

Center for Rare Diseases Karolinska University Hospital

Skeletal dysplasia workshop 13-14 March 2014, Skandia Lecture Hall, Astrid Lindgrens Children Hospital Solna, Karolinska University Hospital, Stockholm

Thursday 13/3			
		Moderator	Outi Mäkitie
08.30-08.50	Coffee		
08.50-09.00		Welcome	Giedre Grigelioniene & Ann Nordgren
9.00-10.00		Introduction to skeletal dysplasias: concept of endochondral and intramembranous ossifications. Pathogenesis, radiology and evolution of common skeletal dysplasias.	Gen Nishimura
10.00-10.05		Short break.	
10.05-11.05		Genomic studies of bone and joint diseases: past, present and future.	Shiro Ikegawa
11.05-12.00		Dysmorphology and genetics hand in hand with radiology, a panel of rare cases	Giedre Grigelioniene & Stefan Geiberger
12.00-13.00	Lunch		
		Moderator	Giedre Grigelioniene
13.00-13.45		Marginal cases in bone dysplasia clinics	Gen Nishimura
13.45-14.15		Spondyloepimetaphyseal dysplasia, a familial case with unknown diagnosis	Karen Rosendahl
14.15-14.45		Presentation of a Japanese case with unknown skeletal dysplasia	Atsuhiko Handa
14.45-15.15	Coffee		
15.15-16.00		Foetal diagnostics with ultrasound and low dosage CT	Peter Conner and Stefan Geiberger
16.00-16.45		Genetic studies on congenital hand malformations	Tobias Laurell & Ann Nordgren
18.00	Dinner		

Friday 14/3			
		Moderator	Ann Nordgren
08.30-09.15		Auxology in achondroplasia/new growth chart	Lars Hagenäs
9.15-10.00		Cartilage hair hypoplasia: from gene discovery to clinical challenges	Outi Mäkitie
10.00-10.30	Coffee		
10.30-11.30		Sclerosing bone dysplasias	Gen Nishimura
11.30-12.00		Two cases of sclerosing bone dysplasia	Giedre Grigelioniene and Outi Mäkitie
12.00-12.45	Lunch		
		Moderator	Lars Hagenäs
12.45-13.30		New genes in osteoporosis	Outi Mäkitie
13.30-14.30		Hypophosphatasia: diagnostics, clinical picture and treatment	Wolfgang Högler
14.30-16.00		Session for unknown cases from the participants	
15.30-16.00	Coffee		

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General information to those who consider participating in the workshop:

The workshop is an advanced course on congenital skeletal diseases and is intended for paediatricians, geneticists, orthopaedic surgeons, endocrinologists and radiologists. The number of participants is limited to 50.

For the unknown diagnosis session, please prepare a short presentation with 5-6 pictures; we will select 3 or 4 cases.

For registration, please send an e-mail to Giedre.Grigelioniene@ki.se, providing your name, work address and your clinical speciality as well information on food allergies/special diets. We also need information on the institutional billing address (fakturaadress) and cost centre (kostnadställe). Please indicate if you would like to present an unsolved case.

The participation fee for the workshop is 2.300 SEK for both days.

The last day to register is the 1st of February 2014.