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CURRICULUM VITAE

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EDUCATION:

1987-1991	B.S.	Biology	Istanbul University, Turkey
1992-1996	M.S.	Molecular Biology	Istanbul University
1997-2002	Ph.D.	Genetics	Istanbul University School of Medicine
2002-2004	Postdoctoral Fellow	Human Genetics	Cold Spring Harbor Laboratory, Woodbury, NY
2004-2007	Research Fellow	Human Genetics	Massachusetts General Hospital, Boston, MA
2004-2006	Clinical Molecular Genetics Fellow	ABMG Genetics Training Program	Harvard Medical School, Boston, MA
2006-2007	Clinical Cytogenetics Fellow	ABMG Genetics Training Program	Harvard Medical School

LICENSURE:

2012	Clinical Cytogenetics Scientist, State of California, California Department of Public Health, License #: MTO 663
2013	Clinical Genetic Molecular Biologist Scientist, State of California, California Department of Public Health, License #: MTP 395
2013	Molecular Pathology Director, Florida Department of Health, License #: DI 45586
2013	Cytogenetics Director, Florida Department of Health, License #: DI 45586

BOARD CERTIFICATION:

2007	Clinical Molecular Genetics	American Board of Medical Genetics and Genomics
2009	Clinical Cytogenetics	American Board of Medical Genetics and Genomics
2011	Technologist in Cytogenetics	American Society for Clinical Pathology, Board of Certification
2012	Technologist in Molecular Biology	American Society for Clinical Pathology, Board of Certification

PROFESSIONAL EXPERIENCE:

Feb. 1992-March 1996	Clinical Technologist, Duzen Laboratory
Mar. 1995-Mar. 1996	Research Assistant, Department of Molecular Biology and Genetics Bogazici University, Istanbul
Apr. 1996-Jan. 2002	Clinical Molecular Geneticist, Duzen Laboratory, Istanbul
Sept. 2007- Feb. 2012	Director, Clinical Cytogenetics Laboratory Beth Israel Deaconess Medical Center
Sept. 2007- Feb. 2012	Instructor, Department of Pathology Harvard Medical School
Mar. 2012-May 2017	Associate Director, Clinical and Molecular Cytogenetics Laboratory Department of Pathology and Laboratory Medicine, UCLA
Mar. 2012-May 2017	Faculty, Clinical Microarray Laboratory Department of Pathology and Laboratory Medicine, UCLA
Mar. 2013-May 2017	Faculty, Clinical Exome Sequencing Genomic Data Board, UCLA Clinical Genomics Center
Mar. 2012-June2015	Health Sciences Assistant Clinical Professor Department of Pathology and Laboratory Medicine, UCLA
Jul. 2015-May 2017	Health Sciences Associate Clinical Professor Department of Pathology and Laboratory Medicine, UCLA
June 2017-ongoing	Senior Cytogenetics Director Quest Diagnostics Nichols Institute

Committee service:

2011-2012 Academy Member, The Academy for Medical Educators at Beth Israel Deaconess Medical Center

2012-2017 Genomic Data Board, UCLA Clinical Genomics Center

2013-2017 Clinical Cytogenetics Fellowship Search Committee, Department of Pathology and Laboratory Medicine, UCLA

2013-2017 UCLA Pathology Clinical Alumni Committee, Department of Pathology and Laboratory Medicine, UCLA

2013-2017 Clinical Quality Improvement Committee, Department of Pathology and Laboratory Medicine, UCLA

2013-2014 Member, International Standards for Cytogenomic Arrays (ISCA) Consortium Evidence-Based Review Committee

2013-ongoing Member, American College of Medical Genetics and Genomics (ACMG) Maintenance of Certification Committee

2014-2017 Member, Clinical Pathology Education Committee, Department of Pathology and Laboratory Medicine, UCLA

2014-ongoing Member, The Clinical Genome Resource (ClinGen): Dosage Sensitivity Curation Working Group

2015-ongoing Member, Cancer Genomics Consortium (CGC): Technical Standards and Reporting Guidelines for Genetic Testing for Chronic Lymphocytic Leukemia (CLL) Working Group

2015-ongoing Member, Cancer Genomics Consortium (CGC): Building Consensus in Microarray Reporting Practices in Constitutional Genetics Working Group

2015-ongoing Member, The Clinical Genome Resource (ClinGen): Copy Number Variant Interpretation Guidelines Working Group

2016-ongoing Member, American College of Medical Genetics and Genomics (ACMG) Interpretation of Constitutional Copy Number Variations Working Group

2016-ongoing Member, American College of Medical Genetics and Genomics (ACMG) Laboratory Quality Assurance (QA): Selection of the Genes in a Gene Panel Subcommittee Working Group

2017-ongoing Member, Genetics & Genomics Education Committee, The Turkish Society of Medical Genetics, Turkey

Other professional service:

2012-ongoing Molecular Pathology Team Member Inspector, College of American Pathologists (CAP)

2012-ongoing Cytogenetics Team Member Inspector, College of American Pathologists (CAP)

2/4/2015 Tutor, Pathology Board Review Course (PTH 92) Cytogenetics, the Osler Institute, Los Angeles, CA

2/24/2016 Tutor, Pathology Board Review Course (PTH 95), Molecular Pathology, the Osler Institute, Los Angeles, CA

2/24/ 2016 Tutor, Pathology Board Review Course (PTH 95), Cytogenetics, the Osler Institute, Los Angeles, CA

2/22/2017 Tutor, Pathology Board Review Course (PTH 98), Molecular Pathology, the Osler Institute, Los Angeles, CA

2/22/ 2017 Tutor, Pathology Board Review Course (PTH 98), Cytogenetics, the Osler Institute, Los Angeles, CA

Professional and scholarly associations:

1997-ongoing	The Turkish Society of Medical Genetics	Member
2002-2005	The New York Academy of Sciences	Member
2002-2007	European Society of Human Genetics	Member
2002-2017	The American Society of Human Genetics	Member
2007-ongoing	American College of Medical Genetics and Genomics	Member
2008-2012	New England Regional Genetics Group	Member
2010-ongoing	American Cytogenetics Conference	Member
2011-ongoing	Association for Molecular Pathology	Member
2012-ongoing	American Society for Clinical Pathology	Member

Editorial service:

Editor (2011-ongoing)
Pathobiology of Human Disease: A Dynamic Encyclopedia of Disease Mechanisms. Section: Molecular Pathology, Elsevier Inc. [database online] Science Direct

Ad Hoc Reviewer (2010-ongoing)
The American Journal of Medical Genetics: Part A
TheScientificWorldJOURNAL
Journal of Biomedicine and Biotechnology

Consulting activities

Nov. 2013-May 2017 Consultant, Ivigen, LLC, 1200 NW 78th Avenue, Suite 103, Miami, FL

Community service:

2011-2012	Volunteer	Reiki Program at Brigham and Women's Hospital, Boston, MA
Jan.-Aug. 2013	Volunteer	Chase Child Life Program at Mattel Children's Hospital, UCLA
Spring 2014	Academic Advisor for Baibing Qin, Volunteer K-8 Student at UCLA Cytogenetics Laboratory, Beverly Hills Academic After School Tutoring Program	
2014-2015	Academic Advisor for Selin Sindel, Volunteer Student at UCLA Cytogenetics Laboratory, Palisades Charter High School	
Nov. 2014-ongoing	Member of the Board of Directors, Galatasaray California	
Jan-May 2016	Secretary of the Board of Directors, Los Angeles American Turkish Association	
Dec. 2016-ongoing	Member of Turkish American Ladies League (TALL) (http://tallglobal.org/)	

HONORS AND SPECIAL AWARDS:

2010 The Eleanor and Miles Shore 50th Anniversary Fellowships for Scholars in Medicine
Beth Israel Deaconess Medical Center Department of Pathology Fellowship ($30,000)
"Gene Discovery in Autosomal Recessive Non-syndromic Intellectual Disability"

RESEARCH GRANTS AND FELLOWSHIPS RECEIVED:

2007- 2012 Research Start-up Package, Beth Israel Deaconess Medical Center Pathology Foundation
PI ($500,000- in addition to salary)

LECTURES AND PRESENTATIONS:

International Invited Lectures:

"Congenital diaphragmatic hernia (CDH): Fryns syndrome and Donnai-Barrow syndrome". Sophia's Children Hospital, 2006, Rotterdam, The Netherlands

"Mutations in *LRP2*, encoding the multiligand receptor megalin, cause Donnai-Barrow syndrome". VIII. National Medical Genetics Congress, 2008, Canakkale, Turkey

"Identification of the genetic basis of nonsyndromic intellectual disability in large consanguineous families by exome sequencing". IX. National Medical Genetics Congress, 2010, Istanbul, Turkey

"Interpretation and reporting guidelines of postnatal constitutional copy number variants". Clinical Genetics Summit sponsored by Chinese Medical Doctor Association, 2015, Shanghai, China

"Sharing genomic copy number variant data to improve patient care". Istanbul University School of Medicine, Division of Medical Genetics, 2015, Istanbul, Turkey

"Clinical significance of genomic copy number variations". Akdeniz University School of Medicine, Department of Pediatrics and Medical Genetics, 2015, Antalya, Turkey

"Cytogenetics to Cytogenomics". The 3rd National Pediatric Genetics Symposium (3. Ulusal Çocuk Genetik Sempozyumu), 2017, Akdeniz University School of Medicine, Antalya, Turkey

National Invited Lectures:

"Megalin mutations cause Donnai-Barrow and Facioocculoacousticorenal (FOAR) syndromes". 5th Structural Birth Defects Meeting, NIH/NICHD, 2007, Bethesda, MD

"Detection of somatically-derived copy number variants (CNVs) using a targeted CNV-enriched array-based comparative genomic hybridization (aCGH) platform". Vincent Center for Reproductive Biology at MGH, 2010, Boston, MA

Regional Invited Lectures:

"Clinical applications of chromosomal microarray analysis". Grand Rounds, Department of Pathology at Harbor-UCLA Medical Center, 2012, Torrance

"Identification of genetic basis of nonsyndromic intellectual disability in large families by exome sequencing". Medical Genetics Seminar Series in Medical Genetics Institute at Cedars-Sinai Medical Center, 2013, Los Angeles

"Pediatric cytogenetics". Grand Rounds, Department of Pathology, University of California, Irvine Medical Center, 2013, Irvine

"Clinical Chromosomal Microarray Analysis". UCLA Fall Laboratory Seminar, 2013, Los Angeles

"Genomic copy number matters". Grand Rounds, Department of Pathology and Laboratory Medicine, Los Angeles County+USC Medical Center, 2014, Los Angeles

"Interesting microdeletion/microduplication cases". Molecular Pathology and Genetics, Children's Hospital Los Angeles, 2014, Los Angeles

"A matter of copy number". Journal Club of the Cytogenetics Department, Quest Diagnostics Nichols Institute, 2014, San Juan Capistrano

"FISH technology and nomenclature". Grand Rounds, Department of Pathology and Laboratory Medicine, Los Angeles County+USC Medical Center, 2014, Los Angeles

"New developments in chromosomal microarray analysis". Division of Genetics & Genomic Medicine, Department of Pediatrics, University of California, Irvine Medical Center, 2014, Irvine

"Chromosomal abnormalities & interpretation of cytogenetics nomenclature". Grand Rounds, Department of Pathology and Laboratory Medicine, Los Angeles County+USC Medical Center, 2015, Los Angeles

"Clinical utility of chromosomal microarray analysis". Pacific Southwestern Regional Genetics Network (PSGN) Conference, 2015, Genoptix, Carlsbad

"Introduction to cytogenetics". Grand Rounds, Department of Pathology and Laboratory Medicine, Los Angeles County+USC Medical Center, 2015, Los Angeles

"Cytogenetics of hematologic neoplasms". Grand Rounds, Department of Pathology and Laboratory Medicine, Los Angeles County+USC Medical Center, 2015, Los Angeles

"Introduction to clinical cytogenetics and nomenclature". Grand Rounds, Department of Pathology, University of California, Irvine Medical Center, 2015, Irvine

"Fluorescence in situ hybridization (FISH) technology and nomenclature". Grand Rounds, Department of Pathology, University of California, Irvine Medical Center, 2015, Irvine

"Constitutional chromosome disorders". Grand Rounds, Department of Pathology, Harbor-UCLA Medical Center, 2015, Torrance

"Chromosomal Microarray Analysis and Interesting Cases". Prenatal Diagnosis Center (PDC) Meeting, Department of Pediatrics, UCLA, 2015, Los Angeles

"Chromosomal abnormalities and nomenclature". Grand Rounds, Department of Pathology, Harbor-UCLA Medical Center, 2015, Torrance

Complementary Therapies for Cancer Patients: Reiki" Simms/Mann-UCLA Center for Integrative Oncology, Ronald Reagan UCLA Medical Center Auditorium, UCLA, 2015, Los Angeles

"Chromosomal abnormalities & interpretation of cytogenetics nomenclature". Grand Rounds, Department of Pathology and Laboratory Medicine, Los Angeles County+USC Medical Center, 2016, Los Angeles

"Clinical genomics: providing a resolution for patients on a diagnostic odyssey". Pacific Southwestern Regional Genetics Network (PSGN) Conference, 2016, Genoptix, Carlsbad

"The use of chromosomal microarray analysis in postnatal diagnosis". Grand Rounds, Department of Pathology and Laboratory Medicine, Los Angeles County+USC Medical Center, 2016, Los Angeles

"Updates on Cell-free Prenatal Screening". Prenatal Diagnosis Center (PDC) Meeting, Department of Pediatrics, UCLA, 2016, Los Angeles

"Cytogenetics and Molecular Testing for Prader-Willi and Angelman Syndromes". Dysmorphology Rounds, Department of Pediatrics, UCLA, 2017, Los Angeles

Platform Presentations:

"Development of routine DNA diagnostic techniques for common inherited disorders in Turkish population". 7th National Congress of Perinatology, 1999, Belek, Turkey

"Haplotype analysis in Turkish autosomal recessive polycystic kidney disease (ARPKD) families". 6th National Congress of Prenatal Diagnosis and Medical Genetics, 2000, Izmir, Turkey

"Mutations in megalin cause Donnai-Barrow and Facioocculoacousticorenal (FOAR) syndromes characterized by overlapping features of corpus callosum, diaphragm, neurosensory, and craniofacial defects". American College of Medical Genetics Annual Clinical Genetics Meeting, 2007, Nashville, TN

"The first report of a *de novo* heterozygous missense *DISP1* mutation in a patient with congenital diaphragmatic hernia (CDH) and additional malformations". The American Society of Human Genetics 57th Annual Meeting, 2007, San Diego, CA

"A novel *BCL2* gene rearrangement, t(5;18)(q31;q21), in follicular lymphoma". The Cancer Cytogenomics Microarray Consortium CCMC/CAGdb Meeting, 2013, Chicago, IL

"A novel 1p35.1p34.3 microdeletion in a baby boy with multiple congenital anomalies and developmental delay". The Cancer Cytogenomics Microarray Consortium CCMC/CAGdb Meeting, 2014, Chicago, IL

"Congenital heart defects in neonates: Determining the incidence of genetic testing and follow up consultation at UCLA". The Cancer Genomics Consortium (CGC) and the Cytogenomics Array Group (CAGdb) Meeting, 2015, Denver, CO

Moderator:

Highlights Plenary Session: "The Exome Strategy for Gene Discovery and Medical Practice". American College of Medical Genetics Annual Clinical Genetics Meeting, 2011, Vancouver, BC Canada

Panel Discussion:

"The pathologist's post-genome practice". Cold Spring Harbor Laboratory, Personal Genomes Meeting, 2010, Cold Spring Harbor, NY

Keynote speaker at the Chromosomal Microarray Chinese Consortium Meeting, 2015, Wyndham East Bund Shanghai Hotel, Shanghai, China

Posters:

"Prenatal diagnosis applications for common inherited disorders in Duzen Laboratory". 7th National Congress of Perinatology, 1999, Belek, Turkey

"Prenatal diagnosis of common inherited disorders in Turkish population". 6th National Congress of Prenatal Diagnosis and Medical Genetics, 2000, Izmir, Turkey

"Haplotype analysis in autosomal recessive polycystic kidney disease (ARPKD) families and the determination of allele frequencies of used markers in the Turkish population". The American Society of Human Genetics 53rd Annual Meeting, 2003, Los Angeles, CA

"Joubert syndrome: a patient with a de novo t(2;22)(q13;q11.1)". The American Society of Human Genetics 53rd Annual Meeting, 2003, Los Angeles, CA

"Identification and characterization of a candidate gene in Joubert syndrome". European Human Genetics Conference, 2004, Munich, Germany

"Molecular genetics approaches in human congenital diaphragmatic hernia". The American Society of Human Genetics 55th Annual Meeting, 2005, Salt Lake City, UT

"Screening of 1q41-q42.12 and 15q26.1-q26.2 regions by multiplex ligation-dependent probe amplification (MLPA) in patients with congenital diaphragmatic hernia (CDH)". European Human Genetics Conference, 2006, Amsterdam, The Netherlands

"Identification of a genetic locus for Donnai-Barrow syndrome". The American Society of Human Genetics 56th Annual Meeting, 2006, New Orleans, LA

"Donnai-Barrow syndrome (DBS/FOAR) in a child with a homozygous *LRP2* mutation due to complete chromosome 2 paternal isodisomy". American College of Medical Genetics Annual Clinical Genetics Meeting, 2008, Phoenix, AZ

"46,XY,i(18)(q10)/46,XY,del(18)(p11.1) mosaicism in an infant with mild congenital anomalies". The American Society of Human Genetics 58th Annual Meeting, 2008, Philadelphia, PA

"A submicroscopic unbalanced translocation resulting in del 4p and dup 17q leads to multiple congenital anomalies in a newborn female". American College of Medical Genetics Annual Clinical Genetics Meeting, 2009, Tampa, FL

"Identification and characterization of somatic copy number variants (CNVs) in embryonic cell lineages using a novel CNV-targeted array comparative genomic hybridization (aCGH) platform". The American Society of Human Genetics 59th Annual Meeting (2009), Honolulu, HI

"Detection of somatically-derived copy number variants (sCNVs) using a targeted CNV-enriched array based comparative genomic hybridization (aCGH) platform". American College of Medical Genetics Annual Clinical Genetics Meeting, 2010, Albuquerque, NM

"A female patient with *FIP1L1-PGDFRA*-positive HES/CEL after receiving R-CHOP chemotherapy for large cell lymphoma. American Cytogenetics Conference, 2010, Niagara Falls, Ontario, Canada

"An exome strategy to identify the genetic basis of nonsyndromic mental retardation in large consanguineous Arab families". The American Society of Human Genetics 60th Annual Meeting (2010), Washington, DC

"X-chromosome exome sequencing of a nonsyndromic intellectual disability family with multiple affected male sibs. The American Society of Human Genetics 12th international congress of human genetics / 61st Annual Meeting, 2011, Montreal, QC, Canada

"Exome sequencing reveals novel compound heterozygous *POMT1* mutations in a non-consanguineous Lebanese family with intellectual disability and microcephaly". Association for Molecular Pathology (AMP) Annual Meeting on Genomic Medicine, 2012, Long Beach, CA

"A de novo intragenic deletion of *AUST2* in a patient with autism spectrum disorder". The American Society of Human Genetics 62nd Annual Meeting, 2012, San Francisco, CA

"Intragenic deletion of STK32B in a family with isolated cleft palate". International Collaboration for Clinical Genomics (ICCG) Meeting. 2013, Bethesda, MD

"A patient with Angelman-like features due to deletion of chromosome 15q26.1q26.2 encompassing *CHD2* and *RGMA"*. The American Society of Human Genetics 63rd Annual Meeting, 2013, Boston, MA

"Exome sequencing identifies a novel mutation in *HEXB* associated with juvenile Sandhoff disease in a consanguineous family with movement disorders and intellectual disability". The Association for Molecular Pathology Meeting, 2013, Phoenix, AZ

"MLH3 gene variants identified by clinical exome sequencing in an infant with multiple benign neuromas, neurofibromas, Schwannomas and hemangiomas: expanding the phenotype of mismatch repair defects". American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting, 2014, Nashville, TN

"First report of 1p36.22 microdeletion in a patient with developmental delay, seizures, and recent onset cardiac and renal failure". American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting, 2014, Nashville, TN

"A ring chromosome 13 with contiguous interstitial duplication and distal deletion in a baby boy with congenital heart malformation". American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting, 2015, Salt Lake City, UT

"Further delineation of segmental uniparental disomy for chromosome 7q (UPD7q): a male patient with 41.7 Mb long contiguous stretch of homozygosity on chromosome 7q21.3q34". American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting, 2016, Tampa, FL

PUBLICATIONS

RESEARCH PAPERS (PEER-REVIEWED)

A RESEARCH PAPERS (PUBLISHED)

1. Koçak U, Gürsel T, Oztürk G, **Kantarci S**. "Thrombosis during all-trans-retinoic acid therapy in a child with acute promyelocytic leukemia and factor VQ 506 mutation" Pediatr Hematol Oncol. 2000; 17(2):177-80.

2. Tadmouri GO, Bilenoglu O, **Kantarci S**, Kayserili H, Perrin P, Basak AN. "A rare mutation [IVS-I-130 (GA)] in a Turkish beta-thalassemia major patient" Am J Hematol. 2000;63(4):223-5.

3. Savas S, Eraslan S, **Kantarci S**, Karaman B, Acarsoz D, Tükel T, Cogulu O, Ozkinay F, Basaran S, Aydinli K, Yuksel-Apak M, Kirdar B. "Prenatal prediction of childhood-onset spinal muscular atrophy (SMA) in Turkish families" Prenat Diagn. 2002; 22(8):703-9.

4. Consugar MB, Anderson SA, Rossetti S, Pankratz VS, Ward CJ, Torra R, Coto E, El-Youssef M, **Kantarci S**, Utsch B, Hildebrandt F, Sweeney WE, Avner ED, Torres VE, Cunningham JM, Harris PC. "Haplotype analysis improves molecular diagnostics of autosomal recessive polycystic kidney disease" Am J Kidney Dis. 2005; 45(1):77-87.

5. **Kantarci S**, Casavant D, Prada C, Russell M, Byrne J, Haug LW, Jennings R, Manning S, Blaise F, Boyd TK, Fryns JP, Holmes LB, Donahoe PK, Lee C, Kimonis V, Pober BR. "Findings from aCGH in patients with congenital diaphragmatic hernia (CDH): a possible locus for Fryns syndrome" Am J Med Genet A. 2006;140 (1):17-23.

6. **Kantarci S**, Al-Gazali L, Hill RS, Donnai D, Black GCM, Bieth E, Chassaing N, Lacombe D, Devriendt K, Teebi A, Loscertales M, Robson C, Liu T, MacLaughlin DT, Noonan KM, Russell MK, Walsh CA, Donahoe PK, Pober BR. "Mutations in LRP2, which encodes the multiligand receptor megalin, cause Donnai- Barrow and facio-oculo-acoustico-renal syndromes" Nat Genet. 2007 39(8); 957-959.
7. **Kantarci S**, Ragge NK, N Thomas S, Robinson DO, Noonan KM, Russell MK, Donnai D, F Raymond L, Walsh CA, Donahoe PK, Pober BR. "Donnai-Barrow syndrome (DBS/FOAR) in a child with a homozygous LRP2 mutation due to complete chromosome 2 paternal isodisomy" Am J Med Genet A. 2008;146A (14):1842-1847.

8. **Kantarci S**, Ackerman KG, Russell MN, Longoni M, Sougnez C, Noonan KM, Hatchwell E, Zhang X, Pieretti-Vanmarcke R, Yeboa K, Dickman P, Wilson J, Donahoe PK, Pober BR." Characterization of the Chromosome 1q41q42.12 region, and the candidate gene DISP1, in patients with CDH" Am J Med Genet A. 2010; 152A (10):2493-504.

9. Park JH, Daheron L, **Kantarci S**, Lee BS, Teixeira JM. "Human endometrial cells express elevated levels of pluripotent factors and are more amenable to reprogramming into induced pluripotent stem cells" Endocrinology. 2011; 152(3):1080-9.

10. Amarillo I, Bui PH, **Kantarci S**, Rao N, Shackley BS, García R, Tirado CA. "Atypical rearrangement involving 3[prime]-IGH@ and a breakpoint at least 400 Kb upstream of an intact MYC in a CLL patient with an apparently balanced t(8;14)(q24.1;q32) and negative MYC expression" Mol Cytogenet. 2013; 6(1):5.

11. Aljinovic N, Bogusz AM, **Kantarci S**, Buck TP, Dewar R. "An unusual case of Philadelphia chromosome positive chronic myelogenous leukemia with trisomy 19 presenting with megakaryoblastosis and myelofibrosis" Arch Pathol Lab Med. 2013; 137(8):1147-51.

12. Bissar Tadmouri N, Donahue WL, Al-Gazali L, Nelson SF, Bayrak-Toydemir P, **Kantarci S**. "X-chromosome exome sequencing reveals a novel ALG13 mutation in a nonsyndromic intellectual disability family with multiple affected male siblings" Am J Med Genet A. 2014; 164(1): 164–69.

13. Amarillo IE, Li WL, Vilain E, Li X, **Kantarci S**. "A de novo intragenic deletion of AUST2 in a patient with autism spectrum disorder and foot deformity: further support for the role of AUTS2 in neurodevelopmental disorders and congenital malformations" Am J Med Genet A. 2014;164A(4):958-65

14. Fogel BL, Lee H, Deignan JL, Strom SP, **Kantarci S**, Wang X, Quintero-Rivera F, Vilain E, Grody WW, Perlman S, Geschwind DH, Nelson SF. "Exome sequencing in the clinical diagnosis of sporadic or familial cerebellar ataxia" JAMA Neurol. 2014; 71(10):1237-46.

15. Lee H, Deignan JL, Dorrani N, Strom SP, **Kantarci S**, Quintero-Rivera F, Das K, Toy T, Harry B, Yourshaw M, Fox M, Fogel BL, Martinez-Agosto JA, Wong DA, Chang VY, Shieh PB, Palmer CG, Dipple KM, Grody WW, Vilain E, Nelson SF. "Clinical exome sequencing for genetic identification of rare Mendelian disorders" JAMA. 2014; 312(18):1880-7.

16. Kallen M, DeNicola M, Pullarkat S, **Kantarci S**, Paquette R, Yang L, Rao N, Tirado CA. "Amplificación de BCR-ABL1 en Leucemia Linfoblástica Agusa tipo B?" Acta Cancerológica, 2014; 43(2):10-13 (Lima, Peru).

17. Arboleda VA, Lee H, Dorrani N, Zadeh N, Willis M, Macmurdo CF, Manning MA, Kwan A, Hudgins L, Barthelemy F, Miceli MC, Quintero-Rivera F, **Kantarci S**, Strom SP, Deignan JL; UCLA Clinical Genomics Center, Grody WW, Vilain E, Nelson SF. "De novo nonsense mutations in KAT6A, a lysine acetyl-transferase gene, cause a syndrome including microcephaly and global developmental delay" Am J Hum Genet. 2015; 96(3):498-506.

18. Jamuar SS, Duzkale H, Duzkale N, Zhang CS, High FA, Kaban L, Bhattacharya S, Crandall B, **Kantarci S**, Stoler JM, Lin AE. "Deletion of chromosome 8q22.1, a critical region for Nablus mask-like facial syndrome: four additional cases support a role of genetic modifiers in the manifestation of the phenotype" Am J Med Genet A. 2015; 167(6):1400-5.

19. Rehm HL, Berg JS, Brooks LD, Bustamante CD, Evans JP, Landrum MJ, Ledbetter DH, Maglott DR, Martin CL, Nussbaum RL, Plon SE, Ramos EM, Sherry ST, Watson MS; **ClinGen (Collaborator: Kantarci S)**. "ClinGen--the Clinical Genome Resource." N Engl J Med. 2015;372(23):2235-42

20. Kansal R, Li X, Shen J, Samuel D, Laningham F, Lee H, Panigrahi G, Shuen A, **Kantarci S**, Dorrani N, Reiss J, Shintaku P, Deignan J, Samuel D, Strom S, Pearson CE, Vilain E, Grody WW. "An infant with MLH3 variants, FOXG1 duplication and multiple, benign cranial and spinal tumors: a clinical exome sequencing study" Genes Chromosomes Cancer. 2016; 55(2):131-42.

21. Ordulu Z, Kammin T, Brand H, Pillalamarri V, Redin CE, Collins R, Blumenthal I, Hanscom C, Pereira S, Bradley I, Crandall BF, Gerrol P, Hayden MA, Hussain N, Kanengisser-Pines B, **Kantarci S,** Levy B, Quintero-Rivera F, Speigel E, Stevens S, Ulm JE, Warburton D, Wilkins-Haug LE, Yachelevich N, Gusella JG, Talkowski ME, Morton CC. "Structural chromosome rearrangements require nucleotide level resolution: Lessons from next-generation sequencing in prenatal diagnosis" Am J Hum Genet. 2016; 99(5):1015-1033.

22. Schoch K, Meng L, Szelinger S, Bearden DR, Stray-Pedersen A, Busk OL, Stong N, Liston E, Cohn RD, Scaglia F, Rosenfeld JA, Tarpinian J, Skraban CM, Deardorff MA, Friedman JN, Akdemir ZC, Walley N, Mikati MA, Kranz PG, Jasien J, McConkie-Rosell A, McDonald M, Wechsler SB, Freemark M, Kansagra S, Freedman S, Bali D, Millan F, Bale S, Nelson SF, Lee H, Dorrani N; **UCLA Clinical Genomics Center**; Undiagnosed Diseases Network, Goldstein DB, Xiao R, Yang Y, Posey JE, Martinez-Agosto JA, Lupski JR, Wangler MF, Shashi V. "A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay" Am J Hum Genet. 2017 Feb 2;100(2):343-351.

23. Mullegama SV, Jensik P, Li C, Dorrani N; UCLA Clinical Genomics Center, **Kantarci S**, Blumberg B, Grody WW, Strom SP. "Coupling clinical exome sequencing with functional characterization studies to diagnose a patient with familial Mediterranean fever and MED13L haploinsufficiency syndromes" Clin Case Rep. 2017 Apr 18;5(6):833-840.

B. BOOK CHAPTERS

1. **Kantarci S.** (Invited Author) "Mechanisms of Cytogenetic Aberrations Resulting in Abnormal Phenotypes" Pathobiology of Human Disease: A Dynamic Encyclopedia of Disease Mechanisms. Section: Cytogenetics, Elsevier Inc. [database online; v.2] Science Direct. January 2015.

2. **Kantarci S,** Cherukuri D, and Vasef M (Invited author) "Chromosomal Microarray Analysis" Diagnostic Pathology: Molecular Oncology, Elsevier Inc. June 2015

C. REVIEWS

1. **Kantarci S**, Donahoe PK. "Congenital diaphragmatic hernia (CDH) etiology as revealed by pathway Genetics" Am J Med Genet C Semin Med Genet. 2007; 145C (2):217-26.

2. **Kantarci S**, Donnai D, Noonan KM, Pober BR. "Donnai-Barrow syndrome" (June 2011) in: GeneReviews at GeneTests: Medical Genetics Information Resource [database online]. Copyright, University of Washington, Seattle, 1997-2010.

D. INVITED ARTICLES

1. Haspel RL, Arnaout R, Briere L, **Kantarci S**, Marchand K, Tonellato P, Connolly J, Boguski MS, Saffitz JE. "A call to action: Training pathology residents in genomics and personalized medicine "Am J Clin Pathol. 2010; 133(6):832-4.

2. Ligon AH, Morton CC, Bieber FR, Fletcher JA, Giersch AB, **Kantarci S**, Leach N, Lee C, Sandstrom M, Weremowicz S, Xiao S, Dal Cin P. "Reporting of Diagnostic Cytogenetic Results" Curr. Protoc. Hum. Genet. 2010; 67:A.1D.1-A.1D.24 by John Wiley & Sons, Inc.

E. ABSTRACTS

1. **Kantarci S**, Apak MY, Eraslan S, Nayir A, Kayserili H, Kirdar B. "Haplotype analysis in autosomal recessive polycystic kidney disease (ARPKD) families and the determination of allele frequencies of used markers in the Turkish population" The American Society of Human Genetics 53rd Annual Meeting, 2003, Los Angeles, CA

2. Hatchwell E, Tommerup N, Kristoffersson U, Stanyon R, **Kantarci S.** "Joubert syndrome: a patient with a de novo t(2;22)(q13;q11.1)" The American Society of Human Genetics 53rd Annual Meeting, 2003, Los Angeles, CA

3. **Kantarci S**, Tommerup N, Kristoffersson U, Stanyon R, Boltshauser E, Hatchwell E. "Identification and characterization of a candidate gene in Joubert syndrome" European Human Genetics Conference, 2004, Munich, Germany

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