
Simeon Boyadjiev Boyd, M.D.

Boyd, Simeon Boyadjiev, M.D. Children's Miracle Network Endowed Chair, Associate Professor and Chief of the Section of Genetics, Department of Pediatrics, School of Medicine.

Biography

Simeon Boyadjiev Boyd, M.D., is a medical geneticist and pediatrician, who joined UC Davis and the M.I.N.D. Institute in June 2006, accepting a position in the Department of Pediatrics as Children's Miracle Network Endowed Chair and Chief of the Section of Genetics. Prior to this appointment, Dr. Boyd was a member of the research faculty of the McKusick-Nathans Institute of Genetic Medicine at Johns Hopkins University. His prior achievements include identification of the gene responsible for Oculo-Dento-Digital dysplasia; establishing a multi-institutional project focused on the analysis of craniosynostosis and bladder exstrophy; and the delineation of several novel dysmorphic syndromes. His laboratory is currently involved in studies of non-Mendelian (multifactorial) birth defects, using approaches that can be applied to the genetic analysis of other complex traits, i.e. autism, developmental delays, and learning disabilities. His ultimate goal is to identify genes and environmental factors contributing to the risk of these disorders. Dr. Boyd is sharing his expertise in analysis of complex traits with other M.I.N.D. Institute researchers interested in these neurobehavioral phenotypes. Dr. Boyd's laboratory is also involved in identification and characterization of genetic syndromes due to defects of the intracellular secretory pathway, and has recently identified and characterized one such syndrome, Cranio-Lenticulo-Sutural dysplasia. Using classical and reverse genetic approaches he plans to identify and characterize other human disorders caused by defects of this pathway. In addition to his research activities, Dr. Boyd directs the clinical activities of the Section of Genetics that provides campus-wide clinical services. The clinical genetics service at the UC Davis Medical Center evaluates infants, children and adults who have or may be at risk for genetic conditions such as birth defects, chromosome abnormalities, mental retardation and developmental delays, muscular dystrophies, inherited skeletal conditions and metabolic disorders. Among the recent initiatives of the Section of Genetics is the initiation of Lysosomal Storage Disease Center that provides enzyme replacement infusion therapy for patients with storage disorders.

Publications

Boyadjiev SA for the International Craniosynostosis Consortium. Genetic analysis of non-syndromic craniosynostosis. *Orthod Craniofacial Res* 10(3):129-137, 2007.

Reutter H, Qi L, Gearhart JP, Boemers T, Ebert A, Utsch B, Rösch W, Ludwig M, **Boyadjiev SA**. Concordance analyses of twins with bladder exstrophy-epispadias complex suggest genetic etiology. *Am J Med Genet* 143A:2751-2756, 2007.

Fromme JC, Ravazzola M, Hamamoto S, Al-Balwi M, Eyaid W, **Boyadjiev SA**, Cosson P, Schekman R, Orci L. The genetic basis of a craniofacial disease provides insight into COPII coat assembly. *Dev Cell* 13(5) 623-634, 2007.

Gambhir L, Höller T, Müller M, Schott G, Vogt H, Detlefsen B, Ebert AK, Fisch M, Beaudoin S, Stein R, **Boyadjiev SA**, Rösch W, Utsch B, Boemers TM, Reutter H, Ludwig M. Epidemiological survey of 214 European families with Bladder Exstrophy-Epispadias Complex (BEEC). *J Urol* 179(4) 1539-1543, 2008.

Presentations

From the clinic to the bench (and back): Developmental phenotypes of ER export Defects, Guest Lecturer, PHA 250 Functional Genomics Course, University of California Davis, May 2007.

Genetics of Craniofacial Development and Dysmorphology, Invited Speaker and Faculty AO, North America Challenges and Advances in the Management of Craniomaxillofacial Surgery, San Francisco, CA, July 2007.

Medical genetics insight into COPII-mediated intracellular trafficking, Invited speaker, Research Series of Shriners Hospital for Children, Sacramento, CA, November 2007

Genetic Analysis of Nonsyndromic Craniosynostosis, Neurosurgery Grand Round, Department of Neurosurgery, University of California Davis, February 2008

Genetic Analysis of Skull Defects, Medical Genetics Grand Rounds, Department of Pediatrics, Stanford University, CA, June 2008

Research Funding

Principal Investigator: Nonsyndromic craniosynostosis: Phenotype/Genotype Study, NIDCR-NIH, 07/01/06 to 06/30/11, \$347,000 annual direct. *The main goal fo this project is to identify the genetic causes on nonsyndromic craniosynostosis and to furhter define the phenotype and the long-term outocmes of this congenital anomaly.*

Principal Investigator: Genetic Analysis of Bladder-Exstrophy-Epispadias Complex, Children's Miracle Network Research Career Development Award, 07/2007 to 06/2009, \$75,000 annual direct. *The aim of this project is to identify candidate genes for bladder exstrophy utilizing expresion studies.*

Community Service

Journal Reviewer: *American Journal of Human Genetics, American Journal of Medical Genetics, European Journal of Human Genetics, Human Mutation, The Cleft Palate-Craniofacial Journal, Molecular Medicine*

Grant reviewer: NIH Special Emphasis Panel, National Institute of Dental and Craniofacial research, Research Grants Council, Hong Kong, China; Ad-hoc Reviewer, NIH, Skeletal Biology Development and Disease Study Section (SBDD)

Awards and Honors

Past-president, Society of Craniofacial Genetics
Children's Miracle Network Endowed Chair
Dean's Nominee for Provost Fellow, UC Davis