

CURRICULUM VITAE

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Citizenship: New Zealand (Permanent Resident: United States; Eligible for Citizenship, 2007)

Current Titles and Affiliations:

Academic Appointment: Professor
Department of Human Genetics
Emory University School of Medicine
August 2013 to present

Professor
Department of Pediatrics (secondary appointment)
Emory University School of Medicine
November 2013 to present

Adjunct Associate Professor
The University of Texas M.D. Anderson Cancer Center
School of Health Sciences, Houston, TX
August 2004 to present

Clinical Appointment: Executive Director, Emory Genetics Laboratory
Emory University School of Medicine
Department of Human Genetics, Division of Medical Genetics
Emory Genetics Laboratory
December 2012 to present

Previous Academic and Professional Appointments:

1992 – 1995	Senior Research Fellow Bombay Hospital, University of Bombay, Bombay, India
1995 – 1996	Research Associate DNA Diagnostic Laboratory, Auckland Hospital, Auckland, New Zealand
1996- 2000	Scientific Officer DNA Diagnostic Laboratory, Auckland Hospital, Auckland, New Zealand
2000– 2003	Postdoctoral Fellow Department of Human Genetics, Baylor College of Medicine, Houston, TX
2003-2006	Assistant Professor Department of Human Genetics, Baylor College of Medicine, Houston, TX
2006-2009	Assistant Professor Department of Human Genetics, Emory University School of Medicine, Atlanta, GA

2009-2013	Associate Professor Department of Human Genetics, Emory University School of Medicine, Atlanta, GA
2013-	Professor Department of Human Genetics, Emory University School of Medicine, Atlanta, GA

Previous Administrative and/or Clinical Appointments:

1996- 2000	Scientific Director, DNA Diagnostic Laboratory, Auckland Hospital, Auckland, New Zealand
2001 – 2006	Assistant Laboratory Director, DNA Diagnostic Laboratory Department of Human Genetics, Baylor College of Medicine, Houston, TX
2006 – 2006	Co- Director, DNA Diagnostic Laboratory Department of Human Genetics, Baylor College of Medicine, Houston, TX
2006-2010	Senior Laboratory Director, Emory Genetics Laboratory, Atlanta, GA
2010-2012	Scientific Director, Emory Genetics Laboratory, Atlanta, GA

Licensures/Specialty Boards:

2005	Clinical Molecular Geneticist, American Board of Medical Genetics (renewed 2013)
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Education:

1987	B.S., Microbiology/Genetics, University of Bombay, Bombay, India
1992	M.Sc., Microbiology/Genetics, University of Bombay, Bombay, India, Advisor: Prof. M.Y. Kamat, Professor and Head, Department of Food Science and Technology, University Institute of Chemical Technology (UICT), University of Bombay, India
2000	Ph.D., Applied Science, University of Auckland, School of Biological Sciences, Auckland, New Zealand, Advisor: Prof. Richard Bellamy, Dean, Faculty of Science, The University of Auckland

Postdoctoral Training:

2000-2003	Department of Human Genetics, Baylor College of Medicine Mentor: Carolyn Sue Richards, Ph.D.
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Committee Memberships:

National and International:

AMP Economic Affairs Committee: NGS Pricing Project Oversight Committee (NPP)	2014-
Chair- Colorectal Cancer Laboratory Guidelines Committee	2008-
Next Generation Sequencing Guidelines committee	2011-
Sequence Interpretation Guidelines committee	2006-
American College of Medical Genetics: CAP Liaison (Molecular)	2013-
American College of Medical Genetics Representative to NIH R25 grant to ASCP	

"Training Residents in Genomics (TRIG)"	2013-
ACMG Task Force on Whole Genome Sequencing	2011-
CDC- Get RM Expert Panel member	2007-
College of American Pathologists (CAP) Next gen sequencing guidelines committee	2011-
CLSI- Document Development Committee on Microarrays for Cytogenetics and Oncology (MM21)	2011-
AMP 2012- 2013 Program Committee	2011-2013
Association of Molecular Pathology, Training and Education Committee	2006- 2009
Scientific Advisory Board- Microarray based assays Neuromuscular diseases- European Union	2008- 2011
American College of Medical Genetics, Quality Assurance Committee	2006-2011
Regional and State:	
Texas Cancer Genetics Network, Houston, TX	2003-2005
Institutional:	
Scientific Advisory Board, Molecular Genetic Technology Committee- M.D. Anderson Cancer Center, School of Health Sciences, Houston, TX	2005-
ACMG Postdoctoral Training Program Committee Division of Medical Genetics, Emory University	2006 –
Clinical Services Utilization Subcommittee Emory Medical Laboratories	2008–
Clinical Services Laboratory Subcommittee Baylor College of Medicine, Houston, TX	2003– 2006
Advisor	
Parent Project Muscular Dystrophy: Duchenne Connect, Registry	2008-
Cure CMD: Congenital Muscular Dystrophy Registry	2009-
Principal Investigator : MutaDataBase (Mutation database)	2010-
Jain Foundation Dysferlin Project- India	2010-
CDC- Next Gen Sequencing Expert meeting	2011-
Founder	
Organization of Rare Disorders (ORD), India	2013-
Consultancy	

Phoenix Children's Hospital	2014-
Ingenuity systems (Qiagen)	2013-
Tessarae	2012-
Oxford Gene Technologies	2011-
RainDance Technologies	2011-2014

Scientific Expert

Myriad VS GeneDx	2013-
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Grant Review:

Belgium Research Council	2013
Canada Cancer Organization	2013
Telethon	2013-
Duchenne Fund, Italy	2013
NHGRI- Genomic Medicine Pilot Demonstration Projects	2012-
Congressionally Directed Medical Research Program (CDMRP)	2012
Wellcome Trust, UK	2012
NIH (ZRG1 IMST-J (15) <i>Genes, Genomes, and Genetics</i> , SBIR/STTR review)	2010-2013
National Medical Research Council, Singapore	2011
Princess Beatrix Research Fund, Netherlands	2011
Medical Research Council, Australia	2010
Medical Research Council, New Zealand	2010

Editor:

Communicating editor- Human Mutation

Manuscript Reviewer:

American Journal of Human Genetics (2011- present)
 American Journal of Medical Genetics (2009; 2010)
 American Journal of Pathology (2011)
 Cancer Detection and Prevention (2011)
 Clinical Genetics (2009; 2011; 2012)
 European Journal of Human Genetics (2011;2012)
 Genetics in Medicine (2006- present)
 Human Mutation (2005- present)
 Human Molecular Genetics (2009, 2011)
 Journal of Clinical bioinformatics (2013)
 Journal of Medical Genetics (2010; 2011)
 Journal of Molecular Diagnostics (2006-present)
 Journal of Pediatrics (2012)
 Journal of Rare and Orphan Diseases (2012)
 Microarrays (2013)
 Nature Reviews Genetics (2012)
 Neuropsychiatry and Neurogenetics (2011)
 Neurogenetics (2011)
 Neuromuscular disorders (2011; 2012)
 Orphanet Journal of Rare Diseases (2011; 2012)
 PLoS Genetics (2011;2012)

PLoS One (2011; 2012; 2013-)
 Prenatal Diagnosis (2011)
 BMC Genetics (2011; 2012)

Honors and Awards:

G.D. Gokhale Trust Fund Fellowship for MSc	1988
Recipient of the Best article, Bombay Technologist (1991)	1992
Publication of University Institute of Chemical Technology, Mumbai, India	
Graduate Aptitude Test GATE/University Grant Commission (National Level) Scholarship for Ph.D	1993
Bronlund Trust Fund Award for Auckland Healthcare employees Auckland Hospital, Auckland, New Zealand	1996
University of Auckland Research Fund Award	1998
M.D. Anderson Cancer Center, Outstanding Faculty Award	2005
ACMGGF –Signature Genomics Laboratories Award	2008
Faculty recognition “Phi Beta Kappa”, Emory College	2011

Society Memberships:

American Society of Human Genetics	1995 –
American College of Medical Genetics, Fellow	2005–
Association for Molecular Pathology	2005–
Baylor College of Medicine, Cancer Center	2005- 2006
Human Genetics Society of Australasia	1998-
Winship Cancer Institute, Emory University	2007-

Leadership Positions:

American College of Medical Genetics, Chair, Colorectal Cancer Laboratory Guidelines	2006-
American College of Medical Genetics, Course Director and Faculty Short Course on Next Generation Sequencing in Clinical Practice (2012)	2011-
Association of Molecular Pathology, Leader, Trainee Exchange Program	2006-2009
Association of Molecular Pathology, Chair (Chair-Elect- 2010): Genetics Subdivision	2011-
Association of Molecular Pathology, Board of Directors	2011-
Nominated to American Board of Medical Genetics (ABMG), Board of Directors	2012
Nominated to American College of Medical Genetics (ACMG), Board of Directors	2012

Co-Chair, Organizing Committee, International conference on Genetics and Genomics
New Delhi, India

2013

Research Focus:

Muscular Dystrophy: Understanding the functional intricacy and the effect of mutation on the protein expression is critical to making the right choice of therapeutic approach for Congenital Muscular Dystrophy (CMD) and Limb Girdle Muscular Dystrophy (LGMD). We are using an integrated approach using next generation genotyping, sequencing and microarray tools, to identify new gene and create a comprehensive map of the CMD and LGMD muscle exome, transcriptome and proteome, which may aid in choosing the most appropriate and focused personalized therapeutic approach.

Newborn screening: Duchenne muscular dystrophy (DMD) is an inherited form of muscular dystrophy which occurs in about 1 out of every 3,600 male infants. In spite of the high frequency of disease, newborn screening (NBS) for DMD is not in practice except in a few countries worldwide. Though, creatine kinase (CK) level testing has been attempted as a NBS method, the high false positive rate and the need for follow-up re-testing have rendered the test unacceptable for NBS. Based on the promising reports from a recent study that introduced a two-tier screening system using dried blood spots from birthing process, we are validating and implementing a comprehensive and low cost next generation sequencing (NGS) approach to detect all types (deletions, duplications and point mutations) in the DMD gene.

Dysferlinopathies: DYSF is a large gene comprising 55 exons and spanning a genomic region of >150 kb. Although mutation detection provides the most definitive diagnosis, it does not provide information about protein expression. We are using a modified monocyte assay method to identify dysferlin deficient patients and compare the dysferlin expression data with the genotype data to define the correlation of protein expression and disease severity. This data will be useful in monitoring patients entering gene therapy trials for dysferlin. We have also implemented this strategy in India.

Clinical Focus:

I devote a significant portion of my time to directing Emory Genetics Laboratory (EGL) within the Division of Medical Genetics in the role of Executive Director. I have the responsibility for the overall operation and management of EGL as well as setting strategic goals to maintain EGL as one of the nation's leading academic genetic testing laboratories. Besides overseeing the administrative sections of EGL, I also oversee development and transition of new state of the art tests into clinical practice. The focus of my clinical research and development is to develop and perform comprehensive mutation analysis and interpretation for complex or challenging genetic disorders using multiple approaches. The primary focus of my clinical work is the development of high-throughput assays for rare disorders using next generation sequencing and microarrays for panels, exome and genomes. I also direct the bioinformatics efforts for variant interpretation and automation for streamlining testing pipelines.

Clinical Sequence Variant Classification and EMR integration: Current technology allows clinical laboratories to rapidly translate research discoveries from small patient cohorts into clinical genetic tests; therefore, a potentially large proportion of sequence variants identified in individuals with clinical features of a genetic disorder remain unpublished. Clinical laboratories willing to share this data face technological and practical barriers perpetuating a current problem: inconsistent interpretation of sequence variants from clinical laboratories working with different information. We have developed an in-house data management system, which is a highly-curated clinical grade variant database with a data structure designed to facilitate sharing of variants identified at [EGL](#). This system also tracks changes in variant classifications, generating notifications for the laboratory about which cases are in need of a review and possible report update. The second component, [EmVClass](#), is a web-based tool that allows any user to view and request a review of variants classified at [EGL](#). These software tools provide a solution to two pressing problems in clinical genetic testing: how to make sequence variants identified in a clinical laboratory freely available to the community and how to communicate changes in variant classification to healthcare providers.

Medical Exome: Next generation sequencing (NGS) technologies are increasingly gaining acceptance in clinical laboratories in the form of targeted gene panels and exome sequencing. These targeted tests are used extensively in the testing for Mendelian diseases with locus and allelic heterogeneity and cancer. While their moderate size allows for high coverage, technical sensitivity and the ability to generate data for every interrogated base, their clinical sensitivity is usually suboptimal. In contrast, exome sequencing offers enhanced detection rates but does not return

data for the entire target region, thereby limiting its clinical utility. To address these challenges, in collaboration with Dr. Birgit Funke (Harvard) and Dr. Avni Santani (CHOP) we are performing an in-depth and iterative curation of all genes that are currently known to be medically relevant. We are also developing a clinically validated enhanced exome assay where coverage of medically relevant genes is optimized. The assay design will be shared with the genetic testing community to promote standardization of medical sequencing. This information will be critical for clinical laboratories, physicians, and researchers that interpret data within the context of a patient's clinical findings and are rapidly adopting exome and genome wide sequencing, where access to curated knowledge is critical to analyze the large number of variants returned.

Grant Support:

Active Support:

Federal:

NIH ORD	06/11-05/13
Role: Co-PI	Effort: 0.2 calendar-years month(s)
Congenital disorders of glycosylation (PI: He)	\$128,000
Training Program in Human Disease Genetics. 1T32MH087977 (PI: Warren)	01/11-12/15
Role: Co-PI	Effort: 0.12 calendar-years month(s)
1 U41 HG006834-01 (PIs: Rehm/Martin/Nussbaum)	08/01/13 – 07/31/16
Role: Co-Investigator	Effort: 0.2 calendar-year month(s)
NIH/NHGRI	\$1,653,965
Development and Curation of a Universal Human Genomic Variant Database	
Hundreds of thousands of disease-causing variants have been identified in patients with disease, yet only a small fraction of that data, and the interpretation of it, is accessible to researchers and clinicians. This project will serve to collect and organize genomic data from many sources into a free and publically accessible environment and enable expert curation of that data for use in improving healthcare and biomedical research.	
1 UM1 (PIs: Berg)	08/01/13-07/31/17
Role: Co-investigator	Effort: 0.2 calendar-year
month(s)	
NIH/NHGRI	\$ 2,000,000

Clinically Relevant Variants Resource (CRVR)

The overarching goal of the Clinically Relevant Variants Resource (CRVR) is to generate a knowledge base of genes and variants for use in the clinical setting. During the first year of the project, we will establish taskforces for the assessment of genes and variants, and coordinate the establishment of the informatics infrastructure required to support the activities of the project and provide points of access to the information in CRVR. During years 2-4, we will convene clinical domain-specific working groups to carry out assessment activities, guided by members of the gene assessment and variant assessment taskforces. Ethical, legal, and social implications (ELSI) will be addressed by providing contextual assessments and exploring the positions of various stakeholders with regard to the education and reporting process for the information generated. Finally, recognizing that integration of genomic information with electronic health records (EHRs) is critical to the future of clinical genomics, we will initiate pilot studies in years 3-4 to ensure interoperability between the CRVR and EHRs.

Private Foundation Funded

MDA	07/01/12-06/30/15
Role: PI	Effort: 0.15 calendar-year month(s)
Identification of new genes for Congenital Muscular Dystrophy	\$390,000
MDA	01/01/13-01/01-14
Role: PI	Effort: 0.15 calendar-year month(s)
Newborn screening for Duchenne Muscular Dystrophy	\$88,000
Jain Foundation	02/12- 03/13
Role: PI	\$47,000
Monocyte assay for Dysferlinopathies	Effort: 0.1 calendar-years month(s)

Submitted

1U01HG007773-01 (PIs: Hegde)	01/01/14-02/28/17
Role: PI	Effort: 0.3 calendar-year month(s)
NIH/NHGRI	\$ 4,000,000
Medical Exome: From Concept to Implementation	

Over the past few year's tremendous technological advances have pushed massively parallel sequencing based technology from the research to the clinical realm and have enabled simultaneous testing of unprecedented numbers of genes. Along with this technical advancement, massive paradigm shifts have taken place and the field is moving away from the traditional one gene-one disorder concept. Many genes have been shown to be related to one disorder or one gene can cause many disorders. Gene panels, already in place in many diagnostic laboratories, are based on this concept and are derived from evidence- based review of the literature. Their main disadvantage is their static nature that cannot keep up with the rapid expansion of knowledge. Exome/Genome sequencing (ES/GS) is now feasible and beginning to be widely implemented in diagnostic laboratories despite limitations including incomplete coverage of medically significant genes. In addition, interrogating all genes is associated with significant challenges as to date <25% have been established to be associated with disease. In spite of this in many cases where a clinical phenotype is not clear exome or genome sequencing has proven to be useful. To narrow the large number of sequence variants obtained from ES/GS, in depth and up-to date curation of gene-disease associations is of critical importance but not available at a systematic level. We propose to develop a "medical exome" by developing an algorithm for continuous curation of genes and create a technically complete exome, which covers all regions of the known disease associated genes. Both resources will be made available to the clinical genetics community in order to catalyze standardization of clinical testing.

Previous support:

NINIDS RC1	09/09-09/12
Role: PI	\$1,000,000
Comprehensive methods for Neuromuscular disorders	Effort: 0.4 calendar-years month(s)
Jain Foundation	08/11- 01/12

Role :PI	\$20,000
Copy number variation in the Dysferlin Gene	Effort: 0.1 calendar-years month(s)
P.I., Muscular Dystrophy Association “ Infrastructure Grant for translation new technologies to NMD testing”	09/09 – 09/11
U01 DK56956 (Chapman) Co-investigator NIH/NIDDK The Early Collaborative Clinical Studies in PKD: The Consortium for Radiologic Imaging Studies of PKD (CRISP) Extended Cohort	2/00-12/10
P.I., NIH ORD- CETT Program” X-linked Mental Retardation”	02/09-06/10
P.I., Muscular Dystrophy Association “Microarray based Mutation detection in genes associated with Neuromuscular disorders”	09/08 – 09/09
P.I., Parent Project Muscular Dystrophy “Use of dried blood spots to detect mutation in Duchenne Muscular Dystrophy”, Sub Contract: Emory Genetics Laboratory	01/09 – 06/09
MM-1097-09/09 Center for Disease Control and Prevention (CDC) Association of American Medical Colleges (AAMC), Co-investigator Qualitative and quantitative study to determine the barriers to the early diagnosis of Duchenne and Becker Muscular Dystrophy (DBMD) (Andrew Faucett)	01/09-12/09
P.I., Vision of Children foundation “Genetic analysis of X-linked Ocular Albinism” P.I., Muscular Dystrophy Association 01/02-01/04 “Studies in detection of point mutations in the dystrophin gene”	01/02-01/03
P.I., Baylor Research to Clinical transition Fund (HNPPC) “Analysis of HNPPC carrier identification in an ethnically diverse population”	03-02-02/04
Co-investigator., “Gene Identification for Idiopathic Hyperphosphatasia” School of Medicine, University of Auckland, Auckland, New Zealand (Prof. Tim Cundy)	09/99-01/01

Equipment Grant:

ACTSI Proof of Principle Fund	2010
This fund is to support the rapid evaluation and translation of high-impact emerging technologies (Translational Technologies & Resources (TTR) program). These funds, along with support from the Georgia Research Alliance (GRA), are used to invest in new, promising technologies that are aligned with the ACTSI strategic goals.	
RainDance Technologies RDT 1000 instrument for multiplex PCR to enhance targeted sequencing capabilities in the Emory Genetics Laboratory.	
March Of Dimes (Georgia Branch)	2007
Qiagen Automated DNA extraction System	

Formal Teaching:**Medical Student Teaching:**

Medi 545: Human and Molecular Genetics Lecturer and Small Group Facilitator Emory University School of Medicine	2006 –
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Medical Residents Lecture Series (Pediatrics)	2010-
Graduate Program:	
Clinical Pathology Conference for Residents and Fellows Lecturer, Department of Genetics, Baylor College of Medicine	2002 – 2006
Molecular Genetics Technology Program Lecturer, The University of Texas M.D. Anderson Cancer Center School of Health Sciences	2004-2006
Undergraduate Teaching:	
Undergraduate Genetics Class Georgia Institute of Technology	November 2011
Other:	
ACMG Review course	2013
Course Director and Faculty American College of Medical Genetics, Next Generation sequencing	2012
Course Faculty International Society of Prenatal Diagnosis, Next Generation sequencing	2012
Course Director and Faculty Cambridge Health Institute (CHI), Next Generation sequencing	2012
Composite letter writer, Emory College	2011-
Microbiology Laboratory, Lecturer S.N.D.T Women's University, Bombay, India	1992 – 1993
Supervisory Teaching:	
Post-doctoral Fellows Directly Supervised:	
Alice Tanner Medical Genetics Fellow, ABMG Training program	2011-
Melanie Jones Medical Genetics Fellow, ABMG Training program Research	2010-
Eli Williams Medical Genetics Fellow, ABMG Training program	2012-
Arunkanth Ankala Research	2010-
Heather Mason-Suares Medical Genetics Fellow, ABMG Training program	2012-
Vanessa Horner Medical Genetic Fellow, ABMG Training program	2012-
Past:	

Alexander Valencia Ph.D. Medical Genetics Fellow, ABMG Training program	2009-2012
Victor Zhang, PhD. Medical Genetics Fellow, ABMG Training program, Emory University	2009-2011
Cindy McClomskey M. D Molecular Pathology Fellow, Molecular Pathology, Training program)	2009-2010
Hussian Askree M.D., Ph.D. Medical Genetics Fellow, ABMG Training program, Emory University	2009-2011
Kavita Gokhale Ph.D. (Research)	2008-2010
Marwan Tayeh, PhD. Medical Genetics Fellow, ABMG Training program, Emory University	2007-2010
David Okou, PhD. Medical Genetics Fellow, ABMG Training program, Emory University	2009
Christine Schmotzer M. D Molecular Pathology Fellow, Molecular Pathology, Training program	2009
Bipin Kulkarni, PhD (Research)	2007-2008
Dmitriy Niyazov, M.D. (Medical Genetics Fellow, ABMG Training program)	2006-2008
Arshad Ahsanuddin M.D. Molecular Pathology Fellow, Molecular Pathology, Training program	2007
Justin Cole Molecular Pathology Fellow, Molecular Pathology, Training program	2008
Monica Basehore	2005-2006
Molecular Genetics Technology Program, M.D. Anderson Cancer Center	
Joanne Tran Molecular Technology Internship Baylor College of Medicine	2004
Denise Juroske Molecular Technology Internship Baylor College of Medicine	2004
Treva Badger Molecular Technology Internship Baylor College of Medicine	2004
Adam Casillas Molecular Technology Internship Baylor College of Medicine	2005
Lindsay Meyers Molecular Technology Internship Baylor College of Medicine	2005

Noor Salman Molecular Technology Internship Baylor College of Medicine	2006
Justin Otchere Molecular Technology Internship Baylor College of Medicine	2006
Crystal Simien Molecular Technology Internship Emory University	2008
Tam Tran Molecular Technology Internship Emory University	2008
Tran Ngyuen Trainee- Laboratory operations Baylor College of Medicine, Houston, TX	2007
Paul Keller Scientific Internship Program Senior, Clements High School, Sugarland, TX	2005
Param Karandikar Scientific Internship Program Westford Academy, Westford, Massachusetts	2012

Lectureships, Seminar Invitations, Conference presentations : 2008 - current

Greenwood Genetics Laboratory." High-throughput DNA testing" Greenwood, SC	February 2008
Annual meeting of American College of Medical Genetics. Application of Microarray Genomic Selection to DNA testing. Phoenix, Arizona 2008 *Recipient of ACMGGF –Signature Genomics Laboratories travel award"	March 2008
Association of Molecular Pathology- American Association of Clinical Chemistry Molecular Pathology Course faculty, Baltimore, MD	May 2008
Corporate Symposia. AMP 2008. Roche. CGH array application to detect deletions and duplications in Disease associated genes", Dallas, TX	November 2008
Genetic Counselor Education meeting, Emory Genetics Laboratory "Use of array technology in diagnostics"	June 2008
Association of Public health laboratories, "Newborn screening- What to order and when" San Antonio, TX	July 2008
Association of Molecular Pathology- Outreach Course Dallas, TX	November 2008
DMD workshop Naarden- DMD test guidelines work shop- "New methods for mutations detection in DMD", Amsterdam, Netherlands	November 2008
Blood Center of Wisconsin, "High throughput DNA testing" Milwaukee, Wisconsin	November 2008
Foundation of Genetic Technologist, "High throughput Mutations detection using microarrays", Atlanta, GA	November 2008

American College of Medical Genetics, Annual meeting "XLMR: Application of High throughput technologies", Orlando, FL	March 2009
Parent Project Muscular Dystrophy "The first Asian Conference on Duchenne Muscular Dystrophy", Philadelphia	March 2009
Association of Genetic Technologists "Importance of comprehensive mutation detection in a clinical laboratory", Jacksonville, FL	June 2009
American Society of Human Genetics "Mutations in the DMXL1 cause a Prader Willi like syndrome", Hawaii	October 2009
Association of Molecular Pathology- Outreach course Association of Molecular Pathology- Annual meeting "Clinical Utility of microarrays", Dallas, TX	November 2009
ABI European User meeting "Clinical application of next generation sequencing", Antwerp, Belgium	February 2010
American College of Medical Genetics, Annual meeting "Deep clinical next generation sequencing", Phoenix, AZ "Emory Exhibit Theater" "OGT exhibit Theater"	March 2010
Human Genome Variation Society "Gene nomenclature issues in a clinical laboratory setting", Paris, France	May 2010
Association of Molecular Pathology course in Pathology "Microarrays in clinical diagnostics", Orlando, FL	May 2010
Hunter Hope Foundation, Annual Meeting Application of microarray to mutation detection in the GALC gene, Buffalo, NY	July 2010
Asuragen- DNA Diagnostics, Austin, TX	July 2010
Southeast Regional Genetics Group NBS Lab Performance group- "New methods in DNA Diagnostics", Ponte Vedra Beach, FL	July 2010
Association of Molecular Pathology "Translation next generation sequencing to clinical laboratory", Dallas, TX	November 2010
Neurology Symposium, Bombay Hospital "Recent advances in clinical molecular diagnostics for Neuromuscular disorders", Bombay, India	December 2010
SRL laboratories- Mumbai, India "Microarrays in research and clinical diagnostics"	December 2010
CHI Tri Molecular Medicine Conference "The true value of next generation sequencing in a clinical setting", San Francisco, CA	February 2011
American College of Medicine Genetics, Vancouver, 2011 "Emory Exhibit Theater" "OGT exhibit Theater"	March 2011
Oregon Health and Science University, Department Seminar "Next generation sequencing for Gene Discovery and Clinical diagnostics", Portland, OR	April 2011
Gen-Probe Inc "Translation next generation sequencing to a clinical laboratory", San Deigo, CA	May 2011

FDA- Ultra High Throughput Sequencing for Clinical Diagnostic Applications Public Meeting. Representing Association of Molecular Pathology, Washington DC	June 2011
Genetic Alliance Annual Meeting "Accelerating adoption of medical genetics Innovations", Washington DC	June 2011
Institute of Medicine of the National Academies "Roundtable on Translating Genomic-Based Research for Health", Washington DC	July 2011
Southeast Regional Genetics Group "Next generation sequencing in clinical diagnostics"- Mol/Cyto Breakout session, Asheville, NC	July 2011
AACC/ ASCLS Joint Annual Meeting " Next Generation sequencing and microarray in clinical genetics", Atlanta, GA	July 2011
8 th International Congress of Pharmaceutical Sciences- Brazil "New approaches to DNA diagnostics: Microarrays and Next generation sequencing", Rebeirao Preto, Brazil	August 2011
Albert Einstein College of Medicine and Montefiore Medical Center, New York " Implementation of next gen sequencing in a clinical setting", New York	September 2011
NMD-Chip European Union Scientific Meeting "The true value of next gen sequencing in a clinical setting", University College, London	September 2011
INSERM, " New gene discovery for congenital disorders of glycosylation", Marseille, France	September 2011
FAOBMB, "NGS for novel gene discovery for rare diseases/Mendelian disorders", Singapore	October 2011
Plenary Session "Emerging techniques and approaches for clinical diagnostics"	
ICHG (ASHG) Postdoc- Melanie Jones " Mutations in the oligosaccharyltransferase complex cause CDG type I", Montreal, Canada	October 2011
Affymetrix, "Carrier screening for an expanded Ashkenazi Jewish panel and 90 recessive diseases using a targeted resequencing array", San Francisco	October 2011
Life Technologies, "The True value of Next gen sequencing in a clinical setting", San Francisco	October 2011
Hong Kong Sanatorium and Hospital, " Clinical Next generation Sequencing: from sequencing panel to ex/ome", Hong Kong	November 2011
TREAT-NMD Annual Curator Meeting "Chips and Next generation sequencing in diagnosis of NMDs", Geneva, Switzerland	November 2011
Association of Molecular Pathology (AMP) " Complimentary assays in interpretation of Next generation sequencing", Dallas	November 2011
Children's Hospital of Atlanta, Genetics Research in Pediatrics "Clinical Utility of Next Generation Sequencing", Atlanta	December 2011
International Symposium on Molecular Pathology "Advanced technologies in Molecular Pathology", New Delhi, India	January 2012
Tata Memorial Hospital "Basics of NextGen sequencing and application in Clinical Practice", Bombay	January 2012

CHI Tri Molecular Medicine Conference "Carrier Detection Using Digital PCR and Resequencing Array", San Francisco	February 2012
United States and Canadian Academy of Pathology (USCAP) "Laying the Groundwork for Personalized Genomic Studies", Vancouver	March 2012
UK Neuromuscular Translational Research Conference "Advancing from targeted resequencing to whole exome sequencing: a perspective on neuromuscular disorders", New Castle, UK	March 2012
American College of Medical Genetics, Annual meeting "Short course on Next Generation Sequencing" "Next Generation Sequencing Applications from Gene Panels, Exome and Whole Genome Sequencing: Technologies and Target Selection", Charlotte, NC	March 2012
American Cytogenetics Conference- Affymetrix Workshop "Monogenic disorders: Carrier detection using digital PCR and resequencing array", Austin, TX	April 2012
International Society of Prenatal Diagnosis "Implementation of next generation sequencing in the clinic", Miami, FL	June 2012
CHI Innovative Tissue based Diagnostics "A course in next generation sequencing" "Tissue based diagnostics using next generation sequencing", Philadelphia	June 2012
The 5 th Annual workshop on Krabbe disease "ArrayCGH improves detection of mutations in the GALC gene in Krabbe disease", Buffalo, New York	July 2012
Baylor College of Medicine, Department of Human Genetics "Laying the groundwork for personalized genomic medicine", Houston, TX	August 2012
Mutation detection workshop Australasia "Implementation of NGS in clinical laboratories", Brisbane, Australia	September 2012
Prince of Wales Hospital "Laying the groundwork for personalized genomic medicine", Sydney, Australia	October 2012
Royal Children's hospital "Implementation of NGS in clinical laboratories", Melbourne, Australia	October 2012
World muscle society "Gene by gene sequencing to whole genome analysis: A perspective on Neuromuscular disorders", Perth, Australia	October 2012
Illumina corporate workshop, AMP "From gene panels to exome in the clinic", Long Beach, CA	November 2012
Roche corporate workshop, AMP "Exome sequencing in the clinic", Long Beach, CA	November 2012
AMP analyst day, AMP "Advances in the sequencing technologies", Long Beach, CA	November 2012
ASHG, 2012 "Addressing the fundamentals: NGS validation and implementation in a clinical setting", San Francisco	November 2012
Association of Maharashtra Obstetrics and Gynaecological Societies "Why my DNA is important: A primer for ob/gyn" "Case studies in genetics", Kolhapur, India	December 2012
International conference on Next Revolution in Genetics and Genomics	January 2013

"NGS technology basics"	
"NGS in neuromuscular disorders"	
"Laying the groundwork for personalized genomic medicine", New Delhi, India	
CHI Molecular Medicine conference, San Francisco	February 2013
" From gene panels to exomes in cancer"	
Oxford Gene Technologies, Oxford, UK	
" Emory Genetics Laboratory"	February 2013
From gene panels to exome sequencing	March 2013
ACMG, EGL exhibit theatre	
Exome sequencing in a clinical setting	April 2013
UMDNJ symposium	
NGS in Neuromuscular disorders	April 2013
International meeting on Glycosylation, Carolinas Medical Center, Charlotte	
CPT codes: The Basics	April 2013
ABMG Fellows meeting , Emory University	
Exome sequencing in a clinical setting	May 2013
CHI Biomarkers and Diagnostics world congress, Philadelphia	
From single genes to panels to exomes in a clinical setting	May 2013
UT Southwestern, McDermott Center for Human Growth and Development	
Targeted gene panels and exome sequencing	July 2013
InVita, San Francisco, CA	
A CLIA based approach to Cancer testing using NGS	August 2013
CHI Biomarkers and NGS meeting, Washington DC	
A Laboratory Directors perspective to NGS	September 2013
European Quality Network (EQMN), Dublin, Ireland	
Winship Annual Retreat	September 2013
Emory Conference Center, Atlanta	
Exome and Genome Sequencing	September 2013
CHiDren's NIH network, Boston	
Medical Exome Arrays	
ASHG Oxford Gene Technologies	October 2013
Boston	
Incidental Findings	
AMP Annual Meeting,	November 2013
Phoenix	
Ingenuity workshop	
AMP Annual Meeting,	November 2013
Phoenix	
Basics of NGS 101	
Cambridge Health Institute	February 2014
San Francisco	
Interpretation of sequence variants: Short course	
ClinVar: Molecular Genetics platform	
EGL: Exhibit Theatre	

OGT: Exhibit Theatre American College of Medical Genetics Nashville	March 2014
AMP webinar: Interpretation of Sequence variants	April 2014
American Scientist webinar: Personalized Genomic Medicine	May 2014

Other Activities:

Faculty Service and Development:

Faculty Development course participant Emory University School of Medicine	2006
AAMC Mid Career Women Professional Development Seminar	2011
Member, Search Committee- Director, Division of Medical Genetics	2010-2011

Guidelines Review:

CDC EGAPP Guidelines for testing for Colorectal cancer	2008
TREAT-NMD Guidelines for DMD, Montpellier, France	2008
TREAT-NMD Guidelines for DMD, Atlanta	2008
KRAS mutation analysis for CRC Comments to Wellstone proposal	2009
AMA CPT codes committee	2013

CAP Laboratory Inspection:

College of American Pathologists - Lead Inspector

Spectrum Health Laboratories Grand Rapids, Michigan	2009
University of Nebraska Health Center , Omaha, NE	2009
Wake Forest University Laboratory, Wake Forest	2010
Hematologics, Seattle, WA	2012
Broad Institute, Boston, MA	2013

Community Service:

Muscular Dystrophy Association Annual Fundraiser Walk	2002-2003
Parent Project Muscular Dystrophy Clinical consultant- DuchenneConnect	2007-

CURECMD- CMDIR	2009-
CYSTIC FIBROSIS ANNUAL FUNDRAISER WALK	2012

Featured articles:

Genome Technology	2008
CAP Today (Official Magazine of College of American Pathologists)	2011

Patents submitted:

Genetic analysis of Seladin-1, Galaktionov K, Hegde MR, June 2005

GLOBE analysis program for sequence based testing, November 2006

A two step high resolution methodology for dystrophin testing, November 2006

Intellectual Know-How licenses

Oxford Gene Technologies- Targeted Gene Arrays

Tessarae- Pan Ethnic Carrier Screening Assay

Agilent- Medical Exome

Published and Accepted Research Articles:

1. Pishawikar M.S., Singhal R., Kulkarni P.R., Rapid Non-Microbiological Methods for enumeration of bacteria in foods, Trends in Foods Science and Technology, (1992), 165-169.
2. Pishawikar M.S., Biosurfactants: An alternative to Chemically synthesized surfactants, Bombay Technologists Vol.48 (1990-91), 163-67.
3. Pishawikar M.S., Kamat M.Y., Microbiological Quality Assurance in pharmaceuticals, cosmetics & toiletries, Ind. J. Pharm Sci, 14 (1992), 27-32.
4. Williams LC, Hegde MR, Herrera G, Stapleton PM and Love DR. Comparative semi-automated analysis of (CAG) repeats in the Huntington disease gene: use of internal standards. Mol Cellular Probes.1999, 13(4): 283-289.
5. Hegde MR, Williams LC, Nagappan R, Bullock J, Faull RLM, Winship I, S Snow K and Love DR Analysis of non-amplifying (CAG)_n alleles at the Huntington Disease locus. Genetic Testing. 2000, 4(1): 55-60.
6. Cardon LR, Garner C, Bennett ST, Mackay IJ, Edwards RM, Cornish J, Hegde M, Murray MAF, Reid IR, Cundy T. Evidence for a major gene for bone mineral density in idiopathic osteoporotic families. American Journal Bone and Mineral Research. 2000, 15 (6): 1132-1136.
7. Hegde MR, Chong B, Fawcner MJ, Leary J, Shelling AN, Culling B, Winship I, Love DR. Hierarchical mutation screening protocol for the BRCA1 gene. Hum Mutat. (2000) Nov; 16(5): 422-30.
8. Hegde MR, Fawcner M, Chong B, McGaughan J, Gilbert D and Love DR Compound heterozygosity at the FMR1 gene. Genetic Testing (2001), 5(2): 135-138.

9. Hegde MR, Chong B, Fawkner M, Lambiris H, Kenneson A, Warren ST, Love DR and McGaughran J. Microdeletion in the FMR-1 gene: an apparent null allele using routine clinical PCR amplification. *J Med Genet* (2001), 38(9): 624-629.
10. Hegde MR, Chong B, Stevenson C, Laing NG, Khadilkar S, Love DR. Clinical & genetic analysis of four patients with distal upper limb spinal muscular atrophy. *Indian J Med Res* (2001). Oct; 114:141-7.
11. Wu G, Wu W, Hegde M, Fawkner M, Chong B, Love D, Su L-K, Lynch P, Snow K, Richards CS. Detection of sequence variations in the Adenomatous Polyposis Coli (APC) gene using denaturing high-performance liquid chromatography (dHPLC). *Genet Test*. (2001) Winter; 5(4): 281-90.
12. Hegde M, Lewis RA, Richards CS. Diagnostic testing for X-linked ocular albinism (X-OA1) using a hierarchical mutation screening protocol. *Genet Test*. (2002). Vol 6 no 1, 7-13.
13. Cundy T*, Hegde M*, Noat D, Chong B, , Wallace R, Mulley J, Love DR, Seidel J, Fawkner M, Banovic T, Callon K, Grey A, Middleton C, Reid I, Cornish J . A Mutation in the gene TNFRSF11B encoding osteoprotegerin cause idiopathic hyperphosphatasia phenotype. *Hum Mol Gen* (2002), Vol 11 (18) 1-9. (*equal authorship)
14. B.Chong, M.Hegde, M.Fawkner, C.Middleton, J.Seidel, B.Tüysüz, Bilgin, M.Coker, C.Tau, H.Cassinelli, D.Love, T.Cundy. Idiopathic hyperphosphatasia and TNFRSF11B mutations: relationships between phenotype and genotype. *J Bone Miner Res*. 2003 Dec; 18(12): 2095-104.
15. Hegde M, Chong B, Chin E, Khadilkar S, Hutchinson D, Richards CS, Love D. Limb girdle muscular dystrophy: use of dHPLC and direct sequencing to detect sarcoglycan gene mutations in a New Zealand cohort. . *Clin Genet*. 2004: 65: 55–60
16. Skinner J, Chong B, Fawkner M, Webster D, Hegde M. Use of the newborn screening card to define cause of death in a 12 year old diagnosed with epilepsy. *J. Paediatr. Child Health*. (2004) 40, 651–653
17. Roberto Mendoza-Londono, Edward Lammer , Rosemarie Watson , John Harper, Atsushi Hatamochi , Saori Hatamochi , Dobrawa Napierala , Pia Hermanns , Sinead Collins , Benjamin B. Roa , Madhuri R. Hedge , Keiko Wakui , Diep Nguyen , David W. Stockton , Brendan Lee. Characterization of a new syndrome associating craniosynostosis, delayed fontanel closure, parietal foramina, imperforate anus and skin eruption- CDAGS. *Am J Hum Genet*. 2005 Jul; 77(1): 161-8. Epub 2005 May 27.
18. Hegde MR, Chong B, Blazo ME, Chin LH, Ward PA, Chintagumpala MM, Kim JY, Plon SE, Richards CS. A homozygous mutation in MSH6 causes Turcot syndrome. *Clin Cancer Res*. 2005 Jul 1; 11(13): 4689-93.
19. Hegde M, Blazo M, Chong B, Prior T, Richards C. Assay validation for identification of hereditary nonpolyposis colon cancer-causing mutations in mismatch repair genes MLH1, MSH2, and MSH6. *J Mol Diagnostics*. 2005 Oct; 7(4): 525-34.
20. Ionescu DN, Papachristou G, Schoen RE, Hedge M, Richards CS, Monzon FA. Attenuated familial adenomatous polyposis: a case report with mixed features and review of genotype-phenotype correlation. *Arch Pathol Lab Med*. 2005 Nov; 129 (11): 1401-4.
21. Khadilkar SV, Menezes KM, Singh RK, Hegde MR. Severe childhood autosomal recessive muscular dystrophy, mental subnormality and chorea. *Neurol India*. 2006 Sep; 54(3):293-5.
22. Noah A. Rosenberg, Saurabh Mahajan, Catalina Gonzales, Laura Nino-Rosales, Vasiliki Ninis, Parimal Das, Madhuri Hegde, Laura Molinari, Gladys Zapata, James L. Weber, John W. Belmont and Pragna I. Patel. Low levels of genetic differentiation across Indian populations. *PLOs Genetics*. December 2006. Volume 2 (12): e215
23. A Phase 1, Pharmacokinetic and Pharmacodynamic Study of Temozolomide in Pediatric Patients with Refractory/Recurrent Leukemia: A Children's Oncology Group Study. Terzah M. Horton, Patrick A. Thompson, Stacey L. Berg, Peter C. Adamson, Ashish M. Ingle, M. Eileen Dolan, Shannon M. Delaney, Madhuri Hedge, Heidi L. Weiss, Meng-Fen Wu, And Susan M. Blaney. *J Clin Oncol*. 2007 Nov 1; 25(31): 4922-8.
24. Richards CS, Bale S, Bellissimo DB, Das S, Grody WW, Hegde MR, Lyon E, Ward BE; Molecular Subcommittee of the ACMG Laboratory Quality Assurance Committee. ACMG recommendations for standards for interpretation and reporting of sequence variations: Revisions 2007. *Genet in Med*. 2008 Apr; 10(4): 294-300.

25. Plon, S.E., Pirics, M.L., Agrawal, S., Zbuk, K., Nuchtern, J., Hicks, J., Hegde, M., Lip-Hon Chin, E., Eng, C., and H. Russell (2008). Multiple Tumors in a Child with Germline Mutations in TP53 and PTEN. *New England J of Medicine*, 2008 Jul 31; 359(5):537-9..
26. Madhuri R. Hegde, ELH Chin, Jennifer G. Mulle, David T. Okou, Stephen T. Warren and Michael E. Zwick. Microarray-based comprehensive mutation detection in the dystrophin gene. *Human Mutation*. 2008 Sep; 29(9): 1091-9.
27. Marwan K. Tayeh, Lip-Hon Chin, Vanessa R. Miller, Lora JH. Bean, Bradford Coffee, Madhuri Hegde. Microarray based testing for single and multi- exon deletion or duplication mutations. *Genet in Med*. 2009 Apr; 11(4):232-40.
28. Terzah M. Horton, Gaye Jenkins, Linna Zhang, M. Eileen Dolan, Albert Ribes-Zamora, Susan M. Blaney, Madhuri Hegde, Alison A. Bertuch, and Stacey L Berg. PARP Inhibitor ABT-888 potentiates the in vitro Anti-Tumor Activity of Temozolomide in Leukemia Cells: Influence of Mismatch Repair Status and MGMT activity. *J Clin Oncol. Molecular Cancer Therapy*. 2009 Aug 8(8): 2232-42 Epub, Aug 11
29. Khadilkar SV, Singh RK, Urtizberea A, Love DR, Chong B, Hegde M. Spectrum of mutations in sarcoglycan genes in the Mumbai region of western India: high prevalence of 525delT. *Neurol India*. 2009 Jul-Aug; 57(4): 406-10.
30. Mulle JG, Patel VC, Warren ST, Hedge MR, Cutler DJ, Zwick ME, Empirical evaluation of probe selection for DNA microarrays. *PLoS One*. 2010 Mar 29; 5(3): e9921.
31. Hjelm LN, Chin EL, Hegde MR, Coffee BW, Bean LJ. A simple method to confirm and size deletion, duplication, and insertion mutations detected by sequence analysis. *J Mol Diagnostics*. 2010 Sep; 12(5): 607-10. Epub 2010 Jul 15.
32. Hussain Askree S, Hjelm LN, Ali Pervaiz M, Adam M, Bean LJ, Hegde M, Coffee B. Allelic dropout can cause false-positive results for prader-willi and angelman syndrome testing. *J Mol Diagnostics*. 2011 Jan; 13(1): 108-12.
33. Phenylbutyrate therapy for maple syrup urine disease. Brunetti-Pierri N, Lanpher B, Erez A, Ananieva EA, Islam M, Marini JC, Sun Q, Yu C, Hegde M, Li J, Wynn RM, Chuang DT, Hutson S, Lee B. *Hum Mol Genet*. 2011. 20(4): 631-640
34. Patrick Yu-Wai-Man, Suma P. Shankar, Valerie Biousse, Neil R. Miller, Lora J.H. Bean, Bradford Coffee, Madhuri Hegde, Nancy J. Newman. Genetic Screening for OPA1 and OPA3 Mutations in Patients with Suspected. Inherited Optic Neuropathies. *Ophthalmology*. 2011 Mar; 118(3): 558-63.
35. Pervaiz Mohammed, He Miao, Hegde Madhuri and Singh Rani. MCT oil-based diet reverses Hypertrophic Cardiomyopathy in a Patient with Very Long-Chain Acyl-CoA Dehydrogenase Deficiency. *Indian Journal of Human Genetics*. 2011, Jan-April, Vol 17 (1): 24-27
36. Targeted PCR-based enrichment and next generation sequencing for diagnostic testing of congenital disorders of glycosylation (CDG). Melanie Jones, Shruti Bhide; Ephrem Chin; Bobby Ng; Devin Rhodenizer; Victor Zhang; Jessica Sun; Hudson Freeze; Madhuri Hegde. *Genet Med*. 2011 Nov; 13(11):921-32.
37. The virtual diagnostic laboratory: A new way of teaching undergraduate medical students about genetic testing. Lora J. Bean, Judith Fridovich-Keil, Madhuri Hegde, M. Katharine Rudd, Kathryn B. Garber, PhD, *Genet Med*. 2011 Nov; 13 (11): 973-937.
38. Identification of Deletions and Duplications in the DMD Gene and female Carrier Status in Indian Population using combined methods of mPCR and MLPA. Rashna S. Dastur, Munira Y Kachwala, Satish V. Khadilkar, Madhuri R Hegde, Pradnya S. Gaitonde. *Neurology India*. Dastur RS, Kachwala MY, Khadilkar SV, Hegde MR, Gaitonde PS. *Neurol India*. 2011 Nov-Dec; 59(6): 803-9.
39. Reply to Clarity and claims in variation/mutation databasing. *Human Mutation Databases*. Reply to the letter of Dalgleish et al. Sherri Bale, Heidi L. Rehm, Robert Nussbaum, Madhuri Hegde, Johan Den Dunnen, Patrick Willems. *Nature Biotechnology*, Volume: 29, 792–794 (2011)

40. Arunkanth Ankala, Jordan N. Kohn, Anisha Hegde, Arjun Meka, Chin Lip Hon Ephrem, Syed H. Askree, Shruti Bhide, Madhuri R. Hegde. Aberrant firing of replication origins explains nonrecurrent rearrangements in the human genome. *Genome Research*. November 2011. *Genome Res.* 2012 Jan 22(1): 25-34. Epub 2011 Nov 16
41. S Martinez A, Alexander C, Chin E., Hegde M. Assessment of Solution and Highly Multiplexed PCR-based Target Enrichment Using Next Generation Sequencing for Mutation Detection for Congenital Muscular Dystrophies. *Journal of Molecular Diagnostics. J Mol Diag.* 2012 May; 14(3): 233-46.
42. Jones MA, Ng Bobby, Bhide S, Rhodenizer D, Chin E, Freeze H, Hegde M. DDOST Mutations Identified by Whole-Exome Sequencing Are Implicated in Congenital Disorders of Glycosylation. *Am J Hum Genet.* 2012 Feb 10; 90 (2), 363- 368.
43. Ghrooda E, Borys A, Spriggs E, Hegde M and Mhanni A. SETX Novel mutations in a patient with ataxia with oculomotor apraxia type 2. *Parkinsonism Relat Disord.* 2012 Jun; 18(5): 700-1.
44. Robert C. Green, MD, MPH, Jonathan S. Berg, MD, PhD, Leslie Biesecker, MD, David Dimmock, MD, James P. Evans, MD, PhD, Wayne W. Grody, MD, PhD, Madhuri Hegde, PhD, Sarah Kalia, ScM, Bruce R. Korf, MD, PhD, Ian Krantz, PhD, Amy L. McGuire, JD, PhD, David Miller, MD, PhD, Mike Murray, MD, Robert Nussbaum, MD, PhD, Sharon Plon, MD, Heidi L. Rehm, PhD, FACMG, Howard J. Jacob, PhD. Expert Concordance and Discordance for Return of Incidental Findings from Whole Genome Sequencing. *Genetics in Medicine. Genet Med.* 2012 Apr 14 (4): 405-10.
45. Bobby G. Ng, Karl Hackmann, Melanie A. Jones, Alexey M. Eroshkin, Ping He, Roy Williams, Shruti Bhide, Vincent Cantagrel, Joseph G. Gleeson, Amy S. Paller, Rhonda E. Schnur, Sigrid Tinschert, Janice Zurich, Madhuri R. Hegde, Hudson H. Freeze. Mutations in the glycosylphosphatidylinositol biosynthesis gene PIG-L cause CHIME syndrome. *Am J Hum Genetics. Am J Hum Genet.* 2012 Apr 6; 90(4): 685-8. Epub 2012 Mar 22.
46. Tanner A, Chin E, Garber K, Hegde M. Microarray based detection of deletion and duplication in the GALC gene. *Orphanet Journal of Rare Diseases. Orphanet J Rare Dis.* 2012 Jun 15; 7: 38.
47. Ulrike Schwarze, Tim Cundy, Shawna Pyott, Helena Christiansen, Madhuri R. Hegde, Ruud Bank, Gerard Pals, Arunkanth Ankala, Laurie Seaver, Suzanne Yandow, Helen Raney, Dusica Babovich-Vicsanovic, Joan Stoler, Ziva Ben-Neriah, Reeval Segal, Aida Al-Aqeel, Liesbeth Siderius, Mark Hannibal, Louanne Hudgins, Elizabeth McPherson, Michelle Clemens, Michael D. Sussman, Robert Steiner, John Mahan, Rosemarie Smith, Kwame Anyane-Yeboah, Karen Chong, Tami Uster, Salim Aftimos, V. Reid Sutton, Elaine C. Davis, Mary Ann Weis, David Eyre and Peter H. Byers. Mutations in FKBP10, the first Bruck syndrome locus, which encodes a 65kD prolyl cis-trans isomerase, result in recessive forms of osteogenesis imperfecta with contractures and fractures and underhydroxylated lysyl-derived cross-linking sites in bone collagen. *Hum Mol Genet.* 2013 Jan 1; 22(1):1-17
48. Amy Gargis, Lisa Kalman, Meredith Berry, David Bick, David Dimmock, Tina Hambuch, Fei Lu, Elaine Lyon, Karl Voelkerding, Barbara Zehnbauser, Richa Agarwala, Sarah Bennett, Bin Chen, Ephrem Chin, John Compton, Soma Das, Dan Farkas, Matt Ferber, Adam Felsenfeld, Birgit Funke, Manohar Furtado, Lilia Ganova-Raeva, Ute Geigenmuller, Sandra Gunselman, Madhuri Hegde, Philip Johnson, Andrew Kasarskis, Shashikant Kulkarni, Thomas Lenk, Jonathan Liu, Megan Manion, Teri Manolio, Elizabeth Mansfield, Elaine Mardis, Jason Merker, Mangalathu Rajeevan, Martin Reese, Heidi Rehm, Brigitte Simen, Joanne Yeakley, Justin Zook, Ira Lubin. Next Generation Sequencing in medical genetics: Approaches to Quality Assurance and Compliance with Regulatory and Professional Standards. *Nat Biotechnol.* 2012 Nov; 30(11): 1033-6.
49. Martinez A, Chin E, Hegde M. Clinical next generation sequencing panel for Congenital muscular dystrophy. *Annals of Neurology. PLoS One.* 2013;8(1): e53083.
50. Ankala, A, Kohn, J. N, Khadilkar, S. V, Gaitonde P, Dastur R, Hegde, M. R. Ancestral founder mutations in calpain-3 in the Indian Agarwal community: Historical, clinical, and molecular perspective. *Muscle and Nerve. Muscle Nerve.* 2013 Jun;47(6):931-937
51. Michael F. Wangler, Rishikesh Chavan, M. John Hicks, Jed.G. Nuchter, Madhuri Hegde, Sharon E. Plon, Patrick A. Thompson. Unusually early presentation of small-bowel adenocarcinoma in a patient with Peutz-Jeghers syndrome. *J Pediatr Hematol Oncol.* 2013 May; 35(4):323-8.
52. Ephrem L.H. Chin, Cristina da Silva, Madhuri Hegde. Assessment of clinical analytical sensitivity and specificity of next generation sequencing for detection of simple and complex mutations. *BMC Genet.* 2013 Feb 19; 14:6.

53. Lisa Kalman, Jack Tarleton, Monica Hitch, Madhuri Hegde, Nick Hjelm, Elizabeth Berry-Kravis, Lili Zhou, James Hilbert, Richard Moxley III, Elizabeth Luebbe, Lorraine Toji. Quality Assurance for Myotonic Dystrophy type 1 (DM1) Genetic Testing: Development of a Genomic DNA Reference Material Panel. *Journal of Molecular Diagnostics*. J Mol Diagn. 2013 May 13. doi:p11: S1525-1578
54. Sandhya Iyer, Arunkanth Ankala, Rekha Singh, Madhuri Hegde. Determination of common genetic variants in cytidine deaminase (CDA) gene in Indian ethnic population. *Gene*. 2013 Jul 15; 524(1): 35-9.
55. Rong Fu, Madhuri Hegde, H. A. Jinnah. Genotype-Phenotype Correlations in Lesch-Nyhan Disease and its Attenuated Variants. *Brain*. Accepted
56. Jones M, Rhodenizer D, Da Silva C, Bean LJB, Coffee B, Tanner A, Collins C, Hedge M. Molecular Diagnostic Testing For Congenital Disorders Of Glycosylation (CDG): Detection Rate For Single Gene Testing And Next Generation Sequencing Panel Testing. *Mol Genet Metab*. Mol Genet Metab. 2013 May 28. S1096-7192(13) 00178-9.
57. Lora J. H. Bean, Stuart W. Tinker, Cristina da Silva, Madhuri R. Hegde. Free the Data: One Laboratory's Approach to Knowledge-based Genomic Variant Classification and Preparation for EMR Integration of Genomic Data. *Human Mutation*. Hum Mutat. 2013 Jun 11. doi: 10.1002/humu.22364
58. Heidi L. Rehm, Sherri J. Bale, Pinar Bayrak-Toydemir, Jonathan S. Berg, Kerry K. Brown, Joshua L. Deignan, Michael J. Friez, Birgit H. Funke, Madhuri R. Hegde and Elaine Lyon; A Working Group of the American College of Medical Genetics and Genomics Laboratory Quality Assurance Committee. ACMG clinical laboratory standards for next-generation sequencing. *Genetics in Medicine*. In press.
59. Ian S. Goldlust, Karen E. Hermetz, Lisa M. Catalano, Rebecca A. Cozad, Richard T. Barfield, Karen N. Conneely, Jennifer G. Mülle, Shikha Dharamrup, Madhuri Hegde, Katherine Kim, Brad Angle, Alison Colley, Amy E. Webb, Erik C. Thorland, Jay Ellison, Jill Rosenfeld, Blake C. Ballif, Lisa G. Shaffer, Laurie A. Demmer, Unique Rare Chromosome Support Group, M. Katharine Rudd. Mouse model implicates *GNB3* copy number in a novel childhood obesity syndrome. *Proc Natl Acad Sci U S A*. 2013 Sep 10; 110(37):14990-4.
60. Alice K Tanner, C. Alexander Valencia, Devin Rhodenizer, Marina Espirages, Cristina Da Silva, Lisa Borsuk, Sara Caldwell, Edward Gregg, Elizabeth Grimes, Agnieszka M Lichanska, Leah Morris, Anjan Purkayastha, Brian Weslowski, Clark Tibbetts, Matthew C Lorence, and Madhuri Hegde. Development and Performance of a Comprehensive Targeted Sequencing Assay for Pan-Ethnic Screening of Carrier Status. *J Mol Diag*. Accepted
61. Ankala A, Hegde MR. Validation of a blood-based protein assay for dysferlinopathy diagnosis: a study of test specificity and sensitivity. *Muscle and Nerve*. Accepted
62. Iyer Sandhya N, , Singhal Rekha S, Hegde Madhuri R. Arunkanth Ankala Genetic Variation in the Dihydropyrimidine dehydrogenase (DPYD) gene in healthy adult Indian population. *Annals of Human Biology*. Accepted
63. Askree SH, Chin EL, Bean LH, Coffee B, Tanner A, Hegde M. Detection limit of intragenic deletions with targeted array comparative genomic hybridization. *BMC Genet*. 2013 Dec 5;14(1):116.
64. Hegde M, Rong M, Ferber M, Samowitz W, Ganguly A. ACMG Guidelines for Inherited Colorectal Cancer. *Genetic In Medicine*. Accepted.
65. Lieu MT, Ng BG, Rush JS, Wood T, Basehore MJ, **Hegde M**, Chang RC, Abdenur JE, Freeze HH, Wang RY.. Severe, fatal multisystem manifestations in a patient with dolichol kinase-congenital disorder of glycosylation. *Mol Genet Metab*. 2013 Dec; 110(4): 484-9.
66. Aziz Nazneen, Zhao Qin, Bry Lynn, Driscoll Denise K, Gibson Jane S, Grody Wayne W, Hegde, Madhuri R, Hoeltge Gerald A, Leonard Debra G.B, Merker, Jason Derek, Palicki Linda A, Robetorye Ryan S, Schrijver Iris, Weck Karen E, and Voelkerding Karl V. College of American Pathologists' Laboratory Standards for Next Generation Sequencing Clinical Tests. *Archives of Pathology & Laboratory Medicine*. Accepted.
67. Sadat, Mohammed, Moir, Susan Chun, Tae-Wook, Lusso, Paolo; He Miao, Hedge Madhuri, Boerkeel N, Rozenweig S. A Glycosylation Defect with Hypogammaglobulinemia and Resistance to Viral Infections. *New Engl J Med*. Accepted.

68. Gregory M. Enns, Vandana Shashi, Matthew Bainbridge, Michael J. Gambello, Farah R. Zahir, Thomas Bast, , Rebecca Crimian, , Kelly Schoch, Julia Platt, , Rachel Cox, , Jonathan Bernstein, Mena Scavina, , Rhonda S. Walter, , Audrey Bibb, , Melanie Jones, Madhuri Hegde, , Brett H. Graham, , Anna C. Need, , Angelica Oviedo, , Christian P. Schaaf, Sean Boyle, , Atul J. Butte, Rong Chen, PhD, Michael J. Clark, Rajini Haraksingh, , Tina M. Cowan, , FORGE Canada Consortium, Ping He, Sylvie Langlois, Huda Y. Zoghbi, , Michael Snyder, Richard Gibbs, Hudson H. Freeze, , David B. Goldstein, Mutations in *NGLY1* Cause an Inherited Disorder of the Endoplasmic Reticulum-Associated Degradation (ERAD) Pathway. PMID: 24651605
69. Morrone A, Tylee KL, Al-Sayed M, Brusius-Facchin AC, Caciotti A, Church HJ, Coll MJ, Davidson K, Fietz MJ, Gort L, **Hegde M**, Kubaski F, Lacerda L, Laranjeira F, Leistner-Segal S, Mooney S, Pajares S, Pollard L, Ribeiro I, Wang RY, Miller N. Molecular testing of 163 patients with Morquio A (Mucopolysaccharidosis IVA) identifies 39 novel GALNS mutations. *Mol Genet Metab*. 2014 Mar 20. pii: S1096-7192(14)00092-4. doi: 10.1016/j.ymgme.2014.03.004. [Epub ahead of print] PMID: 24726177
70. Gallardo E, Ankala A, Núñez-Álvarez Y, Hegde M, Diaz-Manera J, De Luna N, Pastoret A, Suelves M, Illa I. Genetic and epigenetic determinants of low dysferlin expression in monocytes. *Hum Mutat*. 2014 May 17. doi:10.1002/humu.22591.
71. Janice A. Dominov, Özgün Uyan, MS, Peter C. Sapp, BS, Diane M. McKenna-Yasek, Babi Ramesh Reddy Nallamilli, Madhuri Hegde, and Robert H. Brown, Jr. A Novel Dysferlin Mutant Pseudoexon Bypassed with Antisense Oligonucleotides. *Annals of Clinical and Translational Neurology*. Accepted.
72. Yuan Xue, Arunkanth Ankala, William Wilcox, Madhuri Hegde. Solving the molecular diagnostic testing conundrum for Mendelian disorders in the era of next-generation sequencing: single-gene, gene panel, or exome/genome sequencing. *Genetics In Medicine*. Accepted.
73. Arunkanth Ankala, Parag Tamhankar, Madhuri Hegde. Clinical applications and implications of common and founder mutations in Indian sub-populations. *Human Mutation*. Accepted.
74. Ankala, Arunkanth; da Silva, Cristina; Gualandi, Francesca; Ferlini, Alessandra; Bean, Lora; Coffee, Bradford; Collins, Christin; Tanner, Alice; Hegde, Madhuri. A comprehensive genomic approach for NMDs gives a high diagnostic yield. *Annals of Neurology*. Accepted.

Reviews:

1. Madhuri Hegde. Marching towards personalized genomic medicine. *J Pediatr*. 2013 Jan; 162 (1): 10-1.
2. Eli Williams, Madhuri Hegde. Implementing genomic medicine in Pathology. *Advances in Anatomic Pathology*. *Adv Anat Pathol*. 2013 Jul; 20(4): 238-44.

Editorial:

1. Hegde M. New Tools of the Trade for Large-scale Collaborative Genome Analysis. *Hum Mutat*. 2013 Jun; 34(6).
2. Ankala A and Hegde M. Genomic technologies and the new era of genomic medicine. *J Mol Diagn*. 2013 Nov 20

Book Chapters:

1. Hegde Madhuri and Richards Carolyn S. Familial Adenomatous Polyposis. *Encyclopedia of genetics, genomics, proteomics and bioinformatics*. 2005.
2. Hegde Madhuri and Richards Carolyn S. Inherited Genetic Disorders. *DNA and RNA testing for Human Disease*. June 2006

3. Hegde Madhuri and Roa Benjamin. Familial Adenomatous Polyposis. Current Protocols in Human Genetics. June 2006
4. Hegde Madhuri and Roa Benjamin. Hereditary Non Polyposis Colon Cancer. Current Protocols in Human Genetics. April 2009.
5. Askree Hussain Mehdi and Hegde Madhuri. Microarrays in clinical Practice. April 2011
6. Melanie Jones and Madhuri Hegde. Molecular pathology 2nd edition: Congenital disorders of glycosylation. 2012
7. Melanie Jones and Madhuri Hegde. Translational next generation sequencing. Diseases involved in multiple genes of a common pathway (congenital disorders of glycosylation) 2013.
8. Arunkanth Ankala and Madhuri R. Hegde (2013). Implementation of Exome Sequencing Assays. Genomic Applications in Clinical and Anatomic Pathology, Dr. George J. Netto and Dr. Iris Schrijver (Ed.), ISBN: Springer. (in press)
9. Horner Vanessa and Madhuri Hegde (2014). Clinical Genomics (in press)

Books edited

1. Editor- Compendium of Molecular Biology Techniques, 2012
2. Editor- Neuromuscular Disorders, 2012

Other Publications (published abstracts): 421