Professor Nick Bishop is an internationally recognised expert in the field of paediatric bone research with a particular focus on experimental medicine/early phase studies in osteogenesis imperfecta, steroid-induced or disease-associated osteoporosis and hypophosphatasia, as well as having a long-standing interest in early life influences on later skeletal health. He is based at Sheffield Children's Hospital, a member of the BOND ERN, and the lead designated centre for the nationally-commissioned Highly Specialised Severe, Complex and Atypical Osteogenesis Imperfecta Service. He is an Associate Director of the Versus Arthritis Experimental Arthritis Treatment Centre for Children, leading the bone theme, immediate Past President of the Academic Paediatric Association of Great Britain and Ireland and Director of the Clinical Research Facility at Sheffield Children's Hospital.

Alison Boyce is a pediatric endocrinologist and Associate Research Physician in the Skeletal Diseases and Mineral Homeostasis Section, National Institute of Dental and Craniofacial Research, National Institutes of Health in Bethesda MD, USA. Dr Boyce's work focuses on bone and mineral metabolism, using rare and pediatric skeletal disorders as models through which to understand human biology and physiology. She leads investigations in fibrous dysplasia/McCune-Albright syndrome, a rare disease affecting the skeletal and endocrine systems, and has characterized many aspects of this disorder and its treatment. Additional studies focus on disorders of fibroblast growth factor-23 and primary connective tissue disorders. Dr Boyce is a faculty member in the joint NIH and Children’s National Pediatric Endocrinology fellowship training program. She holds an adjunct position at Children’s National Health System in Washington, D.C., where she attends in the multidisciplinary Bone Health Program in partnership with the Division of Orthopaedics. She is a member of the Fibrous Dysplasia/McCune-Albright Syndrome International Consortium and serves as a Medical Advisor to the Fibrous Dysplasia Foundation.

Professor Valérie Cormier-Daire is a medical geneticist (MD, PhD), Professor of Genetics (University Paris Descartes). She is at the head of the clinical genetics unit in Necker Enfants Malades Hospital and coordinator of the French Reference Center for Skeletal Dysplasia. She is also responsible for a team working on osteochondrodysplasia in INSERM Unit 1163, (Imagine Institute) since 1999. In the last ten years, the team has identified the genetic bases of more than 25 skeletal highlighting the crucial role of cilia, proteoglycan and TGFβ signaling in endochondral ossification. The team has developed appropriate cellular and mouse models to further study their function in the ossification process and develop therapeutic approaches. Her track-record includes 410 publications and 2 patents.

Professor Federico Di Rocco graduated from the Catholic University Medical School, Rome, in 2000 and has completed his neurosurgical training in the Neurosurgical Institute of the same university.

During his studies he attended the Brigham and Women’s Hospital, Children’s Hospital, Harvard Medical School laboratory, (Prof. P. Black), the Chicago Institute of Neurosurgery and Neuroresearch Laboratory, Northwestern University Medical School, Chicago (Prof D. McLone), the Jikei University of Tokyo (Prof S Oi) and the International Neuroscience Institute-Hannover (Prof M Samii) for further clinical and research education.

He obtained a PhD degree in Pediatric Neurosciences from the Catholic University Medical School, Rome and a Master Degree in Genetics from the University Paris 5.

After completing the residency in neurosurgery and the Postgraduate Training Course, of the European Society for Pediatric Neurosurgery he was offered a fellowship in pediatric neurosurgery at the hospital Necker Enfants Malades, Paris, led by Prof C. Saint-Rose in 2005 where he earned a permanent staff position a few years later.

He was nominated full Professor of Neurosurgery at the University of Lyon Claude Bernard in 2016

His main fields of interest are the congenital CNS malformations, antenatal treatment of spinal dysraphisms, craniosynostosis, hydrocephalus, crania-cervical junction anomalies, cerebral and spinal tumors, and traumas, in particular head injuries related to sport activities.

Prof Di Rocco is author of more than 160 articles published in peer reviewed international journals and chapters in neurosurgical textbooks.
François Fassier

Dr François Fassier was born and raised in France. He completed his medical school in Lyon and his orthopaedic residency in Grenoble and Paris. After a year of fellowship in pediatric orthopedic surgery at Ste Justine Hospital (University of Montreal), Dr Fassier went back to Grenoble as a young staff (Chef de clinique) for one year. In 1982, he immigrated to Canada and became a member of the Ste Justine Hospital staff until 1993.

He was then appointed at McGill University and became Chief of Orthopaedics at the Montreal Children’s Hospital (MCH). He played a role in the development of complementarity between the MCH and the Shriners Hospitals for Children® - Canada. Director of Pediatric Orthopaedic Surgery at McGill University, he became Chief of Staff of the Shriners Hospitals Canada in 2001 until September 2010. He is now Emeritus Chief of Staff, Shriners Hospitals Canada.

Dr Fassier has many points of interest in pediatric orthopaedics: he introduced the Ilizarov method for bone lengthening in Canada and developed a telescopic implant for children with bone fragility (The Fassier-Duval rod for Osteogenesis Imperfecta) and the Free-Gliding screw for young SCFE patients.

With Dr Francis H. Glorieux, he created a Multidisciplinary Clinic for OI patients which celebrated its 25th anniversary in 2017 which has been copied/imitated in numerous countries.

Antonella Forlino

Dr Antonella Forlino is Associate Professor of Biochemistry at the Department of Molecular Medicine, Unit of Biochemistry, University of Pavia. She has a PhD in Biochemistry and the Speciality in Genetics. She spent 5 years of post-doc training in NIH, Bethesda, USA.

Her research activity has been focused on the molecular, biochemical, and functional study of genetic diseases of the connective tissue in particular Osteogenesis Imperfecta (OI), using in vitro and in vivo models (mice and Zebrafish). She is combining basic science with translational approaches.

She is particularly interested on the intracellular effects of retained aberrant collagen type I in modulating the bone phenotype in both dominant and recessive OI.

Rudolf Ganger

Rudolf Ganger, MD, PhD, Associated Professor of Orthopaedics, Orthopaedic Hospital Vienna-Speising, Department of Paediatric Orthopaedics and Adult Foot and Ankle Surgery

Since 1989 working at the department of Paediatric Orthopaedics in Vienna - Speising (Head: Prof. Franz Grill). Since 2001 deputy of the head of the department. Since 2013 head of the department of Paediatric Orthopaedics and adult Foot and Ankle Surgery. 2007–2010 PhD-post gradual study at the Charles University Prague.

Topic: Computer assisted deformity correction using the Taylor Spatial Frame.

2012 Associated Professor of Orthopaedics

Clinical interests:
Paediatric orthopaedic surgery – especially hip joint and lower limb.
External fixation, limb lengthening, deformity correction and reconstruction surgery for congenital deficiencies.
Computer – assisted surgery (Taylor Spatial Frame, navigation systems, intramedullary lengthening nails).

Additional functions:
Board Member of the German Speaking Paediatric Orthopaedic Society (VKO).
Member of the Educational Board of EPOS (European Paediatric Orthopaedic Society), head of the Lower Limb Study Group.

Vicente Gilsanz

Vicente Gilsanz is a Professor of Radiology, Pediatrics, and Orthopaedic Surgery at the Keck School of Medicine of USC and Director of the Imaging Research Program at Children’s Hospital Los Angeles. He is board certified in internal medicine, diagnostic radiology, and pediatric radiology, and based on his training, his research centers on the use of imaging technologies to identify children at risk for common adult diseases, such as osteoporosis. He has extensive experience in utilizing digital data from all imaging modalities for the development of imaging biomarkers. For the past decades, he has collaborated with a multi-disciplinary group of individuals with an extensive expertise in imaging, pediatrics, bioengineering, and cell biology to investigate sex differences in bone growth and their lifelong implications. He has published more than 250 peer-reviewed publications, 16 book chapters, and one textbook.
Symposium Speaker Profiles

Ingo Grafe
Ingo Grafe, M.D., is a physician scientist currently working in the group of Brendan Lee, M.D., Ph.D. in the Department of Molecular and Human Genetics at the Baylor College of Medicine, Houston, Texas, USA. He received clinical training in the Department of Medicine I and Clinical Chemistry, University Clinic of Heidelberg, Germany, on the treatment of patients with disorders of the endocrine systems and with various bone disorders including Osteogenesis Imperfecta (OI). His research is focused on understanding the molecular mechanisms of bone disorders, with the goal to improve the treatment options for patients with bone diseases. In his research in OI mouse models he discovered that increased TGFβ signaling is an important contributor to the OI pathophysiology in both dominant and recessive forms of OI, and that treatment with a TGFβ-inhibiting antibody can improve the bone phenotype in OI mice.

Catherine Hawrylowicz
Catherine Hawrylowicz is Professor of Immune Regulation in Allergic Disease, Head of the Peter Gorer Department of Immunobiology and Co-Director Asthma UK Centre for Allergic Mechanisms in Asthma at King's College London. She trained in immunology at the National Institute for Medical Research (Mill Hill, London) and Washington University Medical School, St Louis, Missouri, USA. Her research investigates immunological mechanisms that ensure respiratory health, and how these change in chronic respiratory disease, including severe asthma and COPD. A major focus of the work is to understand the role of vitamin D, and intermediaries from vitamin D stimulated cells, in promoting respiratory health. The work has a translational focus, incorporating studies in blood and respiratory tissue, and investigating neonatal, pediatric and adult patient cohorts.

Wolfgang Högler
Professor Wolfgang Högler, Department of Paediatrics, Johannes Kepler University, Linz, Austria
Wolfgang Högler is Chief of Paediatrics at the Johannes Kepler University Hospital in Linz, Austria. He received paediatric & endocrine training at the Medical University Innsbruck, Austria and at The Children's Hospital at Westmead, Sydney, Australia. Following academic clinical appointments in Innsbruck and Birmingham, he was appointed chair in 2018.

Prof Högler’s research group focuses clinical and translational research, including mechanism of disease, novel diagnostic tools and treatments of rare bone and growth disorders such as osteogenesis imperfecta, hypophosphatasia, rickets as well as disorders of vitamin D and growth hormone metabolism. Prof. Högler chaired the 7th International Conference on Children’s Bone Health (ICCBH) and the Global Consensus for the Prevention and Management of Rickets. He organises postgraduate endocrine seminars for the Ipokrates Foundation, regularly hosts international fellows and enjoys mentoring young researchers.

Alex Ireland
Dr Alex Ireland is a Senior Lecturer in Physiology in the Musculoskeletal Science and Sports Medicine Research Centre at Manchester Metropolitan University. His work focuses on studies of bone and joint adaptation to mechanical loading in humans, particularly in the prenatal period and early childhood. Given the key role of muscular forces in skeletal loading, he is also interested in determinants of neuromuscular health and function across lifespan. Alex completed a three-year MRC-funded postdoctoral project leading a study examining the contribution of motor unit loss and remodelling to age-related loss of muscle mass and function. Recent work has also examined the effects of skeletal diseases on neuromuscular health and function, and development of novel exercise therapies for clinical populations.

Masanobu Kawai
Masanobu Kawai M.D., Ph.D. is a Chief Scientist at Research Institute for Osaka Women’s and Children’s Hospital (Osaka, Japan). Dr Kawai earned M.D. and Ph.D. from Osaka University Graduate School of Medicine (Osaka, Japan) and completed his postdoctoral fellowship at Maine Medical Center Research Institute (Maine, USA) under the mentorship of Clifford J Rosen. Dr Kawai now studies bone and mineral metabolisms, especially their regulation by circadian clock network using in vitro and in vivo models.

Klaus Klaushofer
Klaus Klaushofer is a Professor of Internal Medicine at the Medical University of Vienna, Austria. In 2018 he retired from both positions Head of the 1st Medical Department at Hanusch Hospital in Vienna and Director of the Ludwig Boltzmann Institute of Osteology, which he had been holding for 17 and 27 years, respectively. He is a member of the Austrian Advisory Committee for Public Health Issues, Consulting Physician of the Main Association of Austrian Social Insurance Institutions and a member of its Drug Evaluation Committee. His national and international cooperations and research activities have resulted in more than 330 original papers on various topics in osteology.
Symposium Speaker Profiles

Craig B Langman
Professor Craig B Langman, is the Isaac A Abt, MD, Professor of Kidney Diseases at the Feinberg School of Medicine, Northwestern University, Head of Kidney Diseases at the Ann and Robert H Lurie Children’s Hospital of Chicago. His research focuses on the basic and clinical expression of inherited or acquired disorders of calcium, phosphorus, vitamin D, and FGF23 metabolism, inherited genetic diseases (cystinosis, oxalosis, kidney stones, atypical HUS, hypophosphatasia), and the rehabilitation of patients around the world with chronic kidney disease. Professor Langman has published more than 235 articles, reviews and chapters in his discipline.

Agnès Linglart
Agnès Linglart is Professor of Paediatrics, Bicêtre Paris Sud Hospital, APHP and Paris-Sud University, Paris, France. Professor Linglart’s area of interest is paediatric endocrinology, particularly rare diseases affecting calcium and phosphate metabolism. She is the national coordinator of the French National Reference Centre for rare disorders of calcium and phosphate metabolism. Professor Linglart is also the chair of the strategic and finance committee of the European Society of Paediatric Endocrinology.

Outi Mäkitie
Professor Outi Mäkitie, Professor of Pediatric Endocrinology, Children’s Hospital, University of Helsinki, Helsinki, Finland
Prof. Outi Mäkitie received her MD degree at University of Helsinki in 1986 and completed specialty training in Helsinki for pediatrics in 1998 and for paediatric endocrinology in 2000. After a three-year clinical and research fellowship at the Hospital for Sick Children, Toronto, Canada in 2000-2003, Prof Mäkitie worked as the Head of Metabolic Bone Clinic, Division of Pediatric Endocrinology at the Children’s Hospital, University of Helsinki until 2013. In 2013-2015 she was visiting scientist at Karolinska Institutet, Sweden. Since 2015, she is Professor of Pediatric Endocrinology and Chief Physician at Children’s Hospital, University of Helsinki, Finland, and Visiting Professor of Pediatric Endocrinology at Karolinska Institutet, Sweden. For the academic year 2018-2019 she has joined Prof. Valérie Cormier-Daire’s research group at Imagine Institute, Descartes University, Paris, France. Prof. Mäkitie has carried out clinical and translational research on skeletal disorders and has published more than 260 scientific papers.

Zulf Mughal
Professor Zulf Mughal graduated in medicine from the University of Liverpool, UK, in 1978. He received postgraduate training in Paediatrics at hospitals in Manchester, Liverpool, in the UK and in Cincinnati, USA. He was appointed a Consultant Pediatrician in 1988. He is currently employed as a Consultant in Paediatric Bone Diseases at the Royal Manchester Children’s Hospital. In 2013, he was appointed Honorary Clinical Professor in Child Health at the University of Manchester. He has extensive experience in management of childhood bone & mineral disorders. Professor Mughal has published >160 peer reviewed scientific papers, invited reviews and book chapters. In June 2015, he was awarded the Charles Siemenda Award by ICBBH.

Noriyuki Namba
Noriyuki Namba, MD, PhD is a pediatric endocrinologist working at Osaka Hospital, Japan Community Healthcare Organization (JCHO). He received his MD and PhD from Okayama University and completed pediatric residency at Okayama University Hospital, followed by a research fellowship in bone cell biology at Washington University. Upon returning to Japan, he undertook a pediatric endocrinology fellowship at Okayama University Hospital. He subsequently served as associate professor of pediatrics at Osaka University, acting as principal investigator or co-researcher in chondrocyte biology research. After relocating to Osaka Hospital as chief of pediatrics, his main interest in recent years has been clinical research concerning patients with disorders of calcium/phosphate metabolism as well as skeletal dysplasias. He continues to serve as visiting associate professor of pediatrics at Osaka University and is the chair of the Bone and Mineral Metabolism Committee of the Japanese Society for Pediatric Endocrinology.

Dr Niamh Nowlan is Reader in Developmental Biomechanics in the Department of Bioengineering at Imperial College London. The goal of her laboratory is to understand how fetal and postnatal movements direct skeletal development, including that of the joints, spine and bone. This research is advancing our understanding of congenital musculoskeletal conditions and informing tissue engineering and regenerative medicine approaches. Dr Nowlan has received a number of prestigious awards, including a Fulbright Scholarship, a New Investigator Recognition Award from the Orthopaedic Research Society, and was senior author on the 2018 S.M. Perren Award-winning paper of the European Society of Biomechanics. Dr Nowlan’s research is funded by an ERC Starting Grant, and grants from the Leverhulme Trust, the Anatomical Society and the Royal Society.
Frank Rutsch

Frank Rutsch, MD, is a consultant and Professor in Pediatrics at Münster University Children’s Hospital, Münster, Germany. He graduated from Münster University Medical School in 1992 and took part in the Pediatric residency program in Dresden and Dortmund, Germany. After spending a postdoctoral research fellowship at the Department of Rheumatology/Immunology, UCSD, San Diego, USA, he became the leader of an independent research group at Münster University Children’s Hospital, Germany in 2004. His main research interests are focused on the discovery of the underlying genetic defects and on translational aspects in rare Pediatric metabolic and autoimmune disorders. He is co-chair of the metabolic department of Münster University Children’s Hospital and vice chair of the Center for Rare Diseases of Münster University Hospital. He runs an outpatient clinic for children and adults with rare metabolic disorders including connective tissue diseases.

Oliver Semler

Oliver Semler is head of the department of rare skeletal diseases in childhood at the university Cologne, Germany.

PD Dr. Semler studied medicine at the universities in Cologne and Freiburg and completed his thesis on ‘Surgical treatment of femur fractures in Osteogenesis imperfecta’ at the university Heidelberg and finished his PhD in paediatrics with the thesis ‘Translationale research in Osteogenesis imperfecta: From Pathophysiology to personalized treatment’.

PD Dr. Semler is certified as paediatric rheumatologist and focusses clinical and scientifically on different skeletal dysplasias. In this topic a main interest is the improvement and quantification of mobility and muscle function. He is Principal investigator in a number of clinical trials and partner in international research networks. He published more than 70 peer-reviewed articles and book chapters and was awarded with the „Young Investigator Award“ of the „European Society of Pediatric Endocrinology“ and with the „Eva Luise Köhler Forschungspreis für Seltene Erkrankungen“.

Nick Shaw

Nick Shaw is a Consultant Paediatric Endocrinologist at Birmingham Children’s Hospital and Honorary Professor, Institute of Metabolism and Systems Research at the University of Birmingham. He developed an interest in paediatric calcium and bone metabolism whilst a Lecturer at the Universities of Leeds and Liverpool between 1985 and 1992 before completing his endocrine training in Birmingham where he has been a consultant since 1994. He established a service for children with metabolic bone disease in 1996 which in 2011 was designated as a national centre for complex childhood Osteogenesis Imperfecta.

He organises a postgraduate training course in paediatric calcium and bone metabolism and is the co-editor of the book “Calcium and Bone Disorders in Children and Adolescents” published in 2009 and 2015.

He was a founder member of the British Paediatric and Adolescent Bone Group and the Bone Club of the European Society for Paediatric Endocrinology.

Cristina Sobacchi

Dr. Cristina Sobacchi graduated in Pharmaceutical Chemistry and Technologies, University of Milan (1998), with a Training in Molecular Biology. She is CNR Tenured Researcher since 2010, and Junior Principal Investigator at the Humanitas Clinical and Research Center (Milan, Italy).

Her main interest are molecular and cellular mechanisms involved in bone pathophysiology. She contributed to identifying several genes responsible for human osteopetrosis. She has also carried out different studies aimed at the development of new therapies for osteopetrosis in animal models, by using pharmacological, cell-based and gene therapy approaches. Specifically, she has a longstanding interest in the investigation of bone pathophysiological mechanisms and therapies related to RANKL cytokine.

Another recent topic of her research is the role of oxidative stress in bone pathophysiology.

She is Member of the European Calcified Tissue Society (ECTS), where she has been actively serving in different Committees, and Member of the Forum in Bone and Mineral Research. She has been invited speaker to several National and International Scientific Meetings in the field.

She is coauthor of 61 papers in peer-reviewed journals in which she features mainly as first or last author.

Jonathan Tobias

Jonathan Tobias is Professor of Rheumatology at the University of Bristol, UK, and Consultant Rheumatologist at North Bristol Trust. Following undergraduate studies in medicine at Cambridge University and London University from where he qualified in 1984, he completed MD and PhD theses in bone biology in 1990 and 1994, at St George’s Hospital in London. He was appointed as Consultant Senior Lecturer at the University of Bristol in 1995. He manages a diverse research programme into the causes and treatment of musculoskeletal conditions, particularly osteoporosis, with over 240 publications (including 180 peer-reviewed original research papers) in this field. He also has extensive clinical experience in treating patients with osteoporosis, and in running DXA-
Symposium Speaker Profiles

Leanne Ward
Dr Leanne Ward is an Associate Professor of Pediatrics at the University of Ottawa where she has held a Research Chair in Pediatric Bone Health since 2010. She is the Medical Director of the Pediatric Bone Health Clinical and Research Programs at the Children’s Hospital of Eastern Ontario. Dr Ward’s research program is dedicated to the study of bone development and the treatment of pediatric bone disorders, with particular emphasis on bone health in children with chronic illnesses. She has served as an advisor to numerous national and international organizations on various aspects of skeletal health in children, including the Centres for Disease Control Clinical Care Guidelines for Duchenne Muscular Dystrophy. 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, a Canadian Institutes for Health Research New Investigator Award and a Canadian Child Health Clinician Scientist Career Enhancement Award. She has published over 90 papers in peer-reviewed journals, and has delivered over 150 lectures at national and international scientific conventions.

Matt 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. 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.

Hope Weiler
Dr Hope Weiler is a Professor and Registered Dietitian in the School of Human Nutrition in the Faculty of Agricultural and Environmental Sciences at McGill University and recently has joined Health Canada as a Research Scientist. Dr Weiler’s research focus is on fat soluble vitamins, including vitamin D and musculoskeletal health. To date, Dr Weiler has authored over 170 peer-reviewed publications and provided graduate training to over 80 graduate trainees and 50 undergraduate students. Dr Weiler has been awarded prestigious awards from the Canada Research Chairs program, the New Investigator Award from the Canadian Nutrition Society and the Wiebe Visser International Nutrition Dairy Prize from the International Dairy Federation for her work in nutrition and musculoskeletal health. She has served numerous societies, organizations and areas of nutrition. Dr Weiler is a member of the editorial boards of Nutrition Research and The Journal of Nutrition.