



Simeon Boyd, M.D.

Clinical Interests	Dr. Boyd's clinical interests focus on: dysmorphology; delineation of new syndromes; congenital anomalies - craniosynostosis, oral clefts, facial asymmetry syndromes, exstrophy-epispadias complex; and lysosomal storage disorders. His research interests focus on: clinical and molecular dysmorphology; genes causing congenital defects; genetic analysis of multifactorial diseases; genetic epidemiology; and disorders of intracellular trafficking. Philosophy of Care: Dr. Boyd's mission is to further the understanding of human heredity and genetic medicine, and to use that knowledge to treat and prevent disease.
Title	Professor of Pediatrics
Specialty	Pediatric Genetics and Genomic Medicine
Department	Pediatrics
Division	Genomic Medicine
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Languages	Bulgarian, Russian
Education	M.D., Leningrad Pediatric Medical Institute, Russia, Leningrad, Russia, 1985
Internships	State University of New York (SUNY), New York, New York, 1996
Residency	State University of New York (SUNY), New York, New York, 1997
Fellowships	The Johns Hopkins University, McKusick-Nathans Institute of Genetic Medicine, Baltimore, Maryland, 1997-2000
Board Certifications	American Board of Medical Genetics - Clinical Genetics (M.D.), 1999 American Board of Pediatrics, 1997
Professional Memberships	American Academy of Pediatrics



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American Society for Cell Biology

American Society of Human Genetics

European Society of Human Genetics

Society of Craniofacial Genetics, President (2002-2004)

Society of Craniofacial Genetics, Vice President (2000-2002)

Select Recent Publications

Paznekas WA, Karczeski B, Vermeer S, Lowry RB, Delatycki M, Laurence F, Koivisto PA, Van Maldergern L, Boyadjiev SA, Bodurtha IN, Wang Jabs E. GJAI mutations, variants, and connexin 43 dysfunction as it relates to the oculodentodigital dysplasia phenotype. *Hum Mutat.* 30(5):724-33. 2009.

Ludwig M, Ching B, Reutter H, Boyadjiev SA. Bladder exstrophy-epispadias complex. *Birth Defects Res A Clin Mol Teratol.* 85(6):509-22. 2009

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Boyadjiev SA, Fromme JC, Nauta C, Hur DJ, Zhang G, Schekman R, Orci L, Eyaid W. Cranio-lenticulo-sutural dysplasia is caused by a SEC23A mutation leading to abnormal ER-to-Golgi trafficking. *Nat Genet*, 38(10):1192-1197, 2006.

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Simeon Boyd, M.D.

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- Hur DJ, Raymond GV, Kahler SG, Riegert-Johnson DL, Cohen BA, Boyadjiev SA. A novel MGP mutation in a consanguineous family: Review of the clinical and molecular characteristics of Keutel syndrome. *Am J Med Genet*, 135(1):36-40, 2005.
- Jehee FS, Johnson D, Alonso LG, Cavalcante DP, de Sa Moreira E, Alberto FL, Kok F, Kim C, Wall SA, Jabs EW, Boyadjiev SA, Wilkie OA, Passos-Bueno MR. Molecular screening for microdeletions at 9p22-p24 and 11q23-q24 in a large cohort of patients with trigonocephaly. *Clin Genet*, 67(6): 503-510, 2005.
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- Boyadjiev SA, South ST, Radford CL, Patel A, Zhang H, Hur D, Thomas GH, Gearhart GP, Stetten G. A reciprocal translocation 46,XY,t(8;9)(p11.2;q13) in a bladder exstrophy patient disrupts CNTNAP3 and presents evidence of a pericentromeric duplication on chromosome 9. *Genomics*, 85(5):622-629, 2005.
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- Boyadjiev SA, Justice CM, Eyaid W, McKusick VA, Lachman RS, Chowdry AB, Jabak M, Zwaan J, Wilson AF, Jabs EW. A novel dysmorphic syndrome with open calvarial sutures and sutural cataracts maps to chromosome 14q13-q21. *Hum Genet*, 113:1-9, 2003.

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