Please send your registration before the 29th of March to Linda Olsson at <u>linda.olsson@bmrn.com</u>

Be sure to mention your name and professional title

Please let us know if you plan to attend the dinner after the symposium and also possible food restrictions

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Invitation **Metabolic Symposium** Reykjavik, 9th April 2019



13:30 – 19:00 Hilton Reykjavik Nordica Suðurlandsbraut 2, Reykjavik

GZDK.GD.19.02.0052 /UNB-H/19/0005 EU/MPS/1215/0028

Program

13:30 - 14:00	Registration and welcome coffee
14:00 - 14:20	Welcome and introduction Dr Leifur Franzson, Landspítali University Hospital
14:20 - 15:00	Neuronal Ceroid Lipofuscinose Dr Brian Nauheimer Andersen, Center for Rare Diseases, Aarhus University Hospital, Skejby
15:00 - 15:40	Gaucher disease Dr Allan Meldgaard Lund, Center for Inherited Metabolic Diseases, Rigshospitalet, Copenhagen
15:40 - 16:20	CLIR- Collaborative Laboratory Integrated Reports Dr Piero Rinaldo, M.D., Ph.D. Mayo Clinic in Rochester, Minnesota
16:20 - 16:35	Break
16:35 - 17:15	Fabry disease Dr Allan Meldgaard Lund, Center for Inherited Metabolic Diseases, Rigshospitalet, Copenhagen
17:15 - 17:55	Hypophosphatasia – A metabolic bone disease Dr. Raja Padidela, Dept. of Paediatric Endocrinology Royal Manchester Children's Hospital, Manchester
17:55 - 18:35	Hyperammonemia Dr Allan Meldgaard Lund, Center for Inherited Metabolic Diseases, Rigshospitalet, Copenhagen
18:35 – 19:00	General discussion and closure Dr Leifur Franzson, Landspítali University Hospital
19:00	Dinner

Metabolic Symposium

The incidence of each inherited metabolic disease is low, but since there are many different diseases the total number of patients is relatively large.

The diseases can appear at any age and with symptoms from different organs. Many are treatable with a good prognosis if the diagnosis is made in time.

With this symposium we aim to increase the awareness regarding how and when to suspect that a patient suffers from a treatable metabolic disease. The aim is also to guide clinicians regarding the diagnostic and laboratory work-up when investigating a patient for metabolic diseases.

This symposium is organized in collaboration with the Department of Genetics and Molecular Medicine, Landspitali, Reykjavik, Iceland and is supported by unrestricted educational grants from Alexion, BioMarin, Sanofi Genzyme and Orphan Europe.



