

Meet the Experts

Nick Bishop

Nick Bishop is Professor of Paediatric Bone Disease at the University of Sheffield and Sheffield Children's Hospital. His clinical and research work focuses mainly on children with osteogenesis imperfecta, osteoporosis and inherited forms of rickets, as well as research into these conditions.

He is Director of the Clinical Research Facility at Sheffield Children's Hospital and Associate Director of the Arthritis Research UK-funded Experimental Arthritis Treatment Centre for Children, leading the Bone Health theme. He was elected President of the Academic Paediatric Association of Great Britain and Ireland in 2015.



Alison Boyce

Dr Alison Boyce is a pediatric endocrinologist and staff physician scientist in the Skeletal Clinical Studies Unit, National Institute of Dental and Craniofacial Research, National Institutes of Health (NIH). After completing undergraduate studies at the University of Virginia, she received medical and general pediatrics training at Eastern Virginia Medical School, followed by an endocrinology fellowship at the NIH. She holds a faculty appointment at Children's National Health System, and is active in the joint NIH and Children's National Health System's Pediatric Fellowship training program, as well as the Children's National Bone Health Program. Dr. Boyce's research focuses on rare disorders of bone and mineral metabolism, including fibrous dysplasia/McCune-Albright syndrome, disorders of fibroblast growth factor-23, and hypoparathyroidism.



Nicola Crabtree

Dr Nicola Crabtree is a Principal Clinical Scientist presently employed as a Research Clinical Scientist based at Birmingham Children's Hospital. She has had an interest in bone research for over 20 years. However, for the last 17 years her research has concentrated predominately on bone development in the healthy child and in children with chronic diseases. As part of this work, in 2007 she completed her PhD thesis "Interpretation of Paediatric Bone Evaluation by DXA". In 2010 she was awarded a NIHR Research fellowship to prospectively evaluate fracture risk in children with chronic inflammatory and/or disabling conditions. In addition to the prospective study Dr Crabtree was also awarded an Arthritis Research-UK grant to collate bone density data in 3500 healthy children from across the UK, including data from Sheffield, London, Manchester, Birmingham Leeds and Glasgow. The overriding theme of her research is the development of imaging techniques which can improve the diagnosis of poor bone health and increased fracture risk in chronically sick children. However, she has a special interest the muscle-bone relationship and how disruption of this relationship affects bone development in both a paediatric and an aging population.



Rachel Gafni

Rachel I Gafni received her BA from Barnard College and her MD from Temple University. She completed a pediatric residency at the Children's Hospital of Philadelphia followed by a pediatric endocrinology fellowship at the National Institutes of Health (NIH), serving as an officer in the Public Health Service from 1996-2002. She subsequently served as an Assistant Professor at the University of Maryland. Dr. Gafni returned to NIH in 2007 as a staff clinician in the National Institute of Dental and Craniofacial Research. She is also faculty in the NIH Pediatric Endocrinology and NIH Inter-institute Adult Endocrinology Training Programs. She is an investigator on several protocols studying and treating patients with endocrine disorders including hypoparathyroidism, McCune-Albright Syndrome, hypophosphatemic rickets, hyperphosphatemic familial tumoral calcinosis, generalized arterial calcification of infancy, and other metabolic bone diseases.



Heike Hoyer-Kuhn

Dr Heike Hoyer-Kuhn is paediatrician at the Children's Hospital, University of Cologne, Germany. She is member of the Paediatric Bone Dysplasia Outpatient Department and was trained in a fellowship for pediatric endocrinology and osteology by Professor Eckhard Schoenau and PD Dr Oliver Semler.

Dr Hoyer's interest in clinical care is focused on children and adolescents with Osteogenesis Imperfecta and skeletal dysplasia. With her background of pediatric nephrology she is well experienced in all diseases affecting calcium and phosphate homeostasis. Her main clinical research interest includes evaluating new drug treatment approaches within clinical trials. Based on the large sample size of patients in the outpatient department she is involved in basic research deciphering the molecular causes and pathophysiology of the disease by identifying clinical differences between classical collagen associated types and rare subtypes of skeletal diseases.



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.



Mary Leonard

Dr Leonard is a Professor of Pediatrics and Medicine and Director of the Bone Health and Nutrition Research Center at Stanford. Her multidisciplinary research program is focused on the impact of childhood chronic diseases on bone quality and muscle function across the life course, with an emphasis on the effects of glucocorticoid therapy, inflammation and chronic kidney diseases. She has served in leadership roles on international committees to develop clinical practice guidelines for the assessment of bone health in children, and the management of metabolic bone disease in children and adults with chronic kidney disease.



Agnès Linglart

Agnès Linglart, MD, PhD, is Professor of Pediatrics at the medical school of Paris Sud University (France) and the head of the french Reference center for rare disorders of calcium and phosphorus metabolism at Bicêtre-Paris-Sud Hospital. Dr. Linglart was a Postdoctoral Fellow at Endocrine Unit at Massachusetts General Hospital and Harvard Medical School, Boston, USA. She is involved in the management of children with endocrine disorders and bone



diseases, including therapeutic studies involving patients with in hypophosphatemic rickets, osteoporosis, hypoparathyroidism and growth disorders. Her research activities are focused on the mechanisms of parathormone resistance and parental imprinting.

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 Paediatrician in 1988. He is currently employed as a Consultant in Paediatric Bone Disorders at the Royal Manchester Children's Hospital. In 2013, he was appointed Honorary Clinical Professor in Child Health at the University of Manchester. He is a Fellow of the Royal College of Physicians and a Fellow of the Royal College of Paediatrics and Child Health. He has extensive experience in management of childhood bone & mineral disorders. Professor Mughal has published >150 peer reviewed scientific papers, invited reviews and book chapters.



Oliver Semler

PD Dr Oliver Semler is Head of the Paediatric Bone Dysplasia Outpatient Department at the Children's Hospital, University of Cologne, Germany. He also works in a paediatric rehabilitation centre for children with motor deficits, where he focuses on the diagnosis and evaluation of motor function in disabled children. Dr Semler's interest in clinical care is focused on children and adolescents with Osteogenesis Imperfecta. He is involved in basic research elucidating the molecular causes of the disease and developing new drug treatments. Additionally he participates in clinical research investigating new diagnostic strategies and evaluating the effect of different therapeutic concepts in patients with Osteogenesis Imperfecta.

