



COST Action CA18139

GEMSTONE

Genomics of Musculoskeletal
Traits Translational Network



GEMSTONE Annual Meeting Program

21-23 September 2022, Rotterdam, Netherlands

	Time	Speaker/ Chair	Location
21.09.2022 Wednesday			
MC meeting, WG meetings and Round Table			
Coffee and registration	9.00-09.15		Natural History Museum, Rotterdam Or Virtually Meeting ID: 892 1481 6236 Passcode: 145322
MC 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	
Meeting WG6	12.15-13.00	Ms I. Alves Prof E. Duncan	
Lunch Break	13.00-14.00		
Meeting WG2	14.00-15.00	Prof B. Obermayer Ms I. Foessl	
Meeting WG3	15.00-16.00	Prof O. Makitie Dr M. Formosa	
Meeting WG4, WG1 and WG5	16.00-17.00	Prof D. Karasik Dr N. Alonso Dr I. Soldatovic Dr K. Curran Dr G. Roshchupkin	
Social networking event	17.00-19.00		
22.09.2022 Thursday			
WG2 Workshop: Phenotyping			
Coffee and registration	9.00-09.15	Welcome and information about group work, assignment to small groups	Natural History Museum Rotterdam Or Virtually Meeting ID: 886 4566 0443 Passcode: 024886
WG2 session 1	9.15-10.30		
Lecture animal muscle phenotyping 1	9.15-9.45	Erika Kague, UK	
Lecture animal muscle phenotyping 2	9.45-10.15	Cheryl Ackhert- Bicknell, USA	
Coffee break	10.15-10.30		
WG2 session 2	10.30-12.30		
Lecture human muscle phenotyping 1	10.30-11.00	Faidra Laskou, UK	
Lecture human muscle phenotyping 2	11.00-11.30	Jorn Rittweger, Germany	
WG2 session 3	11.30-12.30		
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		



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Lunch break	12.30-13.30		
WG2 session 3	13.30-15.30		
Second round of Workshop			
Coffee break	15.30-15.45		
LEGENDARE project presentation	15.45-18.30		
Introduction to the project	15.45-16.10	Prof F. Rivadeneira	
WP2-EOS	16.10-16.30	Ms F. Koromani	
WP3-OSteoprobe	16.30-16.50	TBA	
Clinical study	16.50-17.10	TBA	
Ethics in ML research	17.10-17.30	TBA	
Dinner and networking activity	18.30 onwards		
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	
From Families to pop	12.15-12.35	Dr M. Formosa	
Q&A	12.35-12.45		
Lunch Break	12.45-13.30		Kunsthall cafe
Session 3: Case presentations of Monogenic Bone Disorders	13.30-15.15	<i>Chair: Dr C. Medina Gomez</i>	Natural History Museum Or Virtually
Rare variant analysis in bone metabolism in a truncated cohort of post-menopausal women	13.30-13.45	Prof K. Rabionet	



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A homozygous missense variant in SP7/Osterix causes a new form of craniodiaphyseal dysplasia	13.45-14.00	Dr G. Hendrickx	Meeting ID: 880 5850 2523 Passcode: 606334
WNT pathway variants at the frontier between monogenic and polygenic bone disorders	14.00-14.15	Prof J. Riancho	
The Role of SEC16B in Osteogenesis Imperfecta	14.15-14.30	Dr A. El Gazzar	
KIF24 variants as a novel cause of skeletal dysplasia	14.30-14.45	Prof O. Mäkitie	
Q&A	14.45-15.00		
Break	15.00-15.15		
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	15.15-15.35	Dr I. Duran	
The Musculoskeletal knowledge portal	15.35-15.55	Prof D. Kiel	
Using Zebrafish to identify osteoanabolic agents	15.55-16.15	Ms G. McDonald	
Q&A	16.15-16.30		
Wrap up / Conclusions	16.30	Dr M. Formosa	

This is the **suggested programme for Sept. 22nd 2022**

- 9:00 Welcome and information about group work, assignment to small groups (by lot? ;))
- 9:15 **Lecture** animal muscle phenotyping 1 (Erika Kague, UK, Fish)
- 9:45 **Lecture** animal muscle phenotyping 2 (Cheryl Ackert-Bicknell; USA, Mice)
- 10:15 Coffee/Tea Break
- 10:30 **Lecture** human muscle phenotyping 1 (Faidra Laskou, UK)
- 11:00 **Lecture** human muscle phenotyping 2 (Jörn Rittweger, Germany)
- 11:30 First round of **Workshop** in small groups, one person dedicated as the “writing secretary” to fix the most important ideas/questions
- 12:30 Lunch
- 13:30 Second round of **Workshop**
- 14:30 Coffee/Tea Break



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15:00 final round and workup for the **upcoming musculoskeletal paper**