CURRICULUM VITAE

Simeon Antonov Boyadjiev Boyd, M.D.

Current Appointment: Professor of Medical Genetics and Pediatrics (tenured)
Section of Genetics, Department of Pediatrics
University of California, Davis, USA

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Citizenship: United States of America and Republic of Bulgaria

Birth date and Place: April 13, 1959; Sofia, Bulgaria

Education and Professional training:

1979-1985	M.D., graduated with honor Pediatric Medical Institute St. Petersburg, Russia
1985-1988	Postdoctoral Fellow in Medical and Molecular Genetics Section of Clinical Genetics, Research Institute of Pediatrics Sofia, Bulgaria
1988-1991	Resident in Pediatrics Research Institute of Pediatrics Medical Academy, Sofia, Bulgaria
1991-1995	Non-matriculating student Department of Medical and Molecular Genetics Indiana University School of Medicine, Indianapolis, IN
1995-1997	Resident in Pediatrics Department of Pediatrics State University of New York, Stony Brook, NY
1997-2000	Postdoctoral Fellow in Medical Genetics McKusick-Nathans Institute of Genetic Medicine Johns Hopkins School of Medicine, Baltimore, MD

Professional Experience:

1991	Assistant Professor of Pediatrics and Genetics, Section of Clinical Genetics, Sofia, Bulgaria
1991-1995	Research Associate Indiana University School of Medicine Department of Medical and Molecular Genetics Indianapolis, IN
1997-2000	Clinical Instructor in Medical Genetics McKusick-Nathans Institute of Genetic Medicine Department of Pediatrics, the Johns Hopkins Hospital Baltimore, MD
2000-2006	Assistant Professor of Genetic Medicine and Pediatrics McKusick-Nathans Institute of Genetic Medicine Department of Pediatrics, the Johns Hopkins Hospital Baltimore, MD
2007-Current	Adjunct Professor Brody Urological Institute, Department of Surgery, the Johns Hopkins Hospital, Baltimore, MD
2006-2011	Founding Chief, Section of Genetics Children's Miracle Network Endowed Chair Department of Pediatrics University of California, Davis
2006-2012	Associate Professor Children's Miracle Network Endowed Chair Department of Pediatrics University of California, Davis
2012-Current	Professor of Medical Genetics and Pediatrics Department of Pediatrics University of California, Davis
2015- current	Adjunct Professor of Genetics, honorary position The Catholic University, Rome, Italy

Clinical activities: Approximately 50% of the professional effort is dedicated to direct clinical care and 50% on sponsored clinical and molecular genetics research. Both outpatient and inpatient activities include evaluation, diagnostic workup and treatment of patients with

various genetic disorders, dysmorphic syndromes, congenital anomalies and metabolic disorders. Approximately 40% of the clinical effort involves adult patients.

Licensure and Certifications:

1991	Bulgarian National Board of Pediatrics
1994	Educational Commission for Foreign Medical Graduates, USA
1997-2014	American Board of Pediatrics
1998-2006	Medical License for the State of Maryland
1999	American Board of Medical Genetics, Clinical Genetics
2005	Fellow American Academy of Pediatrics, elected
2006	Medical License for the State of California

Professional Affiliations:

1995-present	American Academy of Pediatrics - member
2000-present	American Society of Human Genetics - member
2004-present	European Society of Human Genetics - member
2006-present	American Society for Cell Biology -member
2000-2002	Vice President of the Society of Craniofacial Genetics
2002-2004	President of the Society of Craniofacial Genetics

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
Human Molecular Genetics
Molecular Genetics & Genomic Medicine

Grant reviewer:

Special Emphasis Panel, National Institute of Dental and Craniofacial research, NIH, June 2006

Invited reviewer, Research Grants Council, Hong Kong, China, 2006-2007

Ad-hoc reviewer for NIH study section Skeletal Biology Development and Disease (SBDD), October 11, 2008

Invited reviewer, the Face Base consortium, RFA U01, NIDCR, NIH, May 7, 2009

Ad-hoc reviewer for NIDCR, NIH Special Emphasis Panel, RFA “Genome-wide Studies of Craniofacial, Dental and Oral conditions”, July 23, 2009

Invited principal reviewer for Face Base consortium, RFA DE 09-003 U01, July 17, 2011
Invited reviewer for Sparks Charity, UK based foundation for children's health, July 2014

Training and Research Support:

1997-2000	Public Health Service Training Grant GM07471-23, NIH
1998-2000	Collaborator, Center for Inherited Disease Research (CIDR) NHGRI, NIH "Search for a gene involved in a new autosomal recessive syndrome with craniofacial anomalies and cataracts"
1999-2003	Co-Investigator, R01 DE13849, NIDCR-NIH "Genetic studies of oculodentodigital dysplasia"
2000-2005	Principal investigator, K-23 DE 00462, NIDCR-NIH "Genetic epidemiology of craniosynostosis"
2002- 2006	Principal investigator, GCRC protocol "Genetic studies of congenital anomalies of the urinary system and external genitalia" Johns Hopkins University
2003- 2006	Principal investigator, GCRC protocol "Genetic epidemiology of craniosynostosis" Johns Hopkins University
2005-2010	Co-Investigator, R01 DE014581-02, International Genetic Epidemiology of Oral Clefts, P.I. Terri Beaty
2005-2007	Principal Investigator, R03 DE016342, NIDCR-NIH "Candidate gene analysis of nonsyndromic craniosynostosis"
2006-2012	Principal Investigator, R01 DE016886, NIDCR-NIH "Nonsyndromic craniosynostosis: Phenotype/Genotype Study" (scored at 3rd percentile).
2007-2009	Principal Investigator, Children's Miracle Network Research Career Development Award "Genetic Analysis of Bladder-Exstrophy-Epispadias Complex"
2008-2010	Co-Investigator, The National Children's Study, NICHHD, NIH, P.I. Irva Hertz-Picciotto

2009-2010	Principal Investigator, M.I.N.D. Pilot Grant "Candidate Gene Association Analysis of the FMR1 Pathway in Idiopathic Autism Spectrum Disorders"
2009-2010	Principal Investigator, CTSC award " DNA biorepository from multigenerational famileis with nonsyndromic craniosynostosis"
2010	Prinical Investigator "Genome-wide association study of Sagittal Craniosynostosis", X01 grant application to the Center for Inherited Disease Research, NIH
2009-2012	Co-Investigator, Hartwell Foundation Individual Biomedical Research Award "Engineered Composite Materials for Treating Premature Suture Fusion in Infants", P.I. Kent Leach
2014-2019	Principal Investigator, 2R01 DE016886-06, competing renewal application NIDCR-NIH "Nonsyndromic craniosynostosis: Phenotype/Genotype Study". This is international consortium research project with major subcontracts to Oxford University, UK; the Catholic University of Rome, Italy; the New South Wales University Sidney Australia; the Universty of Iowa; and the University of Seattle, WA ($3,750,000)
2014-2019	Co-Investigator, 1R01GM110373-01A1 (PI: Kim) "Pathogenesis of diseases caused by aberrant COPII megavesicle assembly". The major goals of this project are to dissect the molecular mechanism of COPII megavesicle assembly and to define the in vivo consequences of abnormal COPII megavesicle assembly. ($1,480,000)
2014-2019	Co-investigator and PI for UC Davis for P01 program project "Craniosynostosis network" in response to PAR-12-034 "Developmental Mechanisms of Human Structural Birth Defects (P01)". The PI for this application is Dr. Inga Peter from the Mount Sinai School of Medicine, NY. The goal of this study is to: identify and characterize candidate genes associated with syndromic and non-syndromic coronal craniosynostosis and to characterize a large cohort of human coronal craniosynostosis cases using imaging and genotyping ($4,264,567)
2015-2016	Prinical Investigator "Genetic Studies of Metopic Craniosynostosis", X01 grant application to the Center for Inherited Disease Research, NIH ($640,000)

Clinical trials:

2010- ongoing	Principal Investigator: "An open label multicenter extension study to evaluate the long-term safety, tolerability and efficacy of sbc-102 in adult subjects with liver dysfunction due to lysosomal acid lipase deficiency who previously received treatment in study lal-cl01"; Sponsor - Synageva
2014-2015	Principal investigator: "A Phase 1/2, Open Label, Dose Ranging Study To Evaluate the Safety, Tolerability, Pharmacokinetics and Exploratory Efficacy Parameters of PRX-102 Administered By Intravenous Infusion Every 2 Weeks For 12 Weeks To Adulty Fabry Patients "; Sponsor – Protalix.

Invited Presentations:

1998	Center for Inherited Disease Research "Genetic studies of a new dysmorphic syndrome with craniofacial anomalies and cataracts"
1999	Pediatric Grand Round –Johns Hopkins Bayview "Acute metabolic crisis"
2000	Kennedy Krieger Institute Fellow series "Clinical Dysmorphology"
2001	University of Maryland, Genetic counseling program "Molecular basis of human congenital anomalies"
2002	Johns Hopkins Hospital, 30th Annual Pediatric Trends "Approach to the Dysmorphic Patient"
2003	Harvy Institute of Human Genetics, GBMC, Baltimore "Enzyme Replacement Therapy for Lysosomal Disorders"
2004	Harvy Institute of Human Genetics, GBMC, Baltimore "Craniofacial anomalies - diagnosis and management"
2003	National Children Study workshop, CDC Atlanta, invited expert presentation "Evaluation of the face"
2004	Pediatric Grand Rounds, University of California at Davis "Analysis of complex genetic traits"
2005	7th meeting of the International Skeletal Dyspasia Society

	Martigny, Switzerland "Cranio-lenticulo-sutural dysplasia is caused by abnormal COPII mediated intracellular trafficking
2005	Urology Grand Round, Brady Urologic Institute, Johns Hopkins "Genetic analysis of bladder exstrophy"
2006	Seminar at Department of Molecular and Cell Biology, Berkeley, CA: "Characterization of a SEC23A mutation in patients with Cranio-Lenticulo-Sutural Dysplasia"
2007	Keynote Speaker, COAST meeting "Developmental defects of the Craniofacial Skeleton" Asilomar, California. September 8
2007	Invited Speaker and Faculty "Genetics of Craniofacial Development and Dysmorphology" AO North America Challenges and Advances in the Management of Craniomaxilofacial Surgery, San Francisco, July 27-29
2007	Invited speaker for Research Series of Shriners Hospital for Children, Sacramento, CA "Medical genetics insight into COPII-mediated intracellular trafficking", November 26
2008	Neurosurgery Grand Round, Department of Neurosurgery, University of California Davis, 'Genetic Analysis of Nonsyndromic Craniosynostosis", February 11
2008	Guest Lecturer, PHA 250 Functional Genomics Course, University of California Davis "From the clinic to the bench (and back): Developmental phenotypes of ER export defects, May 27
2009	Medical Genetics Grand Rounds, Department of Pediatrics, Stanford University, CA "Genetic Analysis of Skull Defects", June 20
2010	Invited Speaker, International Association for Dental Research, Miami, Fl, "Clinical and Genetics correlates of Craniosynostosis" April 10,
2009	Grand round presentation, School of Veterinary Medicine, UC Davis "A novel syndrome, Cranio-Lenticulo-Sutural dysplasia implicates ER export in skeletal and craniofacial development"
2009	Invited Speaker, 3rd International Symposium on Exstrophy & Epispadias, Johns Hopkins Univ, Baltimore, October 14

2010	Neurology Grand Round, Department of Neurology, University of California Davis, "Lessons from the Genetics Clinic",April 8
2010	Invited Speaker, Prospective in Clinical Cardiology Seminar, Department of Surgery, University of California Davis, "Connective Tissue Syndromes – Management of Cardiac Problems", April 30
2010	ENT Grand Round, Department of ENT, University of California Davis, "Congenital Craniofacial Disorders - Genetics and Resources in Northern California", June 1
2010	Invited Speaker, Prospective in Clinical Cardiology Seminar, Department of Surgery, University of California Davis, "Cardiac Manifestations of Lysosomal Storage Disorders" September 24
2010	Dermatology Grand Round, Department of Dermatology, University of California Davis, "Clinical Dysmorphology: Clues from the Skin" October 6
2011	Pediatric Grand Rounds, Department of Pediatrics University of California Davis, "Lysosomal Disorders – a model for etiologic treatment in genetics" October 10
2012	Invited Speaker, Department of Anatomy and Cell Biology, Catholic University of Rome, Italy, "Genome-wide association studies of craniosynostosis" July 11 (host Dr. Wanda Lattanzi)
2012	Invited Speaker Department of Orofacial Sciences, Center for Craniofacial and Mesenchymal Biology, University of California, San Francisco, "Abnormal BMP2 and BBS9 expression in nonsyndromic sagittal craniosynostosis" August 22 (host Dr. Ophir Kline)
2012	Invited Speaker, Craniofacial Unit, John Radcliffe Hospital, Oxford University, UK, "Genetic etiology of sagittal craniosynostosis", September 4 (host Dr. Andrew Wilkie)
2012	Invited speaker, National University of Singapore, Department of Pediatrics) "An international effort to identify the causes of craniosynostosis", December 18 (host Dr. Samuel Chong)

2012	Invited speaker, National University of Malay, Kuala Lumpur, Department of Pediatrics "Genetics of nonsyndromic craniosynostosis", December 21, (host Dr. M.K. Thong)
2013	KK Hospital, Singapore, Department of Pediatrics " International Craniosynsotosis Consortium", January 4 (host Dr. Angela Lai)
2013	Lessons from the Clinic: "Identification of novel Genetic Syndromes" Genetics Grand Rounds, UC Davis
2013	Advances in Cleft Lip and Palate Including Velopharyngeal Dysfunction, November 15-17, Napa, California (host Dr. Craig Senders)

Postdoctoral fellows, students mentored and staff supervised:

1997 - 1998 Joseph Kamal, undergraduate researcher (Johns Hopkins University), currently M.D.
1998 - 1999 David Choi, undergraduate researcher (Johns Hopkins University), currently MPH
1999 - 2006 George Zhang, Research Assistant
1999 - 2001 Arnab Chowdry, Undergraduate Researcher, currently Ph.D. candidate
2001 - 2003 Cristi Radford, Research Assistant, currently Genetic Counselor
2001 - 2005 Nisha Isaac, Genetic Counselor
2002 - 2004 David Hur, Research Assistant, currently medical student
2004 - 2005 Gerald Ashrafi, Research Assistant, currently dental student
2004 - 2005 Reem Saadeh, Genetics Fellow, currently Clinical Geneticist
2004 - 2005 Veronica Mardo, Genetics Fellow, currently Clinical Geneticist
2005 - 2009 Christopher Nauta, Research Assistant and Lab Manager
2007 - 2008 Serena Chan, undergraduate researcher (University of California Davis)
2007 - 2008 Bonnie Ching, M.Sc. student (thesis advisor), graduated
2007 - 2008 Vijaya Ramachandran, Ph.D., Postdoctoral Fellow
2007 - 2008 Erica Goude, Research Assistant
2008 - 2009 Osman Ozes, Ph.D., project scientist
2007 - 2009 Kathleen Angkustiri, M.D., Fellow, Developmental Behavioral Pediatrics
2009 - 2010 Linda Keyes, graduate student in epidemiology (thesis advisor)
2008 - 2011 Lisa Lit, Ph.D., postdoctoral researcher (principal mentor)
2008 - 2012 Elijah Cherkez, clinical research coordinator
2009 - 2011 Jialie Liu, senior research associate
2009 - 2012 Christina Stevens, high school student, currently undergraduate aat Berkeley (biology)
2009 - 2012 Pedro Sanchez, M.D., K12 award recipient, (co-mentor)
2010 - 2012 Sundon Kim, Ph.D., postdoctoral researcher (principal mentor)
2008 - 2012 Garima Yagnick, Ph.D. Student in Genetics (thesis committee chairman)
2011 - 2012 Jaclyn Greenwood, genetic counselor student (thesis committee member)
2010 - 2014 Carolyn Yrigollen, Ph.D. Student in Genetics (thesis committee member)
2013 - 2014 Anna-Marie Tuazon, Ph.D. student in Genetics (lab rotation)
2013 - now Vicki Hwang, Ph.D. student in Genetics (thesis committee member)

2014 – now Araceli Cuellar, Ph.D., postdoctoral researcher (principal mentor)
2015 – now Kevin Fausto, B.SC., research associate
2015 – now Toni McKinney, B.Sc., clinical research coordinator

Patents

International Patent Application - Docket No: 68335(71699), filed May 22, 2007, registered April 18, 2009: "Diagnostic Screening Methods for Disorders of the Endoplasmic Reticulim-to-Golgi Trafficking of Proteins"

Awards

1979-1985	M.D., graduated with honor
2000-2002	Vice President Elect of the Society of Craniofacial Genetics
2002-2004	President Elect of the Society of Craniofacial Genetics
2007	Keynote Speaker, COAST meeting "Developmental defects of the Craniofacial Skeleton"
2007	Dean nomination for Provost Fellow, UC Davis
2012	Dean Nomination Clinical Research Forum award – US Congress

Other activities:

1. Bulgarian Chamber of Commerce – Founding board member
2. Volunteer soccer coach – Davis CA
3. Hobbies – guitars, yoga, philosophy, windsurfing, snowboarding

Peer Reviewed Publications:

1. Boyadjiev S, Simeonov E, Varon R. Case of metachromatic leukodystrophy. **J Pediatr** (Bulg.) 2: 17-20, 1990

2. Gupta PK, Sahota A, Boyadjiev SA, Bye S, O'Neill JP, Hunter TC, Albertini RJ, Tischfield JA. Analysis of in vivo somatic mutations at the APRT locus. **Adv Exp Med Biol,** 370: 657-660, 1994

3. Boyadjiev SA, Sahota A, Tischfield JA. Identification of polymorphic markers flanking the human APRT gene. **Adv Exp Med Biol,** 370: 653-656, 1994

4. Sahota A, Chen J, Boyadjiev SA, Gault MH, Tischfield JA. Missense mutation in the adenine phosphoribosyltransferase gene causing 2,8-dihydroxyadenine urolithiasis. **Hum Mol Genet,** 3(5): 817-818, 1994

5. Tischfield JA, Engle SJ, Gupta PK, Bye S, Boyadjiev S, Shao C, O'Neill JP, Albertini RJ, Stambrook PJ, Sahota A. Germline and somatic mutation at the APRT locus of mice and man. **Adv Exp Med Biol** 370:661-664, 1994.

6. Pratt VM, Boyadjiev S, Dlouhy SR, Silver K, Der Kaloustian VM, Hodes ME. Pelizaeus-Merzbacher disease in a family of Portuguese origin caused by a point mutation in exon 5 of PLP. **Am J Med Genet,** 55: 402-404, 1995

7. Pratt VM, Boyadjiev S, Green K, Hodes ME, Dlouhy SR. Pelizaeus-Merzbacher disease caused by de novo mutation that originated in exon 2 of the maternal great-grandfather of the propositus. **Am J Med Genet,** 58: 70-73, 1995

8. Boyadjiev SA, Sahota A, Tischfield JA. Identification and application of polymorphisms flanking the human adenine phosphoribosyltransferase gene. **Hum Mutat,** 8: 214-215, 1996

9. Nance MA, Boyadjiev S, Pratt VM, Taylor S, Hodes ME, Dlouhy SR. Adult-onset neurodegenerative disorder due to proteolipid protein gene mutation in the mother of a man with Pelizaeus-Merzbacher disease. **Neurology.** 47: 1933-1935, 1996

10. Gupta PK, Sahota A, Boyadjiev SA, Bye S, Shao C, O'Neill P, Hunter TC, Albertini RJ, Stambrook PJ, Tischfield JA. High frequency in vivo loss of heterozygosity is primary a consequence of mitotic recombination. **Cancer Res,** 57: 188-193, 1997

11. Flanegan N, Boyadjiev SA, Harper J, Kyne L, Earley M, Watson R, Jabs EW, Geraghty MT. Familial craniosynostosis, anal anomalies and porokeratosis - CAP syndrome. **J Med Genet,** 35: 1998

12. Boyadjiev SA, Jabs EW, LaBuda M, Jamal JE, Torbergsen T, Ptacek LJ 2nd, Rogers RC, Nyberg-Hansen R, Opjordsmoen S, Zeller CB, Stine OC, Stalker HJ, Zori RT, Shapiro RE. Linkage analysis narrows the critical region for oculodentodigital dysplasia to chromosome 6q22-q23. **Genomics,** 58(1): 34-40, 1999**.**

13. Kovach MJ, Lin JP, Boyadjiev S, Campbell K, Mazzeo L, Herman K, Rimer LA, Frank W, Llewellyn B, Gelber D, Kimonis VE. A unique point mutation in the PMP22 gene is associated with Charcot-Marie-Tooth disease and deafness. **Am J Hum Genet,** 64(6): 1580-93, 1999

14. Glaser RL, Jiang W, Boyadjiev SA, Tran AK, Zachary AA, Johnson D, Walsh S, Oldridge M, Wall SA, Wilkie AOM, Jabs EW. Paternal origin of FGFR2 mutations in sporadic cases of Crouzon and Pfeiffer syndromes. **Am J Hum Genet,** 66(3):768-77, 2000

15. Prophanphoj V, Boyadjiev SA, Waber LJ, Brusilow SW, Geraghty MT. Three case of sodium benzoate and sodium phenylacetate toxicity occurring in the treatment of acute hyperammoniemia. **J Inherit Metab Dis,** 23:129-136, 2000

16. Boyadjiev SA, Jabs EW. OMIM as a knowledgebase for human developmental disorders, In Developmental Biology: Frontiers for Clinical Genetics **Clin Genet** 57:253-266, 2000.

17. Boyadjiev SA, Chowdry AB, Shapiro RE, Wandstrat AE, Choi JW, Kasch L, Zhang G, Wollnik B, Burgess CE, Schalling M, Lovett, Jabs EW. Physical Map of the Chromosome 6q22 Region Containing the Oculodentodigital Dysplasia Locus: Analysis of Thirteen Candidate Genes and Identification of Novel cDNAs and SNPs. **Cytogenet Genome Res,** 98:29-37, 2002

18. Paznekas WA, Boyadjiev SA, Shapiro RE, Daniels O, Wollnik B, Keegan CE, Innis JW, Dinulos MB, Christian C, Hannibal MC, Jabs EW. Connexin 43 (GJA1) Mutations Cause the Pleiotropic Phenotype of Oculodentodigital Dysplasia. **Am J Hum Genet,** 72:408-418, 2003

19. 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

20. Al-Hassnan ZN, Boyadjiev SA, Praphanphoj V, Hamosh A, Braverman NE, Thomas GH, Geraghty MT. The relationship of plasma glutamine to ammonium and of glycine to acid-base balance in propionic acedimia. **J Inherit Metab Dis,** 26(1):89-91, 2003

21. Boyadjiev SA, Dodson JL Radford .L, Ashrafi GH, Beaty TH, Mathews RI, Broman KW, Gearhart J.P. Clinical and molecular characterization of the bladder exstrophy-epispadias complex: analysis of 232 families. **Brit J Urology International,** 94:1337-1343, 2004

22. 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

23. 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

24. 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

25. Aldridge K, Kane AA, Marsh JL, Panchal J, Boyadjiev SA, Yan P, Govier D, Ahmad W, Richtsmeier JT. Brain morphology in non-syndromic unicoronal craniosynostosis. **Anat Rec A Discov Mol Cell Evol Biol,** 285(2):690-698, 2005**.**

26. Aldridge K, Boyadjiev SA, Capone GT, DeLeon VB, Richtsmeier JT. Precision and error of three-dimensional phenotypic measures acquired from 3dMD photogrammetric images. **Am J Med Genet,** 138A (3):247-253, 2005

27. Jehee FS, Alonso LG, Cavalcanti DP, Kim C, Wall SA, Mulliken JB, Sun M, Jabs EW, Boyadjiev SA, Wilkie AO, Passos-Bueno MR. Mutational screening of FGFR1, CER1, and CDON in a large cohort of trigonocephalic patients. **Cleft Palate Craniofac J,** 43:148-151, 2006

28. Cohn, RD, Eklund E, Bergner AL, Casella JF, Althaus J, Blakemore KJ, Fox HE, Hoover-Fong JE, Hamosh A, Braverman NE, Freeze HH , Boyadjiev SA. Intracranial Hemorrhage as the Initial Manifestation of a Congenital Disorder of Glycosylation. **Pediatrics,** 118(2):514-521, 2006

29. Beaty TH, Hetmanski JB, Fallin MD, Park JW, Jabs EW, McIntosh I, Liang KY, Vander Kolk CA, Boyadjiev SA, Chong SS, Cheah CS, Wu-Chou YH, Chen KT, Chiu YF, Yeow V, Ng Ivy SL, Cheng J, Huang S, Ye X, Wang H, Ingersoll R, Scott AF. Analysis of 64 Candidate Genes on Chromosome 2 in Cleft Case-Parent Trios from Three Populations. **Hum. Genet,** 120(4):501-18, 2006.

30. Boyadiev 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. **(Corresponding author), Nat Genet,** 38(10):1192-1197, 2006

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

32. 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

33. Kimonis V, Gold J-A, Hoffman TL, Panchal, J and Boyadjiev SA. Genetics of Craniosynostosis. **Seminars in Pediatric Neurology,** 14(3):150-161, 2007

34. 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

35. 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

36. Ludwig M, Rüschendorf F, Saar K, Hübner N, Siekmann L, Boyadjiev SA, Reutter H. Genome-wide linkage scan for bladder exstrophy-epispadias complex**. Birth Defects Res A Clin Mol Teratol.** 85(2):174-178, 2008

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

38. Paznekas WA, Karczeski B, Vermeer S, Lowry RB, Delatycki M, Laurence F, Koivisto PA, Van Maldergem L, Boyadjiev SA, Bodurtha JN, Wang Jabs E. GJA1 mutations, variants, and connexin 43 dysfunction as it relates to the oculodentodigital dysplasia phenotype. **Hum Mutat,** 30(5):724-33. 2009

39. Monitto CL, Hamilton RG, Levey E, Jedlicka AE, Dziedzic A, Gearhart JP, Boyadjiev SA, Brown RH. Genetic predisposition to natural rubber latex allergy differs between health care workers and high-risk patients. **Anesth Analg,** 110(5):1310-7, 2010

40. Heuzé Y, Boyadjiev SA, Marsh JL, Kane AA, Cherkez E, Boggan JE, Richtsmeier JT. New insights into the relationship between suture closure and craniofacial dysmorphology in sagittal nonsyndromic craniosynostosis. **J Anat, (Journal cover),** 217(2):85-96. 2010

41. Ching BJ, Wittler L, Proske J, Yagnik G, Qi L, Draaken M, Reutter H, Gearhart JP, Ludwig M, Boyadjiev SA. p63 (TP73L) a key player in embryonic urogenital development with significant dysregulation in human bladder exstrophy tissue. **Int J Mol Med,** 26(6): 861-7. 2010

42. Boyadjiev SA, Kim SD, Hata A, Zackai EH, Naydenov C, Hamamoto S, Schekman RW, Kim JA. Cranio-lenticulo-sutural dysplasia associated with defects in collagen secretion. **Clin Genet,** 80:169-176, 2011

43. Qi L, Chen K, Hur DJ, Yagnik G, Lakshmanan Y, Kotch LE, Ashrafi GA, Martinez-Murillo F, Kowalski J, Naydenov C, Wttler L, Gearhart GP, Draaken M, Reutter H, Ludwig M, Boyadjiev SA. Genome-wide expression profiling of urinary bladder implicates desmosomal and cytoskeletal dysregulation in the bladder exstrophy-epispadias complex. **Int J Mol Med,** 27:755-765, 2011

44. Reutter H, Boyadjiev SA, Gambhir L, Ebert AK, Rösch WH, Stein R, Schröder A, Boemers TM, Bartels E, Vogt H, Beckers GM, Bökenkamp A, Kajbafzadeh AM, Jaureguizar E, Draaken M, Lakshmanan Y, Gearhart JP, Ludwig M, Nöthen MM, Jenetzky E. Phenotype Severity in the Bladder Exstrophy-Epispadias Complex: Analysis of Genetic and Nongenetic Contributing Factors in 441 Families from North America and Europe. **(Co-corresponding author) J Pediatr**, doi:10.1016/j.jpeds.04.042, 2011

45. Vissers L, Cox TC, Smyth A, Maga AM, Janssen IM, Short KM, Wirdajaja F, Jehee F, Bertola D, Liu J, Yagnik G, Bokhoven H, Marcelis C, Cunningham MC, Anderson PA, Boyadjiev SA, Passos-Bueno MR, Veltman JA, Buckley MF and Roscioli T. Heterozygous Mutations of FREM1 are Associated With an Increased Risk of Isolated Metopic Craniosynostosis in Humans and Mice. **PLoS Genet**. 2011 Sep;7(9):e1002278. Epub 2011 Sep 8.

46. Pham NS, Rafii A, Liu J , Boyadjiev SA, Tollefson TT. Clinical and Genetic Characterization of Frontorhiny: report of three novel cases and discussion of the surgical management. **Arch Facial Plast Surg.** 2011 Nov;13(6):415-20.

47. Bhat A, Boyadjiev SA, Senders C, Leach K. Differential Growth Factor Adsorption to Calvarial Osteoblast-Secreted Extracellular Matrices Instructs Osteoblastic Behavior. **PLoS,** 2011 Oct: 6 (10):e25990. Epub 2011 Oct 8.

48. Heuzé Y, Martínez-Abadías N, Stella JM, Senders CW, Boyadjiev SA, Lo LJ, Richtsmeier JT. Unilateral and bilateral expression of a quantitative trait: asymmetry and symmetry in coronal craniosynostosis. **J Exp Zool B Mol Dev Evol.** 2012 Mar;318(2):109-22.

49. Kim SD, Pahuja KB, Ravazzola M, Yoon J, Boyadjiev SA, Hammamoto S, Schekman R, Orci L, Kim J. The SEC23-SEC31 interface plays a critical role for export of procollagen from the endoplasmic reticulum. **J Biol Chem.** 2012 Mar 23;287(13):10134-44.

50. Chassaing N, Sorrentino S, Davis EE, Martin-Coignard D, Iacovelli A, Paznekas W, Webb BD, Faye-Petersen O, Encha-Razavi F, Lequeux L, Vigouroux A, Yesilyurt A, Boyadjiev SA, Kayserili H, Loget P, Carles D, Sergi C, Puvabanditsin S, Chen CP, Etchevers HC, Katsanis N, Mercer CL, Calvas P, Jabs EW. OTX2 mutations contribute to the otocephaly-dysgnathia complex. **J Med Genet**. 2012, Jun;49(6):373-9.

51. Kim S-D, Liu JL, Roscioli T, Buckley MF, Yagnik G, Boyadjiev SA **(co-corresponding author)**, Kim J. Leucine-rich repeat, immunoglobulin-like and transmembrane domain 3 (LRIT3) is a modulator of FGFR1. **FEBS Letters**, 586 (2012): 1516-1521.

52. Yagnik R, Gunman A, Stevens C, Liu J, Kim, J, Boyadjiev SA. ALX4 mutations is nonsyndromic craniosynostosis. **Hum Mut ,** 2012 Dec;33(12):1626-9.

53. Justice CM, Yagnik Y, Kim Y, Peter I, Jabs EW, Ye Y, Shi L, Cunningham ML, Kimonis V, Roscioli T, Wall SA, Wilkie AOM, Stoler J, Richtsmeier JT, Heuzé Y, Sanchez-Lara SA, Buckley MF, Druschel CM, Mills JL, Caggana M, Romitti PA, Kay DM, Senders C, Taub PJ, Klein OD, Boggan J, Zwienenberg-Lee M, Naydenov C, Kim J, Wilsom AF, Boyadjiev SA. A genome-wide association study identifies susceptibility loci for non-syndromic sagittal craniosynostosis near BMP2 and within BBS9. **Nat Genet,** 2012 Nov 18;44(12):1360-4.

54. Kim S-D, Liu JL, Yagnik G, Cunningham ML, Kim J, Boyadjiev SA. MAPK/ERK Signaling Pathway Analysis in Primary Osteoblasts from Patients with Non-Syndromic Sagittal Craniosynostosishop. **Cleft Palate Craniofac J**. 2013 Apr 8. [Epub ahead of print], PMID: 23566293

55. Wiszniewski W, Hunter JV, Hanchard NA, Willer JR, Shaw C, Tian Q, Illner A, Wang X, Cheung SW, Patel A, Campbell IM, Hixson P, Ester AR, Azamian MS, Potocki L, Zapata G, Hernandez PP, Ramocki MB, Santos-Cortez RL, Wang G, York MK, Justice MJ, Chu ZD, Bader PI, Omo-Griffith L, Madduri NS, Scharer G, Crawford HP, Yanatatsaneejit P, Eifert A, Kerr J, Bacino CA, Franklin AI, Goin-Kochel RP, Simpson G, Immken L, Haque ME, Stosic M, Williams MD, Morgan TM, Pruthi S, Omary R, Boyadjiev SA, Win KK, Thida A, Hurles M, Hibberd ML, Khor CC, Van Vinh Chau N, Gallagher TE, Mutirangura A, Stankiewicz P, Beaudet AL, Maletic-Savatic M, Rosenfeld JA, Shaffer LG, Davis EE, Belmont JW, Dunstan S, Simmons CP, Bonnen PE, Leal SM, Katsanis N, Lupski JR, Lalani SR. TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. **Am J Hum Genet**. 2013 Jun 26. [Epub ahead of print]; PMID: 23810381.

56. Greenwood J, Flodman P, Osann K, Boyadjiev SA **(co-corresponding author)**, Kimonis V. Familial incidence and associated symptoms in a population of individuals with non-syndromic craniosynostosis", **Genetics in Medicine**, 013 Sep 26. doi: 10.1038/gim.2013.134

57. Qi L, Wang M, Yagnik G, Mattheisen M, Gearhart JP, Lakshmanan Y, Ebert A-K, Rosch W, Ludwig M, Draaken M, Reutter H, Boyadjiev SA. Candidate Gene Association Study Implicates p63 in the Etiology of Nonsyndromic Bladder-Exstrophy-Epispadias Complex; **Birth Defects Research** (Part A) 2013 Aug 2. doi: 10.1002/bdra.23161.

58. Weaver KN, El Hallek M, Hopkin RJ, Sund K, Del Gaudio D, Yuksel A, Bober M, Kim J, Boyadjiev SA. Keutel Syndrome: Report of 2 new *MGP* mutations and discussion of clinical overlaps with arylsulfatase E deficiency and relapsing polychondritis. **Am J Med Genet,** accepted, 2014 Jan 23. doi: 10.1002/ajmg.a.36390. [Epub ahead of print].

59. Heuzé Y, Martínez-Abadías N, Stella JM, Arnaud E, Collet C, Fructuoso GG, Alamar M, Lo L-J, Boyadjiev SA, Di Rocco F, Richtsmeier JT. Quantification of facial skeletal shape variation in fibroblast growth factor receptor-related craniosynostosis syndromes. **Birth Defects Res A Clin Mol Teratol.** 2014 Feb 27. doi: 10.1002/bdra.23228. [Epub ahead of print]

60. Boyadjiev Boyd SA, Dvorak CC, Bivina L, Wahlstrom J, Horn B, Bailey-Olson M, Cowan MJ. Umbilical cord blood transplantation in Hurler syndrome using busulfan, fludarabine, clofarabine, rATG conditioning regimen **MOL GENET METAB**. Publisher: Elsevier ISSN: 1096-7192-2.83

61. Reutter H, Draaken M, Pennimpede T, Wittler L, Ebert A-K, Bartels E, Rösch W, Boemers TM. Hirsch K, Schmiedeke E, Meesters C, Becker B, Stein R, Utsch B, Mangold E, Nordenskjöld A, Barker G, Boyadjiev SA, Zwink N, Holmdahl H. Classic bladder exstrophy: frequent 22q11.21 duplications and definition of a 414 kb phenocritical region. acceptable for publication in **Birth Defects Research Part A: Clin Mol Teratol** , March 27, 2014

62. Reutter H, Draaken M, Pennimpede T, Wittler L, Brockschmidt FF, Ebert AK, Bartels E, Rösch W, Boemers TM, Hirsch K, Schmiedeke E, Meesters C, Becker T, Stein R, Utsch B, Mangold E, Nordenskjöld A, Barker G, Kockum CC, Zwink N, Holmdahl G, Läckgren G, Jenetzky E, Feitz WF, Marcelis C, Wijers CH, Van Rooij IA, Gearhart JP, Herrmann BG, Ludwig M, Boyadjiev SA, Nöthen MM, Mattheisen M. Genome-wide association study and mouse expression data identify a highly conserved 32 kb intergenic region between WNT3 and WNT9b as possible susceptibility locus for isolated classic exstrophy of the bladder. **Hum Mol Genet** 2014 May 22. pii: ddu259

63. Vassili Valayannopoulos V, Malinova V, Honzík T, Balwani M, Breen C, Deegan PB, Enns GM, Jones SA, Kane JP, Stock EO, Tripuraneni R, Eckert S, Schneider E, Hamilton G, Middleton MS, Sirlin C, Kessler B, Bourdon C; Boyadjiev SA; Sharma R, Twelves C, Whitley CB, Quinn AG. Sebelipase alfa over 52 weeks reduces serum transaminases, liver volume and improves serum lipids in patients with lysosomal acid lipase deficiency. **J of Hepatol** 2014 Nov;61(5):1135-42.

64. Garbes L, Kim K, Rieß A, Hoyer-Kuhn H, Beleggia F, Bevot A, Kim MJ, Huh YH, Kweon HS, Savarirayan R, Amor D, Kakadia PM, Lindig T, Kagan KO, Becker J, Boyadjiev SA, Wollnik B, Semler O, Bohlander SK, Kim J, Netzer C.. Mutations in *SEC24D*, Encoding a Component of the COPII Machinery, Cause a Syndromic Form of Osteogenesis Imperfecta. Submitted to **Am J Hum Genet** 2015 Mar 5;96(3):432-9

65. Kruszka P, Addissie YA, Yarnell CM, Hadley DW, Guillen Sacoto MJ, Platte P, Paelecke Y, Collmann H, Snow N[4], 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 Jan 6. doi: 10.1002/ajmg.a.37528. [Epub ahead of print]

Publications under review:

1. Justice CM, Kim J, Kim SD, Yagnik G, Carrington B, Sood R, Boyadjiev SA, Wilson AF. A nucleotide change at a non-coding polymorphism associated with sagittal nonsyndromic craniosynostosis causes differential expression in zebrafish. **Nat Genet**, submitted July 15, 2015

2. Taché V, Bivina L, White S, Gregg J, Boyadjiev SA, Dungan J, Poulain F Fatal lactic acidosis in two siblings with LIPT1 gene defect – third report of a new disease. **Eu J Pediatrics,** submitted November 2015

Book Chapters and Reviews:

1. Simeonov E, Kratchunova M, Boyadjiev S. Fragile X syndrome. **New Problems in Pediatrics** (Bulg.), 8-12, 1988.

2. Lewanda AF, Boyadjiev SA, Jabs EW. Dysmorphology: Genetic Syndromes and Associations. **Oski's Pediatrics**, 4th Ed. Chapter 462.

3. Boyadjiev SA. SEC23A and Cranio-Lenticulo-Sutural Dysplasia**. Inborn Errors of Development**, 2nd Edition. Oxford University Press, New York (2008)

4. Kim J and Boyadjiev SA. SEC23A and Cranio-Lenticulo-Sutural Dysplasia**. Inborn Errors of Development**, 3nd Edition. Oxford University Press, New York (2013)

5. Boyadjiev SA. "Congenital Craniofacial and Musculoskeletal Abnormalities". Book chapter **Merck Manual** (2014), web publication

6. Kim J and Boyadjiev SA. SEC23A and Cranio-Lenticulo-Sutural Dysplasia**. Inborn Errors of Development**, 3nd Edition, Chapter update. Oxford University Press, New York (2008) IN PRESS.

Published Abstracts and Presentations:

1. Simeonov E, Boyadjiev S. Registration of congenital malformations among newborns. National Congress of Pediatrics, Burgas, Bulgaria, 1987.

2 Simeonov E, Licharska K, Khadjiev A, Boyadjiev S. Our experience in prenatal diagnosis of neural tube defects. 1st Bulgarian-Russian Symposia of Genetic Counseling, Varna, Bulgaria, 1987.

3. Simeonov E, Khadjiev A, Boyadjiev S. Prenatal diagnosis in Bulgaria. 2nd Symposia of Prenatal Diagnosis, Prague, Czech Republic, 1988.

4. Simeonov E, Boyadjiev S. Neural tube defects prevention. Jubilee Session of the Medical Academy, Sofia, Bulgaria, 1988.

5. Simeonov E, Boyadjiev S. Biochemistry of dysmorphic features. 5th International Symposia of Inherited Errors of Metabolism, Brno, Czech Republic, 1988.

6. Simeonov E, Hyanek I, Boyadjiev S, Sinigerska I. Mental retardation, unusual hair and renal tubular acidosis in two sibs. A new entity? 5th International Symposia, Brno, Czech Republic, 1988.

7. Boyadjiev S, Tomova S, Simeonov E. Use of ultrasound for fetal development study and prenatal diagnosis of fetal malformations. 5th National Conference of Biomedical Physics and Techniques, Sofia, Bulgaria, 1988.

8. Boyadjiev S, Dimova E, Simeonov E, Mintchev M. A computer program for genealogic records. 4th National Conference of Medical Cybernetics, Varna, Bulgaria, 1989.

9. Zlatareva N, Sinapova L, Poptchev I, Boyadjiev S, Simeonov E. Expert system for diagnosis of congenital malformation. 5th National Conference of Medical Cybernetics, Varna, Bulgaria, 1989.

10. Andreev A, Kintcheva V, Boyadjiev S. Pseudomosaicism in prenatal chromosomal diagnosis. 4th National Conference of Cytogenetics, Sofia, Bulgaria, 1989.**.**

11. Boyadjiev S, Simeonov E. Intellectual characteristics of individuals with specific fragility of the chromosomes. 4th National Conference of Cytogenetics, Sofia, Bulgaria, 1989.

12. Boyadjiev S, Dimova E, Mintchev M, Simeonov E. EMI-genetic counseling management computer system. 3rd National Congress of Medical Biology and Genetics, Sofia, Bulgaria, 1990.

13. Boyadjiev S, Simeonov E, Tincheva R. Case of ring 13 chromosome- phenotypic similarity with Smith-Lemli-Opitz syndrome. 3rd National Congress of Medical Biology and Genetics, Sofia, Bulgaria, 1990.

14. Simeonov E, Schulzova J, Starka I, Boyadjiev S. Genetic prevention of congenital adrenal hyperplasia. 3rd National Congress of Medical Biology and Genetics, Sofia, Bulgaria, 1990.

15. Simeonov E, Boyadjiev S, Zlatareva N. Computer diagnosis in dysmorphology. 3rd National Congress of Medical Biology and Genetics, Sofia, Bulgaria, 1990.

16. Simeonov E, Khadjiev A, Licharska K, Brankova M, Boyadjiev S. Prenatal diagnosis of genetic defects. 3rd National Congress of Medical Biology and Genetics, Sofia, Bulgaria, 1990.

17. Simeonov E, Boyadjiev S. Genetics of polycystic ovary syndrome. 3rd National Congress of Medical Biology and Genetics, Sofia, Bulgaria, 1990.

18. Simeonov E, Boyadjiev S, Tincheva R. Prevention of dysmorphic syndromes. 5th International Congress of Early Fetal Diagnosis, Prague, Czech Republic, 1990.

19. Simeonov E, Boyadjiev S. Registration of congenital anomalies and prenatal diagnosis. 5th International Congress of Early Fetal Diagnosis, Prague, Czech Republic, 1990.

20. Boyadjiev S, Khadjiev A, Simeonov E. Prenatal ultrasound diagnosis of skeletal dysplasias. 5th International Congress of Early Fetal Diagnosis, Prague, Czech Republic, 1990.

21. Kratchunova M, Simeonov E, Boyadjiev S. Prevalence of Fra-X syndrome among mentally retarded children. 5th International Congress of Early Fetal Diagnosis, Prague, Czech Republic, 1990.

22. Licharska K, Simeonov E, Boyadjiev S. AFP screening in prevention of genetic defects. 5th International Congress of Early Fetal Diagnosis, Prague, Czech Republic, 1990.

23. Kintcheva V, Kratchunova M, Simeonov E, Boyadjiev S. Second trimester prenatal cytogenetic diagnosis in Bulgaria. 5th International Congress of Early Fetal Diagnosis, Prague, Czech Republic, 1990.

24. Kintcheva V, Andreev A, Boyadjiev S. Interstitial deletion del(q31,q33) and formation of an extra stable ring chromosome. 5th Annual Meeting of German Geneticists, Ulm, Germany, 1991.

25. Boyadjiev SA, Sahota A, Pratt VM, Dlouhy SR, Hodes ME. Detection of PLP gene mutations in patients with Pelizaeus-Merzbacher disease by single strand conformational analysis. Am J Hum Genet 53(3) A1131, 1993.

26. Tores W, Sahota A, Boyadjiev SA, Vance GH, Weaver DD. Fragile X full mutation in two clinically unaffected brothers. Am J Hum Genet 53(3):A1241, 1993.

27. Boyadjiev SA, Shapiro RE, Wandstrat A, Jamal JE, LaBuda M, Torbergsen T, Ptcek LJ, Curtis Rogers R, Nyberg-Hansen R, Opjordsmoen S, Zeller CB, Stine OC, Stalker HJ, Zori RT, Lovett M, Jabs EW. Oculodentodigital dysplasia – clinical update and positional cloning effort. Platform presentation, 22nd Annual Meeting of the Society of Craniofacial Genetics, San Francisco, 1999.

28. Boyadjiev SA, Zwaan J, Justice CM, McKusick VA, Jabak M, Wilson AF, Jabs EW. Cranio-lenticulo-sutural dysplasia: Clinical and molecular studies of a new syndrome. Platform presentation, 49th Annual Meeting of the American Society of Human Genetics, San Francisco, 1999.

29. Glaser RL, Jiang W, Boyadjiev SA, Johnson D, Walsh S, Qldridge M, Wall SA, Wilkie AOM, Jabs EW. Paternal origin of FGFR2 mutations in sporadic cases of Crouzon and Pfeiffer syndromes. Am J Hum Genet 65(4):A35, 1999.

30. Justice CM, Boyadjiev SA, Zwaan J, Jabak M, Jabs EW, Wilson AF. Statistical genetic analysis of cranio-lenticulo-sutural dysplasia. NIH NHG RI Annual Retreat, 1999.

31. Al-Hassan ZN, Boyadjiev SA, Prophanphoj V, Hamosh A, Braverman NE, Thomas, GH, Geraghty MT. Hyperammoniemia and amino acid profiles in propionic acidemia. Am J Hum Genet 67(4):A1585, 2000.

32. Boyadjiev SA, Shapiro RE, Wandstrat A, Chowdry AB, Choi JW, LaBuda M, Lovett M, Burgess C, Schalling M, Jabs EW. Genetic Studies of Oculodentodigital Dysplasia. Eur J Hum Genet 9(1):P1518, 2001.

33. Boyadjiev SA, Gearheart JP. Genetic Aspects of the Exstrophy-Epispadias Complex. Platform presentation. Annual Meeting of the Bladder Exstrophy support group, Baltimore, 2002.

34. Boyadjiev SA, Zhang G, Ingersoll R, Isaac I, Kasch L, Hur D, Jabs EW, Beaty T, Scott AF. Analysis of candidate genes for non-syndromic craniosynostosis. Am J Hum Genet 71(4):A1795, 2002.

35. Isaac N, Beaty TH, Carson B, Jabs EW, Richtsmeier J, Vander Kolk C, Scott AF, Boyadjiev SA. Comparison of Web based vs. hospital based retrospective recruitment of participants for a craniosynostosis study. Am J Hum Genet 71(4):A990, 2002.

36. Boyadjiev SA: Genetic epidemiology of non-syndromic craniosynostosis. Platform presentation, 25th Annual Meeting of the Society of Craniofacial Genetics, Baltimore, 2002.

37. Boyadjiev SA, Dodson JL, Radford C, Beaty TH, Mathews RI, Gearhart JP. Genetic epidemiology of the exstrophy-epispadias complex. Am J Hum Genet 73(5):A670. 2003.

38. Boyadjiev SA, Ashrafi GH, Radford CL, South S, Stetten G, Lakshmanan Y, Dodson JD, Gearhart JP. Bladder exstrophy-epispadias complex: analysis of 232 families. Am Soc Hum Genet 54th annual meeting. A754, 2004.

39. Aldridge K, Boyadjiev Boyd SA, DeLeon VB, Richtsmeier JT. Anthropometric landmarks from 3dMD surface images: a study of accuracy and repeatability. Am Soc Hum Genet 54th annual meeting. A750, 2004.

40. Jehee FS, Johnson D, Abe K, Alonso L, Alonso N, Kim C, Cavalcante D, Wall SA, Boyadjiev Boyd SA, Jabs EW, Wilkie OA, Passos-Bueno MR. Genetic and molecular analysis of a large cohort of patients with syndromic and non-syndromic trigonocephaly - screening for loss of heterozigosity in 9p and 11q regions. Am Soc Hum Genet 54th annual meeting. A745, 2004.

41. Boyadjiev SA, Zhang GX, Ingersoll RG, Hur DJ, Vander Kolk CA, Carson BS, Marsh JL, Panchal J, Cutler D, Scott AF. Molecular characterization of nonsyndromic craniosynostosis Euro J Hum Genet, Suppl 1, v.13., P1025 2005.

42. Boyadjiev SA, Hur D, Zhang G, Eyaid W. Cranio-Lenticulo-Sutural dysplasia is caused by SEC23A mutation resulting in abnormal COPII-mediated intracellular trafficking. Am Soc Hum Genet 55th annual meeting, platform presentation #262, 2005.

43. Boyadjiev SA. Cranio-Lenticulo-Sutural dysplasia is caused by SEC23A mutation disruptin ER-to Golgi Trafficking. 38th meeting of the European Society of Human Genetics. Platform presentation. Euro J Hum Genet suppl. 1 vol. 14, C70, 2006.

44. Mardo, V, Lisi E, Riegert-Johnson D, Boyadjiev SA. Novel autosomal dominant syndrome in a three-generation family with broad first digits, oral anomalies, and speech delay. . Am Soc Hum Genet 56th annual meeting, P683, 2006.

45. Fromme JC, Nauta C, Hur DJ, Zhang G, Hamamoto S, Ravazzola M, Orci L, Eyaid E, Boyadjiev SA, Schekman R. Defective ER-to-Golgi Transport Caused by a Mutation in the COPII Protein SEC23A Is Responsible for the Human Disease Cranio-Lenticulo-Sutural Dysplasia. Am Soc Cell Biology, 46th annual meeting, platform presentation #1412, 2006.

46. Reutter H., Qi L., Boemers T., Ebert A.-K., Gearhart J.P., Rösch W., Boyadjiev S.A., Ludwig M. Concordance for the Bladder Exstrophy-Epispadias Complex (BEEC) in Twins: Suggestive of Genetic Susceptibility. 18 Jahrestagung der Deutschen Gesellschaft für Humangenetik, Bonn, 2007, P313

47. Boyadjiev SA, Saadeh R, Scott A, Tian W, Mian A, Wulfsberg E, Beatty T, Vander Kolk C, Redett R. Microstomia- cleft palate: a new syndrome? Poster A971, Euro Soc Hum Genet 2007 annual meeting, Nice, France.

48. Ludwig M, Reutter H, Nauta C, Lakshmanan Y, Gearhart JP, Boyadjiev SA. Analysis of *p63* as a candidate gene for bladder exstrophy. Poster A456, Euro Soc Hum Genet 2007 annual meeting, Nice France.

49. Ching BJ, Ludwig M, Reutter H, Nauta C, Gearhart JP, Boyadjiev SA. Potential role of *p63* in human bladder exstrophy. Am Soc Hum Genet 57th annual meeting, P2545, 2007.

50. Moghaddam B, Yuksel A, Bober M, Bolton K, Chan S, Liu J,Nauta C, Boyadjiev SA. Is Keutel syndrome genetically heterogeneous? Am Soc Hum Genet 57th annual meeting, P507, 2007.

51. Nauta C, Dong Y, Drissi H, Boyadjiev SA. Potential Role of RUNX2 in Nonsyndromic Sagittal Craniosynostosis. Am Soc Hum Genet 57th annual meeting, P2564, 2007.

52. Qi L, Kun Chen K, Hur DJ, Lakshmanan Y, Kotch LE, Ashrafi GH, Martinez-Murillo F, Blackford A, Kowalski J, Gearhart JP, Boyadjiev SA. Genome-wide expression profiling of urinary

bladder identifies candidate genes for the bladder exstrophy-epispadias complex (BEEC). Am Soc Hum Genet 57th annual meeting, P2350, 2007.

53. Nauta C, Zackai E, Kim J, Boyadjiev SA. Cranio-Lenticulo-Sutural Dysplasia – a new genetic syndrome with late-closing fontanels, platform presentation, Pediatric Academic Society meeting, Hawaii, May 2008

54. Boyadjiev SA , Nauta C, Zackai E, Kim J. New case of Cranio-Lenticulo-Sutural Dysplasia – a recently described syndrome with late-closing fontanels. Euro Soc Hum Genet 2008 annual meeting, P01.268, Barcelona, Spain

55. Ching. B, Yagnik G. Qi L, Nauta C, Ludwig M, Reutter H, Gearhart JP, Boyadjiev SA. Dysregulation of p63 (TP73L) expression and desmosomal assembly as etiologic factors for Bladder-Exstrophy-Epispadias Comples, Am Soc Hum Genet 58th annual meeting, P2223, 2008
56. Angkustsiri K, Camilleri K., Nordahl C.,. Li C, Simon T., Hansen R.,. Amaral D, Boyadjiev Boyd SA. Three dimensional evaluation of facial structures in autism spectrum disorders.¨ Am Soc Hum Genet 58th annual meeting, P502, 2008

57. Boyadjiev S, Nauta C, Hata A, Naydenov C, Zackai E, Kim J. A new case of Cranio-Lenticulo-Sutural dysplasia. Am Soc Hum Genet 58th annual meeting, P474, 2008

58. Barber T, Ludwig L, Gargollo P, Draaken M, Zinn A, Reutter H, Boyadjiev SA, Baker L. Goltz syndrome and Classic Bladder Exstrophy due to PORCN mutation: report of the fourth patient and analysis of isolated exstrophy cohort. AAP meeting, 2009, Boston, MA

59. Lisi E, Mardo V, Hata A, Riegert-Johnson D, Boyadjiev SA. Broad first digits, facial and oral anomalies, and developmental delay in a three-generation family: A Novel Syndrome? Euro Soc Hum Genet 2009 annual meeting, Vienna, Austria P02.014

60. Ching B, Yagnik G, Qi L, Hata A, Ludwig M, Reutter H, Naidenov C, Gearheart JP, Boyadjiev SA; Abnormal Expression of p63 (TP73L) in Bladder Exstrophy-Epispadias Complex. Euro Soc Hum Genet 2009 annual meeting, Vienna, Austriam P02.083.

61. Hata A, Getman CE, Kahle RA , Lackman RS, Boyadjiev SA. Neonatal manifestations of Mucolipidosis type II: Longitudinal review of skeletal findings and generalized melanocytosis. Am Soc Hum Genet 59th annual meeting, P474, 2009

62. Angkustsiri K, Camilleri K, Cochran L, Nordahl CW, Barnet LA, Hansen RH, Mastergeorge AM, Rogers SJ, Boyadjiev SA. Poor Verbal and Non-Verbal Skills in Children with Abnormal Craniofacial Variability Indices (CVI) and ASD. IMFAR meeting 2009 Paper #3531.

63. Boyadjiev SA S. D. Kim SD, Helms J, Kim J; Craniofacial syndromes caused by endoplasmic reticulum export defects. Euro Soc Hum Genet 2010 annual meeting, Gothenburg, Sweden, Poster A-962

64. Boyadjiev SA, Liu J, Bober M, Yuksel A, Karaca E, Lambie L, Bolton K; Evidence for genetic heterogeneity of Keutel syndrome. Euro Soc Hum Genet 2010 annual meeting, Gothenburg, Sweden , Poster A-976.

65. Cunningham M, Roscioli T, Passos-Bueno M, Wollnik B, Wilkie A, Boyadjiev SA. International Craniosynostosis Consortium: finding the causes of nonsyndromic craniosynostosis Euro Soc Hum Genet 2010 annual meeting, Gothenburg, Sweden, Poster A-1990.

66. Vissers LM, Cox TC, Janssen IM, Short KM, Smyth I, Jehee F, Yagnik G, Boyadjiev SA, Marcelis C, Anderson PJ, Cunningham LM, Passos-Bueno M, Veltman JA, Buckley MF, Roscioli T. Copy Number and Sequence Variants in *FREM1* are Associated With an Increased Risk of Isolated Metopic Craniosynostosis in Humans and Mice. Euro Soc Hum Genet 2010 annual meeting, Gothenburg, Sweden, Platform presentation A-1986

67. Boyadjiev SA, Decaris ML, Horner C, Bhat A, Cherkez E, Jiu J, Boggan J, Tollefson T, Senders C, Cunningham M, Leach JM. Calvarial Osteoblasts from Infants with Nonsyndromic Craniosynostosis Exhibit Increased Osteogenic Potential. Euro Soc Hum Genet 2010 annual meeting, Gothenburg, Sweden, Poster A-2003.

68. Yagnik G, Drissi H, Stevens C, Boyadjiev SA. RUNX2 Polymorphisms in Patients with Nonsyndromic Sagittal Craniosynostosis. Poster 967. Am Soc Hum Genet 60^{h} annual meeting 2010, Washington D.C.

69. Ghuman AS, Qi L, Liu J, Yagnik G, Cherkez E, Cunningham ML, Kimonis V, Boyadjiev SA. ALX4 Variants in Nonsyndromic Sagittal Craniosynostosis. Poster 954. Am Soc Hum Genet 60th annual meeting 2010, Washington D.C.

70. Kim SD, Liu J, Cherkez E, Boyadjiev SA, Kim J. A Novel SEC23A Missense Mutation Causing Cranio-Lenticulo-Sutural Dysplasia Is Involved in The MAPK Signaling Pathway. Poster 723. Am Soc Hum Genet 60th annual meeting 2010, Washington D.C.

71. McGinniss MA, Wang H, Young3, Calhoun R, Mayberg S, Martinezmoles J, Grix AW, Westerfield BA, Pena LS, Penny DJ, Fan Y, Boyadjiev SA. Novel *ACTA2* mutation in a large family with familial thoracic aortic aneurysms and dissections exhibiting reduced penetrance in a female carrier. Poster 380W. Am Soc Hum Genet 61th annual meeting 2011, Montreal Canada

72. Kim Y, Yagnik G, McMullen IA, Ling H, Sanchez-Lara PA, Kimonis V, Stoler J, Cunningham M, Wilson AF, Justice CM, Boyadjiev SA. Case-parent trio genome-wide association study identifies several candidate loci for nonsyndromic sagittal craniosynostosis. Poster 987W. Am Soc Hum Genet 61th annual meeting 2011, Montreal Canada

73. Yagnik G, Liu J, C. Stevens C, Ghuman A, Kim S, Kim J, Cherkez E, Soung D, Drissi H, Buckley M, Roscioli T, Boyadjiev SA. Candidate gene functional analysis in nonsyndromic sagittal craniosynostosis. Poster 1126F. Am Soc Hum Genet 61th annual meeting 2011, Montreal Canada

74. Heuzé Y, Martínez-Abadías N, Stella JM, Di Rocco F, Collet C, Fructuoso GG, Alamar M,

Lo L-J, Boyadjiev SA, Richtsmeier JT. Is the craniofacial phenotype sufficient to characterize FGFR-related craniosynostosis syndromes? 34th Annual Meeting of the Society of Craniofacial Genetics and Developmental Biology, 2011, Montreal, Canada

75. Boyadjiev SA for the International Craniosynostosis Consortium. A genome-wide association study identifies susceptibility loci for non-sundromic sagittal craniosynostosis near BMP2 and within BBS9. Platform presentation at the 35th Annual Meeting of the Society of Craniofacial Genetics and Developmental Biology, San Francisco, November 6th, 2012

76. Greenwood J, Boyadjiev SA, Osann K, Flodman P, Kimonis V. Familial occurrence and associated symptoms in a population of individuals with non-syndromic craniosynostosis. Poster 1791W. Am Soc Hum Genet 62th annual meeting 2012, San Francisco, USA.

77. G. Yagnik, C. M. Justice, Y. Kim, I. Peter, E. W. Jabs, X. Ye, L. Shi, M. L. Cunningham, V. Kimonis, T. Roscioli, S. A. Wall, A. O. M. Wilkie, J. Stoler, J. T. Richtsmeier, Y. Heuzé, P. A. Sanchez-Lara, M. F. Buckley, C. M. Druschel, J. L. Mills, M. Caggana, P. A. Romitti, D. M. Kay, C. Senders, P. J. Taub, O. D. Klein, J. Boggan, C. Naydenov, J. Kim, A. F. Wilson, S. A. Boyadjiev. A genome-wide association study identifies susceptibility loci for non-syndromic sagittal craniosynostosis on chromosomes 20 and 7. Poster 2151W, Am Soc Hum Genet 62th annual meeting 2012, San Francisco, USA.

78. M. L. Cunningham, P. A. Romitti, C. M. Justice, A. F. Wilson, T. Roscioli, E. Oláh, B. Bessenyei, M. R. Passos-Bueno, B. Wollnik, A. O. M. Wilkie, S. A. Boyadjiev, International Craniosynostosis Consortium. Poster 3070F, Am Soc Hum Genet 62th annual meeting 2012, San Francisco, USA.Clinical and molecular characterization of non-syndromic craniosynostosis: An international consortium approach.

79. Boyadjiev-Jabs syndrome: Clinical and molecular characterization of three new patients. S. A. Boyadjiev, S. Kim, L. Bivina, E. Zackai, P. L. Crotwell, K. Õunap, J. Kim. Poster 3151 W Am Soc Hum Genet 62th annual meeting 2012, San Francisco, USA.

80. Manisha Balwani, Vera Malinova, Reena Sharma, Vassili Valayannopoulos, Eveline O. Stock, Simeon A Boyadjiev, Bruce Kessler, Patrick Deegan, Gregory M. Enns, Catherine Breen, John P. Kane, Eugene Schneider, Anthony G. Quinn. Long Term Safety and Clinical Activity of SBC-102, a Recombinant Human Lysosomal Acid Lipase (rhLAL), in Patients with Late Onset LAL Deficiency. Platform presentation at the 9th Annual World Symposium of the Lysosomal Disease Network, February 15, 2013, Miami, FL, USA

81. R Sharma, M Balwani, C Bourdon, SA Boyadjiev, C Breen, J Burg, P Deegan, G Enns, T Honzik, J Kane, B Kessler, V Malinova, V Valayannopoulos, E Schneider, E Stock, C Whitley, AG Quinn Sebelipase alfa Normalizes Transaminases and Reduces Liver Fat Content in Patients with Late Onset Form of LAL Deficiency. Platform presentation at the European Study of Pediatric Gastroenterology Hepatology and Nutrition, London, May 8-11 2013.

82. S. A. Boyadjiev, M. Zwienenberg-Lee, J. Boggan, C. Senders. Craniosynostosis and Heart defects: Possibly a new autosomal recessive syndrome due to ZDHHC13 mutations Euro Soc Hum Genet annual meeting, Milan, Italy, May 29-June 3, 2014

83. CM Justice, J Kim, S-D Kim, G Yagnik, B Carrington, R Sood , AF Wilson[1],S A Boyadjiev Non-coding variant in the BMP2 locus that is associated with sagittal non-syndromic craniosynostosis causes differential GFP expression in zebrafish. Poster 889S Am Soc Hum Genet 64th annual meeting 2014, San Diego, USA