



Photo: Jean-Marc Good

# Genetic Medicine: developments and perspectives

**Vendredi 1er décembre  
13.00 - 19.00**

**CHUV**

**Auditoire Jequier-Doge  
*entrée libre***

Symposium pour le départ de  
Sheila Unger et Andrea Superti-Furga

Ce petit symposium en anglais, organisé pour le départ de la Pr Sheila Unger et du Pr Andrea Superti-Furga, fait le point sur quelques aspects actuels de recherche et de pratique clinique en médecine génétique avec des orateurs prestigieux. Nous serons ravis de vous accueillir pour un moment scientifique et amical!

Christel Tran et François Clergue, Service de médecine génétique, CHUV

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# Programme

<b>Manuel Pascual</b>	Professor and Dean of Faculty; Faculty of Biology and Medicine, University of Lausanne, CH	<b>Welcome and Opening remarks</b>
<b>Sheila Unger</b>	former Associate Professor, University of Lausanne; Genetica AG, Lausanne, CH	<b>Introduction</b>
<b>Christine Hall</b>	emerita Professor of Radiology, Great Ormond Street Hospital, London, UK	<b>The future of radiology, or: does radiology have a future?</b>
<b>Gen Nishimura</b>	former Director, Department of Radiology, Tokyo Metropolitan Children's Medical Center; Radiology Department, Musashino-Yowakai Hospital, Tokyo, Japan	<b>A review of the history of bone dysplasias from a radiologist's viewpoint</b>
<b>Dan Cohn</b>	Professor of Molecular, Cell, and Developmental Biology, and Orthopaedic Surgery; UCLA, Los Angeles, USA	<b>Trespassing in the genome: Gene discovery in the skeletal dysplasias</b>
<b>Antonio Rossi</b>	Professor of Biochemistry, Department of Molecular Medicine, University of Pavia, IT	<b>Intracellular sulfate – so simple, so important, so difficult to provide</b>
<b>Francesco Ramirez</b>	Professor of Pharmacological Sciences, Orthopedics, Medicine and Cardiology; Icahn School of Medicine at Mount Sinai Hospital, New York, USA	<b>TGF<math>\beta</math> signaling in Marfan syndrome: a tale of two stories : the long side</b>
<b>Valérie Cormier-Daire</b>	Professeur de Genetique, Université Paris Descartes, INSERM, AP-HP ; Institut IMAGINE et Hôpital Necker-Enfants Malades, Paris, FR	<b>TGF<math>\beta</math> and Fibrillin pathways: the short side</b>
<b>Carlos Ferreira</b>	Staff Clinician, Metabolic Medicine Branch, and Head, Skeletal Genomics Unit, NHGRI/NIH, Bethesda, USA	<b>Genomics, models and therapeutic targets of genetic bone diseases</b>
<b>Livia Garavelli</b>	Director, Medical Genetics Unit, Obstetrics and Pediatrics Department, AUSL IRCCS Reggio Emilia, Italy	<b>The importance of networking in diagnosing rare disorders</b>
<b>*** Coffee break ***</b>		
<b>Marco Tartaglia</b>	Head, Molecular Genetics and Functional Genomics, Ospedale pediatrico Bambino Gesù, Rome, Italy; and Dept. of Pediatrics, Mount Sinai School of Medicine, New York, USA	<b>From Noonan syndrome to the Rasopathies – a long and winding pathway</b>
<b>Carlo Rivolta</b>	Professor for Ophthalmic Genetics, University of Basel, CH; Professor of Medical Genetics, Department of Genetics and Genome Biology, University of Leicester, UK	<b>Genetic bioinformatics from bench to bedside: a fruitful exchange</b>
<b>Matt Warman</b>	Professor of Genetics, Harvard Medical School, and Department of Orthopedics, Boston Children's Hospital Precision Medicine Service, Boston, USA	<b>Efficient in vivo testing for variants of uncertain significance (VUS)</b>
<b>Marc Tischkowitz</b>	Professor and Head of Department, Department of Medical Genetics, University of Cambridge, UK	<b>Polygenic – no longer Pollyanna?</b>
<b>Christel Tran</b>	Privat-Dozent and Head of the Metabolic Disease Unit, Division of Genetic Medicine, Lausanne University Hospital, Lausanne, CH	<b>Application of molecular therapies in genetic and metabolic disorders</b>
<b>Luisa Bonafé</b>	former Associate Professor, University of Lausanne; Head, Pediatric Medical Team, Regional Medical Services, National Invalidity Insurance, Vevey, CH	<b>Molecular treatments for rare genetic diseases: a challenge for the social insurance system</b>
<b>Marc Abramowicz</b>	Professor and Head, Division of Genetic Medicine, HUG et UniGE, Geneva, CH	<b>Look back, look forward - the perspective of a neighbour and friend</b>
<b>Andrea Superti-Furga</b>	Professor emeritus, University of Lausanne and Lausanne University Hospital; Genetica AG, Lausanne, CH	<b>Thanks and farewell</b>

**\*\*\* Apéritif \*\*\***



sponsorship gratefully acknowledged:

