GEnomics of MusculoSkeletal traits TranslatiOnal Network (GEMSTONE): Research focused on Real-World needs

COST Action CA18139
GEMSTONE
Genomics of Musculoskeletal Traits Translational Network

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Introduction

- Whole-genome and whole-exome sequencing work have accelerated diagnosis and uncovered functions and mechanisms of several new genes and pathways
- Children and adults with prevalent and rare musculoskeletal conditions demand for multidisciplinary approach such as translational results
- The COST action "GEnomics of MusculoSkeletal traits TranslatiOnal NEtwork" (GEMSTONE) aims to 1) make a functional characterization of discovered genes and pathways; 2) understand the correspondences between molecular and clinical assessments; and 3) implement novel methodological approaches towards translational research

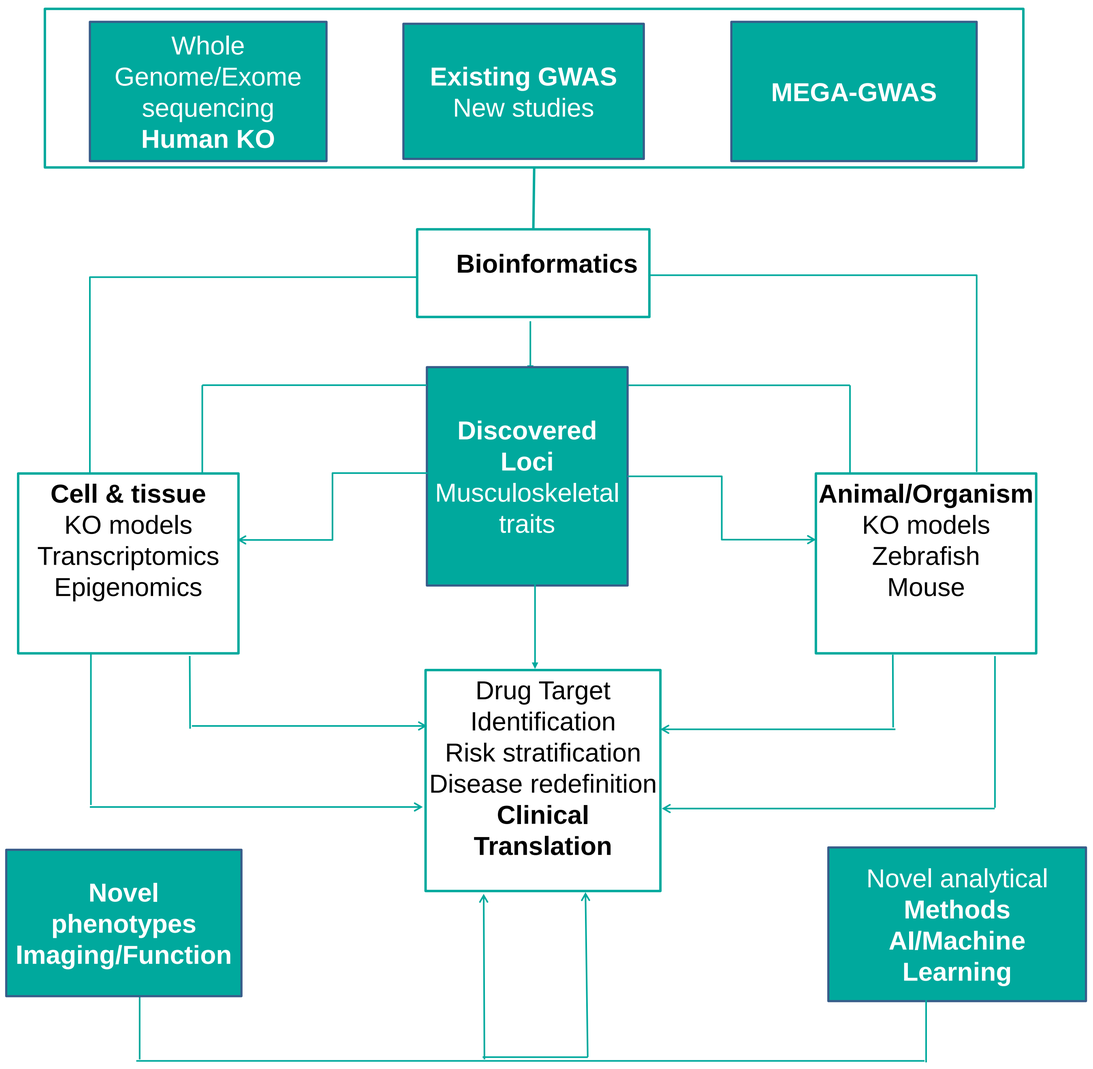

Figure1. The abundance of genetic discoveries needs the creation of a roadmap seeking to organize the functional assessment of genes and biological pathways underlying musculoskeletal metabolism, ultimately highlighting the opportunities to translate these discoveries into clinical applications. This figure shows the rationale behind GEMSTONE scientific objective to translate current and future genetic findings in the clinic (reprinted with permission from Koromani et al. Front. Endocrinol.,2021 |https://doi.org/10.3389/fendo.2021.709815)

Methods

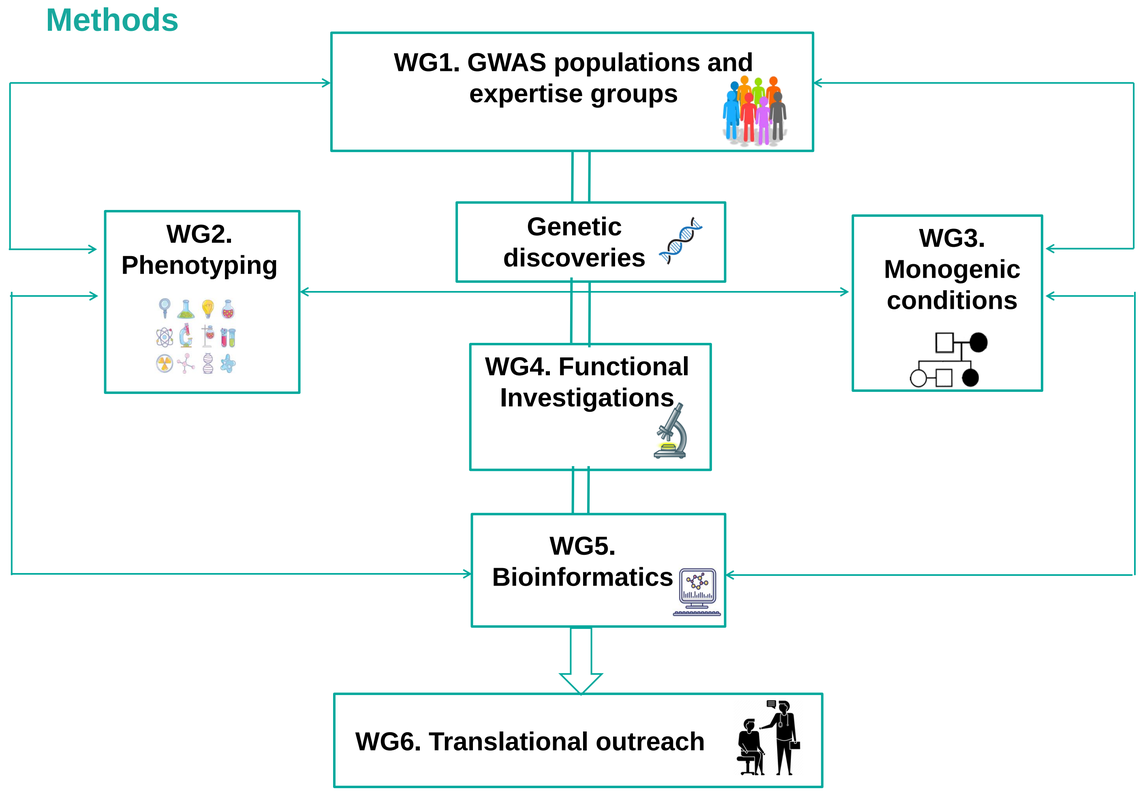

Figure 2 shows the organization of GEMSTONE across six working groups. Collaboration between researchers working in the genomic, fundamental and clinical fields is crucial for successful translational efforts. The figure shows the bridging of the different fields of science and disciplines of their members (genetic epidemiology, molecular biology, bioinformatics and clinical medicine), into one network that will allow advancing the field of musculoskeletal conditions by using such multidisciplinary approach. WG - working group:(reprinted with permission from Koromani et al. Front. Endocrinol.,2021 |https://doi.org/10.3389/fendo.2021.709815)

Networking tools in GEMSTONE

TRAINING SCHOOL
- ◆capacity building
- ◆intensive training on a new topic
- ◆webinar

MEETINGS
- ◆management committee
- ◆working group
- ◆core group

DISSEMINATION
- ◆MC members conference presentation
- ◆website updates
- ◆manuscript publications

ITC CONFERENCE GRANT
- ◆YRI
- ◆participate in a conference to present work related to GEMSTONE

SHORT TERM SCIENTIFIC MISSION (STSM)
- ◆institutional research visits
- ◆YRI
- ◆individual mobility grant

VIRTUAL NETWORKING TOOLS
- ◆Grant for Virtual Networking Support
- ◆Grant(s) for Virtual Mobility

RESULTS & CONCLUSIONS

155 Members

55 founding members
50 new members

Actions
Completed

- 4 papers published
- 6 Short Term Scientific Missions (STSMs)
- 2 Virtual Mobility Projects (VM)
- 4 Training Schools (TS)
- 1 annual meeting

Planned

- 3 Training Schools (TS)
- 5 STSM
- 1 annual meeting

Join GEMSTONE network

- Create your research account in e-cost
- Join GEMSTONE using your research account

Countries	31
ITC countries	40%
YRI MC	15%
Female MC	52%

Table1. Statistics and Representation in GEMSTONE

ITC: Inclusiveness Target Countries; YRI: Young Researcher and Investigator; MC: Management Committee

GEMSTONE website

Publications

Perspective of the GEMSTONE Consortium on Current and Future Approaches to Functional Validation for Skeletal Genetic Disease Using Cellular, Molecular and Animal-Modeling Techniques

Martina Rauner [1 2], Ines Foessl [3], Melissa M Formosa [4 5], Erika Kague [6], Vid Prijatelj [7 8 9], Nerea Alonso Lopez [10], Bodhisattwa Banerjee [11], Dylan Bergen [6 12], Björn Busse [13], Ângelo Calado [14], Eleni Douni [15 16], Yankel Gabet [17], Natalia García Giralt [18], Daniel Grinberg [19], Nika M Lovsin [20], Xavier Nogues Solan [18], Barbara Ostanek [20], Nathan J Pavlos [21], Fernando Rivadeneira [22], Ivan Soldatovic [23], Jeroen van de Peppel [8], Bram van der Eerden [8], Wim van Hul [24], Susanna Balcells [19], Janja Marc [20], Sjur Reppe [25 26 27], Kent Søe [28 29 30], David Karasik [31 32]

Affiliations + expand
PMID: 34938269 PMCID: PMC8686830 DOI: 10.3389/fendo.2021.731217

Genomic Medicine: Lessons Learned From Monogenic and Complex Bone Disorders

Katerina Trajanoska [1], Fernando Rivadeneira [1]

Affiliations + expand
PMID: 33162933 PMCID: PMC7581702 DOI: 10.3389/fendo.2020.556610

A Roadmap to Gene Discoveries and Novel Therapies in Monogenic Low and High Bone Mass Disorders

Melissa M Formosa [1 2], Dylan J M Bergen [3 4], Celia L Gregson [4], Antonio Maurizi [5], Anders Kämpe [6 7], Natalia Garcia-Giralt [8], Wei Zhou [9], Daniel Grinberg [10], Diana Ovejero Crespo [8], M Carola Zillikens [9], Graham R Williams [11], J H Duncan Bassett [11], Maria Luisa Brandi [12], Luca Sangiorgi [13], Susanna Balcells [10], Wolfgang Högler [14 15], Wim Van Hul [16], Outi Mäkitie [17 18 19]

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PMID: 34539568 PMCID: PMC8444146 DOI: 10.3389/fendo.2021.709711

The "GEnomics of Musculo Skeletal Traits TranslatiOnal NEtwork": Origins, Rationale, Organization, and Prospects

Fjorda Koromani [1 2 3], Nerea Alonso [4], Ines Alves [5], Maria Luisa Brandi [6], Ines Foessl [7], Melissa M Formosa [8], Milana Frenkel Morgenstern [9], David Karasik [9], Mikhail Kolev [10], Outi Makitie [11 12 13], Evangelia Ntzani [14 15], Barbara Obermayer Pietsch [7], Claes Ohlsson [16], Martina Rauner [17], Kent Soe [18 19 20], Ivan Soldatovic [21], Anna Teti [22], Amina Valjevac [23], Fernando Rivadeneira [1]

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PMID: 34484122 PMCID: PMC8415473 DOI: 10.3389/fendo.2021.709815

Bone Phenotyping Approaches in Human, Mice and Zebrafish – Expert Overview of the EU Cost Action GEMSTONE ("GEnomics of MusculoSkeletal traits TranslatiOnal NEtwork")

Ines Foessl [1], J H Duncan Bassett [2], Åshild Bjørnerem [3 4], Björn Busse [5], Ângelo Calado [6], Pascale Chavassieux [7], Maria Christou [8], Eleni Douni [9 10], Imke A K Fiedler [5], João Eurico Fonseca [6 11], Eva Hassler [12], Wolfgang Högler [13], Erika Kague [14], David Karasik [15], Patricia Khashayar [16], Bente L Langdahl [17], Victoria D Leitch [18], Philippe Lopes [19], Georgios Markozannes [8], Fiona E A McGuigan [19], Carolina Medina-Gomez [20], Evangelia Ntzani [8 21], Ling Oei [22], Claes Ohlsson [22 23], Pawel Szulc [7], Jonathan H Tobias [24 25], Katerina Trajanoska [20], Şansın Tuzun [26], Amina Valjevac [27], Bert van Rietbergen [28], Graham R Williams [2], Tatjana Zekic [29], Fernando Rivadeneira [20], Barbara Obermayer-Pietsch [1]

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PMID: 34925226 PMCID: PMC8672201 DOI: 10.3389/fendo.2021.720728

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