JARIYA UPADIA, MD

Curriculum Vitae

PERSONAL INFORMATION

Email: jupadia@tulane.edu

Language: English, Thai

EDUCATION AND TRAINING

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

POST-GRADUATE OR OTHER TRAINING

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

Date	Board	Specialty	Certification #	Renewal and Maintenance
2019	American Board of Medical Genetics and Genomics (ABMGG)	Medical Biochemical Genetics	2019235	Current: Cycle 2021-2023
2017	American Board of Medical Genetics and Genomics (ABMGG)	Clinical Genetics and Genomics	2017212	Current: Cycle 2021-2023 Previous: Cycle 2018-2020
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

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

LECTURES

Semester	Lecture	Course	Program	Institution
Fall 2019-present	Muscular dystrophy	Intro to Human Genetics (HMGN-7020)		
Fall 2019-present	Disorder of branched chain amino acid and Tyrosinemia			
Fall 2019-present	Homocystinuria/ MTHFR			
Fall 2019-present	Urea cycle disorders			

Fall 2019-present	Fatty acid oxidation disorders	Medical Biochemistry (HMGN-7050)	Master Program in Medical Genetics&Genomics	Tulane University School of Medicine
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		

GRAND ROUNDS AND INVITED TALKS (1)

1. *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, February 2, 2024

MENTORING/SUPERVISION (4)

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, 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, 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 assistant and she is applying for MD programs.
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.

RESEARCH PROJECT (7)

Date	Project Description
2023-present	**Principal Investigator (PI):** 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 (PI):** Expanded Newborn screening in Louisiana 2006-2023: Results and Outcomes-study 2023-1114- TUHSC (IRB)

2023-present	**Principal Investigator (PI):** 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)
2023-present	**Principal Investigator (PI):** A Multicentre, Observational Study to Evaluate the Real-World Outcomes of Palynziq® (Pegvaliase) in Subjects with Phenylketonuria (OPAL)-study 2023-1035
2023-present	**Principal Investigator (PI):** 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)
2022-present	**Collaborator (International):** The clinical and neuroradiological spectrum of variants in *MACF1.*
2022-2023	**Principal Investigator (PI):** 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)
2022-2023	**Co-PI:** A Family Review of Phenotypic Spectrum Associated with the interstitial duplication at 4q31.1 and/or 16p13.3- study 2022-1103-TUHSC (IRB)
2023-present	**Collaborator (National):** newborn screening VLCADD project

EXPERT GROUP/COMMITTEE POSITIONS

Institutional and local

Date	Title	Institution
7/2020-present	Member of Phase 1 Curriculum subcommittee	Tulane University School of Medicine, New Orleans, LA, USA

PEER REVIEW ACTIVITIES AND POSITIONS

Date	Role	Manuscripts
11/2023	Reviewer	A Clinical Case of Atypical Hutchinson-Gilford Progeria Syndrome, submitted to Fetal and Pediatric Pathology
12/2023	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	Reviewer	Newborn Screening and Inherited Metabolic Disorders, submitted to PBM Genetics

Recent Publications from Reviewed Manuscripts

PEER REVIEW MANUSCRIPTS

Review Articles (3)

1. Savant A, Lyman B, Bojanowski C, **Upadia J**. Cystic Fibrosis. 2001 Mar 26 [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/
2. **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
3. **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

Original Articles (17)

1. 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.

2. 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

3**. 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

4**. 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

5. **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

6. 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

7. 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

8**. 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

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. 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

11. Savant A, Lyman B, Bojanowski C, **Upadia J**. Cystic Fibrosis. 2001 Mar 26 [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/

12. 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.

13. **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.

14. **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

15. 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.

16. Liu J, Li Y, Andersson HC, **Upadia J**. Subtelomeric microdeletion in chromosome 20p13 associated with short stature - a case report. *Submitted to Clinical Case Reports Journal.*

17. Dekker J, Schot R, Aldinger K,…..**Upadia J**….The clinical and neuroradiological spectrum of variant in MACF1. *Submitted to Brain Journal.*

ABSTRACT (14)

(*Poster, **Platform presentation)

1**. *Upadia, J.,** Rutledge, S.L., Wille, k., Bean, L., Robin, N.H., 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

2**. *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

3. ***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.

4**. *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.

5**. *Jariya Upadia**, Nicolette Walano. Radiographic findings of periarticular calcification in adult with hypophosphatasia: a case report. SERGG Annual Meeting 2019

6. ***Jariya Upadia**, Nicolette Walano, Kea Crivelly, Hans Andersson. An adult male with combined malonic and methylmalonic aciduria: a case report. SERGG Annual Meeting 2019

7. *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

8. ***Jariya Upadia**, Hans Andersson. Carnitine deficiency: A case report of an adult female presenting with hyperammonemic encephalopathy. 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. *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

11. ****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

12. *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

13. *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.

14. *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