

**NAME:** Vedam Laxmi Ramprasad, PhD

**POSITION TITLE:** Chief Executive Officer, MedGenome Labs (India)

**EDUCATION/TRAINING:**

INSTITUTION AND LOCATION	DEGREE	Completion Date MM/YYYY	FIELD OF STUDY
Osmania University	BS	1998	
BITS, Pilani	MS	05/2001	Human Genetics
BITS, Pilani	PhD	11/2005	Medical Technology

**A. PERSONAL STATEMENT**

I am a scientist in Human Genetics with several peer-reviewed publications to my credit. I believe in harnessing the power of genomics to provide a better outlook to patients and have been with MedGenome Labs since its inception spearheading the company's operations and strategy, currently serving as the Chief Executive Officer in the company.

My focus at MedGenome has been to drive, build and offer diverse genetic testing portfolios, launching some of the critical genetic tests at an affordable price which include the Non-Invasive Prenatal test (NIPT), Carrier Screening test, and Liquid Biopsy to name a few. My commitment to adhere to the highest quality standards and processes has been instrumental in obtaining CAP accreditation for MedGenome. With over two decades of experience, I was previously associated with Vision Research Foundation, Sankara Nethralaya, Spinco Biotech, and SciGenom labs.

**B. POSITIONS, SCIENTIFIC APPOINTMENTS, AND HONORS**

**Positions:**


04/2019- Till date Chief Executive Officer, Human Genomics, MedGenome Labs (India)  
06/2014- 04/2019 Chief Operating Officer, Human Genomics, MedGenome Labs (India)  
12/2012- 06/2014 Principal Scientist, Human Genomics, SciGenom Labs

**HONORS:**

- Healthcare leadership Award, ABP News, 2015
- Start up 50 by Smart CEO, 2019
- Top 25 Health Technology CEOs of India by Health Technology Report, 2020

**C. CONTRIBUTIONS TO SCIENCE**

- Some key publications



**The GenomeAsia 100K Project enables genetic discoveries across Asia**

GenomeAsia100K Consortium

*Nature* 576, 106–111 (2019) | Cite this article

70k Accesses | 64 Citations | 536 Altmetric | Metrics


This article has been updated

**Abstract**

The underrepresentation of non-Europeans in human genetic studies so far has limited the diversity of individuals in genomic datasets and led to reduced medical relevance for a large proportion of the world's population. Population-specific reference genome datasets as well as genome-wide association studies in diverse populations are needed to address this issue. Here we describe the pilot phase of the GenomeAsia 100K Project. This includes a whole-



MedGenome's GenomeAsia100k Published paper featured on Nature cover



Original Investigation

**Validation of a Genome-Wide Polygenic Score for Coronary Artery Disease in South Asians**

Minxian Wang PhD<sup>a</sup>, Ramesh Menon PhD<sup>b</sup>, Sanghamitra Mishra PhD<sup>b</sup>, Aniruddh P. Patel MD<sup>a, c, d</sup>, Mark Chaffin MSc<sup>a</sup>, Deepak Tanneeru MTech<sup>b</sup>, Manjari Deshmukh MSc<sup>b</sup>, Oshin Mathew MSc<sup>b</sup>, Sanika Apte MSc<sup>b</sup>, Christina S. Devanboo MSc<sup>b</sup>, Sumathi Sundaram BSc<sup>b</sup>, Praveena Lakshmi MSc<sup>b</sup>, Sakthivel Murugan PhD<sup>b</sup>, Krishna Kumar Sharma PhD<sup>a</sup>, Karthikeyan Rajendran BPT<sup>f</sup>, Sam Santhosh BTEch, MBA<sup>b</sup>, Rajesh Thachathodiyl MBBS, MD<sup>g</sup>, Hisham Ahamed MD<sup>h</sup> ... Amit V. Khera MD, MSc<sup>a, c, d, g, h</sup>

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**Extensive list of publications:**

#	Name	Journal	Authors	Year	Link
1	Genotype-phenotype correlation and natural history study of dysferlinopathy: a single-centre experience from India	Neurogenetics	Saraswati Nashi , Kiran Polavarapu , Mainak Bardhan , Ram Murthy Anjanappa , Veeramani Preethish-Kumar , Seena Vengalil , Hansashree Padmanabha , Thenral S Geetha , P V Prathyusha , <b>Vedam Ramprasad</b> , Aditi Joshi , Tanushree Chawla , Gopikrishnan Unnikrishnan , Pooja Sharma , Akshata Huddar , Bharathram Uppilli , Abel Thomas , Dipti Baskar , Susi Mathew , Deepak Menon , Gautham Arunachal , Mohammed Faruq , Kumarasamy Thangaraj , Atchayaram Nalini	2022	10.1007/s10048-022-00707-3
2	Molecular epidemiology of SARS-CoV-2 in healthcare workers and identification of viral genomic correlates of transmissibility and vaccine break through infection: A retrospective observational study from a cancer hospital in eastern India	Indian Journal of Medical Microbiology	Sanjay Bhattacharya, Soumyadip Chatterji, Mammen Chandy, Aseem Yogishwar, Mahajan, Gaurav Goel, Deepak Mishra, Priyanka Vivek, Parijat Das, Sudipto Mandal, Anup Chugani, Antra Mittal, Rajadurai Chinnasamy Perumalg, <b>Vedam L. Ramprasad</b> , Ravi Gupta	2022	<a href="https://doi.org/10.1016/j.ijmmb.2022.09.010">https://doi.org/10.1016/j.ijmmb.2022.09.010</a>

3	Genome-Wide Polygenic Score Predicts Large Number of High Risk Individuals in Monogenic Undiagnosed Young Onset Parkinson's Disease Patients from India PMID: 35810474	Advanced biology	Prashanth Lingappa Kukkle, Thenral S. Geetha, Ruchi Chaudhary, Jarupon F. Sathirapongsasuti, Vinay Goyal, Rukmini Mridula Kandadai, Hrishikesh Kumar, Rupam Borgohain, Adreesh Mukherjee, Merina Oliver, Meeta Sunil, Mohammed Faizal Eeman Mootor, Shruti Kapil, Nitin Mandloi, Pettarusp M. Wadia, Ravi Yadav, Soham Desai, Niraj Kumar, Atanu Biswas, Pramod Kumar Pal, Uday B. Muthane, Shymal Kumar Das, Sakthivel M. Sakthivel Murugan, Andrew S. Peterson, Eric W. Stawiski, Somasekar Seshagiri, Ravi Gupta, Vedam L. Ramprasad, Parkinson Research Alliance of India (PRAI)	2022	<a href="https://onlinelibrary.wiley.com/doi/10.1002/adbi.202101326">https://onlinelibrary.wiley.com/doi/10.1002/adbi.202101326</a>
4	Clinical, genetic profile and disease progression of sarcoglycanopathies in a large cohort from India: high prevalence of SGCB c.544A > C	Neurogenetics	Mainak Bardhan, Ram Murthy Anjanappa, Kiran Polavarapu, Veeramani Preethish-Kumar, Seena Vengalil, Saraswati Nashi, Shamita Sanga, Hansashree Padmanabh, Ravi Kiran Valasani, Vikas Nishadham, Muddasu Keerthipriya, Thenral S. Geetha, <b>Vedam Ramprasad</b> , Gautham Arunachal, Priya Treasa Thomas, Moulinath Acharya & Atchayaram Nalini	2022	<a href="https://link.springer.com/article/10.1007/s10048-022-00690-9">https://link.springer.com/article/10.1007/s10048-022-00690-9</a>
5	Chorea-acanthocytosis: 3 New Families with Novel Genetic and Metabolic Findings PMID: 34447025	Annals of Indian Academy of Neurology	Mirza Masoom Abbas <sup>1</sup> , SG Thenral <sup>2</sup> , <b>Vedam L Ramprasad</b> <sup>2</sup> , Ruth H Walker <sup>3</sup> , Prashanth Lingappa Kukkle <sup>4</sup>	2022	<a href="https://www.annalsofian.org/article.asp?issn=0972-2327;year=2021;volume=24;issue=3;page=452;epage=456;aulast=Abbas">https://www.annalsofian.org/article.asp?issn=0972-2327;year=2021;volume=24;issue=3;page=452;epage=456;aulast=Abbas</a>
6	Whole-exome sequencing and variant spectrum in children with suspected inherited renal tubular disorder: the East India Tubulopathy Gene Study PMID: 35006361	Pediatric Nephrology Journal of the International Pediatric Nephrology Association	Himika Gupta, Sivasankar Malaichamy, Ashwin Mallipatna, Sakthivel Murugan, Nallathambi Jeyabalan, Vishnu Suresh Babu, Anuprita Ghosh, Arkasubhra Ghosh, Sam Santhosh, Somasekar Seshagiri, Vedam L. Ramprasad & Govindasamy Kumaramanickavel	2022	<a href="https://bmcmmedgenomics.biomedcentral.com/articles/10.1186/s12920-021-01034-6">https://bmcmmedgenomics.biomedcentral.com/articles/10.1186/s12920-021-01034-6</a>
7	Clinical Study of 668 Indian Subjects with Juvenile, Young, and Early Onset Parkinson's Disease PMID: 33685545	The Canadian journal of neurological sciences	Prashanth L Kukkle <sup>1,2</sup> , Vinay Goyal <sup>3,4</sup> , Thenral S Geetha <sup>5</sup> , Kandadai R Mridula <sup>6</sup> , Hrishikesh Kumar <sup>7</sup> , Rupam Borgohain <sup>6</sup> , Adreesh Mukherjee <sup>8</sup> , Pettarusp M Wadia <sup>9</sup> , Ravi Yadav <sup>10</sup> , Soham Desai <sup>11</sup> , Niraj Kumar <sup>12</sup> , Ravi Gupta <sup>5</sup> , Atanu Biswas <sup>8</sup> , Pramod K Pal <sup>10</sup> , Uday Muthane <sup>2</sup> , Shymal K Das <sup>8</sup> , Niall Quinn <sup>13</sup> , <b>Vedam L Ramprasad</b> <sup>5</sup> , Parkinson Research Alliance of India (PRAI)	2022	<a href="https://pubmed.ncbi.nlm.nih.gov/33685545/">https://pubmed.ncbi.nlm.nih.gov/33685545/</a>

8	Profile of Pathogenic Mutations and Evaluation of Germline Genetic Testing Criteria in Consecutive Breast Cancer Patients Treated at a North Indian Tertiary Care Center	Annals of Surgical Oncology	Abhenil Mittal, MD, DM1 , S. V. S. Deo, MS, MCh2 , Ajay Gogia, MD, DM1 , Atul Batra, MD, DM1 , Akash Kumar, MD, DM1 , Sandeep Bhoariwal, MS, MCh2 , Koushik Sinha Deb, MD3 , Ekta Dhamija, MD4 , Sanjay Thulkar, MD4 , <b>V. L. Ramprasad</b> , PhD5 , Olufunmilayo Olopade, MD, FACP6 , and Raja Pramanik, MD, DM1	2021	<a href="https://www.annalsof oncology.org/article/S0923-7534(21)01047-4/fulltext">https://www.annalsof oncology.org/article/S0923-7534(21)01047-4/fulltext</a>
9	Growth and neurodevelopmental disorder with arthrogyrosis, microcephaly and structural brain anomalies caused by Bi-allelic partial deletion of SMPD4 gene	J Hum Genet.	Bijarnia-Mahay S, Somashekar PH, Kaur P, Kulshrestha S, <b>Ramprasad VL</b> , Murugan S, Sud S, Shukla A.	2021	doi: 10.1038/s10038-021-00981-3.
10	Retinoblastoma genetics screening and clinical management	Retinoblastoma a genetics screening and clinical management	Gupta H, Malaichamy S, Mallipatna A, Murugan S, Jeyabalan N, Suresh Babu V, Ghosh A, Ghosh A, Santhosh S, Seshagiri S, Ramprasad VL, Kumaramanickavel G. PMID: 34294096; PMCID: PMC8296631.	2021	doi: 10.1186/s12920-021-01034-6.
11	Nemaline Rod/Cap Myopathy Due to Novel Homozygous MYPN Mutations: The First Report from South Asia and Comprehensive Literature Review	National Library of medicine	Kiran Polavarapu # 1 2, Mainak Bardhan # 1, Ram Murthy Anjanappa 3, Seena Vengalil 1, Veeramani Preethish-Kumar 1, Leena Shingavi 1, Tanushree Chawla 1, Saraswati Nashi 1, Dhaarini Mohan 1, Gautham Arunachal 3, Thenral S Geetha 4, <b>Vedam Ramprasad 4</b> , Atchayaram Nalini 5	2021	<a href="https://thejcn.com/DOLx.php?id=10.3988/jcn.2021.17.3.409">https://thejcn.com/DOLx.php?id=10.3988/jcn.2021.17.3.409</a>
12	Matrilineal Analysis Of Mutations In The DMD Gene In A Multigenerational South Indian Cohort Using DMD Gene Panel Sequencing	Molecular Genetics and Genomic Medicine	Arun Shastry, Sankaramoorthy Aravind, Meeta Sunil, Keerthi Ramesh, Berty Ashley, Nithyanandan T., <b>Vedam L. Ramprasad</b> , Ravi Gupta, Somasekar Seshagiri, Upendra Nongthomba, Sameer Phalke	2021	<a href="https://doi.org/10.1002/mgg3.1633">https://doi.org/10.1002/mgg3.1633</a>
13	Phenotypic expression and clinical outcomes in a South Asian PRKAG2 cardiomyopathy cohort	Scientific reports	Hisham Ahamed , Aniketh Vijay Balegadde , Shilpa Menon , Ramesh Menon , Aishwarya Ramachandran , Navin Mathew , K U Natarajan , Indu Ramachandran Nair , Rajesh Kannan , Meghna Shankar , Oommen K Mathew , Thong T Nguyen , Ravi Gupta , Eric W Stawiski , <b>V L Ramprasad</b> , Somasekar Seshagiri , Sameer Phalke	2020	10.1038/s41598-020-77124-9
14	Fatal Familial Insomnia: A Rare Disease with Unique Clinico-Neurophysiological Features.	Movement disorders-Clinical Practice	Kukkle PL, Geetha TS, Mahadevan A, Ramprasad VL.	2020	doi: 10.1002/mdc3.13115.
15	NGS-Based Expanded Carrier Screening For Genetic Disorders In North Indian Population Reveals Unexpected Results – A Pilot Study	BMC Medical Genetics	Kanika Singh, Sunita Bijarnia-Mahay, <b>V. L. Ramprasad</b> , Ratna Dua Puri, Sandhya Nair, Sheetal Sharda, Renu Saxena, Sudha Kohli, Samarth Kulshreshtha, Indrani Ganguli, Kanwal Gujral And Ishwar C. Verma	2020	<a href="https://bmcmmedgenet.biomedcentral.com/articles/10.1186/s12881-020-01153-4">https://bmcmmedgenet.biomedcentral.com/articles/10.1186/s12881-020-01153-4</a>

16	Validation Of A Genome-Wide Polygenic Score For Coronary Artery Disease In South Asians	Journal Of the American College Of Cardiology	Minxian Wang, PHD, Ramesh Menon, PHD, Sanghamitra Mishra, PHD, Aniruddh P. Patel, MD, Mark Chaffin, MSC, Deepak Tanneeru, MTECH, Manjari Deshmukh, MSC, Oshin Mathew, MSC, Sanika Apte, MSC, Christina S. Devanboo, MSC, Sumathi Sundaram, BSC, Praveena Lakshmi pathy, MSC, Sakthivel Murugan, PHD, Krishna Kumar Sharma, PHD, Karthikeyan Rajendran, BPT, Sam Santhosh, BTECH, MBA, Rajesh Thachathodiyl, MBBS, MD, Hisham Ahamed, MD, Aniketh Vijay Balegadde, MBBS, MD, Thomas Alexander, MD, Krishnan Swaminathan, MD, Rajeev Gupta, MD, PHD, Ajit S. Mullasari, MBBS, MD, Alben Sigamani, MBBS, MD, Muralidhar Kanchi, MBBS, MD, MBA, Andrew S. Peterson, PHD, Adam S. Butterworth, PHD, John Danesh, DPHIL, Emanuele Di Angelantonio, MD, PHD, Aliya Naheed, MBBS, MPH, PHD, Michael Inouye, PHD, Rajiv Chowdhury, MPH, PHD, <b>Ramprasad L. Vedam</b> , PHD, Sekar Kathiresan, MD, Ravi Gupta, PHD, Amit V. Khera, MD, MSC.	2020	<a href="https://doi.org/10.1016/j.jacc.2020.06.024">https://doi.org/10.1016/j.jacc.2020.06.024</a>
17	Robinow Syndrome and Brachydactyly: An Interplay of High-Throughput Sequencing and Deep Phenotyping in a Kindred	Molecular Syndromology	Ranjana Mishra , Vibha Jain, Deepti Gupta, Renu Saxena , Samarth Kulshreshtha , <b>Vedam L. Ramprasad</b> Ishwar C. Verma Ratna Dua Puri	2020	<a href="https://www.karger.com/Article/FullText/505506">https://www.karger.com/Article/FullText/505506</a>
18	Whole genome enrichment approach for rapid detection of Mycobacterium tuberculosis and drug resistance-associated mutations from direct sputum sequencing.	Tuberculosis	Lakshmi Soundararajan, Priti Kambli, Sushri Priyadarshini, Biswajit Let, Sakthivel Murugan, Chitra Iravatham, Jeffrey A. Tornheim, Camilla Rodrigues, Ravi Gupta, <b>V.L. Ramprasad</b> .	2020	<a href="https://doi.org/10.1016/j.tube.2020.101915">https://doi.org/10.1016/j.tube.2020.101915</a>
19	A Case of Autosomal Dominant Ataxia with Vocal Cord Palsy Attributed to a Mutation in the PRNP Gene.	Movement disorders- Clinical Practice	Kukkle PL, Geetha TS, Mahadevan A, Ramprasad VL.	2020	
20	Factor XIII Deficiency with a Novel Nonsense Mutation.	Indian J Hematol Blood Transfus.	Khandelwal V, Sharma SK, Doval D, Kumar M, Choudhary Khandelwal V, Sharma SK, Doval D, Kumar M, Choudhary D, Sharma R, <b>Ramprasad VL</b> .	2020	doi: 10.1007/s12288-020-01262-3
21	Genetic analysis of familial hypercholesterolemia in Asian Indians: A single-center study	Journal of Clinical lipidology	Nitika Setia, PhD , Sireesha Movva, PhD, Prahlad Balakrishnan, PhD Ishpreet K. Biji, MSc, Jitendra Pal Singh Sawhney, MD, DM, Raman Puri, MD, DM Anjali Arora, MD, Ratna D. Puri, MD, DM, Renu Saxena, PhD, Sanghamitra, Mishra, PhD, Sanika Apte, MSc, Samarth Kulshreshtha, MSc <b>Vedam Lakshmi Ramprasad</b> , PhD, Ishwar C. Verma, MRCP	2020	<a href="https://doi.org/10.1016/j.jacl.2019.12.010">https://doi.org/10.1016/j.jacl.2019.12.010</a>
22	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	Molecular Genetics and Genomic Medicine	Sophia Yohe Malaichamy Sivasankar Anuprita Ghosh Arkasubhra Ghosh Jennifer Holle Sakthivel Murugan Ravi Gupta Lisa A. Schimmenti <b>Ramprasad Vedam</b> Bharat Thyagarajan	2019	<a href="https://doi.org/10.1002/mgg3.1081">https://doi.org/10.1002/mgg3.1081</a>

23	The GenomeAsia 100K Project Enables Genetic Discoveries Across Asia	Nature	Jeffrey D. Wall, Eric W. Stawiski, Aakrosh Ratan, Hie Lim Kim, Changhoon Kim, Ravi Gupta, Kushal Suryamohan, Elena S. Gusareva, Rikky Wenang Purbojati, Tushar Bhangale, Vadim Stepanov, Vladimir Kharkov, Markus S. Schröder, <b>Vedam Ramprasad</b> , Jennifer Tom, Steffen Durinck, Qixin Bei, Jiani Li, Joseph Guillory, Sameer Phalke, Analabha Basu, Jeremy Stinson, Sandhya Nair, Sivasankar Malaichamy, Nidhan K. Biswas, John C. Chambers, Keith C. Cheng, Joyner T. George, Seik Soon Khor, Jong-Il Kim, Belong Cho, Ramesh Menon, Thiramsetti Sattibabu, Akshi Bassi, Manjari Deshmukh, Anjali Verma, Vivek Gopalan, Jong-Yeon Shin, Mahesh Pratapneni, Sam Santhosh, Katsushi Tokunaga, Badrul M. Md-Zain, Kok Gan Chan, Madasamy Parani, Purushothaman Natarajan, Michael Hauser, R. Rand Allingham, Cecilia Santiago-Turla, Arkasubhra Ghosh, Santosh Gopi Krishna Gadde, Christian Fuchsberger, Lukas Forer, Sebastian Schoenherr, Herawati Sudoyo, J. Stephen Lansing, Jonathan Friedlaender, George Koki, Murray P. Cox, Michael Hammer, Tatiana Karafet, Khai C. Ang, Syed Q. Mehdi, Venkatesan Radha, Viswanathan Mohan, Partha P. Majumder, Somasekar Seshagiri, Jeong-Sun Seo, Stephan C. Schuster & Andrew S. Peterson - Nature Volume 576, Pages106 – 111(2019)	2019	<a href="https://www.nature.com/articles/s41586-019-1793-z">https://www.nature.com/articles/s41586-019-1793-z</a>
24	Detection of clinically relevant epidermal growth factor receptor pathway mutations in circulating cell-free tumor DNA using next generation sequencing in squamous cell carcinoma lung.	South Asian Journal of Cancer	Kanakasetty Babu Govind, Deepak Koppaka, Lokanatha Dasappa, Linu Abraham Jacob, Suresh M C. Babu, N Kadabur Lokesh, Rudresha Antapura Haleshappa, LK Rajeev, Smitha Carol Saldanha, Anand Abhishek, Vikas Asati, R Chethan, <b>Vedam Laxmi Ramprasad</b>	2019	<a href="http://journal.sajc.org/article.asp?issn=2278-330X;year=2019;volume=8;issue=4;spage=247;epage=249;aulast=Govind">http://journal.sajc.org/article.asp?issn=2278-330X;year=2019;volume=8;issue=4;spage=247;epage=249;aulast=Govind</a>
25	Resequencing CYP2D6 gene in Indian population: CYP2D6*41 identified as the major reduced function allele	Pharmacogenomics	Aarthi Manoharan , Deepak Gopal Shewade, Pradeep Anand Ravindranath, Ravi Philip Rajkumar, <b>Vedam L Ramprasad</b> , Surendiran Adithan & Solai Elango Damodaran	2019	<a href="https://doi.org/10.2217/pgs-2019-0049">https://doi.org/10.2217/pgs-2019-0049</a>
26	Immunodeficiency, Motor Delay, and Hypouricemia Caused by a Novel Mutation of Purine Nucleoside Phosphorylase Gene in an Indian Infant.	Annals of Indian Academy of Neurology	Nikit Shah, Lokesh Lingappa, Ramesh Konanki, Sirisha Rani, <b>Ramprasad Vedam</b> , and Sakthivel Murugan	2019	<a href="https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6472228/">https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6472228/</a>
27	Simultaneous diagnosis of unilateral retinoblastoma and contralateral optic pathway glioma in a child with neurofibromatosis type 1	Pediatric Hematology and Oncology	Anirban Das ,Priya Ghosh,Lateef Zameer, <b>Vedam L. Ramprasad</b> & Anirban Bhaduri	2019	<a href="https://doi.org/10.1080/08880018.2019.1591550">https://doi.org/10.1080/08880018.2019.1591550</a>

28	Adult-Onset Myoclonus-Dystonia Syndrome Preceding Characteristic Facial Myoclonus In Indian ADCY5-Related Dyskinesia	Movement disorders-Clinical Practice	Pankaj Ashok Agarwal MD, DNB, DM <b>Vedam L. Ramprasad</b> PhD	2019	<a href="https://onlinelibrary.wiley.com/doi/abs/10.1002/mdc.3.12733">https://onlinelibrary.wiley.com/doi/abs/10.1002/mdc.3.12733</a>
29	Novel GNAL mutation in an Indian patient with generalized dystonia and response to deep brain stimulation	Parkinsonism & Related Disorders	Sanjay Pandey, Charulata Savant Sankhla, <b>Vedam L. Ramprasad</b> Thenral S. Geetha	2019	<a href="https://doi.org/10.1016/j.parkrel.2019.01.011">https://doi.org/10.1016/j.parkrel.2019.01.011</a>
30	Actionable Activating Oncogenic ERBB2/HER2 Transmembrane and Juxtamembrane Domain Mutations	Cancer cells	Kanika Bajaj Pahuja Thong T. Nguyen , Bijay S. Jaiswal , Kumar Prabhash , Tarjani M. Thaker ,Kate Senger, Subhra Chaudhuri, Noelyn M. Kljavin, Aju Antony, Sameer Phalke, Prasanna Kumar, Marco Mravic, Eric W. Stawiski Derek Vargas, Steffen Durinck, Ravi Gupta, Arati Khanna-Gupta, Sally E. Trabucco, Ethan S. Sokol, Ryan J. Hartmaier, Ashish Singh, Anuradha Chougule Vaishakhi Trivedi, Amit Dutt, Vijay Patil, Amit Joshi, Vanita Noronha, James Ziai, Sripad D. Banavali, <b>Vedam Ramprasad</b> , William F. DeGrado, Raphael Bueno, Natalia Jura, Somasekar Seshagiri	2018	<a href="https://doi.org/10.1016/j.ccell.2018.09.010">https://doi.org/10.1016/j.ccell.2018.09.010</a>
31	A neoepitope derived from a novel human germline APC gene mutation in familial adenomatous polyposis shows selective immunogenicity	Plos One	Snigdha Majumder ,Rakshit Shah ,Jisha Elias,Yogesh Mistry,Karunakaran Coral,Priyanka Shah,Anand Kumar Maurya,Bharti Mittal,Jason K. D'Silva,Sakthivel Murugan,Lakshmi Mahadevan,Rekha Sathian, <b>V. L. Ramprasad</b> , [ ... ],Arati Khanna-Gupta	2018	<a href="https://doi.org/10.1371/journal.pone.0203845">https://doi.org/10.1371/journal.pone.0203845</a>
32	Noninvasive prenatal testing (NIPT) detects variant of Turner syndrome not detectable by fluorescent in situ hybridization	The Journal of Maternal-Fetal & Neonatal Medicine	Venkataswamy Eswarachari,Priya Kadam,Sireesha Movva,Shruthi Lingaiah,Riyaz M. Akther,Franics X. Kidangan,Kiran C. Gowda,Rudra R. K. Golakoti,Meena Lall,Surbhi Mahajan,Pushpa Saviour,Ratna Puri,Ishwar C. Verma &Ramprasad L. Vedam	2018	<a href="https://doi.org/10.1080/14767058.2018.1481383">https://doi.org/10.1080/14767058.2018.1481383</a>
33	Clinical, Immunological, and Molecular Findings in Five Patients with Major Histocompatibility Complex Class II Deficiency from India	Frontiers In Immunology	Jahnvi Aluri, Maya Gupta, Aparna Dalvi, Snehal Mhatre, Manasi Kulkarni, Gouri Hule, Mukesh Desai, Nitin Shah, Prasad Taur, <b>Ramprasad Vedam</b> and Manisha Madkaikar	2018	<a href="https://doi.org/10.3389/fimmu.2018.00188">https://doi.org/10.3389/fimmu.2018.00188</a>
34	Comprehensive genomic analysis identifies pathogenic variants in maturity-onset diabetes of the young (MODY) patients in South India.	BMC Medical Genetics	Viswanathan Mohan, Venkatesan Radha, Thong T. Nguyen, Eric W. Stawiski, Kanika Bajaj Pahuja, Leonard D. Goldstein, Jennifer Tom, Ranjit Mohan Anjana, Monica Kong-Beltran, Tushar Bhangale, Suresh Jahnvi, Radhakrishnan Chandni, Vijay Gayathri, Paul George, Na Zhang, Sakthivel Murugan, Sameer Phalke, Subhra Chaudhuri, Ravi Gupta, Jingli Zhang, Sam Santhosh, Jeremy Stinson, Zora Modrusan, <b>V. L. Ramprasad</b> , Somasekar Seshagiri & Andrew S. Peterson	2018	<a href="https://bmcmmedgenet.biomedcentral.com/articles/10.1186/s12881-018-0528-6">https://bmcmmedgenet.biomedcentral.com/articles/10.1186/s12881-018-0528-6</a>

35	Single Nucleotide Polymorphism-Based Noninvasive Prenatal Testing: Experience in India.	The Journal of Obstetrics and Gynecology of India	Ishwar Chander Verma,Ratna Puri, Eswarachary Venkataswamy, Tulika Tayal, Sheela Nampoorthiri, Chitra Andrew, Madhulika Kabra, Rashmi Bagga, Mamatha Gowda, Meenu Batra, Sridevi Hegde, Anita Kaul, Neerja Gupta, Pallavi Mishra, Jayshree Ganapathi Subramanian, Shruti Lingaiah, Riyaz Akhtar, Francis Kidangan, R. Chandran, C. Kiran, G. R. Ravi Kumar, <b>V. L. Ramprasad</b> , and Priya Kadam	2018	<a href="https://link.springer.com/article/10.1007%2Fs13224-017-1061-9">https://link.springer.com/article/10.1007%2Fs13224-017-1061-9</a>
36	Validation of liquid biopsy: plasma cell-free DNA testing in clinical management of advanced non-small cell lung cancer	Lung Cancer: Targets and Therapy	Vidya H Veldore, Anuradha Choughule, Tejaswi Routhu, Nitin Mandloi, Vanita Noronha, Amit Joshi, Amit Dutt, Ravi Gupta, <b>Ramprasad Vedam</b> , Kumar Prabhash	2018	<a href="https://doi.org/10.2147/LCTT.S147841">https://doi.org/10.2147/LCTT.S147841</a>
37	A novel splice variant in EMC1 is associated with cerebellar atrophy, visual impairment, psychomotor retardation with epilepsy	Molecular Genetics and Genomic Medicine	Thenral S. Geetha Lokesh Lingappa Abhishek Ravindra Jain Hridya Govindan Nitin Mandloi Sakthivel Murugan Ravi Gupta and <b>Ramprasad Vedam</b>	2017	<a href="https://doi.org/10.1002/mgg3.352">https://doi.org/10.1002/mgg3.352</a>
38	Asparagine Synthetase deficiency-report of a novel mutation and review of literature	Metabolic Brain Disease	Neerja Gupta, Vishal Vishnu Tewari, Manoj Kumar, Nitika Langeh, Aditi Gupta, Pallavi Mishra, Punit Kaur, <b>Vedam Ramprasad</b> , Sakthivel Murugan, Reema Kumar, Manisha Jana & Madhulika Kabra	2017	<a href="https://doi.org/10.1007/s11011-017-0073-6">https://doi.org/10.1007/s11011-017-0073-6</a>
39	Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS)—First Report of Clinical and Imaging Features from India, and a Novel SACS Gene Duplication	Movement disorders-Clinical Practice	Pankaj A. Agarwal MD, DNB, DM, Priti Ate-Upasani MD, DNB, DMRE and <b>Vedam L. Ramprasad</b> PhD	2017	<a href="https://doi.org/10.1002/mdc3.12520">https://doi.org/10.1002/mdc3.12520</a>
40	Next-Generation Sequencing Reveals Novel Mutations In X-Linked Intellectual Disability	NCBI	Muthusamy B1,2, Selvan LDN1, Nguyen TT3, Manoj J4, Stawiski EW3,5, Jaiswal BS3, Wang W6, Raja R1, <b>Ramprasad VL7</b> , Gupta R7, Murugan S7, Kadandale JS8, Prasad TSK 1,9,10, Reddy K1, Peterson A3, Pandey A11,12,13, Seshagiri S3, Girimaji SC4, Gowda H1,9	2017	<a href="https://www.ncbi.nlm.nih.gov/pubmed/28481730">https://www.ncbi.nlm.nih.gov/pubmed/28481730</a>
41	Next-Generation Sequencing Reveals Novel Mutations in X-linked Intellectual Disability	Journal of Integrative Biology	Babylakshmi Muthusamy, Lakshmi Dhevi N. Selvan, Thong T. Nguyen, Jesna Manoj, Eric W. Stawiski, Bijay S. Jaiswal, Weiru Wang, Remya Raja, <b>Vedam Laxmi Ramprasad</b> , Ravi Gupta, Sakthivel Murugan, Jayarama S. Kadandale, T.S. Keshava Prasad, Kavita Reddy, Andrew Peterson, Akhilesh Pandey, Somasekar Seshagiri, Satish Chandra Girimaji, and Harsha Gowda	2017	<a href="https://doi.org/10.1089/omi.2017.0009">https://doi.org/10.1089/omi.2017.0009</a>
42	The Utility and Futility Of Targeted Next-Generation Sequencing For Carrier Detection In 'At Risk' Couples	The Indian Academy of Medical Genetics (IAMG)	Sunita Bijarnia-Mahay 1*, Deepti Gupta 1, <b>V L Ramprasad</b> 2, Sakthivel Murugan 2 , Renu Saxena 1, Sudha Kohli 1, Seiji Yamaguchi 3, Yosuke Shigematsu 4 And I C Verma 1	2017	<a href="http://iamg.in/genetic_clinics/article/pdf/genevista_January_March_2017.pdf">http://iamg.in/genetic_clinics/article/pdf/genevista_January_March_2017.pdf</a>
43	A Splice Site Mutation In HERC1 Leads To Syndromic Intellectual Disability With Macrocephaly And Facial Dysmorphism:	NCBI	Aggarwal S, Bhowmik AD, <b>Ramprasad VL</b> , Murugan S, Dalal A	2016	<a href="https://www.ncbi.nlm.nih.gov/">https://www.ncbi.nlm.nih.gov/</a>



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44	A novel H395R mutation in MKKS/BBS6 causes retinitis pigmentosa and polydactyly without other findings of Bardet-Biedl or McKusick-Kaufman syndrome	Molecular Vision	John D. Hulleman, Annie Nguyen, <b>V.L. Ramprasad</b> , Sakthivel Murugan, Ravi Gupta, Avinash Mahindrakar, Ravi Angara, Chandrasekhar Sankurathri, and V. Vinod Mootha	2016	<a href="https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4734152/">https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4734152/</a>
45	First Report of Kufor-Rakeb Syndrome (PARK 9) from India, and a Novel Nonsense Mutation in ATP13A2 Gene	Movement disorders-Clinical Practice	L.K. Prashanth DM, Sakthivel Murugan PhD, Vikram Kamath DM, Ravi Gupta PhD, Rakesh Jadav MD, DM, S. Sreekantaswamy MD, DM and <b>Vedam L. Ramprasad PhD</b>	2015	<a href="https://doi.org/10.1002/mdc3.12175">https://doi.org/10.1002/mdc3.12175</a>
46	Identification of Novel Mutations in ABCA4 Gene: Clinical and Genetic Analysis of Indian Patients with Stargardt Disease.	BioMed Research International	Rajani Battu, Anshuman Verma, Ramesh Hariharan, Shuba Krishna, Ravi Kiran, Jemima Jacob, Aparna Ganapathy, <b>Vedam L. Ramprasad</b> , Govindasamy Kumaramanickavel, Nallathambi Jeyabalan , and Arkasubhra Ghosh	2015	<a href="https://doi.org/10.1155/2015/940864">https://doi.org/10.1155/2015/940864</a>
47	Evolution of targeted therapies in cancer: opportunities and challenges in the clinic	Future Oncology	Sam Santhosh, Prasanna Kumar, <b>Vedam Ramprasad</b> & Amitabha Chaudhuri	2015	<a href="https://doi.org/10.2217/fon.14.198">https://doi.org/10.2217/fon.14.198</a>
48	Recessive Mutations In SLC38A8 Cause Foveal Hypoplasia And Optic Nerve Misrouting Without Albinism	The American Journal of Human Genetics	James A. Poulter, Musallam Al-Araimi, Ivan Conte, Maria M. Van Genderen, Eamonn Sheridan, Ian M. Carr, David A. Parry, Mike Shires, Sabrina Carrella, John Bradbury, Kamron Khan, Phillis Lakeman, Panagiotis I. Sergouniotis, Andrew R. Webster, Anthony T. Moore, Bishwanath Pal, Moin D. Mohamed, Anandula Venkataramana, <b>Vedam Ramprasad</b> , Rohit Shetty, Murugan Saktivel, Govindasamy Kumaramanickavel, Alex Tan, David A. Mackey, Alex W. Hewitt, Sandro Banfi, Manir Ali, Chris F. Inglehearn, And Carmel Toomes	2013	<a href="https://doi.org/10.1016/j.ajhg.2013.11.002">https://doi.org/10.1016/j.ajhg.2013.11.002</a>