Stanford

Chung Lee

Clinical Assistant Professor, Pediatrics - Medical Genetics

CLINICAL OFFICES

- **Medical Genetics**

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Bio

CLINICAL FOCUS

- Clinical Genetics
- Biochemical Genetics

ACADEMIC APPOINTMENTS

- Clinical Assistant 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)

PROFESSIONAL EDUCATION

- 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)
- Board Certification: Clinical Genetics, American Board of Medical Genetics and Genomics (2013)
- 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

Research & Scholarship

CLINICAL TRIALS

- "The MaP Study": Mapping the Patient Journey in MMA and PA, Recruiting
- A Phase 1/2 Study of AEB1102 in Patients With Arginase I Deficiency, Recruiting

Publications

PUBLICATIONS

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

- **Rare Saposin A deficiency: Novel variant and psychosine analysis.** *Molecular genetics and metabolism*
  Calderwood, L., Wenger, D. A., Matern, D., Dahmoush, H., Watiker, V., Lee, C.
  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., Li, J., Chen, S., Zhang, J., Vetrini, F., Braxton, A., Eng, C. M., Yang, Y., Xia, F., Keller, K. L., Okinaka-Hu, L., Lee, C., 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., Wilnai, Y., Lee, C., Lorey, F., Feuchtbaum, L., Tang, H., Carter, J., Enns, G. M., Packman, S., 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., Almannai, M., Wang, J., Faqeih, E. A., Al Asmari, A., Saleh, M. A., Elamin, M. A., Alfadhel, M., Alkuraya, F. S., Hashem, M., 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