BIOGRAPHICAL SKETCH

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NAME: Jianling Ji

eRA COMMONS USER NAME (credential, e.g., agency login): JIJIANLING

POSITION TITLE: Assistant Professor of Clinical Pathology, Department of Pathology, Keck School of Medicine of USC

EDUCATION/TRAINING *(Begin with baccalaureate or other initial professional education, such as nursing, include postdoctoral training and residency training if applicable. Add/delete rows as necessary.)*

INSTITUTION AND LOCATION	DEGREE *(if applicable)*	Completion Date MM/YYYY	FIELD OF STUDY
Nantong University, Jiangsu Province, China	MD	07/1998	Clinical Medicine
Nantong University, Jiangsu Province, China	MS	07/2004	Internal Medicine-Hematology
Affiliated Hospital of Nantong University, Jiangsu, China	Residency	07/2004	Pediatrics
UCLA Intercampus Medical Genetics Training Program---Cedars Sinai Medical Center and UCLA	Fellowship	06/2014	Clinical Cytogenetics
UCLA Intercampus Medical Genetics Training Program---UCLA	Fellowship	06/2015	Clinical Molecular Genetics

A. Personal Statement.

Growing up in southern China, I received my MD and MS degrees from Nantong University. I then completed a pediatric residency and fellowship training at the affiliated hospital of Nantong University, where I had a strong focus on pediatric oncology research. My research interest centered around the study of bone marrow angiogenesis and the expression of vascular endothelia growth factor in aplastic anemia. This research resulted in two publications in one of the best journals in China, with one published in English. My experience of balancing time between the clinic and the research laboratory laid the groundwork for my future career direction. After relocating to the US, I began working as a research associate at City of Hope National Medical Center, where I developed a strong research interest in cancer genetics under the mentorship of Dr. Marilyn L. Slovak. I actively participated in the genetics aspects of several studies exploring chemotherapeutic approaches to patients with newly diagnosed acute lymphoblastic leukemias. In addition, I contributed to a study on dedifferentiated liposarcoma and undifferentiated high-grade pleomorphic sarcoma, resulting in a paper published in the American Journal of Surgical Pathology. Furthermore, I played an important role in designing, developing, and validating new DNA probes for clinical use, along with multiple in house research projects. All these earlier research experiences laid a solid foundation for my career path.

During my fellowship years at the University of California, Los Angeles, I was awarded three grants for profiling genomic characteristics of plasmablastic lymphoma using whole exome sequencing and array technologies. Although obtaining samples for this specific tumor type was a challenge, I learned to bring together different groups of people from multiple institutions to accomplish tasks.

As a faculty at the CHLA Center for Personalized Medicine (CPM), I am involved in both germline and somatic aspects of clinical responsibilities. I work closely with the bioinformatics team and was instrumental in developing multiple clinical tests at CPM. I led the development and validation of CPM’s clinical exome sequencing test, launched in May 2016. This test has significantly improved clinicians' ability to diagnose a wide range of genetic disorders and currently is the highest volume test in our laboratory. In addition to the exome sequencing test, I have led the development and validation of multiple other clinical tests, including a pediatric cancer predisposition panel to detect sequence variants and copy number alterations, enhancing diagnostic performance beyond that offered by routine sequencing-based tests. With colleagues in

Neuropathology, I led the validation of our methylation profiling assay for the diagnosis of tumors in the central nervous system. Currently, I am working on development of a sequencing-based methylation profiling method for liquid biopsy diagnosis. I am also actively engaged in ongoing efforts to refine our RNA-seq capabilities for pediatric cancer diagnosis.

In 2020, I received a CHLA Department of Pathology & Laboratory Medicine Interdisciplinary Translational Research Pilot Grant for "Solving the Unsolved: New Strategies to Increase Diagnostic Yield in Negative-Exome Cases". We sought to identify a metric for objective quantification of patient-reported clinical features matching the disease phenotype, develop a bioinformatic solution to prioritize variants from exome sequencing, and integrate analysis of DNA sequence variants and copy number variants. The preliminary findings resulted in a manuscript for which I am senior author entitled "Significance Associated with Phenotype (SAP) Score Aids in Variant Prioritization for Exome Sequencing Analysis," currently under revision in the Journal of Molecular Diagnostics. I have continued to study optimization of genomic diagnosis, including most recently performing copy number variant (CNV) analysis on retrospective exome sequencing data to identify causative findings in an additional 7% of cases that were previously missed with our standard workflow. My work and expertise in this area is reflected by invitations to present at national conferences and peer institutions, and to serve as an elected member of the Association for Molecular Pathology (AMP) Clinical Practice Committee and the Whole Exome Sequencing Standards working group. Through the latter roles, I have had the opportunity to significantly contribute to the field through development of practice guidelines, including most recently the development of the practice guidance document “Exome Slice Testing - Considerations from Ordering to Reporting: A Joint Report of the Association for Molecular Pathology, College of American Pathologists, and National Society of Genetic Counselors”.

I have also been very fortunate to be involved in the genomic interpretation and multiple research projects on pediatric brain tumors for the past several years. One of the research studies aimed at elucidating the genetic profiles and clinical outcomes of G34 tumors is something that has excited me. This serves as a pilot study for the design of a larger investigation in the future and can eventually lead to more accurate diagnoses, prognoses, and therapeutic intervention. Recently, using cutting-edge Optical Genome Mapping we have identified a potential subgroup of supratentorial ependymomas with complex rearrangements resulting from balanced chromothriptic events. I was invited to present these novel findings at the recent 2023 Cancer Genomics Consortium (CGC) annual meeting.

Having a research career is not easy, and sometimes it can be very frustrating. However, as I find myself walking to my car rather late every evening, it is always with a smile on my face. I know in my heart that this is what I really want to do. Somehow, research brings me joy. I am a focused and hard-working person, and I am confident that the opportunity will offer me the opportunity to learn, concentrate, and problem-solve.

My goal is to combine my interests in clinical care and research by engaging in research that will advance the application of molecular diagnostic technologies to the understanding and treatment of human disease.

B. Positions and Honors

Positions and Employment

2004-2005	Attending Pediatrician, Department of Pediatrics, Affiliated Hospital of Nantong University, Nantong, Jiangsu, China
2015-present	Assistant Professor of Clinical Pathology, Department of Pathology, Keck School of Medicine of USC

Professional Society Memberships:

2012-present	American Society of Human Genetics (ASHG)
2014-present	Association of Molecular Pathologists (AMP)
2015-present	American College of Medical Genetics (ACMG)
2015-present	Cancer Genomic Consortium (CGC)

Professional Service:

2016	Program committee member, 2016 Cancer Genomic Consortium (CGC) Annual Meeting, Denver, Colorado	Cancer Genomic Consortium (CGC)
2018-2021	Committee member, CLSI Document Development Committee for Nucleic Acid Sequencing (MM09) documentation	Clinical and Laboratory Standards Institute (CLSI)
2018-2019	Genetics Subdivision Representative, AMP Clinical Practice Committee	Association for Molecular Pathology (AMP)
2018-current	Committee member, ClinGen Pediatric Somatic Working Group	ClinGen-The Clinical Genome Resource
2020-2022	Committee member, Whole Exome Sequencing Standards Working Group	Association for Molecular Pathology (AMP)
2021-current	Committee member, Histone H3 Somatic Cancer Variant Curation Expert Panel	ClinGen-The Clinical Genome Resource
2022-2023	Program committee member, plan and moderate four scientific sessions, 2022 and 2023 AMP Annual Meeting	Association for Molecular Pathology (AMP)

Honors

2012	CTSI Clinical Scholars Award
2014	David L. Rimoin Award for Research Excellence in Medical Genetics
2023	Gordon F. Vawter Pathologist-in-Training Award (Jolee Suddock, Neuropathology Fellow), Society for Pediatric Pathology 2023 Fall Meeting

Professional community Service:

2016	Co-moderator	2016 CGC annual meeting, Denver, Colorado	Moderating a scientific session: Constitutional Genomic Testing
2017	Co-Moderator	2017 ACMG annual meeting, Phoenix, Arizona	Moderating a scientific session: Molecular Cytogenomics: The Next Generation in Balanced Rearrangement Detection
2017	Co-Moderator	2017 AMP annual meeting, Salt Lake City, Utah	Moderating a scientific session: Genetics of Spinal Muscular Atrophy and Amyotrophic Lateral Sclerosis: A Timely Review
2021	Moderator	2021 ASHG annual meeting, Virtual meeting	Moderating a scientific session: New approaches for early detection and prognosis of cancer
2022	Moderator	2022 AMP annual meeting	Moderating 3 scientific sessions: 1. Challenges and Considerations of Germline Testing In the Era of Tumor Sequencing, 2. Diagnostic and Therapeutic Updates of Cancer Predisposition, 3. Revealing the Unknown Regions of Genome
2023	Moderator	2023 AMP annual meeting	Moderating 5 scientific sessions: 1. Case Studies in Genetics 2. Beyond the Exome: What's Next in Diagnostic Testing for Rare Genetic Disorders 3. Use of Genomics in Newborn Screening Programs: The Opportunities and Challenges 4. Rapid Molecular Diagnostics 5. Epigenetic Clocks, Aging, and Cancer

C. Contributions to Science

Peer-reviewed publications:

1. **Ji J,** Xu M, Huang F, Liu H. Bone marrow angiogenesis in aplastic anemia. *J Exp Hematol*. 14(1): 79-82, 2006. PMID: 16584597. Conducted study; Generated data; Drafted manuscript; Edited manuscript

2. **Ji J**, Liu H, Sun C, Jiang SH, Ding RS. Expression of vascular endothelial growth factor in patients with aplastic anemia and its significance. *J Exp Hematol.* 2006 Apr;14(2):285-8. Chinese. PMID: 16638198. Conducted study; Generated data; Drafted manuscript; Edited manuscript
3. Chung L, Lau SK, Jiang Z, Loera S, Bedel V, **Ji J**, Weiss LM, Chu PG. Overlapping features between dedifferentiated liposarcoma and undifferentiated high-grade pleomorphic sarcoma. *Am J Surg Pathol.* 2009 Nov;33(11):1594-600. doi: 10.1097/PAS.0b013e3181accb01. PMID: 19574885. Generated data; Edited manuscript.
4. **Ji J**, Loo E, Pullarkat S, Yang L, Tirado CA. Acute myeloid leukemia with t(7;21)(p22;q22) and 5q deletion: a case report and literature review. *Exp Hematol Oncol.* 2014 Mar 19;3:8. doi: 10.1186/2162-3619-3-8. PMID: 24646765; PMCID: PMC4012275. Conducted study; Generated data; Drafted manuscript; Edited manuscript
5. **Ji J,** Lee H, Argiropoulos B, Dorrani N, Mann J, Martinez-Agosto JA, Gomez- Ospina N, Gallant N, Bernstein JA, Hudgins L, Slattery L, Isidor B, Le Caignec C, David A, Obersztyn E, WiÅ›niowiecka-Kowalnik B, Fox M, Deignan JL, Vilain E, Hendricks E, Horton Harr M, Noon SE, Jackson JR, Wilkens A, Mirzaa G, Salamon N, Abramson J, Zackai EH, Krantz I, Innes AM, Nelson SF, Grody WW, Quintero-Rivera F. DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. *Eur J Hum Genet.* 2015 Nov;23(11):1473-81. doi: 10.1038/ejhg.2015.71. Epub 2015 May 6. PMID: 25944381; PMCID: PMC4613469. Conducted study; Generated data; Drafted manuscript; Edited manuscript
6. **Ji J,** Quindipan C, Parham D, Shen L, Ruble D, Bootwalla M, Maglinte DT, Gai X, Saitta SC, Biegel JA, Mascarenhas L. Inherited germline ATRX mutation in two brothers with ATR-X syndrome and osteosarcoma. *Am J Med Genet A.* 2017 May;173(5):1390-1395. doi: 10.1002/ajmg.a.38184. Epub 2017 Mar 28. PMID: 28371217; PMCID: PMC7521841. Conducted study; Generated data; Drafted manuscript; Edited manuscript
7. Menke LA; DDD study; Gardeitchik T, Hammond P, Heimdal KR, Houge G, Hufnagel SB, **Ji J**, Johansson S, Kant SG, Kinning E, Leon EL, Newbury-Ecob R, Paolacci S, Pfundt R, Ragge NK, Rinne T, Ruivenkamp C, Saitta SC, Sun Y, Tartaglia M, Terhal PA, van Essen AJ, Vigeland MD, Xiao B, Hennekam RC. Further delineation of an entity caused by CREBBP and EP300 mutations but not resembling Rubinstein-Taybi syndrome. *Am J Med Genet A.* 2018 Apr;176(4):862-876. doi: 10.1002/ajmg.a.38626. Epub 2018 Feb 20. PMID: 29460469. Generated data; Edited manuscript
8. Oberley MJ, Denton C, **Ji J,** Hiemenz M, Bhojwani D, Ostrow D, Wu S, Gaynon P, Raca G. A neoplasm with FIP1L1-PDGFRA fusion presenting as pediatric T-cell lymphoblastic leukemia/lymphoma without eosinophilia. *Cancer Genet.* 2017 Oct;216-217:91-99. doi: 10.1016/j.cancergen.2017.07.007. Epub 2017 Aug 3. PMID: 29025601; PMCID: PMC7469920. Generated data; Edited manuscript
9. *Hajek CA, **Ji J**, Saitta SC. Interstitial Chromosome 3p13p14 Deletions: An Update and Review. *Mol Syndromol*. 2018 May;9(3):122-133. doi: 10.1159/000488168. Epub 2018 Apr 7. PMID: 29928177; PMCID: PMC6006617. Drafted manuscript; Edited manuscript
10. Hiemenz MC, Ostrow DG, Busse TM, Buckley J, Maglinte DT, Bootwalla M, Done J, **Ji J**, Raca G, Ryutov A, Xu X, Zhen CJ, Conroy JM, Hazard FK, Deignan JL, Rogers BB, Treece AL, Parham DM, Gai X, Judkins AR, Triche TJ, Biegel JA. OncoKids: A Comprehensive Next-Generation Sequencing Panel for Pediatric Malignancies. *J Mol Diagn*. 2018 Nov;20(6):765-776. doi: 10.1016/j.jmoldx.2018.06.009. Epub 2018 Aug 20. PMID: 30138724. Generated data; Edited manuscript
11. **Ji J**, Shen L, Bootwalla M, Quindipan C, Tatarinova T, Maglinte DT, Buckley J, Raca G, Saitta SC, Biegel JA, Gai X. A semiautomated whole-exome sequencing workflow leads to increased diagnostic yield and identification of novel candidate variants. *Cold Spring Harb Mol Case Stud*. 2019 Apr 1;5(2):a003756. doi: 10.1101/mcs.a003756. PMID: 30755392; PMCID: PMC6549575. Conducted study; Generated data; Drafted manuscript; Edited manuscript
12. **Ji J,** Navid F, Hiemenz MC, Kaneko M, Zhou S, Saitta SC, Biegel JA. Embryonal rhabdomyosarcoma in a patient with a germline CBL pathogenic variant. *Cancer Genet*. 2019 Feb;231-232:62-66. doi: 10.1016/j.cancergen.2018.12.006. Epub 2018 Dec 30. PMID: 30803559; PMCID: PMC7528629. Conducted study; Generated data; Drafted manuscript; Edited manuscript
13. Tiwari N, Tamrazi B, Robison N, Krieger M, **Ji J**, Tian D. Unusual radiological and histological presentation of a diffuse leptomeningeal glioneuronal tumor (DLGNT) in a 13-year-old girl. *Childs Nerv Syst*. 2019 Sep;35(9):1609-1614. doi: 10.1007/s00381-019-04074-7. Epub 2019 Feb 15. PMID: 30770994; PMCID: PMC7474550. Generated data; Drafted manuscript; Edited manuscript.
14. Sheppard SE, Lalonde E, Adzick NS, Beck AE, Bhatti T, De Leon DD, Duffy KA, Ganguly A, Hathaway E, **Ji J,** Linn R, Lord K, Randolph LM, Sajorda B, States L, Conlin LK, Kalish JM. Androgenetic chimerism as an etiology for Beckwith- Wiedemann syndrome: diagnosis and management. *Genet Med.* 2019 Nov;21(11):2644-2649. doi: 10.1038/s41436-019-0551-9. Epub 2019 May 31. PMID: 31147633; PMCID: PMC7848850. Generated data; Drafted manuscript; Edited manuscript.
15. *Kaneva K, Yeo KK, Hawes D, **Ji J**, Biegel JA, Nelson MD, Bluml S, Krieger MD, Erdreich-Epstein A. Rare Pediatric Invasive Gliofibroma Has BRAFV600E Mutation and Transiently Responds to Targeted Therapy Before Progressive Clonal Evolution. *JCO Precis Oncol.* 2019;3:PO.18.00138. doi: 10.1200/PO.18.00138. Epub 2019 Mar 27. PMID: 31179415; PMCID: PMC6555144. Generated data; Edited manuscript.
16. Arboleda-Velasquez JF, Lopera F, O'Hare M, Delgado-Tirado S, Marino C, Chmielewska N, Saez-Torres KL, Amarnani D, Schultz AP, Sperling RA, Leyton- Cifuentes D, Chen K, Baena A, Aguillon D, Rios-Romenets S, Giraldo M, Guzmán-Vélez E, Norton DJ, Pardilla-Delgado E, Artola A, Sanchez JS, Acosta-Uribe J, Lalli M, Kosik KS,

Huentelman MJ, Zetterberg H, Blennow K, Reiman RA, Luo J, Chen Y, Thiyyagura P, Su Y, Jun GR, Naymik M, Gai X, Bootwalla M, **Ji J**, Shen L, Miller JB, Kim LA, Tariot PN, Johnson KA, Reiman EM, Quiroz YT. Resistance to autosomal dominant Alzheimer's disease in an APOE3 Christchurch homozygote: a case report. *Nat Med.* 2019 Nov;25(11):1680-1683. doi: 10.1038/s41591-019-0611-3. Epub 2019 Nov 4. PMID: 31686034; PMCID: PMC6898984. Generated data; Edited manuscript.

17. Quindipan C, Cotter JA, **Ji J**, Mitchell WG, Moke DJ, Navid F, Thomas, SM, VanHirtum-Das V, Wang L, Saitta SC, Biegel JA, Hiemenz M. Custom Pediatric Oncology Next-Generation Sequencing Panel Identifies Somatic Mosaicism in Archival Tissue and Enhances Targeted Clinical Care. *Pediatric Neurology*. 114:55-59, 2021. PMID: 33221597. Generated data; Drafted manuscript; Edited manuscript
18. Hiemenz MC, Oberley MJ, Doan A, Aye L, **Ji J**, Schmidt RJ, Biegel JA, Bhojwani D, Raca G. A multimodal genomics approach to diagnostic evaluation of pediatric hematologic malignancies. *Cancer Genet. 2021 Jun;254-255:25-33.* doi: 10.1016/j.cancergen.2021.01.007. Epub 2021 Jan 21. PMID: 33571894. Generated data; Drafted manuscript; Edited manuscript
19. *Takeda MR, Bansal M, Kamerman-Kretzmer RJ, Church J, **Ji J,** Warren M. Bronchiectasis and Bronchiolectasis with Severe Herniating Pattern Associated with STAT1 Gain-of-Function Mutation: Detailed Clinicopathological Findings. *Pediatr Dev Pathol*. 2021 Mar-Apr;24(2):131-136. doi: 10.1177/1093526620985950. Epub 2021 Jan 13. PMID: 33439110. Generated data; Edited manuscript.
20. **Ji J,** Kaneva K, Hiemenz MC, Dhall G, Davidson TB, Erdreich-Epstein A, Hawes D, Hurth K, Margol AS, Mathew AJ, Robison NJ, Schmidt RJ, Tran HN, Judkins AR, Cotter JA, Biegel JA. Clinical utility of comprehensive genomic profiling in central nervous system tumors of children and young adults. *Neurooncol Adv.* 2021 Feb 25;3(1):vdab037. doi: 10.1093/noajnl/vdab037. PMID: 33948563; PMCID: PMC8080244. Conducted study; Generated data; Drafted manuscript; Edited manuscript
21. Chang EK, Smith-Cohn MA, Tamrazi B, **Ji J**, Krieger M, Holdhoff M, Eberhart CG, Margol AS, Cotter JA. IDH-mutant brainstem gliomas in adolescent and young adult patients: Report of three cases and review of the literature. *Brain Pathol.* 2021 Jul;31(4):e12959. doi: 10.1111/bpa.12959. Epub 2021 May 7. PMID: 33960568; PMCID: PMC8412065. Generated data; Edited manuscript
22. ****Ji J,** **Leung ML, Baker S, Deignan JL, Santani A. Clinical Exome Reanalysis: Current Practice and Beyond. *Mol Diagn Ther.* 2021 Sep;25(5):529-536. doi: 10.1007/s40291-021-00541-7. Epub 2021 Jul 20. PMID: 34283395; PMCID: PMC8410709. Conducted study; Generated data; Drafted manuscript; Edited manuscript
23. Keddy C, Neff T, Huan J, Nickerson JP, Beach CZ, Akkari Y, **Ji J**, Moore S, Nazemi KJ, Corless CL, Beadling C, Woltjer R, Cho YJ, Wood MD, Davare MA. Mechanisms of targeted therapy resistance in a pediatric glioma driven by ETV6-NTRK3 fusion. *Cold Spring Harb Mol Case Stud.* 2021 Oct 19;7(5):a006109. doi: 10.1101/mcs.a006109. PMID: 34429303; PMCID: PMC8559620. Generated data; Edited manuscript
24. **Leung ML, ****Ji J**, Baker S, Buchan JG, Sivakumaran TA, Krock BL, Hutchins R, Bayrak-Toydemir P, Pfeifer J, Cremona ML, Funke B, Santani AB. A Framework of Critical Considerations in Clinical Exome Reanalyses by Clinical and Laboratory Standards Institute. *J Mol Diagn*. 2022 Feb;24(2):177-188. doi: 10.1016/j.jmoldx.2021.11.004. PMID: 35074075. Co-first author. Conducted study; Generated data; Drafted manuscript; Edited manuscript
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26. Yeo KK, Alexandrescu S, Cotter JA, Vogelzang J, Bhave V, Li MM, **Ji J**, Benhamida JK, Rosenblum MK, Bale TA, Bouvier N, Kaneva K, Rosenberg T, Lim-Fat MJ, Ghosh H, Martinez M, Aguilera D, Smith A, Goldman S, Diamond EL, Gavrilovic I, MacDonald TJ, Wood MD, Nazemi KJ, Truong A, Cluster A, Ligon KL, Cole K, Bi WL, Margol AS, Karajannis MA, Wright KD. Multi-institutional study of the frequency, genomic landscape, and outcome of IDH-mutant glioma in pediatrics. *Neuro Oncol*. 2023 Jan 5;25(1):199-210. doi: 10.1093/neuonc/noac132. PMID: 35604410; PMCID: PMC9825351. Generated data; Edited manuscript
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38. **Ji J**, Leung ML. Clinical Utility and Long-Term Feasibility of Exome and Genome Reanalysis: From the Perspectives of a Clinical Laboratory. *J Appl Lab Med.* 2024 Jan 3;9(1):162-167. doi: 10.1093/jalm/jfad062. PMID: 38167756. Drafted manuscript; Edited manuscript

Manuscript review:

2015	Molecular Cytogenetics (1)
2017	Prenatal Diagnosis (1)
2017	Molecular Cytogenetics (1)
2017-2018	European Journal of Human Genetics (2)
2018	BMC Medical Genomics (1)
2019	Cold Spring Harbor Molecular Case Studies (1)
2019	The Journal of Molecular Diagnostics (1)
2019-2023	Genetics in Medicine (3)
2019-2023	The Journal of Molecular Diagnostics (6)
2020	Neuropathology and Applied Neurobiology (1)
2020	Human Genetics and Genomics Advances (1)
2021	Pediatric and Developmental Pathology (1)
2021	Genetics in Medicine (2)
2022-2023	Cancer Genetics (2)

D. Additional Information: Research Support and/or Scholastic Performance

MAJOR AREAS OF RESEARCH INTEREST

1. Molecular diagnosis of rare Mendelian disorders using cutting-edge technologies
2. Pediatric cancer predisposition genetics
3. Genetic alterations in pediatric brain tumors and their potential targeted therapy

RESEARCH IN PROGRESS

1. Significance Associated with Phenotype (SAP) Score: A Method for Ranking Genes and Genomic Regions Based on Patient's Phenotype
2. Copy Number Variant Analysis Improves the Diagnostic Yield in a Cohort of Pediatric Patients with Previously Negative Constitutional Exome Sequencing Results
3. Optical Genome Mapping Reveals New Insights into ZFTA Fusion in Supratentorial Ependymomas: Uncovering an Additional Underlying Genetic Mechanism
4. Identification, Genetic Characterization, and Clinical Outcome Analysis of Pediatric High Grade Gliomas with H3 G34R/V Mutation
5. Spatial Genomics on Medulloblastoma with Potential Duo-Pathway Activation

GRANT SUPPORT – PAST

PI: 8231000-TUA010251 (Fund Code 1029) Dates of Award: 6/22/2020-6/22/2021

Agency: CHLA Department of Pathology and Laboratory Medicine, Pilot Award Percent Effort: 15%

Title: Solving the unsolved: A retrospective study of negative-exome cases using new strategies for an increased diagnostic yield

Description: The diagnostic yield using our current routine clinical exome sequencing for suspected Mendelian disorder ranges from 25-45%, leaving over 50% of patients remaining without a molecular diagnosis. In order to capture the missing diagnoses, we propose some novel approaches, including automated exome reanalysis with the automated system using Exomiser and SAP scores, CNV detection using NxClinical, and whole genome and RNA-sequencing analysis for all the undiagnosed patients.

Role: Principal Investigator

$25,875.00

Co-PI: 8231000-TUA010249 (Fund Code 1029) Dates of Award: 1/27/2020-1/27/2022

Agency: CHLA Department of Pathology and Laboratory Medicine, Interdisciplinary Research Award Percent Effort: 10%

Title: Identification, genetic characterization, and clinical outcome analysis of pediatric high grade gliomas with H3 G34R/V mutation

Description: High grade gliomas with H3F3A G34R/V mutations ("G34 tumors") are a recently described subset of pediatric and adolescent/young adult gliomas which occur in the cerebral cortex. In the past G34 tumors have been misdiagnosed as embryonal tumors (ET) or combined with the diverse set of pediatric high grade gliomas, which has hampered understanding of their biology and natural history. Appropriately identifying this group of tumors has a direct impact on patient care since clinical treatment strategies for ET and HGG significantly differ. To date only small case series have been assembled. This project will identify and collect existing archival cases of G34 tumors to further study their genomic profiles and clinical features. In addition, we will better define the histologic features of this subgroup of tumors and in parallel develop computational approaches to recognize them based on histology alone.

Role: Co-principal investigator

$ 99,250.00

Co-PI: 8231000-TUA010249 (Fund Code 1029) Dates of Award: 6/22/2020-6/22/2021

Agency: CHLA Department of Pathology and Laboratory Medicine, Pilot Award Percent Effort: 5%

Title: Genome-wide copy number variation profiling in hepatocellular malignant neoplasm, not otherwise specified (HCN-NOS)

Description: HCN-NOS is a rare and poorly understood type of primary liver cancer and often poses a diagnostic challenge even to experienced pediatric pathologists due to a lack of specific molecular markers. We hypothesize that HCN-NOS has a distinctive CNV profile. We aim to identify recurrent CNVs that distinguish HCN-NOS from both classic HB and HCC and to identify potential oncogenes and tumor-suppressor genes significantly associated with HCN-NOS.

Role: Co-principal investigator

$ 25,000.00

Co-PI: 8231000-TUA010249 (Fund Code 1029) Dates of Award: 1/27/2020-1/27/2021

Agency: CHLA Department of Pathology and Laboratory Medicine, Pilot Award Percent Effort: 0.5%

Title: Identification of Balanced Structural Variants Causing Rare Genetic Disorders

Description: Emerging methods now allow for the identification of balanced structural variants, which also have the potential to turn off gene function and thereby represent a source of disease-causing genetic variants. As many cases of suspected genetic disease remain unsolved, this presents an exciting opportunity for establishing leadership in discovering mechanisms of gene inactivation and poses the potential to enhance genetic diagnosis at CHLA. We propose to implement newly developed methods of balanced structural variant discovery at the CHLA Center for Personalized Medicine and refine them for future clinical genetic diagnosis.

Role: Co-principal investigator

$ 25,000.00

PI, CTSI Clinical Scholars Grant, Project 223216 Dates of Award: 7/20/2013-7/20/2015

Agency: CTSI Clinical Scholars Program, Cedars Sinal Medical Center Percent Efoort: 15%

Title: A comparison study of genetic alterations in plasmablastic lymphomas (PL), plasma cell myelomas (PCM) and diffuse large B cell lymphomas (DLBCL)

Description: The goal of the study was to define the cytogenetic features and genomic profiles of PL, PCM and DLBCL using a variety of genetic techniques, including single nucleotide polymorphism (SNP) chromosomal microarray, exome sequencing, and fluorescence in situ hybridization.

Role: Principal Investigator

$30,000.00

Co-PI: Clinical Translational Research Fund, TRF 401450 YX 62220 Dates of Award: 8/15/2013-8/15/2014

Agency: Department of Pathology and Laboratory Medicine, UCLA Health System Percent Effort: 15%

Title: Characterization of genetic alterations in plasmablastic lymphoma by SNP chromosomal microarray analysis

Description: The current information available on the genetic alterations involved in the pathogenesis of the PL is extremely limited. The proposed research aims to characterize the genetic features of PL (MYC+ and MYC-) using SNP-chromosomal microarray technology on 20 samples.

Role: Co-principal investigator

$10,000.00

Co-PI: UCLA CTT Core Voucher Fund, UL1TR000124, V084 Dates of Award: 5/7/2014-5/7/2015

Agency: Clinical and Translational Science Institute, UCLA Health System Percent Effort: 15%

Title: Characterization of genetic alterations in plasmablastic lymphomas

Description: In the first part of the study, we have characterized the genomic CNV alterations using high resolution chromosomal (Oncoscan) microarrays. The goal of this project is to characterize the genetic alterations of PL using exome sequencing. Six samples were included in the project.

Role: Co-principal investigator

$9,960.00