



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, Department of Pediatrics
Specialty	Pediatrics
Department	Pediatrics
Division	General Pediatrics
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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 NY 1996
Residency	State University of New York (SUNY), New York NY 1997
Fellowships	The Johns Hopkins University, McKusick-Nathans Institute of Genetic Medicine, Baltimore MD 1997-2000
Board Certifications	American Board of Medical Genetics - Clinical Genetics (M.D.) American Board of Pediatrics
Professional Memberships	American Academy of Pediatrics American Society for Cell Biology



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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

Calpena E, Cuellar A, Bala K, Swagemakers SMA, Koelling N, McGowan SJ, Phipps JM, Balasubramanian M, Douzgou S, Morton JEV, Shears D, Weber A, Wilson LC, Lord H, Lester T, Johnson D, Wall SA, Twigg SRF, Mathijssen IMJ, Cunningham ML, Genomics England Research Consortium, Boyadjiev SA, Wilkie AOM. SMAD6 variants in craniosynostosis: genotype and phenotype evaluation. *Genet Med*. 2020 Jun 5;22(9):1498-1506. doi:10.1038/s41436-020-0817-2. Online ahead of print.

Cuellar A, Bala K, Di Pietro L, Barba M, Yagnik G, Liu JL, Stevens C, Hur DJ, Ingersoll RG, Justice CM, Drissi H, Kim J, Lattanzi W, Boyadjiev SA. Gain-of-function variants and overexpression of RUNX2 in patients with nonsyndromic midline craniosynostosis. *Bone*. 2020 Aug;137:115395. doi:10.1016/j.bone.2020.115395. Epub 2020 Apr 30. PMID:32360898.

Justice CM, Cuellar A, Bala K, Sabourin JA, Cunningham ML, Crawford K, Phipps JM, Zhou Y, Cilliers D, Byren JC, Johnson D, Wall SA, Morton JEV, Noons P, Sweeney E, Weber A, Rees KEM, Wilson LC, Simeonov E, Kaneva R, Yaneva N, Georgiev K, Bussarsky A, Senders C, Zwienenberg M, Boggan J, Roscioli T, Tamburrini G, Barba M, Conway K, Sheffield VC, Brody L, Mills JL, Kay D, Sicko RJ, Langlois PH, Tittle RK, Botto LD, Jenkins MM, LaSalle JM, Lattanzi W, Wilkie AOM, Wilson AF, Romitti PA, Boyadjiev SA; National Birth Defects Prevention Study. A genome-wide association study implicates the BMP7 locus as a risk factor for nonsyndromic metopic craniosynostosis. *Hum Genet*. 2020 Aug;139(8):1077-1090. doi:10.1007/s00439-020-02157-z. Epub 2020 Apr 7. PMID:32266521.

Schiffmann R, Goker-Alpan O, Holida M, Giraldo P, Barisoni L, Colvin RB, Jennette CJ, Maegawa G, Boyadjiev SA, Gonzalez D, Nicholls K, Tuffaha A, Atta MG, Rup B, Charney MR, Paz A, Szlaifer M, Alon S, Brill-Almon E, Chertkoff R, Hughes D. Pegunigalsidase alfa, a novel PEGylated enzyme replacement therapy for Fabry disease, provides sustained plasma concentrations and favorable pharmacodynamics: A 1-year Phase 1/2 clinical trial. *J Inher Metab Dis*. 2019 May;42(3):534-544. doi:10.1002/jimd.12080. Epub 2019 Apr 8. PMID:30834538.



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Barba M, Di Pietro L, Massimi L, Geloso MC, Frassanito P, Caldarelli M, Michetti F, Della Longa S, Romitti PA, Di Rocco C, Arcovito A, Parolini O, Tamburrini G, Bernardini C, Boyadjiev SA, Lattanzi W. BBS9 gene in nonsyndromic craniosynostosis: Role of the primary cilium in the aberrant ossification of the suture osteogenic niche. *Bone*. 2018 Jul;112:58-70. doi:10.1016/j.bone.2018.04.013. Epub 2018 Apr 17. Erratum in: *Bone*. 2019 Apr;121:293. PMID:29674126.

Lee E, Le T, Zhu Y, Elakis G, Turner A, Lo W, Venselaar H, Verrenkamp CA, Snow N, Mowat D, Kirk EP, Sachdev R, Smith J, Brown NJ, Wallis M, Barnett C, McKenzie F, Freckmann ML, Collins F, Chopra M, Gregersen N, Hayes I, Rajagopalan S, Tan TY, Stark Z, Savarirayan R, Yeung A, Adès L, Gattas M, Gibson K, Gabbett M, Amor DJ, Lattanzi W, Boyd S, Haan E, Gianoutsos M, Cox TC, Buckley MF, Roscioli T. A craniosynostosis massively parallel sequencing panel study in 309 Australian and New Zealand patients: findings and recommendations. *Genet Med*. 2018 Sep;20(9):1061-1068. doi:10.1038/gim.2017.214. Epub 2017 Dec 7. PMID:29215649.

Justice CM, Kim J, Kim SD, Kim K, Yagnik G, Cuellar A, Carrington B, Lu CL, Sood R, Boyadjiev SA, Wilson AF. A variant associated with sagittal nonsyndromic craniosynostosis alters the regulatory function of a non-coding element. *Am J Med Genet A*. 2017 Nov;173(11):2893-2897. doi:10.1002/ajmg.a.38392. Epub 2017 Oct 6. PMID:28985029.

Zhang R, Knapp M, Suzuki K, Kajioka D, Schmidt JM, Winkler J, Yilmaz Ö, Pleschka M, Cao J, Kockum CC, Barker G, Holmdahl G, Beaman G, Keene D, Woolf AS, Cervellione RM, Cheng W, Wilkins S, Gearhart JP, Sirchia F, Di Grazia M, Ebert AK, Rösch W, Ellinger J, Jenetzky E, Zwink N, Feitz WF, Marcelis C, Schumacher J, Martín-Torres F, Hibberd ML, Khor CC, Heilmann-Heimbach S, Barth S, Boyadjiev SA, Brusco A, Ludwig M, Newman W, Nordenskjöld A, Yamada G, Odermatt B, Reutter H. ISL1 is a major susceptibility gene for classic bladder exstrophy and a regulator of urinary tract development. *Sci Rep*. 2017 Feb 8;7:42170. doi:10.1038/srep42170. PMID:28176844.

Taché V, Bivina L, White S, Gregg J, Deignan J, Boyadjiev SA, Poulain FR. Lipoyltransferase 1



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Gene Defect Resulting in Fatal Lactic Acidosis in Two Siblings. *Case Rep Obstet Gynecol.* 2016; 2016:6520148. doi:10.1155/2016/6520148. Epub 2016 May 10. PMID:27247813.

Kruszka P, Addissie YA, Yarnell CM, Hadley DW, Guillen Sacoto MJ, Platte P, Paelecke Y, Collmann H, Snow N, Schweitzer T, Boyadjiev SA, Aravidis C, Hall SE, Mulliken JB, Roscioli T, Muenke M. Muenke syndrome: An international multicenter natural history study. *Am J Med Genet A.* 2016 Apr;170A(4):918-29. doi:10.1002/ajmg.a.37528. Epub 2016 Jan 6. PMID: 26740388.

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