JARIYA UPADIA, MD

Curriculum Vitae \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

PERSONAL INFORMATION

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Language: English, Thai

**EDUCATION AND TRAINING**

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| --- | --- | --- | --- |
| Year | Degree | Major | Education Site |
| 2003 | Doctor of Medicine  (M.D.) | Medicine | Khon Kaen University Khon Kaen, Thailand |

**POST-GRADUATE OR OTHER TRAINING**

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| --- | --- | --- | --- |
| Date | Training type | Specialty | Training site |
| 2017-2018 | Fellowship training | Medical Biochemical Genetics | Duke University School of Medicine |
| 2015-2017 | Residency training | Medical Genetics | University of Alabama at Birmingham |
| 2006-2009 | Residency training | Pediatrics | Phramongkutklao College of Medicine, Bangkok, Thailand |

**BOARD CERTIFICATION AND STATE LICENSE**

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| --- | --- | --- | --- | --- |
| Date | Board | Specialty | Certification # | Renewal and Maintenance |
| 2019 | American Board of Medical Genetics and Genomics (ABMGG) | Medical Biochemical Genetics | 2019235 | Current: Cycle 2024-2026  Previous: Cycle 2021-2023 |
| 2017 | American Board of Medical Genetics and Genomics (ABMGG) | Clinical Genetics and Genomics | 2017212 | Current: Cycle 2024-2026  Previous: Cycle 2018-2020, 2021-2023 |
| 2018 | Louisiana State Board of Medical Examiners (LSBME) | Physician&Surgeon | 310879 | Current: 2023  Previous: 2022, 2021, 2021, 2020, 2019, 2018 |
| 2018 | Louisiana Board of Pharmacy | CDS License-Physician | 053340 | Current: 2023  Previous: 2022, 2021, 2020, 2019, 2018 |

**ACADEMIC APPOINTMENTS**

|  |  |  |  |
| --- | --- | --- | --- |
| Date | Appointment Rank | Department | Institution |
| 9/2018-present | Assistant Professor  (Clinical Track) | Department of Pediatrics | Tulane University, New Orleans, LA, USA |

**ADMINISTRATIVE APPOINTMENTS**

|  |  |  |
| --- | --- | --- |
| Date | Title | Institution |
| 7/2019-present | Course Director  Medical student (T1)- Medical Genetics  Tulane University School of Medicine | Tulane University, New Orleans, LA, USA |

**PROFESSIONAL ORGANIZATIONS**

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| --- | --- | --- |
| Date | Role | Professional Organizations |
| 2020-present | Member | Association of Professors of Human and Medical Genetics (APHMG) |
| 2018-present | Fellow | American College of Medical Genetics and Genomics (ACMG) |

**INVITED LECTURES**

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| **Date** | **Title** | **Location** |
| **2/2024** | Maximal Dietary Responsiveness After Tetrahydrobiopterin (BH4) in 19 Phenylalanine Hydroxylase Deficiency Patients: What Super-Responders Can Expect | Genetics Grand Rounds, Tulane University School of Medicine |
| **5/2024** | Student Evaluation of Genetics Curriculum  at Tulane University School of Medicine | APHMG Annual Meeting,  Skamania Lodge-Stevenson, WA |
| **4/2025** | Understanding When and Why Genetic Testing Is Indicated | Pediatric Symposium Series: A Focus on Pediatric Genetic and Genetic Testing  Manning Family Children’s Hospital New Orleans |

**TEACHING**

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| --- | --- | --- | --- | --- |
| Semester | Lecture | Course | Program | Institution |
| Fall 2019-present | Muscular dystrophy | Intro to Human Genetics (HMGN-7020) | Master Program in Medical Genetics&Genomics | Tulane University School of Medicine |
| Fall 2019-present | Disorder of branched chain amino acid and Tyrosinemia | Medical Biochemistry  (HMGN-7050) |
| Fall 2019-present | Homocystinuria/  MTHFR |
| Fall 2019-present | Urea cycle disorders |
| Fall 2019-present | Fatty acid oxidation disorders |
| Fall 2019-present | Mitochondrial disorders |
| Fall 2019-present | Hypoglycemia/lactic acidosis |
| Fall 2019-present | Galactosemia/Fructosemia |
| Fall 2019-present | Glycogen storage disorders |
| Fall 2019-present | Peroxisomal disorder/ Copper metabolism |
| Fall 2019-present | Neurodegenerative diseases |
| Fall 2019-present | Heme-related disorders |
| Spring 2019-present | Skeletal dysplasia | Intro to Human Genetics (HMGN-7020) |
| Spring 2020-present | Gene editing and CASPR-Cas9 | Advanced Topics in Genomics (HMGN-7950) |
| Spring 2019 | Advanced paternal age and genetic disorders |
| Spring 2020-present | Intro to Medical Genetics | T1 Course: Fundamental of Human Biology Module | Medical Doctoral (MD) program |
| Spring 2020-present | Skeletal dysplasia | T1 Course: Musculoskeletal system I |
| Spring 2020-present | Mitochondrial disorder | T1 Course: Musculoskeletal system II |  |
| Spring 2020-present | Genetics Board Review-General Genetics | NA | Pediatric residents |
| Spring 2020-present | Genetics Board Review- Metabolic disorders | NA |

**MENTORING/SUPERVISION (5)**

1. Anna LaRussa (2020), a Tulane Master Student of Medical Genetics & Genomics Program at Hayward Genetics Center, studies Effect of the Imprinted Genome on Prenatal and Maternal Physiology. She graduated in May 2020*. Current:* Medical student at Tulane University School of Medicine (T2).
2. Genevieve Pierre (2020), a Tulane Master Student of Medical Genetics & Genomics Program at Hayward Genetics Center, studies Genetic Contribution to Dental Agenesis. She graduated in May 2020. *Current*: Dental student at Harward Dental School.
3. R. Collin Burris, Jr (2021), a Tulane Master Student of Medical Genetics & Genomics Program at Hayward Genetics Center, studies A Clinical and Molecular review of Dentonigenesis Imperfecta & Dentin dysplasia. He graduated in July 2021. *Current*: a dental student.
4. Hasti Movaffaghi, (2022), a Tulane Master Student of Medical Genetics & Genomics Program at Hayward Genetics Center, studies Inherited Retinal Disease. *Current:* she graduated in May 2022. *Current:* medical student at Medical college of Wisconsin.
5. Vu Nguyen (2023), a Tulane Master Student of Medical Genetics & Genomics Program at Hayward Genetics Center, studies the effects of known teratogens on fetal genes in vivo. Current: got accepted at Tulane University School of Medicine.

**SERVICE:**

**Board/Committee Positions**

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| National and International Committees: | | |
| Date | Role | Professional Organization |
| 2023-present | CD-SIG Executive Committee | Genetics Course Directors Special Interest Group,  Association of Professors of Human and Medical Genetics (APHMG) |
| Institutional and Local | | |
| 7/2020-present | Member of Phase 1 Curriculum subcommittee | Tulane University School of Medicine, New Orleans, LA, USA |

**Editorial/Peer Review Activities and Positions**

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| Date | Role | Manuscripts |
| 11/2023 | Journal Ad Hoc Reviewer | A Clinical Case of Atypical Hutchinson-Gilford Progeria Syndrome, submitted to Fetal and Pediatric Pathology |
| 12/2023 | Journal Ad Hoc Reviewer | A pair of compound heterozygous *IARS2* variants manifesting West syndrome and electrolyte disorders in a Chinese patient, submitted to Global Medical Genetics Journal |
| 3/2024 | Journal Ad Hoc Reviewer | **Newborn Screening and Inherited Metabolic Disorders, submitted to PBM Genetics** |
| 2/2025 | Journal Ad Hoc Reviewer | Insights from the Newborn Screening Program for Very Long-Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency in Kuwait  Published in International Journal of Neonatal Screening |
| 3/2025 | Journal Ad Hoc Reviewer | Clinical and Genetic Analysis of Cobalamin C deficiency Complicated by Hydrocephalus  Submitted to **European Journal of Pediatrics** |
| 4/2025 | Journal Ad Hoc Reviewer | Analysis of 110 cases of global developmental delay and assessment of intellectual developmental disabilities based on the multidisciplinary clinical diagnosis  Submitted to Molecular Genetics & Genomic Medicine |

**RESEARCH AND SCHOLARSHIP**

**Grants and Independent Funding**

**Current Funding**

**External**

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| --- | --- | --- |
| Date | Role | Project Description |
| 2023-present | Principal Investigator | A Multi-Center, Observational Study to Evaluate the Long-Term Safety of Subcutaneous Injections of Palynziq®(pegvaliase) in Subjects with Phenylketonuria-study 2023-680 TUHSC (IRB)  Sponsor: BioMarin Pharmaceutical Inc  Grant support: $255,610 |
| 2023-present | Principal Investigator | A Multicenter, Observational Study to Evaluate the Real-World Outcomes of Palynziq® (Pegvaliase) in Subjects with Phenylketonuria (OPAL)-study 2023-1035 TUHSC (IRB)  Sponsor: BioMarin Pharmaceutical Inc  Grant support: $269,960 |
| 2023-present | Principal Investigator | A Multi-Center, Prospective, Longitudinal Study Evaluating Immunologic, Inflammatory, and Laboratory Parameters Associated With Long-Term Palynziq (pegvaliase) Treatment in Subjects With Phenylketonuria (PKU) in the United States-study 2023-1036 TUHSC (IRB)  Sponsor: BioMarin Pharmaceutical Inc  Grant support: $122,430 |

**Non-Funded Research**

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| --- | --- | --- |
| Date | Role | Project Description |
| 2022-2023 | Co-Principal Investigator | A Family Review of Phenotypic Spectrum Associated with the interstitial duplication at 4q31.1 and/or 16p13.3- study 2022-1103-TUHSC (IRB) |
| 2022-2023 | Principal Investigator | Biochemical and molecular characteristics among infants with abnormal newborn screen for very-long-chain acyl-CoA dehydrogenase deficiency: A single center experience- study 2022-1373- TUHSC (IRB) |
| 2023-2024 | Principal Investigator | Maximal Dietary Responsiveness After Tetrahydrobiopterin (BH4) in 20 Phenylalanine Hydroxylase Deficiency Patients: What Super-responders Can Expect- study 2023-1148-TUHSC (IRB) |
| 2023-present | Principal Investigator | Expanded Newborn screening in Louisiana 2005-2024: Results and Outcomes-study 2023-1114- TUHSC (IRB) |
| 2022-present | Collaborator (International) | The clinical and neuroradiological spectrum of variants in *MACF1.* |
| 2023-present | Collaborator (National | Newborn screening VLCADD project |
| 2025-present | Principal Investigator | Academic Outcomes Following Expanded Newborn Screening in Louisiana |
| 2025-present | Principal investigator | Clinical Experience with Pegvaliase in Phenylketonuria: A Retrospective Chart Review of Outcomes, Safety, and Dosing Patterns (2025-715) |

**Peer Review Manuscripts**

**Peer Review Manuscripts Published (16)**

1. **Upadia, J**., Liu, J., Bier, C., Chenevert, M., & Li, Y. (2025). Diverse Clinical Presentation of RAC1-Related Intellectual Developmental Disorder. *American journal of medical genetics. Part A*, e63991. Advance online publication. <https://doi.org/10.1002/ajmg.a.63991>
2. Liu J, Li Y, Andersson HC, **Upadia J**\*. Subtelomeric microdeletion in chromosome 20p13 associated with short stature. Clin Case Rep. 2024;12:e8927. doi:10.1002/ccr3.8927
3. Leo SL, **Upadia J**, Valley S, Wall LA. Undetectable C1Q, hyper-IgM, hypogammaglobulinemia: whole exome sequencing reveals pathogenic variant in magnesium transporter 1 (MAGT1). The American Journal of the Medical Sciences. 2024 Feb 1;367:S327-8. SSPR New Orleans, LA.
4. **Upadia J**, Crivelly K, Noh G, Cunningham A, Cerminaro C, Li Y, Mckoin M, Chenevert M, Andersson HC. Maximal dietary responsiveness after tetrahydrobiopterin (BH4) in 19 phenylalanine hydroxylase deficiency patients: What super-responders can expect. Molecular Genetics and Metabolism Reports, Volume 38, January 2024, 101050, ISSN 2214-4269. https://doi.org/10.1016/j.ymgmr.2024.101050
5. **Upadia J**, Noh G, Lefante JJ, Andersson HC. Biochemical and molecular characteristics among infants with abnormal newborn screen for very-long-chain acyl-CoA dehydrogenase deficiency: A single center experience. Molecular Genetics and Metabolism Reports, Volume 37, July 2023,101002, ISSN 2214-4269. https://doi.org/10.1016/j.ymgmr.2023.101002.
6. Liu J, Gajewski K, **Upadia J\***. Chromosome 12p12.2p11.22 deletion in a patient with ventricular fibrillation, mitral valve prolapses, dilation of aorta and intellectual disability: a case report. Clin Case Rep J. 2023;4(3):1–5.
7. Savant A, Lyman B, Bojanowski C, **Upadia J**. Cystic Fibrosis. Updated 2022 Nov 10. In: Adam MP, Everman DB, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2022. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1250/
8. Scala, M., Wortmann, S. B., Kaya, N., Stellingwerff, M. D., Pistorio, A., Glamuzina, E., van Karnebeek, C. D., Skrypnyk, C., Iwanicka-Pronicka, K., Piekutowska-Abramczuk, D., Ciara, E., Tort, F., Sheidley, B., Poduri, A., Jayakar, P., Jayakar, A., **Upadia, J**., Walano, N., Haack, T. B., Prokisch, H., … Houlden, H. (2022). Clinico-radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. *Human mutation*, *43*(3), 403–419. https://doi.org/10.1002/humu.24326
9. **Upadia J**, Li Y, Walano N, Deputy S, Gajewski K, Andersson HC. Genotype-phenotype correlation in IARS2-related diseases: A case report and review of literature. *Clin Case Rep*. 2022;10(2):e05401. Published 2022 Feb 24. doi:10.1002/ccr3.5401
10. **Upadia J**, Walano N, Noh GS, et al. HSD10 disease in a female: A case report and review of literature. *JIMD Reports*. 2021; 1-9. doi:10.1002/jmd2.12250
11. Halaby, C. A., Young, S. P., Austin, S., Stefanescu, E., Bali, D., Clinton, L. K., Smith, B., Pendyal, S., **Upadia, J**., Schooler, G. R., Mavis, A. M., & Kishnani, P. S. (2019). Liver fibrosis during clinical ascertainment of glycogen storage disease type III: a need for improved and systematic monitoring. *Genetics in medicine : official journal of the American College of Medical Genetics*, *21*(12), 2686–2694. https://doi.org/10.1038/s41436-019-0561-7
12. Potnis KC, Flueckinger LB, Ha CI, **Upadia J**, Frush DP, Kishnani PS. Bone manifestations in neuronopathic Gaucher disease while receiving high-dose enzyme replacement therapy. *Mol Genet Metab*. 2019;126(2):157-161. doi:10.1016/j.ymgme.2018.11.004
13. **Upadia J, Gonzales PR, Atkinson TP, Schroeder HW, Robin NH, Rudy NL, Mikhail FM. A previously unrecognized 22q13.2 microdeletion syndrome that encompasses TCF20 and TNFRSF13C. Am J Med Genet. 2018;1-7.** DOI: 10.1002/ajmg.a.40492
14. **Upadia J**, Gonzales PR, Robin HN. Novel de novo pathogenic variant in the NR2F2 gene in a boy with congenital heart defect and dysmorphic features. Am J Med Genet Part A. 2018;00:1–4. https://doi.org/10.1002/ajmg.a. 282
15. **Upadia J**, Gomes A, Weiser P, Descartes M. A familial case of multicentric carpotarsal osteolysis syndrome and treatment outcome. J Pedistr Genet 2018;00:1-6
16. **Upadia J**, Philips III J, Robin NH. Lose EJ, Mikhail FM. A case report of chromosome 17q22-qter trisomy with distinct clinical presentation and review of the literature. Clinical case reports. 2017 Oct. doi: 10.1002/ccr3.1298
17. **Upadia J, Oakes J, Hamm A, Hurst AC, Robin NH. Foramen magnum compression in Coffin-Lowry syndrome: A case report. Am J Med Genet A. 2007 Apr; 173(4):1087-1089.**

**Manuscripts currently under peer-review (1)**

1. Dekker J, Schot R, Aldinger K,…..**Upadia J**….The clinical and neuroradiological spectrum of variant in MACF1. *Submitted to Brain Journal.*
2. **Jariya Upadia**, Kea Crivelly, Grace Noh, Jennifer Smith, Hans C. Andersson. Thiamina-responsive maple syrup urine disease missed by newborn screen: A case report. Submitted to MGM Reports Journal.
3. Grace Noh and **Jariya Upadia**. Case Report: Lysine Improvement in Siblings with Glutaric Acidemia Type 1 Following Reduced Medical Food Intake: Implications for Amino Acid Absorption and Reabsorption. MGM Reports Journal.

**ABSTRACT (14)**

(\*Poster, \*\*Platform presentation)

1. Jordan Brignac, Greta Geiger, Kennedi Jones, **Jariya Upadia\***. Novel Co-occurrence of Maternal Uniparental Disomy and a Pathogenic Variant in the SHOX Gene in a patient with Langer Mesomelic Dysplasia: A Case Report. Pediatrics Research Day, May 2025, Manning Family Children’s Hospital, New Orleans, LA.
2. David Van, Molly Sonenklar, David Swetland, **Jariya Upadia**, Andrew Abreo. Variable Clinical Phenotype of Takenouchi-Kosaki Syndrome. 2025 AAAAI / WAO Joint Congress on 3/2/2025, San Diego, CA
3. **Upadia J**, Crivelly K, Noh G, Cunningham A, Cerminaro C, Li Y, Mckoin M, Chenevert M, Andersson HC. Maximal dietary responsiveness after tetrahydrobiopterin (BH4) in 19 phenylalanine hydroxylase deficiency patients: What super-responders can expect. Pediatrics Research Day, May 2024, Children’s Hospital New Orleans, LA.
4. \*Noh GS and **Upadia J**. Impact of Arginine-Fortified Formula on Lysine Levels in GA1 Siblings: A Case Report. 2024 GMDI Conference, April 17-20, 2024 in Charlotte, NC
5. \*Leo SL, **Upadia J**, Valley S, Wall LA. Undetectable C1Q, hyper-IgM, hypogammaglobulinemia: whole exome sequencing reveals pathogenic variant in magnesium transporter 1 (MAGT1). The American Journal of the Medical Sciences. 2024 Feb 1;367:S327-8. SSPR New Orleans, LA.
6. \*Kathryn Cyrus, Anne Tufton, **Jariya Upadia**, Nicole Vegh, Mary Johnson. A perinatal case of extreme neuroanatomical abnormalities and end stage renal disease in chromosome 17q12 deletion syndrome. Southern Regional Meeting, New Orleans, Louisiana on February 22-24, 2024
7. \*\***Upadia J**, Noh G, Lefante JJ, Andersson HC. Biochemical and molecular characteristics among infants with abnormal newborn screen for very-long-chain acyl-CoA dehydrogenase deficiency: A single center experience. At SERGG meeting 2023 at Charleston, SC
8. \*Crivelly KS, Noh GS, Cunningham AC, Cerminaro C, **Upadia J**, Andersson HC. PALYNZIQ, IS IT WORTH THE WEIGHT?: Weight gain observed in Palynziq-Treated PKU Patients. SERGG Annual Meeting 2022
9. \*Yuwen Li, **Jariya Upadia**, Madeline Chenevert, Meredith McKoin, Jiao Liu, Tianjian Chen, Hans Andersson. A family Review of Phenotypic Spectrum Associated with the interstitial duplications at 4q31.1 and/or 16p13.3. SERGG Annual Meeting 2022
10. \***Jariya Upadia**, Hans Andersson. Carnitine deficiency: A case report of an adult female presenting with hyperammonemic encephalopathy. SERGG Annual Meeting 2022
11. \*Jiao Liu, **Jariya Upadia**. Chromosome 12p12.2p11.22 deletion in a patient with ventricular fibrillation, mitral valve prolapse, dilatation of aorta and intellectual disability: a case report. SERGG Annual Meeting 2022
12. \***Jariya Upadia**, Nicolette Walano, Kea Crivelly, Hans Andersson. An adult male with combined malonic and methylmalonic aciduria: a case report. SERGG Annual Meeting 2019
13. \***Jariya Upadia**, Nicolette Walano. Radiographic findings of periarticular calcification in adult with hypophosphatasia: a case report. SERGG Annual Meeting 2019
14. \***Upadia J**, Pendyal S, Bailey L, Koeberl DD, Kishnani P. Clinical course, and outcome in adults with propionic acidemia: case series. SIMD annual meeting 2018. Abstract number 119.
15. \***Upadia J**, Austin S, Fluekinger LB, Ravindra KV, Zhang X, Brady CW, Pendyal S, Kishnani P. A first report of cholangiocarcinoma in GSD I. SIMD annual meeting 2018. Abstract number 118.
16. \***Upadia J**, Mikhail F., Nelson K., Robin N. A case of familial William syndrome with atypical deletion. ACMG annual meeting 2017, abstract number 191. https://acmg.expoplanner.com/index.cfm?do=expomap.sess&event\_id=8&session\_id=5259
17. \***Upadia J**., Rutledge SL, Wille K., Bean L., Robin NH, Sharer J.. Fatal late-onset presentation of ornithine transcarbamylase deficiency in a 41-year-old male bodybuilder. ACMG annual meeting 2017, abstract number 63. https://acmg.expoplanner.com/index.cfm?do=expomap.sess&event\_id=8&session\_id=5259