

Albinusdreef 2
P.O. Box 9600
2300 RC Leiden



Invitation

Program

- 9:30 – 10:00 **Registration, coffee/tea**
- Chair: Nienke Biermasz**
- 10:00 – 10:15 **European collaboration of expertise centres for rare endocrine conditions**
Alberto Pereira, LUMC
- 10:15 – 10:45 **IGSF1 deficiency syndrome**
Sjoerd Joustra, LUMC Leiden and Reinier de Graaf hospital Delft
- 10:45 – 11:15 **Exploring mechanisms of central hypothyroidism in a new mouse model of Igsf1 deficiency**
Daniel Bernard, McGill University, Montreal, QC, Canada
- 11:15 – 11:45 **Diagnosis and treatment of central hypothyroidism**
Luca Persani, Istituto di Ricovero e Cura a Carettere Scientifico (IRCCS), Istituto Auxologica Italiano, and Università degli Studi di Milano, Milan, Italy
- 11:45 – 12:15 **Genetic approach to growth disorders: the Cincinnati experience**
Christiaan de Bruin, Cincinnati Children's Hospital Medical Center, USA, and LUMC
- 12:15 – 13:00 **Lunch**
- Chair: Wilma Oostdijk**
- 13:00 – 13:30 **Novel genetic causes of short and tall stature**
Jan Maarten Wit, LUMC
- 13:30 – 14:00 **Genetic testing for growth disorders: from candidate gene approach to SNP arrays and gene panel analysis**
Hermine van Duyvenvoorde, LUMC
- 14:00 – 14:20 **Application of whole exome sequencing in endocrine disorders**
Monique Losekoot, LUMC
- 14:20 – 14:40 **Inactivating and activating mutations of NPR2**
Sabine Hannema, LUMC
- 14:40 – 15:00 **Mutations, deletions and duplications of SHOX and its enhancers**
Sarina Kant, LUMC
- 15:00 – 15:30 **Coffee/tea**
- 15:30 **Walk to Academiegebouw, Rapenburg 73, 2311GJ Leiden**
- 16:15 **PhD thesis defence by Sjoerd D. Joustra**



Leiden University
Medical Center

The departments of Paediatrics (Willem-Alexander Children's Hospital), Medicine, Division of Endocrinology, and Clinical Genetics have the pleasure to invite you to participate in the symposium entitled:

Ten years of Leiden Growth Genetics Working Group: clinical implications of genetic discoveries

on the occasion of the 10th anniversary of the Leiden Growth Genetics Working Group and the PhD thesis defence of Sjoerd Joustra (thesis: Pituitary disorders and their extra-pituitary implications). All presentations are in English.

Organizing committee:

Wilma Oostdijk
Nienke Biermasz
Alberto Pereira
Sarina Kant
Jan Maarten Wit

The symposium takes place on Tuesday September 27, 2016, in "Collegezaal 3", Leiden University Medical Center (LUMC), Albinusdreef 2, 2333ZA Leiden

For this symposium accreditation has been requested at the "Nederlandse Vereniging voor Kindergeneeskunde", "Nederlandse Internisten Vereniging" and "Vereniging Klinische Genetica Nederland".

Registration is necessary because of the limited capacity of the room, but is gratuitous. Please register using the following link <https://forms.lumc.nl/lumc2/leiden-growth-genetics> before September 12, 2016.

For inquiries, please contact Inge Durmus: inge@lumc.nl.

