientists willing to improve their knowledge in the diagnosis and treatment of muscle glycogenosis + differential diagnosis with other metabolic myopathies.

Scientific Organising Committee

- Prof. Pascal Laforêt, Paris
- Prof. Nadine van der Beek, Rotterdam
- Prof. Ans van der Ploeg, Rotterdam
- Prof. John Vissing, Copenhagen

Learning Objectives

- To describe the pathophysiology and clinical features of the main muscle glycogenosis in children and adults (McArdle disease, Pompe disease, GSD3), but also less frequent glycogenosis (polyglucosans storage myopathies and glycolysis disorders) and their differential diagnosis.
- To describe the main diagnostic tools (exercise tests, muscle biopsy, biochemical and molecular analysis) and to discuss their respective positions in the diagnostic work-up in 2023.
- To provide guidance for diagnosis and management of muscle glycogenosis.
- To describe current treatments and emerging therapies.

Venue: Hotel Crown Plaza Paris République

Fees

The fees of 350€ includes :

- 2 nights bed and breakfast,
- Lunches, dinners and coffee breaks during the course,
- Speakers' presentations to take away after the course.

Fee of 245€, excluding accommodation, for local participants.

Participants are responsible for their own travel arrangements to and from the course.

Registration Process and Deadline

- The registration form should be completed on www.rrd-foundation.org and submitted with your curriculum vitae in English.
- No payment is required at this stage.
- Deadline for registration is the 12th of March 2023.

Covid-19

Depending on the sanitary situation, local regulations will apply. Stay updated on local regulations here.

Selection Criteria and Review Process

Candidates will be selected by the scientific organising committee based on background, experience and the geographical breakdown.

CME Accreditation

An application will be made for European CME accreditation.

Click Here to Register

Contact
ckellquist@rrd-foundation.org

Recordati Rare Diseases Fondation d'entreprise

PROGRAMME

Thursday 13 April

13:45 Start of the course

Clinical recognition and diagnostic clues of muscle glycogenosis.

Introduction to the muscle glycogenosis focus course.
Pascal Laforêt

Main muscle glycogenosis in adults: clinical features
Pascal Laforêt

Main muscle glycogenosis in children: clinical features
Ans van der Ploeg

Muscle pathology of muscle glycogenosis
Teresinha Evangelista

Biochemical analysis in blood, liver and muscle
Ralph Wigley

Genetic diagnosis of muscle glycogenosis
Morten Duno

Role of exercise tests for diagnosis and evaluation of treatments
John Vissing

Cardiac manifestations of muscle glycogenosis
Karim Wahbi

Whole body muscle MRI and DEXA
Robert-Yves Carlier

Collective debate: usefulness of biochemical assays, muscle biopsy, exercise tests…

Dinner at the hotel

Friday 14 April

Clinical features and diagnosis of muscle glycogenosis.

McArdle disease: clinical features
Nikoline Løkken, John Vissing

Other muscle disorders of glycogenolysis mimicking McArdle (Phosphorylase b kinase, PFK, PGK, PGAM, β enolase)
Salman Bhai

Clinical features of Pompe disease in children
Ans van der Ploeg

Clinical features of Pompe disease in adults
Nadine van der Beek, Ans van der Ploeg

Diagnostic clues of Pompe disease
John Vissing

GSD3
Amel Ben Chehida, Pascal Laforêt

Phosphoglucomutase deficiency
Dirk Lefeber

Glycogen synthase deficiency
Anders Oldfors

Polyglucosan storage myopathies: clinical and pathological features
Pascal Laforêt, John Vissing, Anders Oldfors

Participant cases with discussion + rare cases presented by teachers
Workshop 1: paediatric cases; Workshop 2: adult cases

Dinner in Paris

Saturday 15 April

Treatments: drugs, diets, exercise therapy and gene therapy.

Current treatments and trials for McArdle disease
Nicoline Løkken

ERT in classical infantile form
Hannerieke van den Hout, *Ans van der Ploeg*

ERT in Pompe disease: long term effects
Nadine van der Beek

Future treatments and clinical trials for Pompe disease
Nadine van der Beek, Ans van der Ploeg

Role of registries for the long-term follow-up of treatments
Pascal Laforêt

Current dietary approaches for treatment of muscle symptoms. Role of exercise.
Elaine Murphy, John Vissing

12:30 End of the course

Note: Participants are encouraged to prepare case reports of complicated diagnosis or unsolved cases. This is not a requirement of registration. More information will follow closer to the course.