Stanford

Jillian Buchan

Clinical Assistant Professor, Pathology

CLINICAL OFFICES

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Bio

BIO

Jillian is a board-certified Clinical Molecular Geneticist working in Stanford Medicine’s Clinical Genomics Program (CGP). She completed a research-based MS at University College Dublin in Ireland and later received her PhD in Molecular Genetics and Genomics in 2014 from Washington University in St. Louis. After her PhD, Jillian joined Harvard Medical School's Genetics Training Program and completed her fellowship in Clinical Molecular Genetics in 2016. Jillian then joined the Department of Pathology at Stanford School of Medicine and became board-certified by the American Board of Medical Genetics and Genomics in 2017. Her focus is on molecular-based diagnostic testing, with the majority of her time spent in the CGP, where she oversees overall laboratory operations, development of new next-generation sequencing-based clinical assays, ensures CAP/CLIA regulatory compliance, and signs out clinical test reports. She and her team launched Stanford's first clinical exome sequencing test, and the first test for the newly created CGP, in early 2018.

CLINICAL FOCUS

- Genetics
- Pathology
- Genomics

ACADEMIC APPOINTMENTS

- Clinical Assistant Professor, Pathology

ADMINISTRATIVE APPOINTMENTS

- Associate Program Director, Laboratory Genetics and Genomics Training Program, (2019- present)
- Assistant Laboratory Director, Stanford Medicine Clinical Genomics Program, (2016- present)

BOARDS, ADVISORY COMMITTEES, PROFESSIONAL ORGANIZATIONS

- Diplomate (Clinical Molecular Genetics), American Board of Medical Genetics and Genomics (2017 - present)
- Contributor, Clinical & Laboratory Standards Institute Document Development Committee on Nucleic Acid Sequencing (MM09) (2018 - present)
- Fellow, American College of Medical Genetics and Genomics (2018 - present)

PROFESSIONAL EDUCATION

- Fellowship, Harvard Medical School , ABMGG Clinical Molecular Genetics Fellowship (2016)
- PhD, Washington University in St. Louis , Molecular Genetics and Genomics (2014)
- MS, University College Dublin , Research-based (2010)
- BS, University of Washington , Molecular Cellular and Developmental Biology (2008)

Publications

PUBLICATIONS

- **Rapid Genome Sequencing in the Critically Ill** *CLINICAL CHEMISTRY*
  Buchan, J. G., White, S., Joshi, R., Ashley, E. A.
  2019; 65 (6): 723–26

- **Adaptation and validation of the ACMG/AMP variant classification framework for MYH7-associated inherited cardiomyopathies: recommendations by ClinGen's Inherited Cardiomyopathy Expert Panel** *GENETICS IN MEDICINE*
  Kelly, M. A., Caleshu, C., Morales, A., Buchan, J., Wolf, Z., Harrison, S. M., Cook, S., Dillon, M. W., Garcia, J., Haverfield, E., Jongbloed, J. H., Macaya, D., Manrai, et al
  2018; 20 (3): 351–59

- **Canine MAS1: monoallelic expression is suggestive of an imprinted gene.** *Animal genetics*
  Nolan, C. M., Shiel, R. E., Buchan, J. G., O'Sullivan, F. M., Callanan, J. J.
  2018; 49 (5): 438–46

- **Long-read genome sequencing identifies causal structural variation in a Mendelian disease.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Merker, J. D., Wenger, A. M., Sneddon, T., Grove, M., Zappala, Z., Fresard, L., Waggott, D., Utiramerur, S., Hou, Y., Smith, K. S., Montgomery, S. B., Wheeler, M., Buchan, et al
  2017

- **Kinesin family member 6 (kif6) is necessary for spine development in zebrafish.** *Developmental dynamics : an official publication of the American Association of Anatomists*
  Buchan, J. G., Gray, R. S., Gansner, J. M., Alvarado, D. M., Burgert, L., Gitlin, J. D., Gurnett, C. A., Goldsmith, M. I.
  2014; 243 (12): 1646–57

- **Are copy number variants associated with adolescent idiopathic scoliosis?** *Clinical orthopaedics and related research*
  Buchan, J. G., Alvarado, D. M., Haller, G., Aferol, H., Miller, N. H., Dobbs, M. B., Gurnett, C. A.
  2014; 472 (10): 3216–25

- **Rare variants in FBN1 and FBN2 are associated with severe adolescent idiopathic scoliosis.** *Human molecular genetics*
  Buchan, J. G., Alvarado, D. M., Haller, G. E., Cruchaga, C., Harms, M. B., Zhang, T., Willing, M. C., Grange, D. K., Braverman, A. C., Miller, N. H., Morcuende, J. A., Tang, N. L., Lam, et al
  2014; 23 (19): 5271–82

- **Copy number analysis of 413 isolated talipes equinovarus patients suggests role for transcriptional regulators of early limb development** *EUROPEAN JOURNAL OF HUMAN GENETICS*
  Alvarado, D. M., Buchan, J. G., Frick, S. L., Herzenberg, J. E., Dobbs, M. B., Gurnett, C. A.
  2013; 21 (4): 373-380

- **MYBPC1 mutations impair skeletal muscle function in zebrafish models of arthrogryposis.** *Human molecular genetics*
  Ha, K., Buchan, J. G., Alvarado, D. M., McCall, K., Vydyanath, A., Luther, P. K., Goldsmith, M. I., Dobbs, M. B., Gurnett, C. A.
  2013; 22 (24): 4967–77

- **Polygenic threshold model with sex dimorphism in adolescent idiopathic scoliosis: the Carter effect.** *The Journal of bone and joint surgery. American volume*
  Kruse, L. M., Buchan, J. G., Gurnett, C. A., Dobbs, M. B.
  2012; 94 (16): 1485–91

- **Exome sequencing identifies an MYH3 mutation in a family with distal arthrogryposis type 1.** *The Journal of bone and joint surgery. American volume*

Alvarado, D. M., Buchan, J. G., Gurnett, C. A., Dobbs, M. B.
2011; 93 (11): 1045–50

- **Familial isolated clubfoot is associated with recurrent chromosome 17q23.1q23.2 microduplications containing TBX4.** *American journal of human genetics*
  Alvarado, D. M., Aferol, H., McCall, K., Huang, J. B., Techy, M., Buchan, J., Cady, J., Gonzales, P. R., Dobbs, M. B., Gurnett, C. A.
  2010; 87 (1): 154–60

- **SEPT9 gene sequencing analysis reveals recurrent mutations in hereditary neuralgic amyotrophy.** *Neurology*
  Hannibal, M. C., Ruzzo, E. K., Miller, L. R., Betz, B., Buchan, J. G., Knutzen, D. M., Barnett, K., Landsverk, M. L., Brice, A., LeGuern, E., Bedford, H. M., Worrall, B. B., Lovitt, et al
  2009; 72 (20): 1755–59

- **Duplication within the SEPT9 gene associated with a founder effect in North American families with hereditary neuralgic amyotrophy.** *Human molecular genetics*
  Landsverk, M. L., Ruzzo, E. K., Mefford, H. C., Buysse, K., Buchan, J. G., Eichler, E. E., Petty, E. M., Peterson, E. A., Knutzen, D. M., Barnett, K., Farlow, M. R., Caress, J., Parry, et al
  2009; 18 (7): 1200–1208