Stanford

Chung Lee

Clinical Associate Professor, Pediatrics - Medical Genetics

CLINICAL OFFICE (PRIMARY)

- **Medical Genetics**

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Bio

CLINICAL FOCUS

- Biochemical Genetics
- Clinical Genetics and Genomics

ACADEMIC APPOINTMENTS

- Clinical Associate Professor, Pediatrics - Medical Genetics

ADMINISTRATIVE APPOINTMENTS

- Program Director, Medical Biochemical Genetics Fellowship Program, (2020- present)
- Associate Program Director, Medical Biochemical Genetics Fellowship Program, (2017-2020)

HONORS AND AWARDS

- Phi Beta Kappa, Northwestern University (2001)
- Summa cum laude, Northwestern University (2002)
- Dr. Koch Memorial Scholarship Recipient, National PKU Alliance (2012)
- Faculty Teaching Award, Stanford University (2025)

PROFESSIONAL EDUCATION

- Board Certification: Clinical Genetics and Genomics, American Board of Medical Genetics and Genomics (2024)
- Residency: Kaiser Permanente Northern California GME Programs (2011) CA
- Internship: Kaiser Permanente Northern California GME Programs (2009) CA
- Board Certification: Clinical Biochemical Genetics, American Board of Medical Genetics and Genomics (2015)
- Fellowship: University of California, San Francisco (UCSF) (2014) CA
- Residency: University of California, San Francisco (UCSF) (2013) CA
- Board Certification: Pediatrics, American Board of Pediatrics (2011)

- Medical Education: University of Illinois at Chicago (2008) IL

Teaching

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Medical Biochemical Genetics (Fellowship Program)
- Medical Genetics (Fellowship Program)

Publications

PUBLICATIONS

- **Refractory lactic acidosis in methylmalonic acidemia unmasking thiamine deficiency: A case report and call for routine thiamine assessment**
  Chilakamarri, L., Enns, G., Wright, J., Lee, C.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2026

- **A multi-center retrospective chart analysis identifying clinical, biochemical, and radiographic signatures of short-chain Enoyl CoA hydratase (ECHS1) deficiency**
  Kong, C., Alves, C., Anselm, I., Berry, G. T., Cabello, J., Cornejo, V., Fernandez, A., Enns, G., Gomez, M., Haldeman-Englert, C., Hollander, S., Innes, M., Jannotta, et al
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2026

- **Characterizing the frequency of clinical events and assessing biomarkers in propionic acidemia: a natural history study.** *Orphanet journal of rare diseases*
  Schwahn, B. C., Berry, G. T., Vernon, H. J., Li, H., Merritt Ii, J. L., Schiff, M., Chabrol, B., De Las Heras, J., Vockley, J., Lee, C., Koeberl, D. D., Burton, B. K., Grunewald, et al
  2026

- **De novo heterozygous variants of the RSF1 gene are responsible for a syndromic neurodevelopmental disorder.** *European journal of human genetics : EJHG*
  Jost, C., Busa, T., Wegner, D., Shinawi, M., Schaefer, E., Piton, A., Schluth-Bolard, C., Charles, P., Keren, B., Mayerhanser, K., Brunet, T., Schatz, U., Neil, et al
  2026

- **Carbonic anhydrase VA deficiency due to a novel CA5A variant.** *Molecular genetics and metabolism reports*
  Keehan, L., Null, E., Chilakamarri, L., Carter, C., Lee, C., Enns, G. M., Matalon, D. R.
  2025; 45: 101259

- **ReclassifyingIDUAc.250G>A (p.Gly84Ser): Evidence for a Possible Pseudodeficiency Allele.** *International journal of neonatal screening*
  Connolly, C., Fisher, R., Yang, C., Schelley, S., Mendelsohn, B. A., Lee, C., Ahmad, A.
  2025; 11 (4)

- **A zebrafish model of crim1 loss of function has small and misshapen lenses with dysregulated clic4 and fgf1b expression.** *Frontiers in cell and developmental biology*
  Le, T., Htun, S., Pandey, M. K., Sun, Y., Magnusen, A. F., Ullah, E., Lauzon, J., Beres, S., Lee, C., Guan, B., Hufnagel, R. B., Brooks, B. P., Baranzini, et al
  2025; 13: 1522094

- **Mitochondrial HMG-CoA Synthase Deficiency: A Cyclic Vomiting Mimic Without Reliable Biochemical Markers.** *Journal of investigative medicine high impact case reports*
  Niehaus, A. D., Cooper, H., Lee, C. U.
  2024; 12: 23247096241267154

- **Clinicopathologic Features of IDEDNIK (MEDNIK) Syndrome in a Term Infant: Histopathologic Features of the Gastrointestinal Tract and Report of a Novel AP1S1 Variant.** *Pediatric and developmental pathology : the official journal of the Society for Pediatric Pathology and the Paediatric Pathology Society*
  Lu, J. G., Namjoshi, S. S., Niehaus, A. D., Tahata, S., Lee, C. U., Wang, L., McDonnell, E., Seely, M., Martin, M. G., Hazard, F. K.
  2023: 10935266231177402

- **MT-ATP6 mitochondrial disease identified by newborn screening reveals a distinct biochemical phenotype.** *American journal of medical genetics. Part A*
  Tise, C. G., Verscaj, C. P., Mendelsohn, B. A., Woods, J., Lee, C. U., Enns, G. M., Stander, Z., Hall, P. L., Cowan, T. M., Cusmano-Ozog, K. P.
  2023
- **Neonatal lupus is a novel cause of positive newborn screening for X-linked adrenoleukodystrophy.** *American journal of medical genetics. Part A*
  Niehaus, A. D., Mendelsohn, B. A., Zimmerman, B., Lee, C. U., Manning, M. A., Cusmano-Ozog, K. P., Tise, C. G.
  2023
- **MaP NATURAL HISTORY STUDY: CLINICAL AND BIOMARKER FINDINGS IN METHYLMALONIC ACIDEMIA DUE TO MUT DEFICIENCY**
  Schiff, M., Schwahn, B. C., Chabrol, B., Merritt, J., Vockley, J., Vernon, H., Berry, G. T., Santra, S., Lee, C., Koeberl, D., Li, H., Burton, B., De las Heras, et al
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2023: 84
- **MaP NATURAL HISTORY STUDY: CLINICAL AND BIOMARKER FINDINGS IN PROPIONIC ACIDEMIA**
  Schwahn, B. C., Berry, G. T., Santra, S., Vernon, H., Li, H., Merritt, L., Schiff, M., Chabrol, B., De las Heras, J., Vockley, J., Lee, C., Koeberl, D., Burton, et al
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2023: 85
- **MITOCHONDRIAL-ATP6-ASSOCIATED DISEASE PRESENTS WITH DISTINCT PATTERN ON NEWBORN SCREENING: SHOULD IT BE INCLUDED AS A SECONDARY CONDITION?**
  Tise, C., Mendelsohn, B., Lee, C., Woods, J., Hall, P., Tang, H., Rinaldo, P., Cowan, T., Cusmano-Ozog, K.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2022: 247-248
- **ANALYSIS OF URINE HEPARAN SULFATE AND ITS NON-REDUCING ENDS FOR THE FOLLOW-UP OF ABNORMAL NEWBORN SCREENING FOR MPS1**
  Kaczmarczyk, A., Lasio, L., Viskochil, D., Longo, N., Lund, T., Orchard, P. J., Yang, A. C., Chang, I., Lee, C., Pedro, H., Aliu, E., Siemon, A., Mori, et al
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2022: 281-282
- **Carnitine-Acylcarnitine Translocase Deficiency**
  Morales, J. A., Lee, C. U., Enns, G. M., et al
  GeneReviews.
  2022
- **Variable clinical severity in TANGO2 deficiency: Case series and literature review.** *American journal of medical genetics. Part A*
  Schymick, J., Leahy, P., Cowan, T., Ruzhnikov, M. R., Gates, R., Fernandez, L., Pramanik, G., Undiagnosed Diseases Network, Yarlagadda, V., Wheeler, M., Bernstein, J. A., Enns, G. M., Lee, C.
  2021
- **Arginine to ornithine ratio as a diagnostic marker in patients with positive newborn screening for hyperargininemia.** *Molecular genetics and metabolism reports*
  Huang, Y., Sharma, R., Feigenbaum, A., Lee, C., Sahai, I., Sanchez Russo, R., Neira, J., Brooks, S. S., Jackson, K. E., Wong, D., Cederbaum, S., Lacbawan, F. L., Rowland, et al
  2021; 27: 100735
- **Profound neonatal lactic acidosis and renal tubulopathy in a patient with glycogen storage disease type IXα2 secondary to a de novo pathogenic variant in PHKA2.** *Molecular genetics and metabolism reports*
  Morales, J. A., Tise, C. G., Narang, A., Grimm, P. C., Enns, G. M., Lee, C. U.
  2021; 27: 100765
- **Unexpected diagnoses in patients with abnormal newborn screening**
  Tise, C., Velez-Bartolomei, F., Morales, J., Lee, C., Bernstein, J., Enns, G.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2021: S354
- **COVID-19 patient impact: A survey of the Gaucher community involving patients, caregivers and family members based in the US to determine impact of the pandemic**
  Ryan, E., Lopez, G., Balwani, M., Barbouth, D., Burrow, T., Ginns, E., Goker-Alpan, O., Grabowski, G., Kartha, R., Kishnani, P., Lau, H., Lee, C., Mistry, et al
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2021: S93

- **Aicardi-Goutières syndrome may present with positive newborn screen for X-linked adrenoleukodystrophy.** *American journal of medical genetics. Part A*
  Tise, C. G., Morales, J. A., Lee, A. S., Velez-Bartolomei, F. n., Floyd, B. J., Levy, R. J., Cusmano-Ozog, K. P., Feigenbaum, A. S., Ruzhnikov, M. R., Lee, C. U., Enns, G. M.
  2021

- **MERRF**
  Velez-Bartolomei, F., Lee, C., Enns, G.
  GeneReviews/University of Washington Seattle. GeneReviews.
  2021 ; GeneReviews

- **Gaucher disease and SARS-CoV-2 infection: Emerging management challenges.** *Molecular genetics and metabolism*
  Mistry, P. n., Balwani, M. n., Barbouth, D. n., Burrow, T. A., Ginns, E. I., Goker-Alpan, O. n., Grabowski, G. A., Kartha, R. V., Kishnani, P. S., Lau, H. n., Lee, C. U., Lopez, G. n., Maegawa, et al
  2020

- **Rare Saposin A deficiency: Novel variant and psychosine analysis.** *Molecular genetics and metabolism*
  Calderwood, L. n., Wenger, D. A., Matern, D. n., Dahmoush, H. n., Watiker, V. n., Lee, C. n.
  2019

- **TREATMENT WITH CHOLIC ACID LEADS TO RESOLUTION OF RENAL CYSTS IN CONGENITAL BILE ACID SYNTHESIS DISORDER TYPE I**
  Leahy, P. J., Lee, C., Schelley, S.
  BMJ PUBLISHING GROUP.2019: 208

- **Two de novo novel mutations in one SHANK3 allele in a patient with autism and moderate intellectual disability.** *American journal of medical genetics. Part A*
  Zhu, W. n., Li, J. n., Chen, S. n., Zhang, J. n., Vetrini, F. n., Braxton, A. n., Eng, C. M., Yang, Y. n., Xia, F. n., Keller, K. L., Okinaka-Hu, L. n., Lee, C. n., Holder, et al
  2018

- **A NOVEL AUTOSOMAL DOMINANT SYNDROME RESULTING FROM VARIANTS IN CDC42**
  Foskett, G. K., Lee, C., Calderwood, L., Stevenson, D.
  BMJ PUBLISHING GROUP.2018: 170–71

- **Biochemical characteristics of newborns with carnitine transporter defect identified by newborn screening in California.** *Molecular genetics and metabolism*
  Gallant, N. M., Leydiker, K. n., Wilnai, Y. n., Lee, C. n., Lorey, F. n., Feuchtbaum, L. n., Tang, H. n., Carter, J. n., Enns, G. M., Packman, S. n., Lin, H. J., Wilcox, W. R., Cederbaum, et al
  2017

- **Molecular and clinical spectra of FBXL4 deficiency.** *Human mutation*
  El-Hattab, A. W., Dai, H. n., Almannai, M. n., Wang, J. n., Faqeih, E. A., Al Asmari, A. n., Saleh, M. A., Elamin, M. A., Alfadhel, M. n., Alkuraya, F. S., Hashem, M. n., Aldosary, M. S., Almass, et al
  2017

- **De novo mutations on maternal alleles in two patients with neuronopathic Gaucher disease**
  Sabbadini, M., Oglesbee, D., Foster-Barber, A., Segal, S., Alhariri, A., Lee, C., Muller, E., Packman, S.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2015: 359–60

- **GAUCHER DISEASE AND LANGERHANS CELL HISTIOCYTOSIS**
  Alhariri, A., Lee, C., Muller, E., Sabbadini, M., Oglesbee, D., Fisher, J., Segal, S., Packman, S.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2014: 267–68

- **Germline loss-of-function mutations in LZTR1 predispose to an inherited disorder of multiple schwannomas** *NATURE GENETICS*
  Piotrowski, A., Xie, J., Liu, Y. F., Poplawski, A. B., Gomes, A. R., Madanecki, P., Fu, C., Crowley, M. R., Crossman, D. K., Armstrong, L., Babovic-Vuksanovic, D., Bergner, A., Blakeley, et al
  2014; 46 (2): 182-?

- **PREGNANCY OUTCOMES IN MAPLE SYRUP URINE DISEASE**
  Sparks, T., Lee, C., Li, B., Packman, D.

LIPPINCOTT WILLIAMS & WILKINS.2014: 199

- **Teaching NeuroImages: Infant with glutaric aciduria type 1 presenting with infantile spasms and hypsarrhythmia** *NEUROLOGY*
  Young-Lin, N., Shalev, S., Glenn, O. A., Gardner, M., Lee, C., Wynshaw-Boris, A., Gelfand, A. A.
  2013; 81 (24): E182–E183

- **Homozygosity for a FBN1 missense mutation causes a severe Marfan syndrome phenotype** *CLINICAL GENETICS*
  HOGUE, J., Lee, C., JELIN, A., Strecker, M. N., Cox, V. A., Slavotinek, A. M.
  2013; 84 (4): 392-393

- **The phenotype of Floating-Harbor syndrome: clinical characterization of 52 individuals with mutations in exon 34 of SRCAP** *ORPHANET JOURNAL OF RARE DISEASES*
  Nikkel, S. M., Dauber, A., de Munnik, S., Connolly, M., Hood, R. L., Caluseriu, O., Hurst, J., Kini, U., Nowaczyk, M. J., Afenjar, A., Albrecht, B., Allanson, J. E., Balestri, et al
  2013; 8