

## Symposium Speakers

### Yasemin Alanay

Yasemin Alanay, MD, PhD is a pediatric geneticist, professor of pediatrics at Acibadem University School of Medicine, Istanbul. She studied medicine at Hacettepe University, Ankara, completed her residency in Pediatrics in 2002. Completed Pediatric Genetics Fellowship at Hacettepe Ihsan Dogramaci Children's Hospital. In 2006, she was mentored by Profs D. Rimoin and D. Krakow as a research fellow at the International Skeletal Dysplasia Registry, Cedars Sinai Medical Center, Los Angeles, USA. She later received her PhD in Genetics in 2009 at Hacettepe University. She has participated and led research in the field of genetic diseases of skeleton and craniofacial malformations. Dr Alanay has authored over 95 peer-reviewed scientific publications and book chapters. She is a board member of ESHG and editorial boards of the American Journal of Medical Genetics and Clinical Dysmorphology. Her current practice involves clinical genetics, dysmorphology with a special interest in clinical, molecular and social aspects of skeletal dysplasias.



### Lynda Bonewald

Dr Lynda F Bonewald is a University of Missouri, Kansas City (UMKC) Curators Professor, the Lefkowitz Professor of Oral and Craniofacial Sciences, and Director of the Mineralized Tissue/Bone Biology Research Program at the UMKC School of Dentistry, Director of the UMKC Center of Excellence in the Study of Dental and Musculoskeletal Tissues and UMKC Vice Chancellor for Clinical and Translational Research. Dr. Bonewald was educated at the University of Texas, Austin, where she earned a BA in Biology and at the Medical University of South Carolina, Charleston, where she received a Ph.D. in Immunology/Microbiology. She is Past-President of the ASBMR and is best known for her work in the study of osteocyte biology and function.



### Steven Boyd

Dr Steven Boyd holds a PhD in Mechanical Engineering, specialized in Biomedical Engineering. He was appointed as a faculty member at the University of Calgary in 2002, and is now a Professor in the Faculty of Medicine (Radiology), and jointly appointed at the Schulich School of Engineering (Mechanical Engineering) and Faculty of Kinesiology. He is a principal investigator at the McCaig Institute for Bone and Joint Health in medicine, and his research uses a multi-disciplinary biomedical engineering approach for development of early detection and monitoring of bone and joint health, with particular focus on osteoporosis and osteoarthritis. The Bone Imaging Laboratory he established in 2004 develops methods for bone quality detection using high-resolution computed tomography and computer methods such as the finite element analysis to investigate bone and joint diseases. He holds an Alberta Innovates – Health Solutions (AIHS) Senior Scholar position, and was appointed in 2010 as the Bob and Nola Rintoul Chair in Bone and Joint Research. His research is supported by the Canadian Institutes for Health Research, the Natural Sciences and Engineering Research Council (NSERC) of Canada, Canada Foundation for Innovation and Alberta Innovates – Health Solutions.



## Mike Briggs

Mike Briggs obtained his PhD at the MRC Clinical Research Centre, Harrow, studying the genetic basis of Osteogenesis Imperfecta. He undertook postdoctoral work at UCLA identifying the genetic basis of chondrodysplasias. In 1996 Mike moved to Manchester as an AR-UK Fellow to continue studying disease mechanisms in chondrodysplasia. In 2004 he was awarded a Wellcome Trust Senior Research Fellowship that was renewed in 2009. In 2012 he was appointed Professor of Skeletal Genetics in the Institute of Genetic Medicine at Newcastle University and continues to work on disease mechanisms in chondrodysplasia with a focus on identifying novel therapeutics for these rare diseases. Mike has been instrumental in establishing several European consortia for the diagnosis and research of rare skeletal diseases. These have included European Skeletal Dysplasia Network, EuroGrow and most recently SYBIL, a large-scale FP7 funded project involving 18 partners over 5 years.



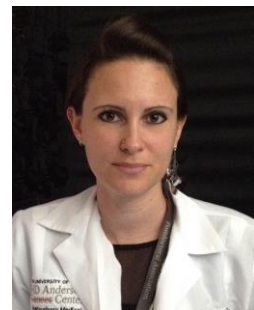
## Sandy Burnham

Sandy Burnham received his MD from the Perelman School of Medicine at the University of Pennsylvania (UPENN) in 1997 and completed his Pediatrics Residency in 2000 and General Pediatrics and Pediatric Rheumatology Fellowships in 2004 at The Children's Hospital of Philadelphia (CHOP). He received his Master of Science in Clinical Epidemiology from UPENN in 2006. Dr Burnham is now Associate Professor of Pediatrics at UPENN and practices Pediatric Rheumatology at CHOP, where he is the Pediatric Rheumatology Training Program Director. His research focuses on the impact of chronic inflammatory conditions on skeletal structure using methods such as DXA, pQCT, and spine QCT. Recent work used spine QCT finite element modeling to characterize vertebral strength deficits in children with systemic lupus erythematosus. In addition, Dr Burnham is interested in developing methods to systematically enhance patient safety and optimize care quality in children exposed to chronic glucocorticoid therapy.



## Eleonora Dondossola

Eleonora Dondossola is a Senior Postdoctoral Fellow in the Intravital Imaging Lab, Koch Center for Applied Research in Genitourinary Cancers, the University of Texas MD Anderson Cancer Center, Houston, TX, USA. She received her PhD in Cell and Molecular Biology from S. Raffaele Vita-Salute University, Milan, Italy, in 2010. Her current research is focused on metastasis to bone and includes bone tissue engineering, bone window models and intravital multiphoton microscopy of metastatic lesions to bone.



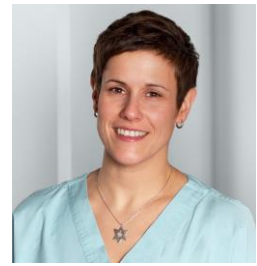
## Peter Fratzl

Peter Fratzl is director at the Max Planck Institute of Colloids and Interfaces in Potsdam, Germany, and honorary professor at Humboldt University Berlin and Potsdam University. He holds an engineering degree from Ecole Polytechnique in Paris, France, and a doctorate in Physics from the University of Vienna, Austria. His scientific interests include the relation between structure and mechanical behaviour of biological and bio-inspired materials, with a special focus on bone material structure and properties in osteoporosis treatment and bone regeneration. Peter Fratzl has been external member and advisor to the director of the Ludwig Boltzmann institute of Osteology in Vienna, Austria, for the last 20 years. He has published over 450 peer-reviewed research publications. He is Fellow of Acatech, the Austrian Academy of Sciences and the Materials Research Society (US), holds an honorary doctorate from Montpellier University and is recipient of the Leibniz Prize from the German Science Foundation.



## Marie Gdalevitch

Dr Marie Gdalevitch completed both her medical and orthopedic surgery degrees at McGill University. Following her residency, Dr Gdalevitch pursued her first fellowship in limb lengthening and deformity correction at the International Center for Limb Lengthening in Baltimore, Maryland. Dr Gdalevitch then embarked on her second fellowship in pediatric orthopedics and basic science research at the Children's Hospital at Westmead in Sydney, Australia. She is currently an assistant professor of surgery in the Division of Orthopedics at McGill University and works at the Shriners Hospital in Montreal as well as the Montreal General Hospital. Her clinical interests include: limb lengthening and deformity correction, osteogenesis imperfecta, hip reconstruction and pediatric orthopedics. Dr Gdalevitch is currently pursuing a PhD in bone regeneration research involving murine models of distraction osteogenesis well as disuse osteopenia models.



## John Herzenberg

Dr John Herzenberg graduated from Boston University Medical School and trained at Duke University for Orthopaedics and Toronto Sick Kids for pediatric orthopaedics. He specializes in the diagnosis and treatment of adult and pediatric patients with congenital abnormalities, joint contractures, neuromuscular disorders, non-unions, malunions, deformity, and bone defects. He is in demand worldwide as a speaker and has more than 110 Pub Med papers listed to his credit. Dr Herzenberg is married to Merrill Chaus, RN. They have three daughters. Since 1998, he and his family have volunteered yearly with Operation Rainbow, providing free orthopaedic surgery to underprivileged children in Central and South America, and Haiti. In 1997, Dr. Herzenberg learned an old, ignored conservative technique for clubfoot treatment called the Ponseti method. He enthusiastically embraced this method, and his efforts in teaching this method have helped turn the tide from surgery to casting for babies with clubfoot.



## Brendan Lee

Brendan Lee is the Robert and Janice McNair Endowed Chair in Molecular and Human Genetics, Professor and interim Chairman of the Department of Molecular and Human Genetics at Baylor College of Medicine. Dr. Lee co-directs the joint MD Anderson Cancer Center and Baylor College of Medicine Rolanette and Berdon Lawrence Bone Disease Program of Texas, and the Baylor College of Medicine Center for Skeletal Medicine and Biology. He is Founder and Director of the Skeletal Dysplasia Clinic at Texas Children's Hospital, and of the Medical Student Research Track at Baylor. As a pediatrician and geneticist, Dr. Lee studies structural birth defects and inborn errors of metabolism. Dr. Lee identified the first genetic causes of human skeletal dysplasias that affect the growth and strength of the skeleton. Most recently, he discovered new causes of brittle bone disease in children. In so doing, he is developing new approaches for diagnosing and treating these disorders.



## Laurence Legeai-Mallet

Laurence Legeai-Mallet is currently director of research at Imagine Institute-Paris Descartes University. She received her PhD in genetic from University of Paris V, she is a member of International skeletal dysplasia Society, European Skeletal Dysplasia Network and the French reference center of bone dysplasias. She has been involved in the field of skeletal disease since 1993. Her research field ranges from identification of disease genes involved in cartilage and bone diseases, in understanding bone development and skeletal diseases to the development of therapeutic approaches. In 2009, she initiated therapeutic approaches for osteochondrodysplasias through collaboration in both academic chemistry laboratory (Paris Descartes university) and pharmaceutical companies. In 2012, in collaboration with BioMarin, she reported the therapeutic potential of a novel natriuretic peptide C analogue (BMN 111) as the first investigational therapy for the most frequent dwarfism, achondroplasia.



## David Little

Professor David Little received his Medical Degree from the University of Sydney where he is Conjoint Professor of Paediatrics and Child Health, specialising in Orthopaedic Surgery. Prof Little is Head of Orthopaedic Research and Biotechnology at The Children's Hospital at Westmead, part of the Sydney Children's Hospital Network. He has broad clinical interests in Children's Orthopaedic Surgery, and has specific expertise in lower limb problems including hip disorders and limb lengthening and deformity correction. His research interests are into the pathophysiology of bone repair in NF1/Congenital Pseudarthrosis, Perthes disease and other forms of Avascular Necrosis of the Hip. He is also renowned for publications on fracture healing, including the role of therapeutic agents in augmentation of bone repair.



## Outi Mäkitie

Dr Outi Mäkitie received her MD and PhD from the University of Helsinki, Finland where she also completed training in Pediatrics and in Pediatric Endocrinology. After a three-year post-doctoral clinical and research fellowship at The Hospital for Sick Children in Toronto, Canada, she returned to Finland and served as Head of the Metabolic Bone Clinic, Children's Hospital, University of Helsinki. In 2013 she moved to Stockholm, Sweden and currently works as Associate Professor at Clinical Genetics, Karolinska Institutet and Karolinska University Hospital. Dr Mäkitie's clinical and translational research focuses on various genetic and acquired skeletal disorders.



## Barbara Misof

Barbara Misof is Staff Scientist at the Ludwig Boltzmann Institute of Osteology, Vienna, Austria. She completed her PhD in Physics and Postgraduate Education in Medical Physics at the University of Vienna in 2000. In 2002, she received the Herbert-Czitober-Research Award of the Austrian Society of Bone and Mineral Research. Her areas of interest are bone material properties, structure-function relation, effects of osteoporosis treatments with a focus on bone matrix mineralization.



## Madhu Misra

Dr Misra is a full Professor of Pediatrics at Harvard Medical School and a pediatric endocrinologist at Massachusetts General Hospital in Boston, Massachusetts. She directs the fellowship training program in pediatric endocrinology at Massachusetts General Hospital, and has a Masters in Public Health from the Harvard School of Public Health. Dr Misra's clinical interests include disorders of bone mineral metabolism (including conditions such as anorexia nervosa, the female athlete triad, celiac disease, inflammatory bowel disorders and autism spectrum disorders), and disorders of growth, puberty, and the pituitary gland. Her past and current research focuses on neuroendocrine and bone changes in conditions that span the nutritional spectrum from anorexia nervosa to the female athlete with amenorrhea to obesity. Her awards include the Janet McArthur Award from Women in Endocrinology and the John Haddad Young Investigator Award from AIMM/ASBMR (amongst others). Dr Misra has been funded consistently by the NIH for the past 10 years, and has authored over 180 original papers, reviews and chapters.



## Craig Munns

Associate Professor Craig Munns is a Senior Staff Specialist in Bone and Mineral Medicine and Endocrinology at the Children's Hospital at Westmead and Conjoint Associate Professor in the Sydney Medical School at the University of Sydney, Australia.

Following the completion of his Paediatric and Endocrinology training at The Royal Children's Hospital, Brisbane, Australia, A/Prof Munns was Clinical Associate in Genetic and Metabolic Bone Disorders at the Shriners Hospital for Children, Montreal, Canada. He was awarded his PhD through the University of Queensland in 2004.

Associate Professor Munns' major clinical and research focus is the diagnosis and management of primary and secondary bone disorders in children.



## Gen Nishimura

Dr Gen Nishimura is Radiologist-in-Chief, Department of Pediatric Imaging, Tokyo Metropolitan Children's Medical Center, Tokyo, Japan. After obtaining the medical degree at Keio University School of Medicine in Tokyo, Dr. Nishimura started his career of diagnostic radiology in the late 70s. During the exposure to pediatric radiology in early 80s, he developed a special interest in a diagnosis of skeletal dysplasias. In 1988, he visited Royal Alexandra Hospital for Children in Sydney and learned the subject from Professor Kazimierz Kozlowski. Thereafter, he was able to develop a number of international scientific collaborations with new friends and mentors, such as Professor Andrea Superti-Furga in Switzerland and Professors Juergen Spranger in Germany. He contributed to several new discoveries of disease-causing genes, such as TGFB in Camurati-Engelmann disease and SLC35D1 in Schneckenbecken dysplasia. He also contributed to recent versions of international nosology and classification of genetic skeletal disorders (2010, 2006 revisions).



## Amaka Offiah

Amaka C Offiah is Senior Lecturer and consultant paediatric radiologist at the University of Sheffield and Sheffield Children's Hospital. She has a specialist interest in the musculoskeletal system. She has co-authored 2 books, 7 book chapters, published 67 peer-reviewed articles and given over 100 invited national and international lectures. She is Convenor of the Skeletal Dysplasia Group for Teaching and Research and Chairperson of the European Society of Pediatric Radiology Child Abuse Taskforce. She was the RCR 2013 Roentgen Professor – being the first female and the first pediatric radiologist to hold this post.



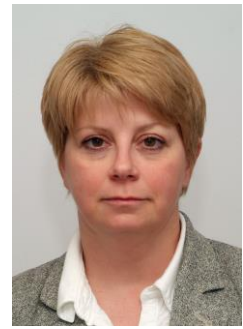
## Frank Rauch

Frank Rauch obtained his MD degree from the Technical University of Munich, Germany, and trained as a pediatrician at the Children's Hospital of Cologne University, Germany. Since 2001 Dr. Rauch has been a clinician-scientist at the Shriners Hospital for Children and is currently a Professor of Pediatrics at McGill University, Montreal, Canada. His clinical and scientific work focusses on heritable bone diseases in children and adolescents, in particular osteogenesis imperfecta. He is the Director of Clinical Laboratories at Shriners Hospital, comprising laboratories for biochemistry, bone histomorphometry and molecular diagnostics. Dr Rauch has published more than 160 original articles in peer-reviewed scientific journals. Since 2009, he has been Editor-in-Chief of the Journal of Musculoskeletal and Neuronal Interactions.



## Susan Schiavi

Susan Schiavi has spent her career as a research scientist within the biotech/pharmaceutical industry. She received her PhD from the University of Massachusetts Medical School and completed a postdoctoral fellowship at Harvard Medical School. As a Senior Scientific Director at Genzyme and Sanofi, her primary role has been the development of scientific strategy for the identification of novel therapeutic targets and translational research associated with genetic and acquired bone and renal diseases. Within this framework, her team's research incorporated a network based bioinformatics approach to complement traditional research strategies centered on the role of critical proteins associated with skeletal health. She has published more than 30 peer research articles, invited review articles and book chapters.



## Nick Shaw

Dr Nick Shaw is a Consultant Paediatric Endocrinologist at Birmingham Children's Hospital and Honorary Senior Clinical Lecturer at the University of Birmingham. He developed an interest in paediatric calcium and bone metabolism whilst a Lecturer at the University of Leeds in 1985 and subsequently as a Lecturer at the University of Liverpool. He completed his endocrine training in Birmingham where he has been a consultant since 1994. He established a multidisciplinary service for children with metabolic bone disease which in 2011 was designated as one of four national centres for complex childhood osteogenesis imperfecta.

He is the organiser of a postgraduate training course in paediatric calcium and bone metabolism and co-editor of the book "Calcium and Bone Disorders in Children and Adolescents" published by Karger in 2009.

He was a founder member and first secretary of the British Paediatric and Adolescent Bone Group and the Bone Club of the European Society for Paediatric Endocrinology. His research interests include secondary osteoporosis in children and vitamin D.



## Anna Villa

Anna Villa is Chief of the Human Genome Unit at UOS/IRGB and is also responsible for a Research Unit at Telethon Institute for Gene Therapy (TIGET). Her group has also extensively contributed towards the molecular dissection of genetic bone disorders, focusing on Autosomal Recessive Osteopetrosis (ARO). In particular she has identified TCIRG1 as the gene responsible for 60% of the ARO patients and contributed towards the characterization of two other forms of ARO due to a defect in the Grey Lethal and PLEKHM1 gene, respectively. More recently her group has described RANKL and RANK as genes responsible for the osteoclast poor ARO. In addition, over the last 5 years, Dr Villa's group has pioneered the use of the *oc/oc* model to test in utero cellular therapy; this has established that prenatal correction of the defect is possible, since almost complete rescue of the phenotype has been achieved. The contribution of Dr Villa in the molecular dissection of ARO has important implications not only for the molecular diagnosis, but also for the treatment of the disease.



## Leanne Ward

Dr Leanne Ward is an Associate Professor of Pediatrics at the University of Ottawa where she holds a Research Chair in Pediatric Bone Health. She is the Medical Director of the Pediatric Bone Health Clinical and Research Programs at the Children's Hospital of Eastern Ontario (CHEO) and a pediatric endocrinologist within the Division of Endocrinology and Metabolism at CHEO. Dr Ward's research program is dedicated to the study of bone development and the treatment of bone disorders in children. She is the principal investigator of the "STOPP" research program (STeroid-induced Osteoporosis in the Pediatric Population), a pan-Canadian project funded by the Canadian Institutes of Health Research to study bone health in children with chronic illnesses. Dr Ward has received a number of awards for her work in pediatric bone health, including a Canadian Child Health Clinician Scientist Career Development Award (2004), a



Canadian Institutes for Health Research New Investigator Award (2004) and a Canadian Child Health Clinician Scientist Career Enhancement Award (2007).

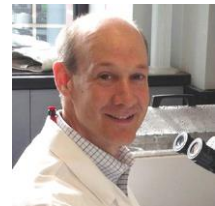
### **Matthew Warman**

Dr Warman is the Harriet M Peabody Professor of Orthopaedic Surgery and Genetics at Harvard Medical School. He attended college at Brown University and medical school at Cornell University. While in medical school, he performed research with Dr Adele Boskey at The Hospital for Special Surgery. After medical school he trained in Pediatrics at the Children's Hospital in Washington, DC, in Genetics at the Children's Hospital in Boston, and he performed post-doctoral research with Professor Bjorn R Olsen at Harvard Medical School. In 1994, Dr Warman established an independent laboratory and clinical program in the Department of Genetics and Center for Human Genetics at Case Western Reserve University and University Hospitals of Cleveland. In 2006, he returned to Boston to become director of the Orthopaedic Research Laboratories at Boston Children's Hospital. Dr Warman is also an investigator with the Howard Hughes Medical Institute. The patients and families, who Dr. Warman has come to know through his clinical work as a pediatrician and geneticist, have often served as the impetus for his research. In addition to working with patients and families, members of Dr Warman's lab try to understand and treat human disease by studying cultured cells, purified proteins, and other organisms. Having benefited from superb mentoring throughout his career, Dr Warman enjoys introducing students (from high school to professional school) to the importance and excitement of Human Genetics. He is proud to have mentored students at all levels, who have gone on to become excellent scientists, physicians, and educators.



### **Kenneth White**

Kenneth E White, PhD is the David D Weaver Professor of Genetics in the Department of Medical and Molecular Genetics at the Indiana University School of Medicine in Indianapolis, IN, USA. He serves as Director of the Division of Molecular Genetics and Gene Therapy. Dr White's research interests focus on the molecular genetics of metabolic bone diseases in regards to phosphate metabolism and control of FGF23. He is the recipient of the American Society for Bone and Mineral Research's 2007 Fuller Albright Award, and in 2013, Dr White was named an inaugural IU School of Medicine Showalter Scholar. He is also a member of the IU Simon Cancer Center.



### **Michael P Whyte**

Michael Whyte is Professor of Medicine, Pediatrics, and Genetics at the Washington University School of Medicine, a staff member of Barnes-Jewish Hospital and St. Louis Children's Hospital, and Medical-Scientific Director at the Center for Metabolic Bone Disease and Molecular Research, Shriners Hospital for Children in St. Louis, Missouri, USA.

Dr Whyte earned his MD degree at Downstate College of Medicine, State University of New York, Brooklyn, New York and then had internship and residency training in Internal Medicine at Bellevue Hospital in New York City. After two years as Clinical Associate at the National Institutes of Health, Bethesda, Maryland, he did his fellowship in the Division of Bone and Mineral Diseases and joined the medical faculty of the Washington University School of Medicine, St. Louis, USA.

Dr Whyte's research interests include especially the cause, outcome, and treatment of heritable disorders of bone and mineral metabolism in children and adults. Included are genetic forms of rickets such as hypophosphatasia and X-linked hypophosphatemia, brittle bone diseases like osteogenesis imperfecta, conditions that cause dense bones such as osteopetrosis, and disorders of accelerated skeletal turnover including juvenile Paget's disease. The Research Center at Shriners Hospital serves as a national and international resource for the diagnosis, treatment, and investigation of disorders of bone and mineral metabolism and skeletal dysplasias in children. Laboratory investigations include searches for the underlying mutated genes of new disorders. Phenotype/genotype



correlations aim to better understand the pathogenesis of established conditions. Bone-targeted alkaline phosphatase replacement therapy is being evaluated for pediatric patients with hypophosphatasia. Dr. Whyte has authored or coauthored more than 300 scientific papers or book chapters concerning these disorders.