

13:30 – 14:30	Room: Topkapi A	FC8
Session type:	Free Communication Session 8	
Session title:	Genetics of Growth	
Moderators:	Jesús Argente, Madrid, Spain Serge Amselem, Paris, France	

Heterozygous defects of the acid-labile subunit gene in idiopathic short stature FC8-110

Alessia David¹; Farideh Miraki-Moud¹; A. Louise Metherell¹; Jenny Jones²; Ray Edwards³; Cecilia Camacho-Hübner⁴; B. Linda Johnston¹; O. Martin Savage¹; J.L. Adrian Clark¹

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Acid-labile subunit gene mutations: clinical, biochemical and molecular study in two unrelated families FC8-111

Alessia David¹; Steve Rose²; Farideh Miraki-Moud³; Louise Metherell³; Adrian Clark³; Martin Savage³; Cecilia Camacho-Hübner⁴

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Promoter Haplotype in insulin-like growth factor-binding protein-3 (IGFBP3) gene: correlation with Serum levels, growth and response to growth hormone treatment in short children born small for gestational age (SGA) FC8-112

Danielle van der Kaay¹; Anita Hokken-Koelega²; Emile Hendriks²; Sandra de Kort²; Ruben Willemsen²; Wietske Ester²; Ralph Leunissen²; Jean Paquette¹; Cheri Deal¹

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Novel SOX3 mutations in patients with various forms of syndromic pituitary anomalies FC8-113

Irina Giurgea¹; Kalotina Machinis²; Marie-Pierre Vié-Luton¹; Graziella Pinto³; Brigitte Mignot⁴; Anne-Marie Bertrand⁴; Catherine Naud-Saureau⁵; Sophie Rose²; François Kurtz⁶; Marie Legendre²; Marie-Laure Sobrier²; Julianne Léger⁷; Pierre Czernichow⁷; Serge Amselem²

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Screening for CUL7 mutations in those born small for gestational age (SGA)

FC8-114

Daniel Hanson¹; Gudrun Moore²; Sayeda Abu-Amero²; Peter Clayton³;Graeme Black⁴¹University of Manchester, Medical Genetics, Manchester, United Kingdom; ²Institute of Child Health, Clinical and Molecular Genetics, London, United Kingdom;³University of Manchester, Endocrine Science Research Group, Manchester, UnitedKingdom; ⁴University of Manchester, Medical Genetics, Manchester, United Kingdom**A novel mutation in GH molecule (GH-R178H) affecting the correct Zn²⁺-induced dimerization and condensation in secretory granules presented in a patient with GH deficiency**

FC8-115

Vibor Petkovic¹; Michela Godi²; Didier Lochmatter¹; James Turton³;Sandy Alatzoglou³; Mehul Dattani³; Christa Fluck¹; Primus Mullis¹¹University Children's Hospital, Pediatric Endocrinology, Bern, Switzerland;²University of Eastern Piedmont, Department of Medical Sciences, Novara, Italy;³Institute for Child Health, Biochemistry, Endocrinology and Metabolism Unit, London, United Kingdom