

Second Karolinska Workshop on Skeletal Dysplasias **12-13 March 2015**

The **Workshop** is intended to provide a forum for collaborative clinical education and networking as well as an introduction to novel diagnostic methods and translational research on rare congenital skeletal disorders.

The number of participants is limited to 50.

The official language is English.

Please register before the 14th of January 2015 by email to: Eva.Ekblom@karolinska.se

Registration fee 3000 SEK includes: tuition, lunch, fruit, coffee/tea, and get-together dinner on Thursday.

Thursday 12/3 2015

8.30–9.00 Registration

Moderator/Giedre Grigelioniene

9.00–9.15 Welcome and introduction/G Grigelioniene

9.15–10.00 Clinical examination and documentation of a patient with skeletal dysplasia/A Nordgren

10.00–10.30 Coffee break

10.30–11.15 Radiological approach to fetal skeletal dysplasias/G Nishimura

11.15–12.00 Metaphyseal chondrodysplasias/O Mäkitie

12.00–13.00 Lunch

Moderator/Thomas Hertel

13.00–13.30 Where do the cells come from for mesenchymal bone formation/Igor Adameyko

13.30–14.15 Clinical-radiographic session: Multiple epiphyseal dysplasias/K Rosendahl

14.15–14.45 Coffee break

14.45–15.30 Norwegian Quiz/E Mercolli & C Rustad

15.30–15.50 Phenotypes related to aggrecan mutations/E-L Stattin

15.50–16.30 Swedish Quiz/G Grigelioniene & E Horemuzova

16.30–17.00 Using achondroplasia growth references to illustrate linear growth of other skeletal dysplasias/L Hagenäs

18.30–21.00 Get Together Dinner

Friday 13/3 2015

Moderator/Eva Horemuzova

8.15–8.45 Short overview of different treatments of achondroplasia/L Hagenäs

8.45–9.30 Therapeutic use of natriuretic peptide in achondroplasia /M Irving

9.30–10.00 Coffee break

10.00–10.45 Clinical-radiographic session: chondrodysplasia punctata/G Nishimura

10.45–11.25 Danish Quiz/I Vogel & A Haegerupp

11.25–12.10 Clinical aspects of cleidocranial dysplasia/M Irving

12.10–13.00 Lunch

Moderator/Ann Nordgren

13.00–13.45 Whole exome sequencing in rare skeletal disorders/S Ikegawa

13.45–14.30 Growth plate, articular cartilage and gene expression/O Nilsson

14.30–15.00 Coffee break

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14.45–15.30 Clinical-radiographic session: *COL2A1*-related skeletal
dysplasias/A Handa & G Grigelioniene
15.30–16.15 Unknown case session/coordinator A Handa
16.15–16.30 Concluding remarks/G Grigelioniene

Course venue: Karolinska University Hospital Solna, Eugeniahemmet,
Leksell Lecture Hall

Lecturers: G Nishimura, S Ikegawa, M Irving, A Handa, I Adameyko, O Mäkitie,
K Rosendahl, G Grigelioniene, O Nilsson, L Hagenäs, A Nordgren

Quiz:

- Denmark: I Vogel & A Hagerupp,
- Norway: E Mercoll & C Rustad,
- Sweden: G Grigelioniene & E Horemuzova

Organizing committee: G Grigelioniene, A Nordgren, M Nordenskjöld, O Mäkitie,
L Hagenäs, E Horemuzova

We are grateful to the pharmaceutical companies supporting this workshop:

