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Division of Medical Genetics
Department of Medicine
Faculty of Medicine Siriraj Hospital
Mahidol University

Certification & Education

2013	Abridged Business Certificate Thammasat Business School Thammasat University
2007	Diplomate, American Board of Medical Genetics (Clinical Molecular Genetics) Diplomate, Thai Board of Internal Medicine
2005	Diplomate, American Board of Medical Genetics (Clinical Genetics)
2003 – 2006	Fellowship in Clinical Genetics and Clinical Molecular Genetics National Human Genome Research Institute National Institutes of Health
2003	Diplomate, American Board of Internal Medicine
2000 – 2003	Internal Medicine Residency University of Illinois at Chicago College of Medicine
1989 – 1995	Doctor of Medicine (First Class Honor) Faculty of Medicine Siriraj Hospital Mahidol University, Thailand

Professional Experience

2017 – Present	Professor Division of Medical Genetics, Department of Medicine Siriraj Genomics, Faculty of Medicine Siriraj Hospital, Mahidol University Leader, Siriraj Center of Research Excellence (SiCORE) in Precision Medicine Leader of Health Cluster, Research University Network Thailand
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Chair of Research and Development Working Group,
Genomics Thailand Initiative

2012 – 2017	Associate Professor
2009 – 2012	Assistant Professor
2006 – 2009	Clinical Instructor

Publications

1. Vichara-anont I, Lumkul L, Taratikhundej S, **Pithukpakorn M**, Roothumnong E, Wongsas C, Krikeerati T, Jameekornrak Taweechue A, Theankeaw O, Limjunyawong N, Sookrung N, Thongngarm T, Sompornrattanaphan M. HLA variants and their association with IgE-Mediated banana allergy: A cross-sectional study Heliyon. 2024 Jun 11;10(12):e32787
2. Suwatthanarak T, Acharayothin O, Thanormjit K, Chaiboonchoe A, Suwatthanarak T, Niyomchan A, **Pithukpakorn M**, Tanjak P, Chinswangwatanakul V. Assessment of long-term stored specimens in the Siriraj Hospital colorectal cancer biobank for RNA sequencing and profiling. J Lab Med. 2024 Jun. doi: 10.1515/labmed-2023-0137
3. Suwatthanarak T, Tanjak P, Chaiboonchoe A, Acharayothin O, Thanormjit K, Chanthercrob J, Suwatthanarak T, Niyomchan A, Tanaka M, Okochi M, Pongpaibul A, Chalermwai WV, Trakarnsanga A, Methasate A, **Pithukpakorn M**, Chinswangwatanakul V. Overexpression of TSPAN8 in consensus molecular subtype 3 colorectal cancer. Exp Mol Pathol. 2024 Jun 10;137:104911. doi: 10.1016/j.yexmp.2024.104911.
4. Thongrattana W, Arigul T, Suktitipat B, **Pithukpakorn M**, Sathornsumetee S, Wongsurawat T, Jenjaroenpun P, GLIMMERS: glioma molecular markers exploration using long read sequencing. Bioinform Adv. 2024; vbae058 doi: 10.1093/bioadv/vbae058
5. Kansuttiviwat C, Lertwilaiwittaya P, Roothumnong E, Nakthong P, Dungort P, Meesamarnpong C, Tansa-Nga W, Pongsuktavorn K, Wiboonthanasarn S, Tititumjariya W, Phuphuripan N, Lertbussarakam C, Wattanarangsan J, Sritun J, Punuch K, Kammarabutr J, Mutirangura P, Thongnoppakhun W, Limwongse C, **Pithukpakorn M**. Germline mutations of 4567 patients with hereditary breast-ovarian cancer spectrum in Thailand. NPJ Genom Med. 2024 Feb 14;9(1):9. doi: 10.1038/s41525-024-00400-4
6. Lertwilaiwittaya P, Tantai N, Maneeon S, Kongbunrak S, Nonpanya N, Hurst ACE, Srinonprasert V and **Pithukpakorn M**. A cost-utility analysis of BRCA1 and BRCA2 testing in high-risk breast cancer patients and family members in Thailand: a cost-effective policy in resource-limited settings. Front Public Health. 2023 Dec 11:1257668. doi: 10.3389/fpubh.2023.1257668

7. Suwatthanarak T, Tanjak P, Suwatthanarak T, Acharayothin O, Thanormjit K, Chaiboonchoe A, Tawantanakorn T, Phalanusitthepha C, Trakarnsanga A, Methasate A, **Pithukpakorn M**, Okamoto R, Chinswangwatanakul V. Exploring extracellular matrix and prostaglandin pathway alterations across varying resection margin distances of right-sided colonic adenocarcinoma. *BMC Cancer* 2023 Dec 23;1202. doi:10.1186/s12885-023-11595-7
8. Angkasekiwinai N, Suputtamongkol Y, Tantibhedhyangkul W, Onlamoon N, Phoompoung P, **Pithukpakorn M**, Karuphong E, Pusuwan P, Angkasekiwinai P. Efficacy of bortezomib for treating anti-interferon-gamma autoantibody-associated adult-onset immunodeficiency (AOID) syndrome. *Clin Infect Dis*. 2023 Nov 8:ciad676. doi: 10.1093/cid/ciad676
9. Anukul N, Jenjaroenpun P, Sirikul C, Wankaew N, Nimsamer P, Roothumnong E, **Pithukpakorn M**, Leetrakool N, Wongsurawat T. Ultrarapid and high-resolution HLA class I typing using transposase-based nanopore sequencing applied in pharmacogenetic testing. *Front Genet* 2023 June; 14. doi:10.3389/fgene.2023.1213457
10. Jinda W, Mounthard H, Limwongse C, **Pithukpakorn M**, Saelee P, Pokkasup N, Khunpukdee S, Sukthaworn S, Jumpasri J. Identification of Genomic Alterations in Thai Patients With Colorectal Cancer Using Next-Generation Sequencing-Based Multigene Cancer Panel. *Cureus*, 2023 May 16;15(5), e39067. doi: 10.7759/cureus.39067
11. Hantaweeapat C, Suktitipat B, **Pithukpakorn M**, Chinthammitr Y, Limwongse C, Tansiri N, Sawatnatee S, Takpradit C, Rotchanapanya W, Pongudom S, Charoenprasert K, Paiboonsukwong K, Thamprasert W, Nolwachai N, Rattanasawat W, Sae-Aeng B, Khorwanichakij N, Saetow P, Saengboon S, Kamjornpreecha K, Pholmoo W, Dujjawan B, Siritanaratkul N. Whole exome sequencing and rare variant association study to identify genetic modifiers, KLF1 mutations, and a novel double mutation in Thai patients with hemoglobin E/beta-thalassemia. *Hematology*. 2023 Dec;28(1):2187155. doi: 10.1080/16078454.2023.2187155.
12. Chiraphappaiboon W, Thongnoppakhun W, Limjindaporn T, Sawasdichai S, Roothumnong E, Prangphan K, Pamornpol B, Limwongse C, **Pithukpakorn M**. STK11 Causative Variants and Copy Number Variations Identified in Thai Patients With Peutz-Jeghers Syndrome. *Cureus*. 2023 Feb 1;15(2):e34495. doi: 10.7759/cureus.34495.
13. Tanjak P, Chaiboonchoe A, Suwatthanarak T, Acharayothin O, Thanormjit K, Chanthercrob J, Suwatthanarak T, Wannasuphaphol B, Chumchuen K, Suktitipat B, Sampattavanich S, Korphaisarn K, Pongpaibul A, Pongvarin N, Grove H, Riansuwan W, Trakarnsanga A, Methasate A, **Pithukpakorn M**, Chinswangwatanakul V. The KRAS-Mutant Consensus Molecular Subtype 3 Reveals an Immunosuppressive Tumor Microenvironment in Colorectal Cancer. *Cancers* 2023, 15, 1098. doi: 10.3390/cancers15041098

14. Jaru-Ampornpan P, Tansirisithikul C, Prukajorn M, Sampattavanich S, **Pithukpakorn M**. Germline ATM mutation and somatic PIK3CA and BCOR mutations found in an infant with aggressive orbital embryonal rhabdomyosarcoma. *Am J Ophthalmol Case Rep*. 2021 Aug 4;23:101189. doi: 10.1016/j.ajoc.2021.101189.
15. Lertwilaiwittaya P, Suktitipat B, Khongthon P, Pongsapich W, Limwongse C, **Pithukpakorn M**. Identification of novel mutation in RANKL by whole-exome sequencing in a Thai family with osteopetrosis; a case report and review of RANKL osteopetrosis. *Mol Genet Genomic Med*. 2021 May 30:e1727. doi: 10.1002/mgg3.1727.
16. Kittikoon S, **Pithukpakorn M**, Pramyothin P. Physician awareness, preparedness, and opinions toward consumer-initiated genetic testing in Thailand: Views from a changing landscape. *J Genet Couns*. 2021. Apr 30. doi: 10.1002/jgc4.1420.
17. Lertwilaiwittaya P, Roothumnong E, Nakthong P, Dungort P, Meesamarnpong C, Tansa-Nga W, Pongsuktavorn K, Wiboonthanasarn S, Tititumjariya W, Thongnoppakhun W, Chanprasert S, Limwongse C, **Pithukpakorn M**. Thai patients who fulfilled NCCN criteria for breast/ovarian cancer genetic assessment demonstrated high prevalence of germline mutations in cancer susceptibility genes: implication to Asian population testing. *Breast Cancer Res Treat*. 2021 Jul;188(1):237-248. doi: 10.1007/s10549-021-06152-4. Epub 2021 Mar 1.
18. Chiowchanwisawakit P, **Pithukpakorn M**, Luangtrakool K, Permpikul P. HLA-B*27:04 associated with enthesitis and younger age of onset, and HLA-B allele profile in patients with ankylosing spondylitis in Thailand: A cross-sectional study. *Int J Rheum Dis*. 2021. Jan 15. doi: 10.1111/1756-185X.14055.
19. Rujirachun P, Junyavoraluk A, **Pithukpakorn M**, Suktitipat B, Winijkul A. Successful treatment of arrhythmia with β -blocker and flecainide combination in pregnant patients with Andersen-Tawil syndrome: A case report and literature review. *Ann Noninvasive Electrocardiol*. 2020 Sep 21;e12798. doi: 10.1111/anec.12798
20. Korphaisarn K, Pongpaibul A, Roothumnong E, Pongsuktavorn K, Thamlikitkul L, Anekpuritanang T, Pongvarin N, Thongnoppakhun W, **Pithukpakorn M**. High Frequency of KRAS Codon 146 and FBXW7 Mutations in Thai Patients with Stage II-III Colon Cancer. *Asian Pacific journal of cancer prevention: APJCP*. 2019;20(8):2319-26. doi: 10.31557/APJCP.2019.20.8.2319
21. Angkasekwinai N, Suputtamongkol Y, Phoompoung P, **Pithukpakorn M**, Wongswat E, Umrod P, Tongyai S, Foongladda S. Clinical outcome and laboratory markers for predicting disease activity in patients with disseminated opportunistic infections associated with anti-interferon-gamma

autoantibodies. PLOS One. 2019;14(4):e0215581. doi: 10.1371/journal.pone.0215581

22. Shotelersuk V, Tongshima S, **Pithukpakorn M**, Eu-Ahsunthornwattana J, Mahasirimongkol S. Precision medicine in Thailand. Am J Med Genet, C Semin Med Genet. 2019. Mar 19. doi: 10.1002/ajmg.c.31694.
23. Niyomnaitham S, Parinyanitikul N, Roothumnong E, Jinda W, Samarnthai N, Atikankul T, Suktitipat B, Thongnoppakhun W, Limwongse C, **Pithukpakorn M**. Tumor mutational profile of triple negative breast cancer patients in Thailand revealed distinctive genetic alteration in chromatin remodeling gene. Peer J. 2019 Feb 7:e6501 doi: 10.7717/peerj.6501
24. Ritthaphai A, Wattanapanitch M, **Pithukpakorn M**, Heepchantree W, Soi-Ampornkul R, Mahaisavariya P, Triwongwaranat D, Pattanapanyasat K, Vatanashevanopakorn C. Derivation of an induced pluripotent stem cell line (MUSIi004-A) from dermal fibroblasts of a 48-year-old spinocerebellar ataxia type 3 patient. Stem Cell Res. 2018 May 21;30:113-116. doi: 10.1016/j.scr.2018.05.012
25. Suktitipat B, Sathirareuangchai S, Roothumnong E, Thongnoppakhun W, Wangkiratikant P, Vorasan N, Krittayaphong R, **Pithukpakorn M**, Boonyapisit W. Molecular investigation by whole exome sequencing revealed a high proportion of pathogenic variants among Thai victims of sudden unexpected death syndrome. PLOS One 2017 Jul 13;12(7):e0180056. doi: 10.1371/journal.pone.0180056.
26. Phoompoung P, Ankasekwinai N, **Pithukpakorn M**, Foongladda S, Umrod P, Suktitipat B, Mahasirimongkol S, Kiartiburanakul S, Suputtamongkol Y. Factors associated with acquired Anti IFN-Gamma autoantibody in patients with nontuberculous mycobacterial infection. PLOS One 2017 Apr 24;12(4):e0176342. doi: 10.1371/journal.pone.0176342.
27. Panichareon B, Seedapan T, Thongnoppakhun W, Limwongse C, **Pithukpakorn M**, Limjindaporn T. Novel Mutation of the TINF2 Gene in a Patient with Dyskeratosis Congenita Case Rep Dermatol 2015 Aug (7):212-219
28. **Pithukpakorn M**, Toro JR. Hereditary Leiomyomatosis and Renal Cell Cancer. 2006 Jul 31 [Updated 2015 Aug 6]. In: Pagon RA, Adam MP, Ardinger HH, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2015. Available from: <http://www.ncbi.nlm.nih.gov/books/NBK1252/>
29. **Pithukpakorn M**, Roothumnong E, Angkasekwinai N, Suktitipat B, Assawamakin A, Luangwedchakarn V, Umrod P, Thongnoppakhun W, Foongladda S, Suputtamongkol Y. HLA-DRB1 and HLA-DQB1 Are Associated with Adult-Onset Immunodeficiency with Acquired Anti-Interferon-Gamma Autoantibodies. PLOS One 2015 May 10(5): e0128481. doi:10.1371/journal.pone.0128481

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31. Tanboon J, Rongsa K, **Pithukpakorn M**, Boonyapisit K, Limwongse C, Sangruchi T. A Novel Mutation of the GNE Gene in Distal Myopathy with Rimmed Vacuoles: A Case with Inflammation. *Case Rep Neurol* 2014 Mar (6):55-59
32. Assawamakin A, Sriratanaviriyakul N, Lalerd Y, Thongnoppakhun W, Praditsap O, Tongsima S, **Pithukpakorn M**. Meta-analysis of the plasminogen activator inhibitor-1 (PAI-1) gene with insertion/deletion 4G/5G polymorphism and its susceptibility to ischemic stroke in Thai population. *Asian Biomed* 2012 Apr 6(2):203-217.
33. Panichareon B, Taweechue K, Thongnoppakhun W, Aksornworanart M, **Pithukpakorn M**, Yenchitsomanus PT, Limwongse C, Limjindaporn T. Six novel ATP7B mutations in Thai patients with Wilson disease. *Eur J Med Genet* 2011 Mar-Apr;54(2):103-7.
34. **Pithukpakorn M**. Unusual presentation as an adult onset painful myopathy in a Thai patient with Becker muscular dystrophy. *J Med Assoc Thai* 2011 Feb (94) Suppl 1:S250-2
35. Chierakul N, Pongprasobchai S, Boonyapisit K, Chinthammitr Y, **Pithukpakorn M**, Maneesai A, Srivijitkamol A, Koomanachai P, Koolvisoot A, Tanwadee T, Shayakul C, Kachintorn U. Can we predict final outcome of internal medicine residents with in-training evaluation? *J Med Assoc Thai* 2011 Feb (94) Suppl 1:S246-9
36. Tanwadee T, **Pithukpakorn M**, Vipatakul N, Charatcharoenwittaya P, Chainuvati S, Nimanong S, Prachayakul V, Pongprasobchai S, Manatsathit S, Leelakusolvong S, Pausawasdi N, Kachintorn U, Limwongse C, Udompunturak S. Genetic polymorphism of low-density lipoprotein receptor did not affect treatment outcome of chronic hepatitis C genotype 3. *J Med Assoc Thai* 2011 Feb (94) Suppl 1:S147-53
37. Pramyothin P, **Pithukpakorn M**, Arakaki RF. A 47, XXY patient and Xq21.31 duplication with features of Prader-Willi syndrome: results of array-based comparative genomic hybridization. *Endocrine* 2010 Jun;37(3):379-82.
38. **Pithukpakorn M**, Chaowalit N. Clinical presentation and echocardiographic findings of Thai patients with Marfan syndrome. *J Med Assoc Thai* 2010 Jan (93) Suppl 1:S43-7.
39. **Pithukpakorn M**, Aksentijevich I, Toro JR Autoinflammatory disease: clinical and dermatologic features, genetics, pathogenesis and therapy. *Adv Dermatol* 2006; 22:67-90
40. **Pithukpakorn M**, Wei MH, Toure O, Steinbach PJ, Glenn GM, Zbar B, Linehan WM, Toro JR Fumarate hydratase enzyme activity in lymphoblastoid

cells and fibroblasts of individuals in families with hereditary leiomyomatosis and renal cell cancer. *J Med Genet* 2006 Sep 43(9):755-762

41. Wei MH, Toure O, Glenn GM, **Pithukpakorn M**, Neckers L, Stolle C, Choyke P, Grubb R, Middleton L, Turner ML, Walther M, Merino M, Zbar B, Linehan WM, Toro JR. Novel mutations in FH and expansion of the spectrum of phenotypes expressed in families with hereditary leiomyomatosis and renal cell cancer. *J Med Genet* 2006 Jan 43(1):18-27
42. **Pithukpakorn M**. Disorders of pyruvate metabolism and the tricarboxylic acid cycle. *Mol Genet Metab* 2005 Aug; 85(4):243-6
43. **Pithukpakorn M**, Toro JR. Birt-Hogg-Dubé syndrome. 2005 In: *GeneReviews at GeneTests Medical Genetics Information Resource* (database online). Copyright, University of Washington, Seattle. 1997-2006.
44. **Pithukpakorn M**, Glenn GM. von Hippel Lindau syndrome. *Comm Oncol* 2004 Nov/Dec; 1(4):232-243

Professional Membership

2002 – Present	Member, The American College of Physicians
1999 – Present	Member, The Royal College of Physicians of Thailand Member, The Genetics Society of Thailand
2019 – Present	Member, The Medical Genetics and Genomics Association Member, Thai Society of Human Genetics