



University Faculty Details Page on DU Web-site

Title	Dr.	First Name	Thelma	Last Name	B.K.	Photograph
Designation	Professor					
Department	Genetics					
Address	Department of Genetics University of Delhi South Campus Benito Juarez road New Delhi- 110 021 011-24118201					
Phone No	Office	91-11-24118201				
	Residence	91-11-24113548				
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	Fax	91-11-2411 2761				
Email	thelmabk@gmail.com , thelmabk@south.du.ac.in					
Web-Page						
Educational						
Subject	Institution	Year	Details			
Post Doctoral Fellow	Human Genetics Lab, Children's Hospital, Basel, Switzerland	1984-1985				
Ph.D.	University of Delhi	1982				
PG	Bangalore University	1975				
UG	Bangalore University	1973				
Career Profile						
Organization / Institution	Designation	Duration	Role			
Department of Genetics, UDSC	Professor	2012- to date				
	Professor & Head	2009-2012				
	Professor	2006-2009				
Department of Zoology, DU	Professor	2003-2006				
Department of Genetics, UDSC	Reader	1998-2003				
Department of Genetics, UDSC	Sr. Lecturer	1990-1998				
	Lecturer	1987-1990				
Administrative Assignments						
Professional/ Other Academic Member Affiliations						
Current						
2015-2020	Team leader, Centre of excellence on Genome Science and Predictive Medicine, (Phase II)					
2014-2017	Team Leader, Newborn Screening for Inborn error of metabolism					
2017-2020	Expert Member, Task Force in Basic research in modern biology, DBT					
2015-2020	Member, Selection committee, INSPIRE faculty program					
2016-	Member, Scientific Advisory Committee, National Institute for Research in Reproductive Health, Mumbai					
2015-2018	Member, Council, Indian National Science Academy, New Delhi					
2013-	Member, Management Committee, Kishore Vaigyanik Protsahan Yojana, IISC					
2012-	Member, Life sciences selection committee, Summer research fellows program, Science Education Panel, Indian Academy of Sciences					
2008-	Trustee, XV International Genetics Congress Trust, New Delhi					
VC nominee/University Representative on Governing Body, University of Delhi Departments/Colleges						
2017 –	Cluster Innovation Centre					

2017 –	Member of Antirragging committee, Department of Genetics
2017 –	Member of Dicipinary Resource Committee, Department of Genetics
2017 –2018	Holy Family College of Nursing
2016 –2018	Departmental Research Committee of Medical Sciences
2015 –2017	Raj Kumari Amrit Kaur College of Nursing
2015 -2017	Board of Research Studies, Faculty of Interdisciplinary &Applied Sciences
2006-	Departmental Research Committee, Genetics
Previous	
2013-2016	Member, Screening and Selection committee, UGC-FRP
2011-2015	Member, Science and Engineering Research Board (SERB)
2009-2014	Member, Scientific Advisory Council to Prime Minister of India
2009-2012	Head, Department of Genetics, University of Delhi South Campus
2008-2012	Prof-in-charge, Central Instrument facility, University of Delhi South Campus
2008-2014	Team leader, Centre of Excellence on Genome Science and Predictive Medicine (Phase I)
2006-2008	Vice President, Indian Society of Cell Biology
2013 –2016	Member, Special committee of Special Centre for Molecular medicine, JNU
2011-	Member, The Netherlands International IBD Genetics Consortium
2011-	Member, The Netherlands International Celiac Disease Consortium
2012-2014	Member, Research Advisory board of National Council of Science museums
2011-2014	Member, National Women Bioscientists Awards Committee, DBT
2014- 2015	Member, Human Development and Disease Biology Task Force, DBT
2009-2012	Expert Member, Pharmacogenetics Task Force, ICMR
2003- 2014	Member, Human Genetics and Genome Analysis Task Force, DBT
2009-2010	UGC-SAP DRS member, Department of Biochemistry, UDSC
VC nominee/Univ Rep, Governing Body, DU Departments/Colleges	
2014-2016	Member, Board of Research Studies Faculty of Medical Sciences
2012-2015	Lady Harding Medical College
2011-2013	Departmental Research Committee, Department of Home Science
2011-2012	Acharya Narendra Dev college
2010-2012	Sri Venkateshwara College
2010-2012	Departmental Research Committee, Department of Anthropology
2007-2008	Amar Jyoti Institute of Physiotherapy
2004-2006	Raj Kumari Amrit Kaur College of Nursing
2004-2007	Lady Irwin College
Others	
2014-2016	EC nominee, Management Committee of Gitanjali hostel for women
2009-2011	EC nominee, Management Committee of Delhi school of social work hostel
Research Interests / Specialization	
<ul style="list-style-type: none"> • Molecular genetic analysis of complex disorders in humans (Schizophrenia, Parkinson’s disease, Rheumatoid arthritis, Inflammatory bowel disorders) • Pharmacogenetics of commonly used antipsychotic, anti-PD, anti rheumatoid drugs • Identification of new gene(s) for X-linked mental retardation and Parkinson’s disease • Generation of control and disease associated SNP databases for Indian population • Functional genomics: Genotype-phenotype correlations • Ayurgenomics • Molecular diagnostics for genetic disorders 	
Teaching Experience (Subject / Courses Taught)	
<ul style="list-style-type: none"> • Human Genetics • Human and Medical Biotechnology • Cytogenetics • Recombinant DNA technology • Prokaryotic and Eukaryotic gene expression 	

<ul style="list-style-type: none"> • Molecular systematics and evolution • Cell biology • Functional genomics 	
Honors & Awards	
2015	The Sanghvi Oration Award, ISHG
2014	The Sunder Lal Hora Medal, INSA
2012	Stree Shakti Science Samman
2011	JC Bose Fellow
2011	Member, Science and Engineering Research Board (SERB)
2009	Member, Scientific Advisory Committee to Prime Minister
2009	Fellow, Indian National Science Academy
2006	Fellow, Indian Academy of Sciences, India
2003	Fellow, The National Academy of Sciences, India
2008	Centre For Excellence In Genome Sciences And Predictive Medicine (GESPREM)- Team Leader , Funded by Department of Biotechnology, Govt. of India
2008	Trustee, XV International Genetics Congress Trust
2006	Vice President, Indian Society of Cell Biology
2003	Member, Human Genetics Task Force, Dept. of Biotechnology Expert Member, Indian Council of Medical Research
2002	Elected member, Guha Research Conference
2006	Visiting scientist, Max Planck Institute for Molecular Genetics, Berlin, Germany
2001	Visiting Scientist, Dept. of Psychiatry, Hadassah Medical Centre, Jerusalem, Israel
2000	Visiting scientist, Dept. of Psychiatry, Western Psychiatric Institute and Clinic, University of Pittsburgh, Pittsburgh, USA
1997	Fogarty International Research Career Award- Univ. Pittsburgh, USA
1995	Visiting Scientist- Dept. of Human Genetics, Memorial Sloan Kettering Cancer Centre, New York, USA
1991	Biotechnology Associateship- Institute of Human Genetics, Berlin, Max-Planck Institute, Munich, and Physical Chemistry Institute, Heidelberg, Germany
1989	Biotechnology Associateship- Centre for Cellular and Molecular Biology, Hyderabad
RESEARCH GUIDANCE:	
Ph.D. Students : Awarded 22 Registered 8	
M.Phil. Students : Awarded 4	
Total Publications : 110	
I. Research papers published in Refereed/Peer Reviewed Journals: (LAST FIVE YEARS)	
<ol style="list-style-type: none"> 1. Mittal K, Kirola L, Hegde S, Shetty M, Kabra M, Thelma BK. A novel de novo heterozygous deletion at 13q14.2-q21.1 in two siblings with mild intellectual disability Gene Reports (2018). S2452-0144(18)30073-6.DOI: doi:10.1016/j.genrep.2018.06.011 2. John J, Sharma A, Kukshal P, Bhatia T, Nimgaonkar VL, Deshpande SN, Thelma BK Rare variants in Tissue Inhibitor of Metalloproteinase 2 (TIMP2) as a risk factor for schizophrenia: Evidence from familial and cohort analysis (Schizophrenia bulletin 2018; doi: 10.1093/schbul/sbx196) 3. Kashyap S, Kumar U, Pandey AK, Kanjilal M, Chattopadhyay P, Yadav C, Thelma BK. Functional characterisation of ADP ribosylation factor-like protein 15 in rheumatoid arthritis synovial fibroblasts. (Clin Exp Rheumatol. 36(4):581-588 (2018) 4. Sood A, Midha V, Makharia G, Thelma BK, Halli SS, Mehta V, Mahajan R, Narang V, Sood K, Kaur K. A simple phenotypic classification for celiac disease. Intest Res. 16(2):288-292. (2018) 5. Singh A, Beniwal RP, Kukshal P, Bhatia T, Thelma BK, Deshpande SN. A preliminary study of association of genetic variants with early response to olanzapine in schizophrenia. Indian J Psychiatry. 60(1):10-16 (2018). 	

6. Prasad S, Bhatia T, Kukshal P, Nimgaonkar VL, Deshpande SN, **Thelma BK**. Attempts to replicate genetic associations with schizophrenia in a cohort from north India. **NPJ Schizophr.** 3(1):28. (2017)
7. Panchaichira TJ, Dey SK, Mukhopadhyay A, Kundu S, **Thelma BK**. Characterization of SNPs in the dopamine- β -hydroxylase gene providing new insights into its structure-function relationship. **Neurogenetics** 18(3):155-168. (2017)
8. Chaudhuri Z, John J, Aneja S, Thelma BK. Pedigree Analysis of Familial Primary Concomitant Horizontal Strabismus in Northern India. **Strabismus.** 26(7):311-23 (2017)
9. John J, Kukshal P, Bhatia T, Chowdari KV, Nimgaonkar VL, Deshpande SN, **Thelma BK**. Possible role of rare variants in Trace amine associated receptor 1 in schizophrenia. **Schizophr Res.** 189:190-195 (2017)
10. Chowdhury MR, Chauhan S, Dabral A, **Thelma BK**, Gupta N, Kabra M. Validation of Polymerase Chain Reaction-Based Assay to Detect Actual Number of CGG Repeats in FMR1 Gene in Indian Fragile X Syndrome Patients. **J Child Neurol.** 32(4):371-378. (2017)
11. Dey SK, Prabhakar P, Saini M, Joseph T, **Thelma BK**, Maulik SK, Kundu S. LBOS 02-05 Inhibitors of dopamine- β -hydroxylase obtained by structure based methods exhibited anti-hypertensive effect in l-name induced hypertensive rats. **J Hypertens.** Sep;34 Suppl 1:e550. (2016)
12. Abbas MM, Govindappa ST, Sudhaman S, **Thelma BK**, Juyal RC, Behari M, Muthane UB Early Onset Parkinson's disease due to DJ1 mutations: An Indian study. **Parkinsonism Relat Disord.** 32:20-24 (2016)
13. Gupta A, Juyal G, Sood A, Midha V, Yamazaki K, Vila A V, Esaki M, Matsui M, Takahashi A, Kubo M, Weersma R K, **Thelma B K**. A cross-ethnic survey of CFB and SLC44A4, Indian ulcerative colitis GWAS hits, underscores their potential role in disease susceptibility. **Eur J Hum Genet,** 25(1):111-122 (2016)
14. Kirola L, Behari M, Shishir C, **Thelma B K**. Identification of a novel homozygous mutation Arg459Pro in SYNJ1 gene of an Indian family with autosomal recessive juvenile Parkinsonism. **Parkinsonism Relat Disord.** 201631:124-128. (2016)
15. John J , Bhatia T, Kukshal P, Chandra P, Nimgaonkar VL, Deshpande S N , **Thelma BK** Association study of MiRSNPs with schizophrenia and related traits **Schizophrenia research** . 174, (1-3), 29-34. (2016)
16. Panchaichira TJ, Prasad S, Deshpande SN, **Thelma BK** Deep sequencing identifies novel regulatory variants in the distal promoter region of the dopamine- β -hydroxylase gene. **Pharmacogenet Genomics.** 26(7):311-23 (2016)
17. Gogtay NJ, Kapileshwar SR, Shah SU, Bendkhale SR, Ramakrishna S, Sridharan K, **Thelma BK**, et al., Evaluation of cytochrome P4502E1 polymorphisms in healthy adult Western Indians and patients with antituberculous drug-induced hepatotoxicity. **Indian J Pharmacol.**48(1):42-6 (2016)
18. Sudhaman S, Muthane UB, Behari M, Govindappa ST, Juyal RC, **Thelma BK** Evidence of mutations in RIC3 acetylcholine receptor chaperone as a novel cause of autosomal-dominant Parkinson's disease with non-motor phenotypes **J Med Genet** 53(8):559-66 (2016)
19. Sudhaman S, Prasad P, Behari M, Muthane UB, Juyal RC, **Thelma BK**. Discovery of a frameshift mutation in podocalyxinlike (PODXL) gene, coding for a neural adhesion molecule, as causal for autosomal-recessive juvenile Parkinsonism **J Med Genet.** 53(7):450-6 (2016)
20. Gupta A and **Thelma BK** Identification of critical variants within SLC44A4, an ulcerative colitis susceptibility gene identified in a GWAS in north Indians **Genes and Immunity**17(2):105-9 (2015).
21. Liu JZ, Sommeren SV, Huang H, Ng SC, Alberts R, Takahashi A, Ripke S, Lee JC, Jostins L, Shah T, Abedian S, Cheon JH, Cho J, Dayani NE, Franke L, Fuyuno Y, Hart A, Juyal RC, Juyal G, Kim WH, Morris AP, Poustchi H, Newman WG, Midha V, Orchard TM, Vahedi H, Sood A, Sung JY, Malekzadeh R, Westra HJ, Yamazaki K, Yang

- SK, The International Multiple Sclerosis Genetics Consortium, The International IBD Genetics Consortium, Barrett JC, Alizadeh BZ, Parkes M, **Thelma BK**, Daly MJ, Kubo M, Anderson CA, Weersma RK Association analyses identify 38 susceptibility loci for inflammatory bowel disease and highlight shared genetic risk across populations. **Nat Genet.** 47(9):979-86 (2015)
22. Juyal G, Negi S, Sood A, Gupta A, Prasad P, Senapati S, Zaneveld J, Singh S, Midha V, van Sommeren S, Weersma RK, Ott J, Jain S, Juyal RC, **Thelma BK** Genome-wide association scan in a north Indian cohort identifies three novel HLA independent susceptibility loci in ulcerative colitis **GUT** 64:571–579 (2015)
 23. Senapati S, Gutierrez-Achury J, Sood A, Midha V, Szperl A, Romanos J, Zhernakova A, Franke L, Alonso S, **Thelma BK**, Wijmenga C, Trynka G. Evaluation of European coeliac disease risk variants in a north Indian population. **Eur J Hum Genet.** 23,530-535 (2014)
 24. Juyal G, Mondal M, Luisi P, Laayouni H, Sood A, Midha V, Heutink P, Bertranpetit J, **Thelma BK**, Casals F. Population and genomic lessons from genetic analysis of two Indian populations. **Hum Genet.** 133(10):1273-87 (2014).
 25. Sood A, Amre D, Midha V, Sharma S, Sood N, Thara A, Bansal M, Juyal G, **Thelma BK**, Seidman E. Low hygiene and exposure to infections may be associated with increased risk for ulcerative colitis in a North Indian population. **Ann Gastroenterol.** 27(3):219-223 (2014)
 26. Taur S R, Kulkarni N B, Gandhe P P, **Thelma BK**, Ravat S H, Gogtay N J, Thatte U M. Association of polymorphisms of CYP2C9, CYP2C19, and ABCB1, and activity of P-glycoprotein with response to anti-epileptic drugs. **J Postgrad Med** 60:265-9 (2014)
 27. Laayouni H, Oosting M, Luisi P, Ioana M, Alonso S, Ricaño-Ponce I, Trynka G, Zhernakova A, Plantinga TS, Cheng S, Meer JWM, Popp R, Sood A, **Thelma BK**, Wijmenga C, Joosten LAB, Bertranpetit J, and Netea MG Convergent evolution in European and Roma populations reveals pressure exerted by plague on Toll-like receptors **Proc Natl Acad Sci** 111(7), 2668-2673 (2014).
 28. Senapati S, Singh S, Das M, Kumar A, Gupta R, Kumar U, Jain S, Juyal RC and **Thelma B.K** Genome-wide analysis of Methotrexate pharmacogenomics in rheumatoid arthritis reveals multiple novel risk variants and leads for TYMS regulation **Pharmacogenet Genomics.** 24(4):211-9 (2014).
 29. Antony Michealraj K, Jatana N, Jafurulla M, Narayanan L, Chattopadhyay A, **Thelma BK**. Functional characterization of rare variants in human dopamine receptor D4 gene by genotype-phenotype correlations. **Neuroscience.** 262:176-89 (2014).
 30. Thenral S. Geetha , K.Antony Michealraj, Kabra M, Kaur G, Juyal RC, **B.K. Thelma** Targeted deep resequencing identifies MID2 mutation for X-linked Intellectual disability with varied disease severity in a large kindred from India **Hum Mutat.** 35(1):41-4 (2014)
 31. Kukshal P, Chowdari KV, Srivastava V, Wood J, McClain L, Bhatia T, Bhagwat A M, Deshpande SN, Nimgaonkar VL, **Thelma BK**. Dopaminergic gene polymorphisms and cognitive function in a north Indian schizophrenia cohort **J Psychiatr Res.** 47(11):1615-22 (2013).
 32. Negi S, Juyal G, Senapati S, Prasad P, Gupta A, Singh S, Kashyap S, Kumar A, Kumar U, Gupta R, Kaur S, Agrawal S, Aggarwal A, Ott J, Jain S, Juyal RC, **Thelma BK**. A genome-wide association study reveals ARL15, a novel non-HLA susceptibility gene for Rheumatoid arthritis in north Indians. **Arthritis Rheum.** 65(12):3026-35 (2013).
 33. Sudhaman S, Behari M, Govindappa ST, Muthane UB, Juyal RC, **Thelma BK**. VPS35 and EIF4G1 mutations are rare in Parkinson's disease among Indians. **Neurobiol Aging.** 34(10):2442.e1-3. (2013).
 34. Kukshal P, Bhatia T, Bhagwat AM, Gur RE, Gur RC, Deshpande SN, Nimgaonkar VL, **Thelma BK** Association study of Neuregulin-1 gene polymorphisms in a north Indian schizophrenia sample **Schizophrenia Research** 144(1-3):24-30 (2013)

II. Other than refereed /Peer Reviewed Journals

1. Kumar S, Yadav N, Pandey S, **Thelma BK** Advances in the discovery of genetic risk factors for complex forms of neurodegenerative disorders - contemporary approaches, success, challenges and prospects *Journal of Genetics* 97:3 625-648(2018).
2. Kapoor S, **Thelma BK**. Status of Newborn Screening and Inborn Errors of Metabolism in India. *Indian J Pediatr*. doi: 10.1007/s12098-018-2681-5(2018).
3. **Thelma BK**, Srivastava V, Tiwari AK Genetic underpinnings of Tardive dyskinesia: Passing the baton to Pharmacogenetics. *Pharmacogenomics* 9:1285-306 (2008)
4. **Thelma BK** and R.C.Juyal, Molecular genetics of Parkinson's disease: Hopes and realisations .In: Brain disorders across life span: Hopes and challenges. Ranbaxy science foundation – Round table conference series, New Delhi, India (14) (2004)
5. **Thelma BK**, Prasad S, Sharma D Complexity of some complex traits. *PINSA B68 No.5 pp479-496 (2002)*
6. Prasad S, Semwal P, Deshpande S, Bhatia T, , Nimgaonkar V L, and **Thelma B K**. Molecular Genetics of Schizophrenia: Past, present and future. *J. of Biosciences* : 27(1) 101-118 (2002)
7. SN Deshpande, **BK Thelma**, VL Nimgaonkar Genetics of Schizophrenia: The Search for Markers *JOSH Vol. VII:61-64 (2000)*
8. **Thelma BK** and Ramesh C. Juyal ;Genetic Basis of Parkinson's Disease. Proceedings of 6th National Workshop on Movement disorders, AIIMS, New Delhi, pp27-46 (1999)
9. **Thelma BK** Human Genetics. Chapter In: National Syllabus of Genetics and Plant Breeding M.C. Kharakwal and R.B. Mehra (Eds) pp.121-131(2001)

Books

1. **T. Kuttapa**, Assistant Editor, Familial cancer, (Muller, Weber, eds.) Karger 1985.
2. **Thelma BK** From Genomics to 'Ayurgenomics'. In: *PHISPC series on History of Science, philosophy and culture in Indian Civilisation Volume: Life and Organicism*, ed N.S. Rangaswamy, Gen. Editor D.P.Chattopadhyaya (2008)

Conference Organised

1. *Organization of a Conference* "Trends in Human Genetics", August 20- 22, 2007, Puri, Orissa, India
2. *Symposium titled* "Recent advances in molecular genetics with new biomedical insights", jointly organized by INSA, New Delhi and the Israeli Academy of Sciences and Humanities held at INSA, New Delhi, February 12- 13, 2018; Funded by IASH, Israel; and INSA, DBT, India

Professional Societies Memberships

1. *Reviewing* – Reviewing manuscripts for several Journals
2. *Memberships* – Life member, Indian Society of Cell Biology and Indian Society of Human Genetics
Life Member, Indian Society of Human Genetics

Project (Major/Grants/Collaborations)

ONGOING RESEARCH PROJECTS:

1. "Centre For Excellence In Genome Sciences And Predictive Medicine (GESPREM)- PHASE II: Systems biology of complex diseases: From genetic findings to lead molecule development for Rheumatoid arthritis" Funded by Department of Biotechnology, Govt. of India
2. "Identification of rare variants and copy number variations in schizophrenia by affected sib pair analysis using next generation sequencing tools" (Principal Investigator)Funded by Dept. of Biotechnology, Govt. of India
3. "J C Bose fellowship" funded by Science and Engineering Research Board, Govt. of India
4. "The Indian Movement Disorder Registry and Biobank: Clinical and genetic evaluation of movement disorders in Indian patients -[26428]" (Co-Principal Investigator) Funded by Department of Biotechnology, Govt. of India
5. "Biomarker for early diagnosis of neonatal sepsis as point of care testing using biosensors", Indo-Spanish Project, (Co-Principal Investigator) Funded by Department of Biotechnology, Govt. of India

COMPLETED RESEARCH PROJECTS: TWENTY THREE

In the last five years:

1. “ A novel feasibility study of newborn screening for the treatable disorders and epidemiologic data generation for inborn metabolic errors in Delhi state” (Principal Investigator) funded by Science and Engineering Research Board, Govt. of India
2. “Unravelling Monogenic Causes of Mental Retardation Using Contemporary tools ” (Principal Investigator) Funded by Department of Biotechnology, Govt. of India
3. “Continuing research for Novel causative gene(s) for Parkinson’s disease” (Principal Investigator) Funded by Department of Biotechnology, Govt. of India
4. Centre For Excellence In Genome Sciences And Predictive Medicine (GESPREM)-
“ Whole Genome Association Analyses in Complex Diseases: An Indian Initiative” (Team Leader)
Funded by Department of Biotechnology, Govt. of India
5. “Functional characterization of SNPs/SNP haplotypes in candidate genes” (Principal Investigator)
Funded by Department of Biotechnology, Govt. of India
6. “Molecular Diagnostics: Development of Cost Effective Pre-prescription and Pre-implantation Genetic Testing” (Principal Investigator) Funded by DST-PURSE grant
7. “Pharmacogenetics of Adverse Drug Reaction : Role of drug metabolizing enzymes and VKORC1 polymorphisms in reducing drug toxicity due to antitubercular drugs and Warfarin” Funded by Department of Biotechnology, Govt. of India
8. “Identification of Novel Gene(s) for familial Parkinson’s Disease” Funded by Department of Biotechnology, Govt. of India
9. “Genetic Susceptibility to Rheumatoid Arthritis using a novel combination of Prakriti based case-control selection and Molecular Analysis Tools” Funded by Ministry of Ayush, Govt. of India
10. “An exploratory analysis of genetic correlates and effects of Yoga on circadian rhythms, cognitive functions and social burden in major mental disorders: schizophrenia, bipolar disorder and depression and their comparison with a cardiac group” Funded by Ministry of Ayush, Govt. of India
11. “Methotrexate in Rheumatoid Arthritis: Pharmacogenetics & Clinico-Immunological correlates” Funded by Department of Biotechnology, Govt. of India
12. “An integrated approach for mutation analysis and identification of new gene(s) for X-linked mental retardation” Funded by Department of Biotechnology, Govt. of India
13. “Pharmacogenetics of anti-epileptic drugs role of ABCB1, CYP2C9, CYP2C19 and SCN2A in optimizing therapy” (Co-Investigator) Funded by Department of Biotechnology, Govt. of India