Anshika Srivastava, Ph.D.
Associate Professor
Department of Medical Genetics
Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow.
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asrivastavapgi@gmail.com

EDUCATION

Ph.D.	Medical Genetics, King George Medical University, Lucknow, India and Department of Medical Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow Supervisor: Dr. Balraj Mittal Thesis Title: Influence of Genetic Variations in ABC Transporters and Hormonal Genes in Susceptibility to Gallstone Disease.	2008-2013
M.Sc.	Biochemistry Department of Biochemistry, University of Lucknow, India.	2005-2007

RESEARCH EXPERIENCE

Postdoctoral Fellow	Department of Human Genetics, University of Michigan, Ann Arbor, Michigan, USA Mentor: Dr. Stephanie Bielas, Ph.D. Postdoc area of work: *Genetics of developmental disorders.*	2013-2019
Junior Research Assistant	Department of Medical Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences (SGPGIMS), Lucknow, India. Project Title: *Effect of genetic variant (rs11887534) in ABCG8 gene in coronary artery disease and response to atorvastatin therapy.*	2007-2008

PUBLICATIONS

2024 Werren, E.A., Peirent, E.R., Jantti, H., Guxholli, A., Srivastava, K.R., Orenstein, N., ... & Bielas, S.L., 2024. Biallelic variants in *CSMD1* are implicated in a neurodevelopmental disorder with intellectual disability and variable cortical malformations. *Cell Death & Disease*, 15(5), p.379. https://doi.org/10.1038/s41419-024-06768-6

2024 Werren, E.A., LaForce, G.R., Srivastava, A., Perillo, D.R., Li, S., Johnson, K., ... & Schaffer, A.E., 2024. TREX tetramer disruption alters RNA processing necessary for corticogenesis in THOC6 Intellectual Disability Syndrome. *Nature Communications*, 15(1), p.1640. https://doi.org/10.1038/s41467-024-45948-y

2024 Ryan, C.W., Regan, S.L., Mills, E.F., McGrath, B.T., Gong, E., Lai, Y.T., Sheingold, J.B., Patel, K., Horowitz, T., Moccia, A., Tsan, Y.C., Srivastava, A., & Bielas, S.L., 2024. RING1 missense variants reveal sensitivity of DNA damage repair to H2A monoubiquitination dosage during neurogenesis. *Nature Communications*, 15(1). https://doi.org/10.1038/s41467-024-52292-8

2023 Werren, E.A., Srinivasan, V.M., Gowda, V.K., Pandey, A., Vaish, S., Kabbur, A.R., Nandeesh, B.N., & Srivastava, A., 2023. A novel biallelic frameshift variant in *C2orf69* causing developmental regression, seizures, microcephaly, autistic features, and hypertonia. *American Journal of Medical Genetics Part A*, 191(9), pp.2446–2450. https://doi.org/10.1002/ajmg.a.63310

Nanjundagowda, V.K., Paikaraya, S., Srinivasan, V.M., & Srivastava, A., 2023. In silico characterization of *RNASEH2A* pathogenic variants and identification of novel splice site donor variant c.549+1G>T in Indian population. *Cureus*. https://doi.org/10.7759/cureus.40366

2023 Kumar, S., Mishra, A., Srivastava, A., Mittal, R., & Mittal, B., 2023. Genetics of left ventricular dysfunction in coronary artery disease. In: *Biomedical Research, Medicine, and Disease*. CRC Press, pp.167–202.

2023 McGrath, B., Tsan, Y., Salvi, S., Ghali, N., Martin, D., Hannibal, M., Keegan, C., Helms, A., Srivastava, A., & Bielas, S., 2022. Aberrant extracellular matrix and cardiac development in models lacking the PR-DUB component *ASXL3*. *bioRxiv (Cold Spring Harbor Laboratory)*. https://doi.org/10.1101/2022.07.14.500124

2023 McGrath, B., Wu, P., Salvi, S., Girgla, N., Chen, X., Zhu, J., Kc, R., Tsan, Y., Moccia, A., Srivastava, A., Zhou, X., & Bielas, S., 2021. *ASXL3* controls cortical neuron fate specification through extrinsic self-renewal pathways. *bioRxiv (Cold Spring Harbor Laboratory)*. https://doi.org/10.1101/2021.07.20.452995

2018 Sarkar, M.K., Tsoi, L.C., Xing, X., Liang, Y., Berthier, C.C., Swindell, W.R., Wolterink, L., Patrick, M., Hile, G.A., Tsou, P.S., Beamer, M.A., Srivastava, A., Bielas, S.L., Liu, J., Harms, P.W., Getsios, S., Johnston, A., Elder, J.T., Voorhees, J.J., Kahlenberg, J.M. & Gudjonsson, J.E., 2018. Photosensitivity and type I IFN responses in cutaneous lupus are driven by epidermal derived interferon kappa. *Annals of the Rheumatic Diseases*, 77(11), pp.1653–1664. https://doi.org/10.1136/annrheumdis-2017-212404

2018 Srivastava, A., Srivastava, K., Hebbar, M., Galada, C., Kadavigrere, R., Cao, X., Su, F., Chinnaiyan, A.M., Girisha, K.M., Shukla, A. & Bielas, S.L., 2018. Genetic diversity of *NDUFV1*-dependent mitochondrial complex I deficiency. *European Journal of Human Genetics*, 26(11), pp.1582–1587. https://doi.org/10.1038/s41431-018-0171-z

2018 Moccia, A., Srivastava, A., Skidmore, J.M., Bernat, J.A., Wheeler, M., Chong, J.X., Nickerson, D., Bamshad, M., Hefner, M.A., Martin, D.M. & Bielas, S.L., 2018. Genetic analysis of CHARGE syndrome identifies overlapping molecular biology. *Genetics in Medicine*, 20(9), pp.1022–1029. https://doi.org/10.1038/gim.2017.236

2018 Bélanger, C., Bérubé-Simard, F., Leduc, E., Bernas, G., Campeau, P., Lalani, S., Martin, D., Bielas, S., Moccia, A., Srivastava, A., Silversides, D.W. & Pilon, N., 2018. Dysregulation of co-transcriptional alternative splicing underlies CHARGE syndrome. *Proceedings of the National Academy of Sciences*, 115(4), pp.E620–E629. https://doi.org/10.1073/pnas.1715830115

2018 Jordan, V., Fregeau, B., Jessica, G., Wapner, J., Balci, T.B., Carter, M., Bernat, J., Moccia, A., Srivastava, A., Martin, D., Bielas, S., Pappas, J., Melissa, S., Rio, M., Cantagrel, V., Lewis, A.M., Scaglia, F., Kohler, J., Bernstein, J.A., Zornio, P.A., Rosenfeld, J.A., Sherr, E.H., Bi, W. & Scott, D.A., 2018. Genotype–phenotype correlations in individuals with *RERE* mutations. *Human Mutation*, 39(5), pp.666–675. https://doi.org/10.1002/humu.23402

2018 Galada, C., Hebbar, M., Lewis, L., Soans, S., Kadavigere, R., Srivastava, A., Bielas, S., Girisha, K.M. & Shukla, A., 2018. Report of four novel variants in *ASNS* causing asparagine synthetase deficiency and review of literature. *Congenital Anomalies (Kyoto)*, 58(5), pp.181–182. https://doi.org/10.1111/cga.12259

2017 Hebbar, M., Girisha, K.M., Srivastava, A., Bielas, S. & Shukla, A., 2017. Homozygous c.359del variant in *MGME1* is associated with early onset cerebellar ataxia. *European Journal of Medical Genetics*, 60(10), pp.533–535. https://doi.org/10.1016/j.ejmg.2017.07.008

2017 Srivastava, A., McGrath, B. & Bielas, S.L., 2017. Role of histone H2A ubiquitination in neurodevelopmental disorders. *Trends in Genetics*, 33(8), pp.566–578. https://doi.org/10.1016/j.tig.2017.05.006

2017 Shukla, A., Hebbar, M., Srivastava, A., Kadavigere, R., Upadhyai, P., Kanthi, A., Brandau, O., Bielas, S.L. & Girisha, K.M., 2017. Homozygous c.259G>A variant in *ISCA1* is associated with a new multiple mitochondrial dysfunctions syndrome. *Journal of Human Genetics*, 62(7), pp.723–727. https://doi.org/10.1038/jhg.2017.33

2016 Srivastava, A., KC, R., Tsan, Y.C., Liao, R., Hannibal, M., Keegan, C., Chinnaiyan, A.M., Martin, D.M. & Bielas, S.L., 2016. De novo dominant *ASXL3* mutations alter H2A deubiquitination and transcription in Bainbridge-Ropers Syndrome. *Human Molecular Genetics*, 25(3), pp.597–608. https://doi.org/10.1093/hmg/ddv502

2016 Kc, R., Srivastava, A., Wilkowski, J.M., Richter, C.E., Shavit, J.A., Burke, D.T. & Bielas, S.L.,

2016. Detection of nucleotide-specific CRISPR/Cas9 modified alleles using multiplex ligation detection. *Scientific Reports*, 6, p.32048. https://doi.org/10.1038/srep32048

2016 Kumar, S., Mishra, A., Srivastava, A., Bhatt, M., Garg, N., Agarwal, S.K., Pande, S. & Mittal, B., 2016. Role of common sarcomeric gene polymorphisms in genetic susceptibility to left ventricular dysfunction. *Journal of Genetics*, 95(2), pp.263–272. https://doi.org/10.1007/s12041-016-0653-3

2015 Moorthy, N., Garg, N., Kapoor, A., Tewari, S., Kumar, S., Sinha, A., Shrivastava, A. & Goel, P.K., 2015. In Reply–Time to Focus on Preventing Coronary Artery Disease Through Exercise Training Among Normoglycemic Individuals. *Mayo Clinic Proceedings*, 90(3), pp.418–419. https://doi.org/10.1016/j.mayocp.2015.01.001

2014 Kumar, S., Mishra, A., Srivastava, A., Mittal, T., Garg, N. & Mittal, B., 2014. Significant role of *ADRB3* rs4994 towards development of coronary artery disease. *Coronary Artery Disease*, 25(1), pp.29–34. https://doi.org/10.1097/MCA.0000000000000053

2014 Sharma, K.L., Rai, R., Srivastava, A., Sharma, A., Misra, S., Kumar, A. & Mittal, B., 2014. A multigenic approach to evaluate genetic variants of *PLCE1*, *LXRs*, *MMPs*, *TIMPs* and *CYP* genes in gallbladder cancer predisposition. *Tumor Biology*, 35(9), pp.8597–8606. https://doi.org/10.1007/s13277-014-2154-7

2014 Mishra, A., Srivastava, A., Mittal, T., Garg, N. & Mittal, B., 2014. Genetic predisposition to left ventricular dysfunction: a multigenic and multi-analytical approach. *Gene*, 546(2), pp.309–317. https://doi.org/10.1016/j.gene.2014.05.047

2014 Garg, N., Moorthy, N., Kapoor, A., Tewari, S., Kumar, S., Sinha, A., Shrivastava, A. & Goel, P.K., 2014. Hemoglobin A1c in nondiabetic patients: an independent predictor of coronary artery disease and its severity. *Mayo Clinic Proceedings*, 89(7), pp.908–916. https://doi.org/10.1016/j.mayocp.2014.03.021

2013 Von Kampen, O. *et al.*, 2013. Genetic and functional identification of the likely causative variant for gallstone disease at the ABCG5/8 lithogenic locus. *Hepatology*, 57(6), pp.2407–2417. https://doi.org/10.1002/hep.26200

2013 Srivastava, A., Mishra, A., Singh, R., Rai, R., Srivastava, N. & Mittal, B., 2013. Multi-analytic approach elucidates significant role of hormonal and hepatocanalicular transporter genetic variants in gallstone disease in North Indian population. *PLoS One*, 8(4), p.e59173. https://doi.org/10.1371/journal.pone.0059173

2013 Mishra, A., Srivastava, A., Mittal, T., Garg, N. & Mittal, B., 2013. Role of inflammatory gene polymorphisms in LVD susceptibility in CAD patients. *Cytokine*, 61(3), pp.856–861. https://doi.org/10.1016/j.cyto.2012.12.016

2013 Mishra, A., Srivastava, A., Mittal, T., Garg, N. & Mittal, B., 2015. Role of Angiotensin II type I (AT1) Receptor Polymorphism in Left Ventricular Dysfunction. *Indian Heart Journal*, 67(3), pp.214–221. https://doi.org/10.1016/j.ihj.2015.03.008

2012 Srivastava, A., Sharma, K.L., Srivastava, N., Mishra, S. & Mittal, B., 2012. Significant role of Estrogen and Progesterone receptor sequence variants in Gallbladder Cancer predisposition: A Multi-analytical strategy. *PLoS One*, 7(7), p.e40162. https://doi.org/10.1371/journal.pone.0040162

2012 Mishra, A., Srivastava, A., Mittal, T., Garg, N. & Mittal, B., 2012. Impact of renin angiotensin-aldosterone system gene polymorphisms on left ventricular dysfunction in coronary artery disease patients. *Disease Markers*, 32(1), pp.33–41. https://doi.org/10.3233/DMA-2011-0856

2012 Mishra, A., Srivastava, A., Mittal, T., Garg, N. & Mittal, B., 2012. Association of matrix metalloproteinases (MMP2, MMP7 and MMP9) genetic variants with left ventricular dysfunction in coronary artery disease patients. *Clinica Chimica Acta*, 413(19–20), pp.1668–1674. https://doi.org/10.1016/j.cca.2012.06.010

2011 Srivastava, A., Srivastava, A., Srivastava, N., Choudhuri, G. & Mittal, B., 2011. Organic anion transporter 1B1 (*SLCO1B1*) polymorphism and gallstone formation: High incidence of Exon4 CA genotype in female patients in North India. *Hepatology Research*, 41(1), pp.71–78.

https://doi.org/10.1111/j.1872-034X.2010.00744.x

2011 Srivastava, A., Garg, N., Mittal, T., Khanna, R., Gupta, S., Seth, P.K. & Mittal, B., 2011. Association of 25 bp deletion in *MYBPC3* gene with left ventricle dysfunction in coronary artery disease patients. *PLoS One*, 6(9), p.e24123. https://doi.org/10.1371/journal.pone.0024123

2010 Srivastava, A., Garg, N., Srivastava, A., Srivastava, K. & Mittal, B., 2010. Effect of genetic variant (rs11887534) in *ABCG8* gene in coronary artery disease and response to atorvastatin therapy. *Disease Markers*, 28(5), pp.307–313. https://doi.org/10.1155/2010/741357

2010 Srivastava, A., Srivastava, A., Srivastava, K., Choudhuri, G. & Mittal, B., 2010. Role of *ABCG8* D19H (rs11887534) variant in gallstone susceptibility in northern India. *Journal of Gastroenterology and Hepatology*, 25(11), pp.1758–1762. https://doi.org/10.1111/j.1440-1746.2010.06389.x

Book Chapters

2024 Pooja, Srinivasan, V. M., & **Srivastava, A**. (2024). Genetics of neuronal and glioneuronal cancers. In Elsevier eBooks (pp. 339–369). https://doi.org/10.1016/b978-0-323-95114-2.00022-4

2014 Mittal B, Mishra A, **Srivastava A**, Garg N. Emerging Role of Genetic Variants of Matrix Metalloproteinases Genes in Left Ventricular Dysfunction. In: Dhalla N., Chakraborti S. (eds) Role of Proteases in Cellular Dysfunction. ***Advances in Biochemistry in Health and Disease***, 2014; vol 8. Springer, New York, NY

2014 Mittal B, Mishra A, **Srivastava A**, Kumar S, Garg N. Matrix metalloproteinase in coronary artery disease. ***Adv Clin Chem.*** 2014; 64:1-72

FELLOWSHIP AND AWARDS

2024 Awarded Best Research Paper Award at Research Day Sanjay Gandhi Post Graduate Institute of Medical Sciences

2023 Secured CSIR A Special Call for Research Grants for Women Scientists (CSIR-ASPIRE) 2023. Awarded to 10% of the female scientist for research in India

2020 Ramalingaswami Fellow 2020, Department of Biotechnology (DBT), Govt. of India.

2019 Innovative Young Biotechnologist Award (IYBA), Department of Biotechnology (DBT), Govt. of India.

2017 Best Postdoctoral Poster award in the Department of Human Genetics, University of Michigan Medical School. Awarded to one postdoctoral fellow in the department at yearly retreat. Awardee receives a cash prize.

2012 Awarded Senior Research Fellowship from Indian Council of Medical Research.

2011 Travel Grant from several funding agencies namely DST, DBT, CSIR and ICMR, India to attend the 12th International Congress of Human Genetics, October 11-15, 2011, Montreal, Canada.

2010 Best Poster Award at 35th Annual Conference of the Indian Society of Human Genetics (ISHG) and an International Symposium on Role of Genomics in Clinical Practice 2010, Lucknow, India.

2004 Stood 1st in quiz competition on CSIR foundation day at CDRI, September 2004

ORAL PRESENTATIONS AND INVITED TALKS

2024 Guest speaker at Manipal Genetics Update VII on Cellular and Animal Models for Rare Genetic Diseases held from January 18th to 20th, 2024, at the Department of Medical Genetics, Kasturba

Medical College, Manipal Academy of Higher Education, Manipal. Presentation Title "Biallelic variants in CSMD1 are implicated in a neurodevelopmental disorder with intellectual disability and variable cortical malformations".

2023 Invited talk titled Role of ASXL3 in Neurodevelopmental Disorders presented at MPAICON2023, the 10th Annual Conference of the Molecular Pathology Association of India (MPAI), 18th and 19thFebruary 2023 at the Convention Center, SGPGI, Lucknow, India.

2018 Oral presentation to pitch the poster entitled as "Translation human genetics to precision medicine poster" at *Young Investigator Meet 2018*; September 8-9, 2018, Chicago, USA.

2017 Oral presentation entitled as "Congenital heart defects in Bainbridge Ropers Syndrome" at *American Society of Human Genetics*; October 17-21, 2017, Orlando, Florida.

2014 Oral presentation entitled as "Novel insights into the varied genetic background of neurodevelopment disorder" at *INDO-US Symposium on Genomic insights into Human Morphogenesis: Prenatal, Postnatal and Molecular Dysmorphology and First Annual Meeting of Society for Indian Academy of Medical Genetics;* November 7-9, 2014, Hyderabad, India.

2014 Invited talk entitled as "A genetic approach to understanding the pediatrics brain disorders" at the *Department of Medical Genetics, SGPGIMS;* October 30, 2014, Lucknow, India.

POSTER PRESENTATIONS

2016 **Srivastava A**, KC R, Moccia A, Bielas SL. Molecular mechanisms of Asxl3 in neurodevelopment. "*Cell Symposia: Transcriptional Regulation in Development and Disease"* June 26-28, 2016, Chicago, USA.

2016 Martin D, Niederriter AN, Sperry, ED, Skidmore, JM, **Srivastava A**, Attardi L, Heller S, Scacheri P. Neuroblast-specific bias for CHD7 with potential roles in regulation of LONG GENES in the inner ear. *"Associate for Research in Otolaryngology"* February 20-24, 2016, San Diego, California, USA.

2015 Bielas SL, **Srivastava A**, KC R, Tsan YC, Chinnaiyan AM, Bielas SL. ASXL3 regulates H2A deubiquitination and gene transcription in Bainbridge-Ropers-Syndrome. *"American Society of Human Genetics"* October 6-10, 2015, Baltimore MD, USA.

2014 **Srivastava A**, Phadke SR, Girisha KM, Martin DM, Leber S, Innis J, Chinnaiyan AM, Nickerson DA, Bielas SL. Novel insights into the varied genetic background of neurodevelopment disorder. *"INDO-US Symposium on Genomic insights into Human Morphogenesis: Prenatal, Postnatal and Molecular Dysmorphology and First Annual Meeting of Society for Indian Academy of Medical Genetics"* November 7-9, 2014, Hyderabad, India.

2013 **Srivastava A**, Mishra A, Mittal B. Significant Role of Combined Alleles of Hormonal Receptors and Hepatocanalicular Transporter Gene in Susceptibility to Cholesterol Gallstone Disease. *"American Society of Human Genetics"* October 22-26, 2013, Boston, USA.

2012 **Srivastava A**, Srivastava N, Sharma K L, Mishra A, Mittal B. Role of *ESR1, ESR2* and *PGR* genetic variants in gallbladder cancer susceptibility in Northern India*."International Conference on Genes, Genetics & Genomics Today & Tomorrow: Human Concerns and 37th Annual Conference of Indian Society of Human Genetics"* March 3-5, 2012 Panjab University, Chandigarh, India*.*

2011 **Srivastava A**, Sharma K L, Srivastava N, Misra S, Kumar A, Mittal B. Role of genetic variants of *ESR1and ESR2* in susceptibility to gallbladder cancer. *"12th International Congress of Human Genetics",* October 11-15, 2011 Montreal, Canada.

2011 **Srivastava A**, Srivastava N, Mittal B. Role of *ABCG8* D19H (rs11887534) variant in gallstone susceptibility in northern India. *"International Conference Genomics, Genetic Disease and Diagnostics and 36th Annual Conference of the Indian Society of Human Genetics (ISHG)"*, Feb 14-16, 2011 Manipal University, India.

2010 **Srivastava A**, Srivastava A, Srivastava N, Mittal B. "Role of *SLCO1B1* transporters in gallstone

susceptibility in North Indian Population. *"35th Annual Conference of the Indian Society of Human Genetics (ISHG) and an International Symposium on Role of Genomics in Clinical Practice"* March 6-8, 2010, Lucknow, India.

2009 **Srivastava A**, Srivastava A, Srivastava K, Choudhuri G, Mittal B. Genetic variant of *ABCG8* and risk of gallbladder diseases *"ISGCON"* December 9-13, 2009, Kolkata, India.