Speaker Profiles

Natasha Appelman-Dijkstra

Natasha Appelman-Dijkstra is an internist-endocrinologist at the Leiden University Medical Center in Leiden, the Netherlands, and she is the chair of the Center for Bone Quality, an international Reference Center for Bone and Mineral Disorders, participating in the steering committee of the European Reference Networks for rare endocrine as well as bone disorders. Since 2022 she is the chair of the academic residency program for internal medicine at the LUMC and has hosted multiple clinical fellowships on endocrinology, bone and mineral metabolism. Natasha's research focus is centered around care for patients with (rare) bone and mineral conditions. Natasha is the coordinator of the European Registries for rare bone and mineral conditions EuRR-Bone (www.eurr-bone.com) where an OI module has been launched including clinician reported outcomes as well as patient reported outcomes.

What are your department/lab/centre's specialties (related to OI-research)?

Clinical studies and PROMS in patients with rare diseases including OI and more specifically measurement of bone quality with impact micro indentation. Furthermore our center is leading the European Registries for rare bone and mineral conditions.

Which OI-related networks does your department/lab/centre belong to?

ERN BOND, ENDO ERN and the Dutch OI group.

Do you contribute to any OI-related registries or databases?

Yes, we contribute to EuRR-Bone and collaborate with the Dutch OI group.

Meena Balasubramanian

Meena Balasubramanian, MBBS, DCH, FRCPCH, MD, is a Senior Clinical Lecturer at the Department of Oncology and Metabolism, University of Sheffield, UK. As a Consultant Clinical Geneticist she is the Lead Geneticist for the highly specialised, severe, complex and atypical Osteogenesis Imperfecta national service.

Dr Balasubramanian has led several projects focused on genetics of rare bone disorders. She has published over 115 principal-author publications and textbooks including a molecular medicine series on OI. She has written >15 patient information leaflets on rare bone and genetic disorders for Unique and the Brittle Bone Society.

Her other roles include Research Director, North East and Yorkshire NHS Genomic Medicine Service Alliance; Bone fragility lead, Genomic Clinical Interpretation Partnership (GeCIP), part of 100,000 Genomes project initiative. She is Secretary for the Clinical Genetics Society and serves on the 'Medical Advisory Board' for the Brittle Bone Society, SATB2 patient support group and Scientific Advisory Committee member for The Children's Hospital Charity.

What are your department/lab/centre's specialties (related to OI-research)?

Genomics in unresolved bone fragility, Deep phenotyping of OI cohorts, Skin biopsy and functional studies for novel genes in OI, BRONE (Brain and Bone) Study, Natural history study on rare forms of OI, Zebrafish models for bone fragility; Developing gene therapy for OI.

Which OI-related networks does your department/lab/centre belong to?

NHS England Highly Specialised OI Service, previously ERN BOND.

Do you contribute to any OI-related registries or databases?

RUDY Study (deep phenotyping, data collection) and hopefully GSD node in future.

Nick Bishop

Nick Bishop is an internationally recognised expert in the field of paediatric bone research. His focus is on experimental medicine/early phase studies in osteogenesis imperfecta, hypophosphatasia and other forms of rickets. He has a long-standing interest in early life influences on later skeletal health. He is Associate Director of the Versus Arthritis Experimental Arthritis Treatment Centre for Children, leading the bone theme and was meeting chair of the 14th International Conference on Osteogenesis Imperfecta, September 2022 in Sheffield, UK.

What are your department/lab/centre's specialties (related to OI-research)?

Clinical trials of new drugs with pharma.

Investigator led clinical studies - observational studies across the life course, development of assessment tools, repurposed drugs, imaging modalities, genetics and phenotyping.

Basic science - OI cell lines (skin, bone). Zebrafish models. Bone implant models in nude mice. Studies of effects of drugs in cell lines.

Which OI-related networks does your department/lab/centre belong to?

The NHS England Highly Specialised Service for Severe, Complex and Atypical OI.

Ex-member of ERN BOND (sadly).

Do you contribute to any OI-related registries or databases?

RUDY - data contribution.

Speaker Profiles

Björn Busse

Björn Busse is Full Professor of Biomedical Sciences and Medical Technology at the Department of Osteology and Biomechanics, University Medical Center Hamburg, Germany. He is director of a Heisenberg Research Group and the Forum Medical Technology Health in Hamburg (FMTHH), Germany. Björn has a strong interest in skeletal imaging, biomechanics and bone quality analyses. His work provides a contribution to our understanding of the fracture mechanisms of bone, specifically by focusing on aspects of bone quality, such as osseous changes with skeletal aging, osteoporosis, osteoporosis treatment strategies, osteomalacia, osteogenesis imperfecta, Paget's disease of bone, etc, from both a medical and a bioengineering perspective.

What are your department/lab/centre's specialties (related to OI-research)?

Multiscale Bone Quality Assessment and High Resolution Imaging of bone samples. Utilization of mouse and zebrafish models for OI. Microcomputed Tomography, Electron Microcopy, Transmission Electron Microscopy, Fourier Transform Infrared Microscopy, Raman Spectroscopy, Micro-mechanical Testing, Nanoindentation, Histology, Histomorphometry. Access to human bone samples via the Hamburg Bone Biopsy Register. Collaboration with the DESY (German Electron Synchrotron) to study OI-related effects on the musculo-skeletal system in high resolution.

Which OI-related networks does your department/lab/centre belong to?

The University Medical Center Hamburg (UKE) is member of the ERN BOND, which includes now 53 European Health Care Providers and their specialist departments on rare bone diseases from 20 EU Member States. Our laboratory is member of the GEMSTONE consortium (GEnomics of MusculoSkeletal Traits TranslatiOnal NEtwork) focusing on the characteristics of rare and common bone diseases. Further on, active membership and engagement in the European Calcified Tissue Society has fostered the collaboration with laboratories in Europe to study the effects of OI in a translational manner. The lab is also belonging to the Interdisciplinary Competence Center for Interface Research (ICCIR) facilitating research on the interfaces between bone, muscle, fat, tendon and vascularization.

Do you contribute to any OI-related registries or databases?

Via the European Calcified Tissue Society (ECTS) and the GEMSTONE consortium we established collaborations with OI researchers in Italy (Antonella Forlino, University of Pavia) in Belgium, (Andy Willaert, Paul Coucke, University of Ghent) and in Israel/USA (Prof. David Karasik, Bar-Ilan University). Further on, we have co-authored several research papers focusing on the mechanical characteristics of bone tissue obtained from animal models resembling OI or bone biopsies from individuals with OI.

Valérie Cormier-Daire

Valérie Cormier-Daire, MD, PhD, is a medical geneticist, Professor of Genetics at the Université Paris Cité, Hôpital Necker Enfants Malades, Paris, France. She is the head of the French Reference Center for Skeletal Dysplasia (SD). She is chief investigator for a number of clinical trials and co-leads the working group dedicated to diagnostics and research in the ERN BOND.

She is also responsible for a research team working on SD in INSERM Unit 1163 (Imagine Institute). She has contributed to highlight the role of cilia, proteoglycan and TGFβ signalling in the ossification processes by studying human cellular and mouse models. Her track-record includes 473 publications.

What are your department/lab/centre's specialties (related to OI-research)?

Clinical genetics and Reference center for skeletal dysplasia, INSERM UMR1163, Paris Cité University, Imagine Institute, Necker Enfants Malades Hospital.

Which OI-related networks does your department/lab/centre belong to?

French skeletal dysplasia network.

ERN BOND.

ISDS.

French patient association (AOI).

Do you contribute to any OI-related registries or databases?

Yes, local database.

Paul Coucke

Paul Coucke, Assistant Professor at the Ghent University Hospital, Belgium, leads the connective tissue lab at the Department of Medical Genetics. One of the focuses of his research group, consisting of a post-doc, 4 PhD students and 3 technicians, is unravelling the pathogenetic mechanisms and exploring the underlying pathways involved in familial bone disorders. Gene identification through linkage analysis, homozygosity mapping and exome sequencing using Next Generation Sequencing technology together with the use of the zebrafish model, are the main expertises of the lab. His group identified several genes for monogenic disorders for which further functional investigations have been performed.

What are your department/lab/centre's specialties (related to OI-research)?

1. *To identify potential biomarkers in dominant OI, we employed three OI zebrafish models, carrying different glycine substitutions in type I collagen (col1a1adc124/+,*

col1a2mh15/+, col1a1amh13/+), and showing variability in phenotypic severity using different omic approaches combined with X-ray, micro-CT and Alizarin red mineral staining.

2. *We investigate inter as well as intra-familial variability in those mutants and also in the fkbp10a knockout mutant. Exome sequencing of the 6 most mildly and 6 most severely affected mutants is performed on a NovaSeq 6000 illumina sequencer, followed by SNP-based linkage analysis. Specific candidate regions are delineated in order to identify candidate modifying OI genes.*
3. *We investigate unsolved OI-families in which no mutation in any existing OI gene have been identified. This is done through whole exome and/or whole genome sequencing combined with CNV analysis.*

Methodologies:

- *Next Generation sequencing technology in order to identify new OI genes.*
- *Proteomic and transcriptomic analysis of the vertebral column of OI mutants and correlation with phenotypic severity.*
- *Cellular composition and function of the vertebral bone in dominant OI models and correlation with phenotypic severity.*
- *The use of CRISPR/Cas9 technology to create crispants and stable mutant zebrafish lines in order to study the underlying molecular mechanisms in OI mutants.*

Which OI-related networks does your department/lab/centre belong to?

Paul Coucke and Sofie Symoens are members of ERN BOND.

Do you contribute to any OI-related registries or databases?

We submit regularly OI mutations to the LOVD database.

Ivan Durán

Ivan Durán is professor at the University of Malaga, Spain, and a PI at IBIMA-BIONAND institute. He worked for 10 years in the US at Cornell University and the International Skeletal Dysplasia Registry at University of California Los Angeles before moving back to Europe where he leads a lab using precision medicine approaches to study skeletal dysplasias with a special focus in OI. His research covers from gene discovery of skeletal dysplasias to development of new precision therapies based in genetic mechanism of disease.

What are your department/lab/centre's specialties (related to OI-research)?

My lab specializes in precision medicine approaches with the goal of matching mechanism of disease and individual genotype with precision treatments. We perform functional studies with patient specific models like skeletal organoids or searching for phenomic markers.

Which OI-related networks does your department/lab/centre belong to?

Our lab is headquarters of the DIAGSKEL consortium (national registry and diagnostic network) and it belongs to the European GEMSTONE cost action (GEnomics of Musculo Skeletal traits TranslatiOnal NEtwork). We coordinate our work with the national patient organization (Fundación AHUCE) and we keep a close collaboration with members of the American organizations OIF, International Skeletal Dysplasia Registry and the Brittle Bone Disease Consortium.

Do you contribute to any OI-related registries or databases?

We coordinate the DIAGSKEL Spanish national registry and had a significant contribution history within the International Skeletal Dysplasia Registry.

Lars Folkestad

Lars Folkestad, MD, PhD, is a clinical associate professor in endocrinology from the Bone and Mineral Unit at the Department of Endocrinology at Odense University Hospital, Denmark. He works primarily with metabolic bone and mineral diseases and thyroid diseases. His research is centred around registery based epidemiology focused on the natural history of osteogenesis imperfecta. Over the past decade he has been part involved in several national and international studies in adults with OI. He is a scientific advisory board member of OIFE and Care 4 Brittle Bones. Furthermore, he was a founding member of the Danish Osteogenesis Imperfecta Consortium - which is a collaboration between the Danish OI Patient society and the pediatric and adult centers of expertise. The DOICon works toward collaborative nationwide research projects and evidence-based treatment and follow-up in OI.

Antonella Forlino

Antonella Forlino is a basic scientist with strong interests in bone biology. Her research activity is focused on the molecular, biochemical, and functional study of genetic diseases of the connective tissue, in particularly the brittle bone disease Osteogenesis Imperfecta (OI). She developed and characterized several in vitro and in vivo models for OI and uses these models for investigating the molecular basis of the disease and for identifying new therapeutic targets and innovative treatments. She is a scientific advisory board member of ASITOI, OIFE and Care 4 Brittle Bones.

What are your department/lab/centre's specialties (related to OI-research)?

Generation and characterization of in vitro (primary and immortalized cells) and in vivo models (mice and zebrafish) for heritable skeletal diseases.

The laboratory has strong expertise in collagen biochemistry and cellular biology. Expression analysis at transcript and protein level of cells and tissue are performed by advanced molecular and biochemical approaches.

Mass spectrometry analysis, high resolution microscopy and in vivo microCT are routinely used.

Which OI-related networks does your department/lab/centre belong to?

ASITOI/OIFE/OIF/ICCBH.

Do you contribute to any OI-related registries or databases?

No.

Cecilia Giunta

Cecilia Giunta is a molecular geneticist working in the field of Connective Tissue Disorders (CTD). She obtained her PhD in Genetics in 1993 at the University of Ferrara, Italy. In 1994 she conducted a postdoctoral research fellowship at the Royal Children's Hospital Melbourne, Australia. In 1995 she was employed as postdoctoral fellow and later as senior research scientist at the Division of Metabolism and Children's Research Center, Children's Hospital Zurich, Switzerland. Since 2008 she runs the diagnostic and research activities on brittle bone disorders and other CTDs, studying their pathomechanisms and investigating diagnostic biomarkers and therapeutic targets (ORCID iD 0000-0002-9313-8257).

I would like to highlight that our research work is the equal contribution of Marianne Rohrbach, Chief Physician, and myself. We are co-leaders of the Connective Tissue Research Unit at the University Children's Hospital Zürich (https://www.kispi.uzh.ch/forschungszentrum/forschungsgebiete/stoffwechsel/angeborene-bindegewebskrankheiten)

What are your department/lab/centre's specialties (related to OI-research)?

Molecular and biochemical research of the entire spectrum of OI with a main focus on the discovery of the genetic basis of new unsolved cases and the understanding of the disease pathomechanisms by acquiring and implementing clinical, morphological, biochemical, cell-biological and -ultrastructural, molecular and OMICS data. Development of 3D cell-based models of OI to advance 3Rs research according to the Swiss National Science Foundation (SNSF).

Which OI-related networks does your department/lab/centre belong to?

SWISS Bone Diseases Network (SG BOND, Swiss Group BONe Disorder). Being based in Switzerland and thus belonging to a non-EU Research Facility, has hampered our participation to larger European Research Networks ERN.

Do you contribute to any OI-related registries or databases?

We have established a i) Patient Registry for Connective Tissue Disorders which includes patients with a clinical and genetic confirmed diagnosis of OI and ii) Biobank consisting of approximately 200 OI patient cells (fibroblasts and osteoblasts).

We contribute genetic data to the OI LOVD3 Database (Global Variome shared LOVD, formerly Osteogenesis Imperfecta Variant Database) supported by OIFE.

ORCID database Cecilia Giunta (0000-0002-9313-8257) (orcid.org).

Cecilia Götherström

Cecilia Götherström is an Associate Professor in Stem Cell Research at Karolinska Institutet in Sweden and her research is on regenerative medicine. Dr Götherström has developed fetal mesenchymal stem cells as a treatment for Osteogenesis Imperfecta with promising results. Dr Götherström leads an academic international multicentre trial, Boost Brittle Bones Before Birth (BOOSTB4), to evaluate the safety and the clinical effect of mesenchymal stem cell transplantation for the treatment of severe OI in young children.

What are your department/lab/centre's specialties (related to OI-research)?

We have developed a new potential therapy (mesenchymal stem cell transplantation) for OI. Our specialty and expertise is basic and translational research on mesenchymal stem cells and OI.

Which OI-related networks does your department/lab/centre belong to?

OIFE, Care for Brittle Bones, and Care4Bones.

Do you contribute to any OI-related registries or databases?

We contribute to the Leiden Open Variation Database (the previous OI Variant Database).

Kassim Javaid

Kassim Javaid is an adult rheumatologist and Associate Professor in metabolic bone disease at the University of Oxford, UK. He specializes in common and rare metabolic bone diseases. He is the clinical lead for Oxford Fracture Prevention Service, and the national Fracture Liaison Service Audit for England and Wales and co-chairs the Capture the Fracture programme. He is the clinical lead for the Oxford Rare Bone Disease Service for adults and the Musculoskeletal Genomic Clinical Interpretation partnership. His research interests include the epidemiology of musculoskeletal diseases with a focus on rare diseases of the bone, vitamin D and secondary fracture prevention.

What are your department/lab/centre's specialties (related to OI-research)?

Epidemiology and Health economics.

Which OI-related networks does your department/lab/centre belong to?

Nil.

Do you contribute to any OI-related registries or databases?

Yes, Rudy and EuRR-Bone.

Brendan Lee

Brendan Lee is the Robert and Janice McNair Endowed Chair in Molecular and Human Genetics, Professor and Chairman of the Department of Molecular and Human Genetics at Baylor College of Medicine, Houston, USA. Dr Lee studies structural birth defects and inborn errors of metabolism, and multi-omic diagnostic approaches in genomic medicine implementation. He currently leads the NIH BCM Undiagnosed Diseases Network Clinical Site at Baylor and NIH Brittle Bone Disorders Consortium. He holds multiple patents in the areas of drug discovery and gene therapy, and several technologies are in clinical trials including in osteogenesis imperfecta, osteoarthritis, and maple syrup urine disease.

What are your department/lab/centre's specialties (related to OI-research)?

Mouse Genetics, Human Genetics, Clinical Genetics (Texas Children's Hospital Skeletal Dysplasia and Baylor Medicine Clinic for Adult Metabolic and Genetics Disorders of Bone).

Which OI-related networks does your department/lab/centre belong to?

Brittle Bone Disorders Consortium (https://www1.rarediseasesnetwork.org/cms/BBD), Undiagnosed Diseases Center (https://www.bcm.edu/research/research-centers/undiagnosed-diseases-center); Lawrence Family Bone Diseases Program of Texas (https://txbonediseaseprogram.org/); Center for Skeletal Medicine and Biology (https://www.bcm.edu/research/research-centers/center-for-skeletal-medicine-and-biology).

Do you contribute to any OI-related registries or databases?

Data access for community via BBDC.

Alexandre Mercier

Alexandre Mercier is a young clinician in the adult rheumatology department of Professor Roland Chapurlat in Lyon, France, and a PhD student at the University Claude Bernard Lyon 1. His research focuses on the genetic and epigenetic regulation of rare bone diseases and their clinical and therapeutic implications, especially in osteogenesis imperfecta.

What are your department/lab/centre's specialties (related to OI-research)?

The department specializes in clinical and research on rare bone diseases in adults, participating in clinical trials and courses and specializing in basic research on epigenetic regulation by non-coding RNAs in OI and fibrous dysplasia.

Which OI-related networks does your department/lab/centre belong to?

The Inserm U1033 laboratory and the rheumatology department are attached to the OSCAR network, Filière OSCAR, the french national network for rare bone, calcium and cartilage diseases.

Do you contribute to any OI-related registries or databases?

Not currently.

Dimitra Micha

Dimitra Micha, PhD, is assistant professor at the Department of Human Genetics in Amsterdam University Medical Centres, Netherlands. She is principal investigator of the Centre for Connective Tissue Diseases which is focused on the study of Osteogenesis Imperfecta and other forms of monogenic bone dysplasias. Main research lines are the improvement of molecular diagnosis, investigation of the disease mechanism and the development of meaningful therapy which is urgently lacking for these patients. In addition to the discovery of new genetic causes, her team has recently taken over the curation of the international Osteogenesis Imperfecta Variant Database.

What are your department/lab/centre's specialties (related to OI-research)?

Human genetics department: We have been for more than 30 years the national reference centre for the molecular diagnosis of OI.

Centre for Connective Tissue Diseases laboratory: We have a lot of experience with investigating the mechanism of genetic causes in OI based on which we have several lines of research for therapy development.

Amsterdam UMC centre: National expert centre for OI. In addition to diagnostics and preclinical research, our centre offers through the endocrinology department (Dr M Eekhoff) specialised care for OI while clinical studies and the TOPAZ trial are also being conducted.

Which OI-related networks does your department/lab/centre belong to?

ERNBOND

Care4Bones Pro (OI Basic Science Network).

Do you contribute to any OI-related registries or databases?

We manage the international database for OI: https://databases.lovd.nl/shared/genes/COL1A1

Stuart Ralston

Stuart Ralston is Professor of Rheumatology at the University of Edinburgh, UK, and Clinical Director of the rheumatology service in NHS Lothian. He serves as the programme director of Edinburgh University's MSc in clinical trials and leads the Kennedy Trust funded intercalated MB-PhD programme. His research currently focuses on the genetic determinants of osteoporosis and its response to treatment, the pathophysiology of Paget's disease of bone and pregnancy-associated osteoporosis, and the treatment of osteogenesis imperfecta where he is chief investigator on the TOPAZ trial. Additionally, he is joint editor-in-chief of Calcified Tissue International and editor of Davidson's Textbook on the Principles and Practice of Medicine. Stuart served as chair of the Commission for Human Medicines for the Medicines and Healthcare Regulatory Authority of the UK (2013–2021) and continues to advise the MHRA on matters relating to medicines for women's health and rheumatology.

What are your department/lab/centre's specialties (related to OI-research)?

We have experience in treating adult OI and undertake transition care. We are co-ordinating a large scale clinical trial of treatment in OI currently (TOPAZ).

Which OI-related networks does your department/lab/centre belong to?

None specifically, but we work closely with the BBS in UK.

Do you contribute to any OI-related registries or databases?

No, not at present.

Frank Rauch

Frank Rauch, MD, is a Professor of Pediatrics and clinician-scientist at the Shriners Hospital for Children and at McGill University in Montreal, Canada. He obtained his MD degree from the Technical University of Munich and trained as a pediatrician at Cologne University, Germany. At Shriners Hospital and McGill University since 2001, his clinical activities and research program concentrate on improving bone health in children, with a special focus on osteogenesis imperfecta and on the role of the muscle system in bone diseases. Dr Rauch has authored or co-authored more than 400 publications that have been cited more than 27,000 times.

What are your department/lab/centre's specialties (related to OI-research)?

Bisphosphonate treatment, genetic testing, orthopedic surgery, rehabilitation.

Which OI-related networks does your department/lab/centre belong to?

Shriners Hospitals for Children; Brittle Bone Disease Consortium.

Do you contribute to any OI-related registries or databases?

Brittle Bone Disease Consortium: Data collection, paper writing.

Luca Sangiorgi

Luca Sangiorgi, MD, PhD, Director of Department of Rare Skeletal Disorders, Bologna, Italy, is the coordinator of the Rare Diseases Centre of Rizzoli Orthopaedic Institute since 2006 and responsible of 4 National Registers of Rare Disease (Multiple Hereditary Exostoses, Osteogenesis Imperfecta, Ehlers-Danlos Syndrome, Ollier Disease and Maffucci Syndrome). He's the coordinator of BIOGEN, diagnostic and research genetic biobank, and of Telethon Network of Genetic Biobanks. Since March 2017, he has been coordinator of the European Reference Network on Rare Bone Disorders (ERN BOND).

Luca Sangiorgi participates as coordinator or partner in more than 20 national and international research projects, including: European Joint Programme on Rare Disease EJP-RD, MCDS-Therapy Project, the European Registry for Rare Bone and mineral conditions (EuRR-Bone). He's the first author who has contributed to more than 80 articles published in high impact journals (Index H: Scopus 25 Google Scholar 27).

What are your department/lab/centre's specialties (related to OI-research)?

OI Registry, AI studies on data from OI patients, Biobank with OI patients samples, HRpQCT studies on OI patients, OI cardio study, study on pain in OI patients, sponsor for Italy of the study on Losartan in adult OI patients, movement analysis on OI patients.

Which OI-related networks does your department/lab/centre belong to?

Emilia-Romagna regional Rare Bone Diseases network (coordinator) ERN BOND Italian network (coordinator), ERN BOND (coordinator), ECTS Rare Diseases working group, Forum on Bone and Mineral Research, ICCBH.

Do you contribute to any OI-related registries or databases?

ROI (Italian Registry on OI) coordinator, EuRR-Bone (Wp Leader and contributor), Emilia-Romagna Regional registry for Rare Diseases (contributor and co-curator for rare bone diseases), Italian National Registry for Rare Diseases (contributor).

Oliver Semler

Oliver Semler is head of the department of rare skeletal diseases in childhood at the Children´s University Hospital Cologne, Germany, and professor of pediatrics. He studied medicine in Cologne and Freiburg and spent a research term in Canada.

He is certified as paediatric rheumatologist and focusses clinical and scientifically on improvements of mobility and medical treatments in rare bone diseases. He is Principal Investigator in clinical trials and published more than 90 peer-reviewed articles and was awarded with numerous awards. He is chair of the medical advisory board of OIFE and medical advisor of the German OI patient organisation.

What are your department/lab/centre's specialties (related to OI-research)?

We are specialised in phenotyping of patients, bone densitometry, muscle function, mobility assessments. Genetic department specialized in rare bone diseases.

Which OI-related networks does your department/lab/centre belong to?

ERN BOND, German network for rare bone diseases (NetsOs).

Do you contribute to any OI-related registries or databases?

Only our database at our institution.

Frank Zaucke

Frank Zaucke is research director at the Department of Orthopaedics at the University Hospital in Frankfurt, Germany. His laboratory focuses on mechanisms of skeletal development and, in particular, on alterations in rare human skeletal diseases. In addition, his laboratory aims to provide a better understanding of the structure and function of the extracellular matrix in bone and cartilage which plays a crucial role in osteoarthritis. He has published more than 100 papers, is a member of numerous scientific societies and vice-president of the German Society for Matrix Biology. His research is funded by the German Research Foundation (DFG), the European Union and the Dr Rolf M Schwiete Foundation.

What are your department/lab/centre's specialties (related to OI-research)?

We are specialised in phenotyping of patients, bone densitometry, muscle function, mobility assessments. Genetic department specialized in rare bone diseases. In addition, we are studying the impact of OI causing mutations in animal (mouse and zebrafish) and cellular models to unravel disease mechanisms at the molecular level.

Which OI-related networks does your department/lab/centre belong to?

ERN BOND, German network for rare bone diseases (NetsOs).

Do you contribute to any OI-related registries or databases?

Only our database at our institution.