JSS ACADEMY OF HIGHER EDUCATION & RESEARCH
(DEEMED TO BE UNIVERSITY) MYSURU

Special Interest Group

On

Human Genomics and Rare Disorders

An Initiative
of
Research Division
JSS AHER, Mysuru-15.

Team Lead
Dr. Akila Prashant
Professor and Head
Department of Biochemistry
JSS Medical College,
JSS AHER, Mysuru

Started on 2018-19

JSS Academy of Higher Education & Research
(Deemed to be University)
Accredited "A" Grade by NAAC

Ref. No. REG/ACA/SIG/225/2018-19/ 1742.

Date: 24.05.2018

NOTIFICATION

Sub: Special Interest Group on" Human Genomics and Rare Disorders".
Ref: Your letter No. JSSMC/BIO/1056/2018-19 dated 15/05/2018.

Preamble: The constituent Colleges of JSS Academy of Higher Education & Research have shown keen interest on nurturing /accelerating the research activities in their respective colleges and requested to setup a Special Interest Group (SIG) in specific areas of interest/research for preparing research proposals for submitting to various National/International funding agencies for funding. The proposal of formation of SIG is approval in the 9th Board of Management meeting held on 26th November 2010.

Guidelines for the formation of Special Interest Groups (SIG):

Objectives: Top promote research in specific areas by a group of individuals working in the constituent colleges of the JSS AHER, who are desirous of promoting research in a specific areas through.

- Continuous dialogue
- Conducting meetings
- Submitting projects
- Development of new ideas in research in the identified area and translating the same into action/research
- Translating research findings into the clinical applications
- Organizing seminars

The SIG should categorize their objectives as short term and long term objectives and while submitting the progress reports, highlight their achievements under the same.

Structure: The SIG consist of 5-7 members with a group leader. They shall meet periodically and submit the report annually of the group activities and achievements.

The Special Interest Group shall function for a period of 3 years from the date of Notification, which can be extended further, based on the outcomes and reviewed by the Board of Management of the JSS University.

Budget: The budget required for functioning of the SIG shall be allocated from Research and Development fund of the JSS Academy of Higher Education & Research based on the budget prepared and submitted by them annually.

The Budget shall be provided for following aspects:

- Meeting expenses
- Short travels within India for the purpose of group objective.
- Secretarial assistance as required
- Preparation of project proposals/reports

The AHER shall provide in its budget for the expenditure proposed by the Special Interest Groups and make the funds available to the respective group.

Evaluation of outcomes: The AHER will evaluate the SIG periodically in respect to their group objective examining the following:

- Progress in the proposed research projects.
- Publication of research findings in periodicals and presentation.
- Reports.

Special Interest Group (SIG) – Human Genomics and Rare Disorders

The Special Interest Group (SIG) in the area of **"Human Genomics and Rare Disorders"** has been setup for focusing on basic and applied research in the area of Public Health with the following members:

SL.No.	NAME	REMARKS
1	**Dr. Akila Prashant** Professor, Dept. of Biochemistry JSS Medical College, Mysuru.	Group Leader
2	**Dr. Suma M N** Professor and Head, Dept. of Biochemistry JSS Medical College.	Member
3	**Dr. Praveen D** Consultant Biochemist, JSS Hospital.	Member
4	**Dr. Shobha C R** Tutor, Dept. of Biochemistry JSS Medical College.	Member
5	**Dr. Deepa Bhat** Associate Professor, Dept. of Anatomy JSS Medical College.	Member
6	**Dr. Ravi M D** Professor, Dept. of Paediatrics JSS Medical College.	Member
7	**Dr. Srinivasa Murthy D** Professor, Dept. of Paediatrics JSS Medical College.	Member
8	**Dr. Sunil Kumar S** Assistant Professor, Dept. of Cardiology JSS Medical College.	Member
9	**Dr. Ravi K** Assistant Professor, Dept. of Surgical Oncology JSS Medical College.	Member

The SIG "**Human Genomics and Rare Disorders**" Shall function initially for a period of three years.

The SIG may submit their report annually for the approval of the JSS AHER. The budget for the functioning shall be submitted by the SIG and approval has to be obtained from the JSS AHER for carrying out their various activities.

REGISTRAR

To,

All the above members

Copy to:

1. The Vice Chancellor, JSS Academy of Higher Education & Research.
2. The Finance Officer, JSS Academy of Higher Education & Research.
3. The Controller of Examinations, JSS Academy of Higher Education & Research.
4. The Director (Academics), JSS Academy of Higher Education & Research.
5. The Director (Research), JSS Academy of Higher Education & Research.
6. The Deputy Registrar (Senior Grade), JSS Academy of Higher Education & Research.
7. The Principal, JSS Medical College.
8. The Principal, JSS Dental College & Hospital.
9. The Principal, JSS College of Pharmacy, Mysuru.
10. The Principal, JSS College of Pharmacy, Ooty.
11. The Chairman, Faculty of Life Sciences, JSS Academy of Higher Education & Research.
12. The Head, Faculty of Life Sciences, JSS Academy of Higher Education & Research.
13. The Coordinator, Dept. of Health System Management Studies, JSS Academy of Higher Education & Research.
14. Office copy.

Name of the SIG: Human Genomics and Rare Disorders

Approval Reference: JSSMC/BIO/1056/2018-19 dated 15/05/2018

Team Lead: Dr. Akila Prashant

Dr. Akila Prashant **Professor, Dept of Biochemistry** **JSS Medical College, Mysuru**	**Group Leader**
Dr. Suma M N **Professor, Dept of Biochemistry** **JSS Medical College, Mysuru**	**Member**
Dr. Shobha CR **Assistant Professor, Dept of Biochemistry** **JSS Medical College, Mysuru**	**Member**
Dr. Deepa Bhat **Professor, Dept of Anatomy** **JSS Medical College, Mysuru**	**Member**
Dr. Ravi M D **Professor, Dept of Paediatrics** **JSS Medical College, Mysuru**	**Member**
Dr. Srinivasa Murthy D **Professor, Dept of Paediatrics** **JSS Medical College, Mysuru**	**Member**
Dr. Sunil Kumar S **Assistant Professor, Dept of Cardiology** **JSS Medical College, Mysuru**	**Member**
Dr. Ravi K **Assistant Professor, Dept of Surgical Oncology** **JSS Medical College, Mysuru**	**Member**

Objectives

- Facilitate early diagnosis, counselling and management of rare disorders
- Prepare SOPs that facilitate multidisciplinary approach to any genetic disorders
- Organise symposia/workshop/seminars
- Conduct collaborative research projects
- Promote research activities among group members by group discussion and expert advises
- Work together towards establishing a biobank to store clinical specimens of rare inherited disorders

Activities Conducted

Workshops/Seminars:

- Pre-conference workshop on "Basic and advanced techniques in pharmacogenomics"

The Department of Medical Genetics and the Department of Biochemistry, JSS Medical College and Hospital, JSS Academy of Higher Education & Research, Mysuru organized a pre-conference workshop on "Basic and advanced techniques in pharmacogenomics" on 22-02-2023 as a part of IPSCON organized by JSS College of Pharmacy, Mysuru. Twenty-four delegates from various medical and pharmacy colleges participated in this workshop. After the inaugural session at Pharmacy college, participants left for the Dept. of Medical Genetics, JSS hospital for the workshop. The workshop consisted of lectures as well as hands-on sessions on different techniques which are generally used in pharmacogenomics.

Pre-Ethiopian Food and Drug Authority (EFDA)

Department of Medical Genetics and Biochemistry, JSS Medical College hosted a two-day workshop from 18th – 19th December 2023, for the officials of the Ethiopian Drug and Food Authority (EFDA). The objectives of the workshop were to equip and train the participants with different molecular techniques to evaluate Rapid Diagnostic Test (RDT) performances. We could demonstrate DNA isolation, Real-time PCR assay, ELISA, and the associated data analysis of these techniques. The faculties and scholars of both departments participated enthusiastically in the workshop and made it a success. The FDA officials responded positively as feedback of the training. We look forward to engaging in a continuous learning and growth process through conducting more such programs in the future. We thank JSSAHER and Vice-chancellor Dr. Surinder Singh for providing us with this wonderful opportunity.

Clinical, Epidemiological and Experimental Approaches to Assess Air Pollution Related Chronic Health Effects'

Centre of Excellence in Molecular Biology & Regenerative Medicine laboratory, Department of Biochemistry in association with the Department of Medical Genetics organized a three day workshop on **Molecular Biology, Cell Culture and Cytogenetics** from 8th to 10th January 2020. This workshop was a part of post conference activity, the conference theme being **'Clinical, Epidemiological and Experimental Approaches to Assess Air Pollution Related Chronic Health Effects'.** A total of 12 participants from in-and around Mysuru enlisted and actively took part in the workshop.

Guest Lecture

- Guest lecture on "Role of Genomics in Precision Medicine" **by Dr. Suma P.** Shankar, Associate Professor, Director Precision Genetics, Department of Pediatrics and Ophthalmology, UC Davis Health, USA was held at Sri Rajendra Centenary Auditorium, JSS Hospital on 21st August 2018

- Dr. Dhavendra Kumar, Chair- UK India Genomic Medicine Alliance, Director- The Genomic Medicine Foundation (UK); Consultant In-charge: The Genome Clinic Spire Cardiff Hospital Pentwyn, Cardiff, UK on Mendelian Genomics on 13th February 2021.
- Dr. Jayaram S Kadandale, Professor, Centre for Human Genetics, Bangalore "Clinical applications of FISH" 3rd February 2021
- Dr. Jayaram S Kadandale, Professor, Centre for Human Genetics, Bangalore "Applications of FISH in Prenatal Diagnosis and confirming microdeletion syndrome" 10th February 2021

- Dr. Jayaram S Kadandale, Professor, Centre for Human Genetics, Bangalore "Advanced Molecular Cytogenetic Techniques" 13th February 2021
- Dr. Jayaram S Kadandale, Professor, Centre for Human Genetics, Bangalore "In vitro transformation of human peripheral B-lymphocytes by Epstein-Barr virus to establish lymphoblastoid cell lines (LCLs)" 20th July 2021
- S.Sunil Kumar (Manager- Specialty Services, Eppendorf India Pvt Ltd)- Eppendorf Pipette Calibration (EpiC) program on 12-01-2023.
- Dr. Senthilnathan Senguttuvan (Head, Application Scientist Team, Qiagen India Pvt Ltd) - Introduction to Digital PCR on 13-01-2023.
- Amal George (Regional Head – Acquisition Sales -South), Meenakshi Verma (Marketing lead -India) and Vinay Kumar Janga (Regional Head – Technical Application Specialist – South) - Immunoassay, Clinical Chemistry, Haematology, Informatics, Total Lab Automation on 25-01-2023.
- Mr Onkar Rahatkar (Product specialist for Next Generation Sequencing and Automation) – HIVE CLX: AN END-TO-END SOLUTION FOR SINGLE-CELL RNAseq 1-06-2023.

Guest Lecture Delivered

- "Prenatal and newborn diagnosis" at Genomics in current day practice on 31st October 2019 at JSS Hospital, Mysuru
- Dr. Akila Prashant & Dr. Prashant Vishwanath were invited speakers in the CME on Inborn Errors of Metabolism – An approach towards diagnosis and management held on 29th November 2019 at Karwar Institute of Medical Sciences, Karwar – attended by 50
- 2nd Annual State-Level Emergency Medicine Conference on 18th of January, 2020 Topic: Inborn Errors of Metabolism and the ER as a Chairperson
- Dr. Akila P. COVID-19 and vaccines in the International Webinar on "Immunity in Today's world" organized by St. Philomena's College, Mysuru on September 12, 2020.
- Dr. Akila P. Anemia – Research base for public policy decision making, in the National e-conference on Anemia Summit 2021 organized by Department of Biochemistry, Chettinad Hospital and Research Institute (CHRI), Chengalpattu on 25th February 2021.

- Dr. Akila. Diagnostic and therapeutic implications of micro RNA in human diseases, in National e-Conference and Workshop, Genomic Conclave 2021 held on 27th & 28th May 2021, organized by Chettinad Hospital and Research Institute (CHRI).
- Dr. Akila P. Genetic aspects in Diabetes mellitus in the International Webinar entitled "Killing the Killer – Diabetes a Kelideoscope held from 26th to 29th June 2021, organized by the Special Interest Group – Diabetes & Oral Care of JSS Dental College & Hospital.
- Dr. Akila P. Software in references and reviews in the International Virtual Conference – "Time to untangle research held from 12th to 14th August 2021, organized by JSS College of Nursing.
- Dr. Akila P. Importance of newborn screening, in Virtual CME on Update on Diagnostics in Medical Genetics held on 18th August 2021 organised by Raipur Institute of Medical Sciences, Raipur.
- Dr. Akila P. Universal newborn screening: need of the hour. Oration in Cerebration the 3rd Colloqium 2021 held on 24th & 25th September 2021 organised by All India Institute of Medical Sciences, Mangalagiri.
- Dr. Akila P. Competency-based education to Outcome-based education: Means to attain higher-level Bloom's taxonomy in the international e-conference on "Exploring newer modalities in teaching - learning and Research in Basic Medical Sciences during Covid Era" held from 25th-27th November 2021 through online pedagogy by Aarupadai Veedu Medical College, Pondicherry.
- Dr. Akila P. Basics of PCR & Primer Designing. Indo-US workshop on Molecular Epidemiology of Infectious Diseases: Translational Research from Bench to Bedside to the Field held on 21st to 27th March 2022 at JSS Academy of Higher Education & Research under the DST STUTI Scheme.
- Dr. Akila P. Emerging Biological Mechanisms linking Obesity to Cancer. in the State Level CME titled "Current Trends in Cancer Biology" on 11th May 2022 by Sri Devaraj Urs Academy of Higher Education & Research Center, Kolar.
- Dr. Akila P. The scope and services available in the Department of Medical Genetics at the Academic Society meeting at JSS Hospital on 16th June 2022.

- Dr. Akila P. PCR – Basics and Primer Designing. Workshop on Cytogenetics, cell and molecular biology techniques held on 18th to 24th June 2022 at JSS Academy of Higher Education & Research under the DST STUTI Scheme.
- Dr. Akila P. Maternal serum screening in the pre-conference of the 29th Annual meeting of the Association of Medical Biochemists of India held at Vanivilas Women and Children Hospital, Bangalore between 18th to 20th July 2022.
- Dr. Akila P. Use of Karyotype in the diagnosis of chromosomal disorders in the 29th Annual meeting of the Association of Medical Biochemists of India held at Moongate Resort, Bangalore between 21st to 23rd July 2022.
- Dr. Akila P. Diagnostic utility of conventional cytogenetics in identifying chromosomal abnormalities in the First South Zone Conference of Indian Academy of Biomedical Sciences (IABSCON-SOUTH ZONE 2022) held at University of Mysore on 8th & 9th September 2022.
- Dr. Akila P. Think, Articulate and Pen-down: Tips for successful grant writing, in the CME program organized Hassan Institute of Medical Science, Hassan on 19th October 2022.
- Dr. Akila P. Medical Research. Guest lecture in the Department of Pathology, JSS Medical College held on 7th December 2022.

Seminar

- One day seminar on “Rare Diseases-Bridging Health and Social Care” was held on 28th Feb 2019 at Sri Rajendra Centenary Auditorium, JSS Hospital. The brochure highlighting the importance of newborn screening was released on this occasion.




- The SIG-HGRD, JSS Medical College, JSS Academy of Higher Education & Research (JSSAHER) in association with 'Indo-UK Genomic Education forum' of the Genomic Medicine Foundation (UK) had organized one day seminar on "The Impact of Human Genomics in Current Medical Practice" on Thursday 31st October 2019 at Sri Rajendra Centenary Auditorium, JSS Hospital, Mysuru. Maternal serum screening brochure was released on that occasion.

Symposium

- The Department of Biochemistry & Paediatrics, JSS Medical College had jointly organized a one-day symposium on 'Rare Diseases – Bridging Health and Social Care' on 28th February 2019 as a part of International Rare Disease Day through the Special Interest Group on Human Genomics and Rare Disorders at Sri Rajendra Centenary Auditorium, JSS Hospital Mysuru.

Conference

- 5th International Conference on Birth Defects (ICBD 2022)21st-23rd November 2022:The Department of Biochemistry and Department of Medical Genetics, JSS Medical College and Hospital, JSS Academy of Higher Education & Research Mysuru organized a 3-day International Conference on Birth Defects on the theme of "Precision Medicine For Birth Defects" from 21st to 23rd November 2022. It was co-organized by JSS Academy of Higher Education and Research, Mysuru, Karnataka, India & UK India Genomic Medicine Alliance Indo UK Genetic Education Forum with partial Funding from the Department of Science & Technology Promotion of University Research and Scientific Excellence (DST-PURSE).

Inauguration of Rare Disease Care Coordination Center on 25th January 2019 at the Paediatrics ward of JSS Hospital

This would be second center in entire India to provide support to patients and caregivers of rare diseases with information, connection with experts' education and awareness programs to patients and physicians. This will help in accelerating diagnosis and management. This center was inaugurated by Dr C G Betsurmath Executive Secretary of JSS Mahavidyapeeta, Dr B Suresh, Vice Chancellor of JSS Academy of Higher Education and Research, Dr B.Manjunatha Registrar, JSSAHER, Dr H. Basavanagowdappa, Principal, of JSS Medical College. Dr (Col) M. Dayananda Director, JSS Hospital. Dr.P.A Kushalappa, Director (Academics), Mr.Prasanna Shirol, Executive Director of ORDI inaugurated this program.

Awareness Programs

- Red FM program to spread awareness regarding rare diseases – 25th February 2019
- Awareness on Rare Diseases: Race for 7 – marathon to create awareness among public and support rare disease families on 3rd March 2019

Awareness regarding Rare Genetic Diseases on 25th September 2019 Mathru Mandali Shishu Vikasa Kendra Special School for Children Mysore – attended by 45

- Awareness regarding Rare Genetic Diseases on 16th October 2019 Devdaan Foundation – attended by 30

- Awareness regarding Rare Genetic Diseases on 5th December 2019 Spastic Society of India – attended by 25.

- Spreading awareness on availability of genetic counselling and lab services at JSS Hospital on 1st June 2019 at Railway Hospital, Mysuru

Research Projects

Submitted

- "Methylation of SOCS3 gene and its relation with serum leptin levels in young adults with and without family history of obesity" submitted by Dr. Shobha CR to Vision Group of Science and Technology, Govt of Karnataka.
- "Identification of the Beneficial Mutations in selected candidate genes that prevent the occurrence of Chronic Obstructive Pulmonary Disease among smokers and their further use in Translational Research" submitted by Dr. Prashant V to Vision Group of Science and Technology, Govt of Karnataka.
- "DNA methylation pattern of CpG site in the promoter region of CALCA-alpha gene as a putative epigenetic biomarker in neonatal sepsis-a case control study in South India". Carried out as ICMR STS project by Rupesh R Reddy under the guidance of Dr. Prashant V
- Role of Bacopa monnieri in combating cognitive impairment caused due to AGE's through PI3K/Akt/Ataxin1 pathway in type 2 diabetes mellitus" submitted to STARS funding by Dr. Prashant V.
- We are one of the participating centers of a multicentric project by ICMR entitled "National registry for rare and other inherited disorders" PI Dr. Akila Prashant
- Proposal for Centre for Training, Research and Innovation in Tribal Health to India Alliance-DBT Wellcome trust by Dr. Deepa Bhat
- "To investigate the role of SOCS3 and related genes as a potential drug target in obesity induced metabolic syndrome"- Dr. Shobha CR to VGST RGS/F
- "Study the prevalence of metabolic diseases in newborns and understand the reasons for not complying with follow-up with healthcare providers"- Dr. Shobha CR to ICMR STS
- "DNA methylation status of obesity related genes and correlation of gene expression and cardio-metabolic parameters among young adults with and without family history of obesity"- Dr. Shobha CR to Medical Education & Research Trust(MERT)
- "Explore the diagnostic value of urinary proteins as early biomarkers of preeclampsia and develop a low-cost screening tool" by Dr. Sowmya K & Dr. Akila P to DBT.

- "Antenatal screening of Hemoglobinopathies and molecular diagnosis" by Dr. Rajalakshmi & Dr. Akila P to Lady Tata trust Mumbai
- "To establish Center of Excellence in Maternal Screening for Hemoglobinopathies" by Dr. Prashant & Dr. Akila to VGST
- Methylation of genes regulating adipogenesis (PPARγ, CEBPA, SREBF1) and its relation with serum Vitamin D in young adults with and without obesity – ICMR
- Establishment of DBT NIDAN Kendra and Screening of Aspirational district – DBT UMMID
- Establishment of Centre for Advanced Newborn Screening and Wellness (CANWe) – DBT India Alliance.
- To develop a cost-effective risk prediction model integrating clinical features and novel genetic signatures to predict severe COVID -19 infections among Indians. – ICMR
- The role of microRNAs in regulating the γ-globin expression and increasing the fetal hemoglobin levels in patients with beta-thalassemia.
- Netrin-I induced epithelial to mesenchymal transition in colorectal cancer and its possible use as a biomarker and therapeutic target – An exploratory study.
- SeptiScan Rapid and Affordable multiplex PCR based diagnostic solution for the identification of etiology of sepsis
- Studies on the role of small RNAs in epigenetic regulation of Erythroferrone – dependent hepcidin synthesis in beta-thalassemia
- Discover of small molecule that serve as a catalyst with supersede risdiplam for the development of cost-effective combitorial therapeutics for spinal muscular atrophy.
- BUILDER 2021 program- Development of low cost in house CYP2C9/VKORC1 gene polymorphism assay ans asses the clinical utility of CYP2C9/VKORC1 genotyping to guide personalizes anticoagulant therapy in patients with atrial fibrillation asa well as cardiac valve replacement
- PURSE 2021 program- Studies on the role of small RNAs in epigenetic regulation of erythroferrone- dependent hepcidin synthesis in β- thalassemia

Collaborations

- Dr. Dhavendra Kumar, Consultant in Clinical Genetics, Institute of Medical Genetics, University Hospital of Wales, Cardiff University School of Medicine, UK; The Visiting Professor, Genomic Policy Unit, Faculty of Life Sciences and Education, the University of South Wales, Pontypridd, South Wales, UK. has been added as an adjunct faculty of JSS AHER
- Dr. Giriraj Chandak, Group leader, CSIR-CCMB, Hyderabad and Dr. Jayarama S Kadandale, Professor & Head, Clinical and Molecular Diagnostics, Center for Human Genetics, Bangalore as adjunct faculty for our Institution.
- We have entered into MoU with Genomic Medicine Foundation (UK) in association with whom we have conducted conferences and workshops.

Projects and Patents

Ongoing/Completed Projects

- "Improving the capacity of health system and community for sickle cell disease screening and management: an intervention study" sanctioned as PI (Dr. Deepa Bhat) for National Task Force on Sickle Cell Anaemia by ICMR- Completed
- "Association of genotypic pattern of obesity related genes with the cardiometabolic parameters among young adults with and without family history of obesity" by JSS Academy of Higher Education & Research – Dr. Shobha C.R.
- We are one of the participating centers of a multicentric project by ICMR entitled "National registry for rare and other inherited disorders" PI Dr. Akila Prashant from ICMR
- Proposal to establish "Centre for Training, Research, and Innovation in Public Health" sanctioned with Dr. Deepa Bhat as Co-PI by DBT/Wellcome Trust India Alliance.

Patents/Prototypes:

- Multiplex PCR assay with cocktail of primer mix to identify pathogenic bacteria causing neonate sepsis, Patent No. 202041015935 granted.

Achievements

Publications

- Supriya Bevinakoppamath, Akila Prashant, Shobha C.R. An insight into the use of transgenic animal models for conducting research on coronavirus. Int J Health Allied Sci2020;9(5): 18-23□
- Bevinakoppamath S, Ramachandra SC, Yadav AK, Basavaraj V, Vishwanath P and Prashant A. Understanding the Emerging Link Between Circadian Rhythm, Nrf2 Pathway, and Breast Cancer to Overcome Drug Resistance. Front. Pharmacol. 2022;12:719631.IF:5.988
- H. C. Manju, Supriya Bevinakoppamath, Deepa Bhat, Akila Prashant, Jayaram S. Kadandale & P. V. V. Gowri Sairam. Supernumerary derivative 22 chromosomes resulting from novel constitutional non-Robertsonian translocation t(20;22) Case Report. Molecular Cytogenetics 2022;15:14. IF:1.506.
- Kalaigar SS, Rajashekar RB, Nataraj SM, Vishwanath P, Prashant A. Bioinformatic Tools for the Identification of MicroRNAs Regulating the Transcription Factors in Patients with β-Thalassemia. Bioinformatics and Biology Insights. January 2022IF: 2.063.
- Kalaigar SS, Rajashekar RB, Nataraj SM, Vishwanath P, Prashant A. Bioinformatic Tools for the Identification of MicroRNAs Regulating the Transcription Factors in Patients with β-Thalassemia. Bioinformatics and Biology Insights. 2022;16. IF: 2.063
- Manju, H.C., Bevinakoppamath, S., Bhat, D. et al. Supernumerary derivative 22 chromosome resulting from novel constitutional non-Robertsonian translocation: t(20;22)—Case Report. Mol Cytogenet 15, 14 (2022). IF: 1.506
- Bevinakoppamath S, Ramachandra SC, Yadav AK, Basavaraj V, Vishwanath P and Prashant A. Understanding the Emerging Link Between Circadian Rhythm, Nrf2 Pathway, and Breast Cancer to Overcome Drug Resistance. Front. Pharmacol. 2022;12:719631. IF: 5.988
- Yadav AK, Chakkumkollath AK, Helna A, et al. Substantiation of a clopidogrel metabolism-associated gene (CYP2C19) variation among healthy individuals. Indian Heart J. 2023;75(5):343-346. IF: 1.5.

- Birla S, Angural A, Madathumchalil A, Shende RV, Shastry SV, Mahadevappa M, Shambhu SK, Vishwanath P, Prashant A. Redefining the polypill: pros and cons in cardiovascular precision medicine. Front Pharmacol. 2023 Sep 19;14:1268119. IF: 5.988.
- Ramachandra, Shobha C; Prashant, Akila; Shivanna, Akshitha; Bhujanga, Ananth Koushik; Meherunnisa, Syeda; and Vishwanath, Prashant (2023) ""Cost-effective markers to identify metabolically healthy/unhealthy individuals and their future risk for cardiovascular disorders"," International Journal of Health and Allied Sciences: Vol. 12: Iss. 4, Article 7.
- NR Kumar, TA Balraj, SN Kempegowda, A Prashant. Multidrug-Resistant Sepsis: A Critical Healthcare Challenge. Antibiotics 2024; 13 (1), 46. IF: 4.9

Awards and Recognitions

- Dr. Vivek Anand Ojha, 2nd year post graduate student of Biochemistry won the second place in quiz competition held during the 4th International conference on birth defects & 5th Annual conference of the society of Indian Academy of Medical Genetics at CMC Vellore from 12-15th December 2018
- Selected as a member of advisory board during SANOFI GENZYME INDIA – SCIENTIFIC ADVISORY BOARD(SGI-SAB) at the meeting held on 25th May, 2019
- Dr. Vivek Anand Ojha, Postgraduate student, Department of Biochemistry, JSS Medical College was awarded the young scientist travel award of 1500 SGD in the 15th Asia Pacific Federation of Clinical Biochemistry Congress held between 17th-20th November 2019 at Jaipur, Rajasthan
- Best oral presentation for the research entitled “The expression of IREB2, CHRNA3 and CHRNA5 in smokers with and without Chronic Obstructive Pulmonary Disease” in the 5th State level Conference of Association of Medical Biochemists, Karnataka Chapter held on 20th and 21st of September 2019 at Mysuru Medical College and Research Institute, Mysuru.

Academic and Diagnostic Progress

- We have also initiated BSc Medical Genetics and MSc Medical Genetics & Genomics programs. 01 student is currently enrolled and has successfully completed her 1st semester.
- The newborn screening activity is continued and we have screened more than 5000 neonates.
- Maternal serum screening activity is continued

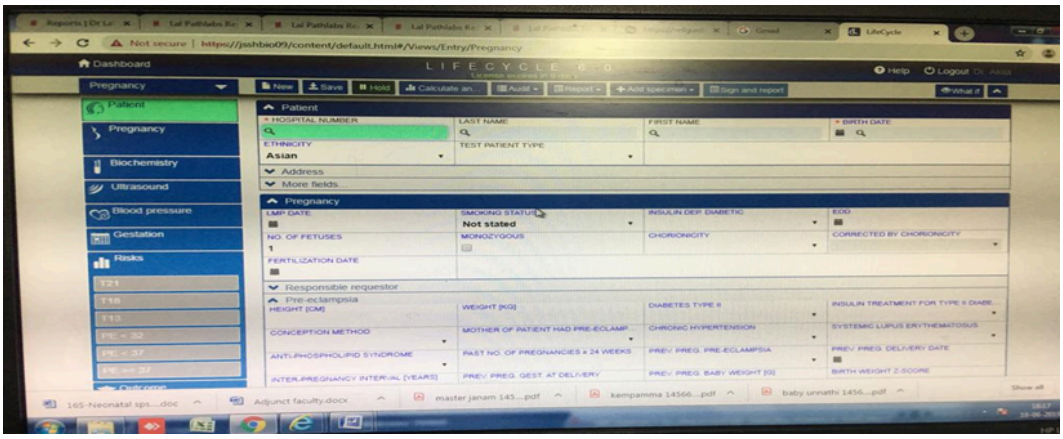
Patient care & Laboratory investigations

New Born Screening

We are screening all the newborns at JSS hospital for 7 disorders regularly. Since January 2019 we have screened more than 5000 newborns till date.We are now offering this service to screen all babies for selected disorders at JSS Hospital. In this program, the seven most common disorders encountered in the Indian population are screened in all newborns viz.: Congenital Hypothyroidism, G-6-PD deficiency, Phenylketonuria, Cystic fibrosis, Biotinidase deficiency, Galactosemia, Congenital adrenal hyperplasia. The test is done twice a week at JSS Hospital, and reports are made available within 3-4 days after the collection of the sample. If the test is positive, the concerned treating physician is alerted immediately. The babies detected positive in the screening program will be recalled for confirmatory tests. Once the diagnosis is confirmed the parents are referred to the concerned doctor for management.

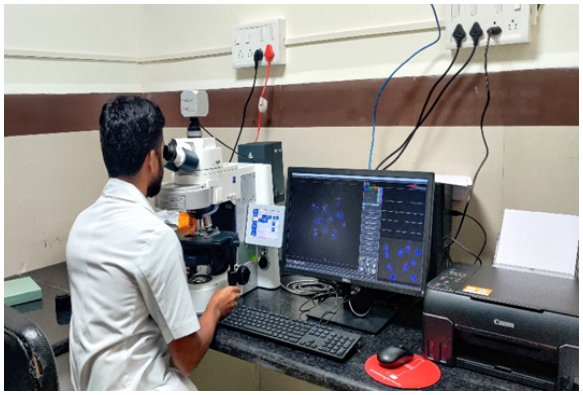
Maternal serum screening

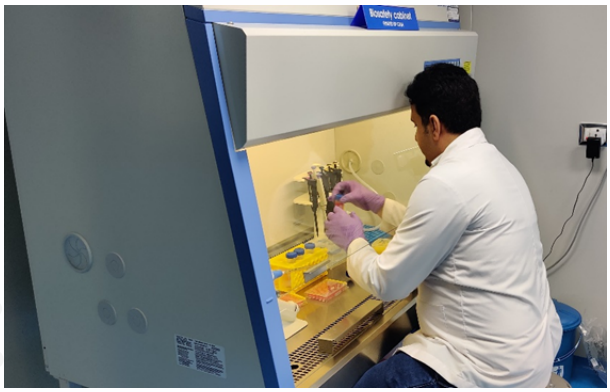
- We have initiated maternal serum screening for the high-risk pregnant ladies with double and triple marker testing at JSS Hospital for which the required instrumentation &software has been installed. We have analyzed more than 250 samples and identified two high-risk pregnancies to date.



Karyotyping and FISH

- The set-up for karyotyping and FISH analysis at JSS hospital is fully functional. We have analyzed blood and amniotic fluid samples from infertility centers and compared our results with that of CCMB for the process of standardization. To date, we have reported more than 200 patient samples and 17 chromosomal abnormalities have been identified.



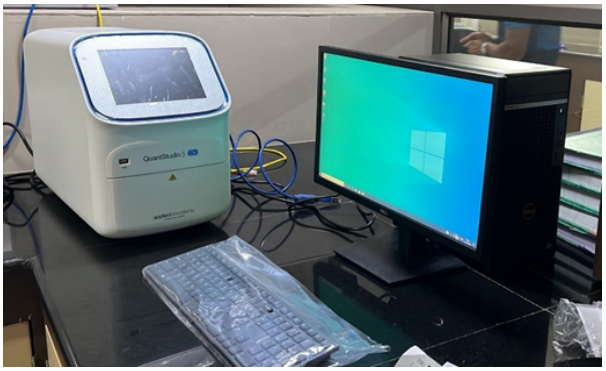


Tandem mass spectrometry

The Institute recently established an LCMS facility, which is used for expanded newborn screening. Apart from this, it has various other applications like biomarker discovery, testing for steroid panels, and toxicology screening.

Facilities for PCR and real-time PCR

We have conventional and real-time PCR facilities currently used for student training, research, and diagnostics (Figure 5). PCR assays like tetra-primer amplification refractory mutation system-polymerase chain (ARMS-PCR), PCR-RFLP (restriction fragment length polymorphism), reaction for genotype single-nucleotide polymorphisms (SNPs), multiplex PCR for identification of common infection causing micro-organisms, microdeletion of the Y chromosome, and SMN gene exon 7 and 8 deletions are being performed. Real-time PCR platform is used for gene expression studies and to study copy number variations.



Drosophila facility

The fly facility at our Institute aids the maintenance of different species of Drosophila and mutants for molecular biology and epigenetics studies (Figure 6). This facility's current activities include using a fly as a model organism to understand gene functions and epigenetic mechanisms in rare diseases. The laboratory is also utilized for training undergraduate and postgraduate students in the Drosophila lifecycle, genetics, and application of this model in research

Establishment of a Genome facility

We are also in the process of establishing the genome facility in consultation with experts from the Center for Cellular and Molecular Biology (CCMB), Hyderabad. The laboratory will comprise three rooms; the reagent preparation room, sample preparation and amplification room, and sequencing room enabled with air handling units to avoid mixing of air between the rooms to minimize the chances of contamination. Dedicated faculty with expertise in sequencing and data analysis have been appointed to run this facility. CCMB will hand-hold us in establishing the genome facility by guiding us during the standardization of various protocols and also training the manpower in attaining the skills for testing. Instruments purchased for this facility are Ion Genestudio S5 Plus, Ion Chef, Seqstudio Genetic Analyzer, and GeneChip Scanner 3000 7G.

Student/Faculty Involvement

PhD students working on topics related to genetics

- Mrs. Lavanya B. R: Understanding the role of Vitamin D on Glomerulus basement membrane constituents during Diabetic Nephropathy – Registered in October 2018.
- Mr. Adel Mohammed Saleh Ahmed: Association of gene polymorphism in obesity-related genes and insulin resistance with the risk of breast cancer – Registered in October 2018 under Dr. Akila Prashant.
- Mrs. Nishitha R. Kumar: Detection of antibiotic resistance pattern in clinical isolates of Escherichia coli and Klebsiella pneumonia using multiplex PCR and its impact in the management of sepsis – Registered in January 2019 under Dr. Akila Prashant.
- Dr. Shobha CR: "DNA Methylation Status of Obesity Related Genes and Correlation of Gene Expression and Cardio-metabolic Parameters among Adults" – Registered in January 2019 under Dr. Prashant Vishwanath.
- Ms. Supriya Bevinakoppamath: Role of Circadian rhythm genes in the progression and response to treatment in Breast cancer – Registered in January 2020 under Dr. Akila Prashant.
- Mrs. Sumayakausar S Kalaigar: Role of microRNAs in mitigating oxidative stress and regulating the gamma-globin expression in Beta-Thalassemia. – Registered in January 2021 under Dr. Akila Prashant.
- Ms. Deepthi: Role of Circadian Rhythm genes in Colorectal Cancer – Registered in July 2021.
- Mr. Siddharth Birla: Pharmacogenomics in cardiovascular diseases – Registered in October 2022.
- Ms. Anju S: Developing a low-cost protocol for screeningnewborns for spinal muscular atrophy usingdried blood spot – Registered in April 2021 under Dr. Prashant Vishwanath

Workshops attended

- Reproductive health solution workshop organized by Thermo Fischer Scientific on 5th & 6th June 2019 at Gurugram, New Delhi
- GA TOUR 2019 held at Taj Hotel Bangalore on 30th August, organised by Thermo Fisher SCIENTIFIC, Life Science Solutions
- Dr. Deepa Bhat attended the Project Investigators Orientation cum project staff training of National Task Force project entitled "Improving the Capacity of Health System and Community for Sickle Cell Disease Screening and Management
- Dr. Deepa Bhat attend the workshop organized by Society for Study of Inborn Error in Metabolism (SSIEM) 2020 on inborn error in metabolism from 6th to 9th January 2020 at Hotel Radisson Blue Plaza, Dwaraka, New Delhi.
- 4th South Asia LSD Symposium organized by Indian Society of Inborn Errors of Metabolism and Indian Academy of Medical Genetics from 17th to 18th April 2021.
- The Indian Myeloma Congress 2021 organized by the Indian Myeloma Academic Group from 23rd to 25th April 2021
- 3 days leadership webinar series organized by Internal Quality Assurance Cell (IQAC), JSS Academy of Higher Education & Research from 3rd to 5th June 2021.
- 3 total hours of Comprehensive course in the molecular testing laboratory on 20th June 2021.
- 6th Annual International Conference on Genomics and Genetic Counseling organized by the Board of Genetic Counseling India from 2nd – 4th July 2021.

Skill development training

- Mr. Manju H C, working as a Research Assistant in the Dept. of Biochemistry at JSS Medical College was deputed for training in Clinical Cytogenetics under the direction of Dr. Jayarama S. Kadandale, Professor and Head, Dept. of Clinical and Molecular Cytogenetics, Center for Human Genetics (CHG), Bengaluru, from 2/11/2019 to 16/11/2019 for a period of 15 days
- Ms. Supriya Bevinakoppamath, working as a Research Assistant in the Dept. of Biochemistry at JSS Medical College was deputed for training to enhance the capabilities and skills for the molecular diagnosis of Hemoglobinopathy under the direction of Dr. Giriraj R Chandak, Group leader, Center for Cellular & Molecular Biology (CCMB), Hyderabad, from 22/01/2020 to 25/01/2020 for a period of 04 days.
- Dr. Giriraj Chandak, Scientist, Centre for Cellular and Molecular Biology (CCMB) and also the adjunct faculty of JSSAHER had visited our Institute on 30th November 2019. He visited the genetic laboratory at JSS Hospital and provided his inputs regarding the infrastructure and the basic tests which can be performed with the currently available instrumentation.
- Application of technologies in Genomics for Clinical Research held from 3rd to 28th May 2021 by ICMR – National Institute for Research in Reproductive health, Mumbai attended by Dr. Akila P and Dr. Shobha CR.

Future Plans

- Initiate diagnostic tests on the real time PCR platform, Sanger sequencing, microarray and NGS platforms.
- Increase the number of publications and research grants

Conclusion

The Special Interest Group (SIG) has now evolved into a dedicated department, focusing on several key areas. In academics, it offers a BSc (Hons) in Medical Genetics & Genomics, with an integrated MSc program aimed at fostering advanced knowledge in the field. In diagnostics, the department has introduced new investigations, including expanded newborn screening, exon sequence analysis, and segregation analysis, enhancing diagnostic capabilities. On the research front, the SIG is actively working to secure grants from external funding agencies to advance its work on rare disease therapeutics.