



Nordic Skeletal Dysplasia Symposium

Copenhagen, March 8. & 9. 2018

Carlsberg Akademi, Copenhagen, Denmark

Program

Thursday March 8th

08.15-08.45 Registration, coffee and tea

08.45-09.00 Welcome Skovby/Hindsø/Hove

Skeletal signs and symptoms associated with metabolic and endocrine disease

09.00-09.30 Lysosomal storage diseases: clinical and skeletal findings, Nishimura G

09.30-10.00 Hypophosphatasia, Bishop N

10.00-10.30 X-linked hypophosphatemia, Beck-Nielsen S

10.30-11.00 Gene panels in identification of skeletal dysplasia, Kangas-Kontio T (sponsor lecture)

11.00-11.30 Break, coffee, tea and fruit

Free lectures, open session

11.30-11.45 Brachyolmia caused by PAPSS2 mutations, Juliusson P

11.45-12.00 Hajdu-Cheney syndrome - phenotypes at different ages, Gregersen P

12.00-12.15 Trial for Hajdu-Cheney syndrome, Irwing M

12.15-12.30 WES in diagnostic setting for skeletal dysplasia, Beleza A

12.30-13.30 Lunch

Jaw and dental aspects of skeletal dysplasia

13.30-14.00 Pycnodysostosis: clinical and skeletal findings, Haagerup A

14.00- 14.30 Symptoms applied to my body, Thilde/Sofie (with pycnodysostosis)

14.30-15.00 Craniofacial, dental and upper airway aspects of some skeletal dysplasias, Kreiborg S

15.00-15.30 Swedish quiz with jaw and dental focus, three-five questions, Horemuzova/Voss

15.30-16.00 Break, coffee, tea and fruit

16.00-17.00 Carlsberg sightseeing tour

17.30- Dinner at Carlsberg Museum, Queens Room





Friday March 9th

Genes and filaminopathies

09.00-09.30 Genomic study of skeletal dysplasias, Ikegawa S

09.30-10.00 Skeletal dysplasia panels and beyond, Grigelioniene G

10.00-10.30 Filaminopathies, Nishimura G

10.30-11.00 Break, coffee, tea and fruit

Recessive OI

11.00-11.30 Recessive OI. Genetic background, signs and symptoms, Wekre L

11.30-12.00 Fracture management and deformity correction, Nygaard T

12.00-12.30 Norwegian quiz with focus on recessive OI, three-five questions, Merckoll E/Rustad C

12.30-13.30 Lunch

Achondroplasia

13.30-14.30 Management and follow-up of achondroplasia, Irwing M

14.30-15.00 Cervical spine compression/instability: diagnoses, signs and symptoms, Borbjerg T

15.00-15.15 Achondroplasia: mechanical axis considerations. Orthosis – true or false, Hindsø K

15.15-15.30 Finnish quiz with focus on achondroplasia three-five questions, Valta H

Conclusions

15.30-16.00 Concluding remarks, evaluation and passing the baton, Skovby/Hindsø/Hove

