



COST Action CA18139

GEMSTONEGenomics of Musculoskeletal
Traits Translational NetworkEUROPEAN COOPERATION
IN SCIENCE & TECHNOLOGY**GEMSTONE Annual Meeting Program****21-23 September 2022, Rotterdam, Netherlands**

21.09.2022 Wednesday MC meeting, WG meetings and Round Table			
Coffee and registration	9.00-09.15		Natural History Museum, Rotterdam
Management Committee meeting	9.15-10.30	Prof F. Rivadeneira	
Coffee break	10.30-10.45		
WG6 Workshop Round Table with patient advocacy organizations	10.45-12.15	Ms. I. Alves	Or Virtually Meeting ID: 892 1481 6236
Meeting WG6	12.15-13.00	Ms. I. Alves Prof E. Duncan	Passcode: 145322
Lunch Break	13.00-14.00		Natural History Museum
Meeting WG2	14.00-15.00	Prof B. Obermayer-Pietsch Ms I. Foessl	Natural History Museum, Rotterdam
Meeting WG3	15.00-16.00	Prof O. Makitie Dr M. Formosa	Or Virtually
Meeting WG4, WG1 and WG5	16.00-17.00	Prof D. Karasik Dr N. Alonso Dr I. Soldatovic Dr K. Curran Dr G. Roshchupkin	Meeting ID: 892 1481 6236 Passcode: 145322
Social networking event: Guided City Tour	17.00-19.00		
22.09.2022 Thursday WG2 Workshop: Phenotyping			
WG2 session 1			
Welcome, registration and information about group work	9.00-9.15		Natural History Museum
Lecture animal muscle phenotyping 1	9.15-9.45	Dr. E. Kague	
Lecture animal muscle phenotyping 2	9.45-10.15	Dr. C. Ackert-Bicknell	
Coffee break	10.15-10.30		Or Virtually
WG2 session 2			
Lecture human muscle phenotyping 1	10.30-11.00	Dr. F. Laskou	Meeting ID: 886 4566 0443
Lecture human muscle phenotyping 2	11.00-11.30	Prof. J. Rittweger	
WG2 session 3			
First round of Workshop in small groups, one person dedicated as the "writing secretary" to note the most important ideas/questions	11.30-12.30		Passcode: 024886



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Lunch break	12.30-13.30		Kunsthal Cafe
WG2 session 3			Natural History Museum Or Virtually Meeting ID: 886 4566 0443 Passcode: 024886
Second round of Workshop	13.30-14.30		
Final round and workup for the upcoming musculoskeletal paper	15.00-15.30		
Coffee break	15.30-15.45		
LEGENDARE project information			
Introduction to the project	15.45-16.00	Prof F. Rivadeneira	
Bone center Rotterdam	16.00-16.15	Prof C. Zillikens	
EOS Edge imaging	16.15-16.30	Ms F. Koromani	
Measuring bone material properties in vivo, the impact micro indentation technique	16.30-16.45	Dr N. Appelman-Dijkstra	
Dinner and networking activity	18.00-20.00		KADE 4
23.09.2022 Friday			
WG3 workshop “Translational Research in Monogenic Bone Disorders: From Bench to Bedside”			
Session 1: Rare Monogenic Bone Disorders – What’s new	9.00-11.15	<i>Chair: Dr J. Dahl</i>	Natural History Museum Or Virtually Meeting ID: 880 5850 2523 Passcode: 606334
Coffee and registration			
Introduction & Welcome to WG3 Workshop	9.00-9.10	Prof O. Mäkitie	
Achondroplasia: Latest updates with a focus on drug discovery	9.10-9.40	Dr M. Irving	
Animal models of Osteopetrosis	9.40-10.10	Dr A. Maurizi	
X-LINKED Hypophosphatemia: From the disease pathogenesis to improved patient care	10.10-10.40	Prof J. Bacchetta	
Q&A	10.40-11.00		
Coffee Break	11.00-11.15		
Session 2: From Rare Bone Disorders to Population Studies	11.15-13.30	<i>Chair: Prof E. Ntzani</i>	
OI Rare Spectrum: From genetic discoveries to novel pathways	11.15-11.45	Prof E. Duncan	
New therapies of genetic bone disorders: CRISPR/Cas9 gene editing and cell therapy in the treatment of osteogenesis imperfecta - where are we now	11.45-12.15	Dr F. van Dijk	
Lunch Break	12.15-13.00		Kunsthal cafe
From Families to population studies: putting together a study collection	13.00-13.20	Dr M. Formosa	Natural History Museum Or
Q&A	13.20-13.30		
Session 3: Case presentations of Monogenic Bone Disorders	13.30-15.15	<i>Chair: Dr C. Medina Gomez</i>	



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Rare variant analysis in bone metabolism in a truncated cohort of post-menopausal women	13.30-13.45	Prof K. Rabionet	Virtually Meeting ID: 880 5850 2523 Passcode: 606334
A homozygous missense variant in SP7/Osterix causes a new form of craniodiaphyseal dysplasia	13.45-14.00	Dr G. Hendrickx	
WNT pathway variants at the frontier between monogenic and polygenic bone disorders	14.00-14.15	Prof J. Riancho	
KIF24 variants as a novel cause of skeletal dysplasia	14.15-14.30	Prof O. Mäkitie	
Q&A	14.30-14.50		
Break	14.50-15.10		
Session 4: Identification of Treatment Options using Cells & Animal Models	15.15-16.30	<i>Chair: Dr D. Bergen</i>	
Tissue engineering systems as translational models in skeletal disorders	15.10-15.30	Dr I. Duran	
The Musculoskeletal knowledge portal	15.30-15.50	Prof D. Kiel	
Using Zebrafish to identify osteoanabolic agents	15.50-16.10	Ms G. McDonald	
Q&A	16.10-16.25		
Wrap up / Conclusions	16.30	Dr M. Formosa	