BIOGRAPHICAL SKETCH

Provide the following information for the key personnel and other significant contributors in the order listed on Form Page 2.
Follow this format for each person. **DO NOT EXCEED FOUR PAGES.**

NAME	POSITION TITLE
Lalani, Seema R.	Assistant Professor
eRA COMMONS USER NAME (credential, e.g., agency login) SEEMAL	

EDUCATION/TRAINING *(Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.)*

INSTITUTION AND LOCATION	DEGREE *(if applicable)*	YEAR(s)	FIELD OF STUDY
St. Joseph's College, Karachi, Pakistan	H.S.C.	1988	Biology
Aga Khan University, Karachi, Pakistan	M.B.,B.S.	1994	Medicine
Hershey Medical Center, Hershey, PA	Diplomate of the ABP	2000	Pediatrics
Baylor College of Medicine, Houston, TX	Diplomate of the ABMG	2002	Clinical Genetics
Baylor College of Medicine, Houston, TX	Diplomate of the ABMG	2005	Clinical Cytogenetics

A. Personal Statement

My research interests involve the study of DNA copy number variations associated with human birth defects with emphasis on congenital cardiovascular malformations. I'm also interested in identifying population-specific genetic variants responsible for speech and language disorders in children. I have additionally worked on the molecular aspects of CHARGE syndrome.

B. Positions and Honors

Positions and Employment

1996-1997	Internship, Dept. of Pediatrics, Hershey Medical Center, Hershey, PA
1997-1999	Residency, Dept. of Pediatrics, Hershey Medical Center, Hershey, PA
1999-2001	Clinical Genetics fellowship, Dept. of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX
2001-2002	Research Associate, Dept. of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX
2003-2005	Clinical Cytogenetics fellowship, Baylor College of Medicine, Houston, TX
2002-2007	Assistant Professor, non-tenure-track, Dept of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX
2007-present	Assistant Professor, tenure-track, Dept of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX
2005-present	Assistant Laboratory Director, Kleberg Cytogenetics Laboratory, Baylor College of Medicine, Houston, TX

Professional Associations and Board Certifications

2004-present Member of the American Society of Human Genetics
2009-present Member of the Society of Pediatric Research

Certification by the American Board of Pediatrics (2000-2007) and recertification (2007-2014) Diplomate of the American Board of Medical Genetics in Clinical Genetics (2002-2012) and Clinical Cytogenetics (2005-2015)

Honors and Awards

2002-2007	Recipient of the Doris Duke Clinical Scientist Development Award (CSDA) to study the genetic basis of CHARGE syndrome
2007-2008	Recipient of the Gillson Longenbaugh Foundation Award to study DNA copy-number alterations in cardiovascular malformations

C. Peer-reviewed publications

1. Edelmann L, Spiteri E, McCain N, Goldberg R, Pandita RK, Duong S, Fox J, Blumenthal D, **Lalani SR**, Shaffer LG, Morrow BE. A common breakpoint on 11q23 in carriers of the constitutional t(11;22) translocation. *Am J Hum Genet.* 1999;65(6):1608-1616.

2. Sutton VR, Coveler KJ, **Lalani SR**, Kashork CD, Shaffer LG. Subtelomeric FISH uncovers trisomy 14q32: lessons for imprinted regions, cryptic rearrangements and variant acrocentric short arms. *Am J Med Genet.* 2002;112(1):23-27.

3. **Lalani SR**, Stockton DW, Bacino C, Molinari LM, Glass NL, Fernbach SD, Towbin JA, Craigen WJ, Graham JM, Jr., Hefner MA, Lin AE, McBride KL, Davenport SL, Belmont JW. Toward a genetic etiology of CHARGE syndrome: I. A systematic scan for submicroscopic deletions. *Am J Med Genet A.* 2003;118A(3):260-266.

4. **Lalani SR**, Safiullah AM, Molinari LM, Fernbach SD, Martin DM, Belmont JW. SEMA3E mutation in a patient with CHARGE syndrome. *J Med Genet.* 2004;41(7):e94.

5. **Lalani SR**, Safiullah AM, Fernbach SD, Phillips M, Bacino CA, Molinari LM, Glass NL, Towbin JA, Craigen WJ, Belmont JW. SNP genotyping to screen for a common deletion in CHARGE syndrome. *BMC Med Genet.* 2005;6:8.

6. **Lalani SR**, Vladutiu GD, Plunkett K, Lotze TE, Adesina AM, Scaglia F. Isolated mitochondrial myopathy associated with muscle coenzyme Q10 deficiency. *Arch Neurol.* 2005;62(2):317-320.

7. **Lalani SR**, Sahoo T, Sanders ME, Peters SU, Bejjani BA. Coarctation of the aorta and mild to moderate developmental delay in a child with a de novo deletion of chromosome 15(q21.1q22.2). *BMC Med Genet.* 2006;7:8.

8. **Lalani SR**, Safiullah AM, Fernbach SD, Harutyunyan KG, Thaller C, Peterson LE, McPherson JD, Gibbs RA, White LD, Hefner M, Davenport SL, Graham JM, Bacino CA, Glass NL, Towbin JA, Craigen WJ, Neish SR, Lin AE, Belmont JW.

Spectrum of CHD7 mutations in 110 individuals with CHARGE syndrome and genotype-phenotype correlation. *Am J Hum Genet.* 2006;78(2):303-314.

9. Sahoo T, Cheung SW, Ward P, Darilek S, Patel A, del Gaudio D, Kang SH, **Lalani SR**, Li J, McAdoo S, Burke A, Shaw CA, Stankiewicz P, Chinault AC, Van den Veyver IB, Roa BB, Beaudet AL, Eng CM. Prenatal diagnosis of chromosomal abnormalities using array-based comparative genomic hybridization. *Genet Med.* 2006;8(11):719-727.

10. Brunetti-Pierri N, Grange DK, Ou Z, Peiffer DA, Peacock SK, Cooper ML, Eng PA, **Lalani SR**, Chinault AC, Gunderson KL, Craigen WJ, Cheung SW. Characterization of de novo microdeletions involving 17q11.2q12 identified through chromosomal comparative genomic hybridization. *Clin Genet.* 2007;72(5):411-419.

11. Kang SH, Scheffer A, Ou Z, Li J, Scaglia F, Belmont J, **Lalani SR**, Roeder E, Enciso V, Braddock S, Buchholz J, Vacha S, Chinault AC, Cheung SW, Bacino CA. Identification of proximal 1p36 deletions using array-CGH: a possible new syndrome. *Clin Genet.* 2007;72(4):329-338.

12. Ben-Shachar S, Ou Z, Shaw CA, Belmont JW, Patel MS, Hummel M, Amato S, Tartaglia N, Berg J, Sutton VR, **Lalani SR**, Chinault AC, Cheung SW, Lupski JR, Patel A. 22q11.2 distal deletion: a recurrent genomic disorder distinct from DiGeorge syndrome and velocardiofacial syndrome. *Am J Hum Genet.* 2008;82(1):214-221.

13. Brunetti-Pierri N, Berg JS, Scaglia F, Belmont J, Bacino CA, Sahoo T, **Lalani SR**, Graham B, Lee B, Shinawi M, Shen J, Kang SH, Pursley A, Lotze T, Kennedy G, Lansky-Shafer S, Weaver C, Roeder ER, Grebe TA, Arnold GL, Hutchison T, Reimschisel T, Amato S, Geragthy MT, Innis JW, Obersztyn E, Nowakowska B, Rosengren SS, Bader PI, Grange DK, Naqvi S, Garnica AD, Bernes SM, Fong CT, Summers A, Walters WD, Lupski JR, Stankiewicz P, Cheung SW, Patel A. Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. *Nat Genet.* 2008;40(12):1466-1471.

14. Lu XY, Phung MT, Shaw CA, Pham K, Neil SE, Patel A, Sahoo T, Bacino CA, Stankiewicz P, Kang SH, **Lalani S**, Chinault AC, Lupski JR, Cheung SW, Beaudet AL. Genomic imbalances in neonates with birth defects: high detection rates by using chromosomal microarray analysis. *Pediatrics.* 2008;122(6):1310-1318.

15. Shao L, Shaw CA, Lu XY, Sahoo T, Bacino CA, **Lalani SR**, Stankiewicz P, Yatsenko SA, Li Y, Neill S, Pursley AN, Chinault AC, Patel A, Beaudet AL, Lupski JR, Cheung SW. Identification of chromosome abnormalities in subtelomeric regions by microarray analysis: a study of 5,380 cases. *Am J Med Genet A.* 2008;146A(17):2242-2251.

16. Erez A, Patel AJ, Wang X, Xia Z, Bhatt SS, Craigen W, Cheung SW, Lewis RA, Fang P, Davenport SL, Stankiewicz P, **Lalani SR**. Alu-specific microhomology-

mediated deletions in CDKL5 in females with early-onset seizure disorder. *Neurogenetics.* 2009;10(4):363-369.

17. **Lalani SR**, Thakuria JV, Cox GF, Wang X, Bi W, Bray MS, Shaw C, Cheung SW, Chinault AC, Boggs BA, Ou Z, Brundage EK, Lupski JR, Gentile J, Waisbren S, Pursley A, Ma L, Khajavi M, Zapata G, Friedman R, Kim JJ, Towbin JA, Stankiewicz P, Schnittger S, Hansmann I, Ai T, Sood S, Wehrens XH, Martin JF, Belmont JW, Potocki L. 20p12.3 microdeletion predisposes to Wolff-Parkinson-White syndrome with variable neurocognitive deficits. *J Med Genet.* 2009;46(3):168-175.

18. Ben-Shachar S, Bidwa BM, Potocki L, **Lalani SR**. Coexistence of an unbalanced chromosomal rearrangement and spinal muscular atrophy in an infant with multiple congenital anomalies. *Am J Med Genet A.* 2009;149A(3):515-518.

19. Layman WS, McEwen DP, Beyer LA, **Lalani SR**, Fernbach SD, Oh E, Swaroop A, Hegg CC, Raphael Y, Martens JR, Martin DM. Defects in neural stem cell proliferation and olfaction in Chd7 deficient mice indicate a mechanism for hyposmia in human CHARGE syndrome. *Hum Mol Genet.* 2009;18(11):1909-1923.

20. Vissers LE, Bhatt SS, Janssen IM, Xia Z, **Lalani SR**, Pfundt R, Derwinska K, de Vries BB, Gilissen C, Hoischen A, Nesteruk M, Wisniowiecka-Kowalnik B, Smyk M, Brunner HG, Cheung SW, van Kessel AG, Veltman JA, Stankiewicz P. Rare pathogenic microdeletions and tandem duplications are microhomology-mediated and stimulated by local genomic architecture. *Hum Mol Genet.* 2009;18(19):3579-3593.

21. Yan J, Zhang F, Brundage E, Scheuerle A, Lanpher B, Erickson RP, Powis Z, Robinson HB, Trapane PL, Stachiw-Hietpas D, Keppler-Noreuil KM, **Lalani SR**, Sahoo T, Chinault AC, Patel A, Cheung SW, Lupski JR. Genomic duplication resulting in increased copy number of genes encoding the sister chromatid cohesion complex conveys clinical consequences distinct from Cornelia de Lange. *J Med Genet.* 2009;46(9):626-634.

22. Dhar SU, del Gaudio D, German JR, Peters SU, Ou Z, Bader PI, Berg JS, Blazo M, Brown CW, Graham BH, Grebe TA, **Lalani S**, Irons M, Sparagana S, Williams M, Phillips JA, 3rd, Beaudet AL, Stankiewicz P, Patel A, Cheung SW, Sahoo T. 22q13.3 deletion syndrome: clinical and molecular analysis using array CGH. *Am J Med Genet A.* 2010;152A(3):573-581.

23. Parri V, Katzaki E, Uliana V, Scionti F, Tita R, Artuso R, Longo I, Boschloo R, Vijzelaar R, Selicorni A, Brancati F, Dallapiccola B, Zelante L, Hamel CP, Sarda P, **Lalani SR**, Grasso R, Buoni S, Hayek J, Servais L, de Vries BB, Georgoudi N, Nakou S, Petersen MB, Mari F, Renieri A, Ariani F. High frequency of COH1 intragenic deletions and duplications detected by MLPA in patients with Cohen syndrome. *Eur J Hum Genet.* 2010;18(10):1133-1140.

24. Nagamani SC, Erez A, Shen J, Li C, Roeder E, Cox S, Karaviti L, Pearson M, Kang SH, Sahoo T, **Lalani SR**, Stankiewicz P, Sutton VR, Cheung SW. Clinical

spectrum associated with recurrent genomic rearrangements in chromosome 17q12. *Eur J Hum Genet.* 2010;18(3):278-284.

25. Szafranski P, Schaaf CP, Person RE, Gibson IB, Xia Z, Mahadevan S, Wiszniewska J, Bacino CA, **Lalani S**, Potocki L, Kang SH, Patel A, Cheung SW, Probst FJ, Graham BH, Shinawi M, Beaudet AL, Stankiewicz P. Structures and molecular mechanisms for common 15q13.3 microduplications involving CHRNA7: benign or pathological? *Hum Mutat.* 2010;31(7):840-850.

26. Boone PM, Bacino CA, Shaw CA, Eng PA, Hixson PM, Pursley AN, Kang SH, Yang Y, Wiszniewska J, Nowakowska BA, del Gaudio D, Xia Z, Simpson-Patel G, Immken LL, Gibson JB, Tsai AC, Bowers JA, Reimschisel TE, Schaaf CP, Potocki L, Scaglia F, Gambin T, Sykulski M, Bartnik M, Derwinska K, Wisniowiecka-Kowalnik B, **Lalani SR**, Probst FJ, Bi W, Beaudet AL, Patel A, Lupski JR, Cheung SW, Stankiewicz P. Detection of clinically relevant exonic copy-number changes by array CGH. *Hum Mutat.* 2010;31(12):1326-1342.

27. Sanchez-Valle A, Wang X, Potocki L, Xia Z, Kang SH, Carlin ME, Michel D, Williams P, Cabrera-Meza G, Brundage EK, Eifert AL, Stankiewicz P, Cheung SW, **Lalani SR**. HERV-mediated genomic rearrangement of EYA1 in an individual with branchio-oto-renal syndrome. *Am J Med Genet A.* 2010;152A(11):2854-2860.

28. Nagamani SC, Erez A, Bader P, **Lalani SR**, Scott DA, Scaglia F, Plon SE, Tsai CH, Reimschisel T, Roeder E, Malphrus AD, Eng PA, Hixson PM, Kang SH, Stankiewicz P, Patel A, Cheung SW. Phenotypic manifestations of copy number variation in chromosome 16p13.11. *Eur J Hum Genet.* 2011;19(3):280-286.

29. Campbell IM, Kolodziejska KE, Quach MM, Wolf VL, Cheung SW, **Lalani SR**, Ramocki MB, Stankiewicz P. TGFBR2 deletion in a 20-month-old female with developmental delay and microcephaly. *Am J Med Genet A.* 2011;155A(6):1442-1447.

30. Tariq M, Belmont JW, **Lalani S**, Smolarek T, Ware SM. SHROOM3 is a novel candidate for heterotaxy identified by whole exome sequencing. *Genome Biol.* 2011;12(9):R91.

31. Liu P, Erez A, Nagamani SC, Dhar SU, Kolodziejska KE, Dharmadhikari AV, Cooper ML, Wiszniewska J, Zhang F, Withers MA, Bacino CA, Campos-Acevedo LD, Delgado MR, Freedenberg D, Garnica A, Grebe TA, Hernandez-Almaguer D, Immken L, **Lalani SR**, McLean SD, Northrup H, Scaglia F, Strathearn L, Trapane P, Kang SH, Patel A, Cheung SW, Hastings PJ, Stankiewicz P, Lupski JR, Bi W. Chromosome catastrophes involve replication mechanisms generating complex genomic rearrangements. *Cell.* 2011;146(6):889-903.

32. Nagamani SC, Erez A, Bay C, Pettigrew A, **Lalani SR**, Herman K, Graham BH, Nowaczyk MJ, Proud M, Craigen WJ, Hopkins B, Kozel B, Plunkett K, Hixson P, Stankiewicz P, Patel A, Cheung SW. Delineation of a deletion region critical for corpus callosal abnormalities in chromosome 1q43-q44. *Eur J Hum Genet.* 2012;20(2):176-179.

33. Andrés Hernández-García, Erwin Brosens, Hitisha P. Zaveri, Elisabeth M. de Jong, Zhiyin Yu, Maria Namwanje, Allison Mayle, Caraciolo J. Fernandes, Brendan Lee, Maria Blazo, **Seema R. Lalani**, Dick Tibboel, Annelies de Klein, Daryl A. Scott. Contribution of *LPP* Copy Number and Sequence Changes to Esophageal Atresia, Tracheoesophageal Fistula, and VACTERL Association. *In Press, Am J Med Genet*

34. **Lalani SR**, Shaw C, Wang X, Patel A, Patterson LW, Kolodziejska K, Szafranski P, Ou Z, Tian Q, Kang SH, Jinnah A, Ali S, Malik A, Hixson P, Potocki L, Lupski JR, Stankiewicz P, Bacino CA, Dawson B, Beaudet AL, Boricha FM, Whittaker R, Li C, Ware SM, Cheung SW, Penny DJ, Jefferies JL, Belmont JW. Rare DNA Copy Number Variants in Cardiovascular Malformations with Extracardiac abnormalities. *Eur J Hum Genet.* 2012 Aug 29. doi: 10.1038/ejhg.2012.155

35. Bi W, Borgan C, Pursley AN, Hixson P, Shaw CA, Bacino CA, **Lalani SR**, Patel A, Stankiewicz P, Lupski JR, Beaudet AL, Cheung SW. Comparison of chromosome analysis and chromosomal microarray analysis: what is the value of chromosome analysis in today's genomic array era? *Genet Med*. 2012 Dec 13. doi: 10.1038/gim.2012.152

36. Wiszniewska J, Bi W, Shaw C, Stankiewicz P, Kang SH, Pursley AN, **Lalani S**, Hixson P, Gambin T, Tsai CH, Bock HG, Descartes M, Probst FJ, Scaglia F, Beaudet AL, Lupski JR, Eng C, Wai Cheung S, Bacino C, Patel A. Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. *Eur J Hum Genet*. 2013 May 22. doi: 10.1038/ejhg.2013.77

37. Boone PM, Campbell IM, Baggett BC, Soens ZT, Rao MM, Hixson PM, Patel A, Bi W, Cheung SW, **Lalani SR**, Beaudet AL, Stankiewicz P, Shaw CA, Lupski JR. Deletions of recessive disease genes: CNV contribution to carrier states and disease-causing alleles. *Genome Res.* 2013 May 16.

38. Dittwald P, Gambin T, Szafranski P, Li J, Amato S, Divon MY, Rodriguez Rojas LX, Elton LE, Scott DA, Schaaf CP, Torres-Martinez W, Stevens AK, Rosenfeld JA, Agadi S, Francis D, Kang SH, Breman A, **Lalani SR**, Bacino CA, Bi W, Milosavljevic A, Beaudet AL, Patel A, Lupski JR, Shaw CA, Gambin A, Cheung SW, Stankiewicz P. NAHR-mediated copy-number variants in a clinical population: mechanistic insights into both genomic disorders and Mendelizing traits. *Genome Res.* 2013 May 8.

39. **Lalani SR**, Ware SM, Wang X, Zapata G, Bray M, Chinault AC, Boggs BA, Scott DA, Brundage EK, Patel A, Fernbach SD, Huang M, Baker M, Hamilton SL, Towbin JA, McBride KL, Potocki L, Belmont JW: Role of MCTP2 in coarctation of the aorta. *Hum Mol Genet.* 2013 Jun 27. [Epub ahead of print]

40. Wojciech Wiszniewski, Jill V Hunter, Neil A Hanchard, Qi Tian, Xueqing Wang, Sau W Cheung, Ankita Patel, Pawel Stankiewicz, Ian Campbell, Patricia Hixson, Audrey R Ester, Lorraine Potocki, Gladys Zapata, Patricia P Hernandez, Melissa B Ramocki, Regie LP Santos-Cortez, Suzanne M Leal, Michele K. York, Monica Justice, Patricia Bader, Lisa Omo-Griffith, Nirupama S Madduri, Gunter Scharer, Heather P Crawford, Apiwat Mutirangura, Pattamawadee Yanatatsaneejit, Anna

Eifert, Jeffery Kerr, Matthew Hurles, Chad Shaw, Adiaha IA Franklin, Robin Goin-Kochel, Gayle Simpson, Ladonna Immken, Muhammad E Haque, Marija Stosic, Arthur L Beaudet, John W Belmont, Misti D Williams, Thomas M Morgan, Simeon Boyd, Emily Hall, Jill Mokry, Martin Lloyd Hibberd, Chiea Chean Khor, Mirjana Maletic-Savatic, NguyenVan Vink Chau, Lisa Shaffer, Sarah Dunstan, Cameron Simmons, Penelope Bonnen, James R Lupski, **Seema R Lalani**. *TM4SF20* deletion and susceptibility to a novel pediatric disorder of cerebral white matter hyperintensities and language delay. *Am J Hum Genet*. 2013 Jun 26. doi:pii: S0002-9297(13)00269-3. 10.1016/j.ajhg.2013.05.027.

41. Ian M. Campbell, Mitchell Rao, Sean D. Arredondo, **Seema R. Lalani**, Zhilian Xia, Sung-Hae L. Kang, Weimin Bi, Amy M. Breman, Janice L. Smith, Carlos A. Bacino, Arthur L. Beaudet, Ankita Patel, Sau Wai Cheung, James R. Lupski, Paweł Stankiewicz, Melissa B. Ramocki, and Chad A. Shaw. Fusion of Large-Scale Genomic Knowledge and Frequency Data Computationally Prioritizes Variants in Epilepsy. In press, *PLOS Genetics*

C. Research Support

Ongoing research support

MOD #6-FY13-167 6/01/13-5/31/16
March of Dimes
Genetic Registry for Pediatric Heart Disease: The CCVM Consortium
Role: Co-Investigator

Completed Research support

CSDA 20020297 7/01/2002-6/30/2007
Doris Duke Charitable Foundation
Genetic Etiology of CHARGE Syndrome
Role: PI

Junior Faculty Seed Funding Program 8/01/2007-7/31/2008
The Gillson Longenbaugh Foundation
Identifying gene copy-number alterations in cardiovascular malformations
Role: PI

RO1-HL091771 2/2008-1/2012
NIH
Novel Genomic Disorders Causing Cardiovascular Malformations
Role: Co-investigator

MOD #1-FY10-401 7/2010-6/2012
March of Dimes
Genetic causes of congenital heart defects
Role: Co-Investigator