Dr. Mehul A. Mistri

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CAREER OBJECTIVE

To contribute to a dynamic organization that will give me chance to exploit my knowledge, skills and experience to explore new avenue in biological science, genomics and public health.

RESEARCH & ACADEMIC EXPERIENCE

- **Scientist (Inherited Genomics and Metabolism) (July, 2018 - Present)**
  Neuberg Center for Genomic Medicine (NCGM), Neuberg Supratech Reference Laboratories, Ahmedabad, Gujarat, India.
- **Research Associate (January, 2015 – June, 2018)**
- **Senior Research Fellow (January 2011 – December 2014)**
- **Junior Research Fellow (May 2008 – December 2010)**
  Foundation for Research in Genetics and Endocrinology (FRIGE), Ahmedabad, Gujarat, India (2008 – 2018)

EDUCATION

- **Doctor of Philosophy (Ph.D.) in Biochemistry with coursework (2010 - 2017)**
  Department of Biochemistry, Faculty of Science
  The M. S. University of Baroda and FRIGE's Institute of Human Genetics.
- **Master of Science (M.Sc.) in Biochemistry with Dissertation (2006 - 2008)**
  BRD School of Bioscience, Sardar Patel University, Anand, Gujarat, India.
- **Bachelor of Science (B.Sc.) in Biochemistry (2004 - 2006)**
  C. U. Shah Science collage, Gujarat University, Ahmedabad, Gujarat, India.

EDUCATIONAL TRAINING

- **International Summit of Human Genetics and Genomics (2018)**
  National Human Genome Research Institute (NHGRI), at Nation Institute of Health (NIH) campus, Bethesda, MD, USA.
- **ICMR Workshop on Basics of Bioinformatics and it Application (2012)**
  National Institute for Research in Reproductive Health (NIRRH), Parel, Mumbai.
- **Introduction and Application of Sanger sequencing (2011)**
  Genetic research centre-NIRRH-ICMR, Parel, Mumbai.
- **Introduction to Real-Time PCR (2011)**
  Life Technologies (India) Pvt. Ltd., Gurgaon, Haryana.

AWARDS & HONOUR

- International Travel Fellowship to attend SSIEM annual symposium (2018)
- Complete Fellowship to attend International Summit in Human Genetics and Genomics (2018)
- International Travel Fellowship to attend SSIEM annual symposium (2015)
- ISHG Young Scientist Award (2014)
- International Travel Award for Developing Country to attend ICHG and ASHG annual meeting (2011)

PUBLICATIONS

- Dipti Deshpande, Shailesh Kumar Gupta, Asodu Sandeep Sarma, Prajnya Ranganath, Jamal Md Nurul Jain S, Jayesh Sheth, **Mehul Mistri**, Neerja Gupta, Madhulika Kabra, Shubha R Phadke, Katta M Girisha, Ratna Dua Puri, Shagun Aggarwal, Chaitanya Datar, Kausik Mandal, Preetha Tilak, Mamta Muranjan, Sunita Bijarnia-Mahay, Radha Rama Devi A, Naresh B Tayade, Akash Ranjan, Ashwin B Dalal. Functional characterization of novel variants in SMPD1 in Indian patients with acid sphingomyelinase deficiency. Hum Mutat. 2021 Jul 17. doi: 10.1002/humu.24263. Epub ahead of print. PMID: 34273913.
- Divya Pasumarthi, Neerja Gupta, Jayesh Sheth, S Jamal Nurul Jain, Ikrormi Rungsung, Madhulika Kabra, Prajnya Ranganath, Shagun Aggarwal, Shubha Phadke, Katta Girisha, Anju Shukla, Chaitanya Datar, IC Verma, Ratna Puri, Riddhi Bhavsar, **Mehul Mistri**, VH Sankar, Kalpana Gowrishankar, Divya Agrawal, Mohandas Nair, Sumita Danda, Jai Soni, Ashwin Dalal (2020). Identification and characterization of 30 novel pathogenic variations in 69 unrelated Indian patients with Mucolipidosis Type II and Type III. J Hum Genet, July 2020, 1-14.
- **Mehul Mistri**, Sanjiv Mehta, Dhaval Solanki, Mahesh Kamate, Neerja Gupta, Madhulika Kabra, Ratna Puri, Girish Katta, VH Sankar, Sheela Nampoothiri, Frenny Sheth, Jayesh Sheth (2019). Identification of novel variants in a large cohort of children with Tay-Sachs disease: An initiative of a multicentric taskforce on lysosomal storage disorders by Government of India. J Hum Genet 64, 985–994.
- Jayesh Sheth, Riddhi Bhavsar, **Mehul Mistri**, Dhairya Pancholi, Ashish Bavdekar, Ashwin Dalal, Prajanya Ranganath, Girisha KM, Anju Shukla, Shubha Phadke, Ratna Puri, Inusha Panigrahi, Anupriya Kaur, Mamta Muranjan, Manisha Goyal, Radha Ramadevi, Raju Shah, Sheela Nampoothiri, Sunita Danda, Chaitanya Datar, Seema Kapoor, Seema Bhatwadekar, Frenny Sheth (2019). Gaucher disease: single gene molecular characterization of one-hundred Indian patients reveals novel variants and the most prevalent mutation. BMC Med Genet. 14;20(1):31.
- Jayesh Sheth, **Mehul Mistri**, Riddhi Bhavsar, Dhairya Pancholi, Mahesh Kamate, Neerja Gupta, Madhulika Kabra, Sanjiv Mehta, Sheela Nampoothiri, Arpita Thakker, Vivek Jain, Raju Shah, Frenny Sheth (2018). Batten disease: Biochemical and Molecular characterization revealing novel PPT1 and TPP1 gene mutations in Indian patients. BMC Neurol. 18(1):203.
- Jayesh Sheth, Dhairya Pancholi, **Mehul Mistri**, Payal Nath, Chitra Ankleshwaria, Riddhi Bhavsar, Ratna Puri, Shubha Phadke and Frenny Sheth (2018). Biochemical and molecular characterization of adult patients with type I Gaucher disease and carrier frequency analysis of Leu444Pro - a common Gaucher disease mutation in India. BMC Medical Genetics 19: 178.
- Jayesh Sheth, **Mehul Mistri**, Lakshmi Mahadevan, Sanjeev Mehta, Dhaval Solanki, Mahesh Kamate, Frenny Sheth (2018). Identification of deletion-duplication in HEXA gene in five children with Tay-Sachs disease from India. BMC Medical Genetics 19: 109.
- Jayesh Sheth, Jijo John Joseph, Krati Shah, Mamta Muranjan, **Mehul Mistri** and Frenny Sheth (2017). Pulmonary manifestations in Niemann-Pick type C disease with mutations in NPC2 gene: case report and review of literature. BMC Medical Genetics 18:5.
- Jayesh Sheth, **Mehul Mistri**, Sheela Namoothiri, Riddhi Bhavsar, Inusha Panigrahi, Mahesh Kamate, Frenny Sheth (2017). Quantitative and Qualitative Analysis of Urinary Glycosaminoglycans (GAGs). Arch Pediatr: JPED-122. DOI:10.29011/2575-825X. 100022.
- Jayesh J Sheth, **Mehul Mistri**, Krati Shah, Mayank Chaudhary, Koumudi Godbole, Frenny Sheth (2016). Lysosomal storage disorders in non-immune hydrops fetalis (NIHF) - An Indian

Experience. JIMD Rep. Dec 8.

- Jayesh Sheth, Chaitanya Datar, **Mehul Mistri**, Riddhi Bhavsar, Frenny Sheth, Krati Shah (2016). GM2gangliosidosis AB variant: Novel mutation from India – a case report with review. BMC Pediatrics 16:88.
- Parag M Tamhankar*, **Mehul Mistri***, Pratima Kondurkar, Daksha Sanghavi and Jayesh Sheth (2016). Clinical, biochemical and mutation profile in Indian patients with Sandhoff disease. J Hum Genet. 61: 163-166. (DOI: 10.1038/jhg.2015.130). (* Joint first author)
- Jayeh Sheth, **Mehul Mistri**, Riddhi Bhavsar, Frenny Sheth, Mahesh Kamate, Harshuti Shah, Chaitanya Datar (2015). Lysosomal storage disorders in Indian children with neuroregression attending a genetic center. Indian Pediatr. 52(12):1029-1033.
- Jayesh Sheth, **Mehul Mistri**, Riddhi Bhavsar, Harsh Patel, Frenny Sheth (2015). Novel mutation in XPC gene: A Case report of patient with Xeroderma Pigmentosum. International Journal of Dermatology 015, 54, e487–e491.
- Jayesh Sheth, **Mehul Mistri**, Chaitanya Datar, Umesh Kalane, Shekhar Patil, Mahesh Kamate, Harshuti Shah, Sheela Nampoothiri, Sarita Gupta, Frenny Sheth (2014). Expanding the spectrum of HEXA mutations in Indian patients with Tay-Sachs disease. Molecular Genetics and Metabolism Reports. 1: 425-430. (DOI: 10.1016/j.ymgmr.2014.09.004).
- Jayeh Sheth, **Mehul Mistri**, Frenny Sheth, Chaitanya Datar, Koumudi Godbole, Mahesh Kamate, Kamal Patil (2014). Prenatal diagnosis of lysosomal storage disorders by enzymes study using chorionic villus and amniotic fluid. J Fetal Med 1(1):17-24. (DOI: 10.1007/s40556-014-0001-3).
- Jayesh Sheth, **Mehul Mistri**, Frenny Sheth, Raju Shah, Ashish Bavdekar, Koumudi Godbole, Nidhish Nanavaty, Chaitanya Datar, Mahesh Kamate, Nrupesh Oza, Chitra Ankleshwaria, Sanjeev Mehta, Marie Jackson (2014). Burden of lysosomal storage disorders in India: Experience of 387 affected children from a single diagnostic facility. JIMD Rep.12: 51-63 DOI;10.1007/8904_2013_244.
- Chitra Ankleshwaria, **Mehul Mistri**, Ashish Bavdekar, Mamta Muranjan, Usha Dave, Parag Tamhankar, Varun Khanna, Eresha Jasinge, Sheela Nampoothiri, Suresh Udayankara Kadangot, Frenny Sheth, Sarita Gupta, Jayesh Sheth (2014). Novel mutations in the glucocerebrosidase gene of Indian patients with Gaucher Disease. J Hum Genet. 59:223-228. (DOI: 10.1038/jhg.2014.5).
- **Mehul Mistri**, Parag M Tamhankar, Frenny Sheth, Daksha Sanghavi, Pratima Kondurkar, Swapnil Patil, Susan Idicula-Thomas, Sarita Gupta, Jayesh Sheth (2012). Identification of novel mutations in HEXA gene in children affected with Tay-Sachs disease from India. PLoS ONE 7(6):e39122. (DOI: 10.1371/journal.pone.0039122).
- Jayesh Sheth, **Mehul Mistri**, Mahesh Kamate, Sashi Vaja, Frenny Sheth (2012) Diagnostic strategy of Mucolipidosis II/III. Indian Pediatr. 49(12):975-977.
- Jayesh Sheth, Chitra Ankleshwaria, **Mehul Mistri**, Nidhish Nanavaty, Sanjiv Mehta (2011). Splenomegaly, Cardiomegaly and Osteoporosis in a child with Gaucher Disease. Case Reports in Pediatrics. 1–4.
- Jayesh Sheth, Nrupesh Oza, **Mehul Mistri**, Premal Naik, Suresh Kumar, Frenny Sheth (2009). Mucolipidosis type II (I-Cell) in two children with skeletal abnormality, dysmorphism and hepatosplenomegaly. Pediatric oncall vol (6). http://www.pediatriconcall.com/fordoctor/casereports/ mucolipidosis.asp

ABSTRACTS PUBLISHED IN VARIOIUS JOURNALS

- **Mehul Mistri**, Ashwin Dalal, Payal Priyadarshini, Sheela Nampoothiri, Sanjeev Mehta, Ankur Singh, Frenny Sheth, Jayesh Sheth (2018). Identification of novel and known mutation in 19 patients with mucolipidosis type II and III from India and validation of novel method of screening for mucolipidosis-II and III screening. Molecular Genetics and Metabolism 123(2): S96. DOI: 10.1016/j.ymgme.2017.12.252.
- Riddhi Bhavsar, Jayesh Sheth, **Mehul Mistri**, Mahesh Kamate, Frenny Sheth (2016). Molecular characterization and identification of novel mutations in the PPT1 gene causing neuronal ceroid

lipofuscinosis-1 (NCL1) in children from India**.** Molecular Genetics and Metabolism; 117(2); S28. DOI: 10.1016/j.ymgme.2015.12.202.

- **Mehul Mistri**, Parag Tamhankar, Pratima Kondukar, Frenny Sheth, Jayesh Sheth (2015). Biochemical study and molecular analysis identifying novel alleles in children affected with Sandhoff disease from India. J of Inh Metab Disease. Vol. 38(Suppl.1); S285.
- Jayesh Sheth, **Mehul Mistri**, Nrupesh Oza, Frenny Sheth (2011). Screening strategy for lysosomal storage disorders. J of Inh Metab Disease. Vol. 34 (2); P15.
- Jayesh Sheth, **Mehul Mistri**, Nrupesh Oza, Frenny Sheth (2011). Occurrence and screening strategy of lysosomal storage disorders in India: Our experience. Molecular Genetics and Metabolism 102(2); S41. DOI:10.1016/j.ymgme.2010.11.138.
- **Mehul Mistri**, Jayesh Sheth, Frenny Sheth, Sarita Gupta (2014). Identification of Novel mutations in HEXA gene in children affected with Tay-Sachs disease from India. Molecular Cytogenetic 7(1); P53.
- Jayesh Sheth, **Mehul Mistri**, Harsh Patel, Chitra Ankleshwaria, Aradhana Parikh (2014). Autosomal dominant mutation in COL7A1 gene causing Epidermolysis Bullosa Dystrophica. Molecular Cytogenetic 7(1); P58.
- Jayesh Sheth, **Mehul Mistri**, Frenny Sheth, Sarita Gupta (2014). Prenatal diagnosis of Tay-Sachs disease: Our Experience in India. Molecular Cytogenetic 7(1); P124.
- **Mehul Mistri**, Harsh Patel, Tanmay Tanna, Chitra Ankleshwari, Frenny Sheth (2014). Prenatal diagnosis of autosomal recessive Osteopetrosis: A case report. Molecular Cytogenetic 7(1); P125.
- **Mehul Mistri**, Nrupesh Oza, Freeny Sheth, Jayesh Sheth (2014) Prenatal Diagnosis of Lysosomal storage disorders: Our experience. Molecular Cytogenetic 7(1); P126.
- Chitra Ankleshwaria, Jayesh Sheth, **Mehul Mistri**, Ashish Bavdekar, Sheela Nampoothiri, Sarita Gupta, Frenny Sheth (2014). Identification of Novel Mutations in Glucocerebrosidase (GBA) Gene in Indian Patients with Gaucher disease (GD). Molecular Cytogenetic 7(1); P52.
- Harsh Patel, **Mehul Mistri**, Chitra Ankleshwaria, Frenny Sheth, Jayesh Sheth (2014). Frequency analysis of Spinocerebellar ataxia types 1, 2, 3 & 6 in patients with ataxia from Gujarat. Molecular Cytogenetic 7(1); P64.

TALKS

- FRIGE-IHG workshop on 'Application of Real Time PCR in Medical Biotechnology', 2016, Ahmedabad, Gujarat, India. ***Workshop Co-ordinator and Faculty***. Primer designing.
- International conference of inborn error of Metabolism (ICIEM), September 2014, Hyderabad, India. ***Oral presentation.*** Identification of GSD type III by debrancher enzyme activity form leucocytes.
- International Conference on Human Genetics and 39th Annual Meeting of the Indian Society of Human Genetics, 2014, Ahmedabad, Gujarat, India. ***Workshop Co-ordinator***. PCR based strategies for genetic testing.
- International Conference on Human Genetics and 39th Annual Meeting of the Indian Society of Human Genetics, January 2014, Ahmedabad, Gujarat, India. ***Oral presentation***. Identification of novel mutations in HEXA gene in children affected with Tay-Sachs disease from India.

POSTERS PRESENTATION

- ~20 in International conferences and one in national conference.

TECHNICAL SKILL

- **Molecular techniques:** DNA, RNA, Protein and Plasmid isolation, PCR, Electrophoresis, SDS-PAGE, DNA synthesis, Gene cloning, Western/Southern blotting, MLPA, Fragment analysis, Sanger sequencing, NGS, Microarray etc.
- **Biochemical techniques:** Quantitative and Qualitative analysis of various bio-molecules, Protein estimation and expression, Chromatography, ELISA, Enzyme analysis etc.
- **Cell culture technique:** Cell culture handling and maintenance from various biological tissues, Cell differentiation etc.
- **Bioinformatics tools and Computer proficiency:** NGS data analysis, annotation and curation, Online blast tool, Sequence alignment tools, Primer designing software, SNP validation software, Restriction enzyme selection tools, Gene mapper, Basic knowledge of protein modelling and docking, Adobe Photoshop, MS office etc.

MANAGERIAL SKILLS

Documentations, Collaborations, Team building, Priority Setting, Resource allocation, Ability to pivot, etc.

PERSONAL INFORMATION

Nationality	:	Indian.
Date of birth	:	10/10/1985
Languages known	:	English, Hindi, and Gujarati
Marital status	:	Married
Hobbies	:	Reading, Writing, Playing cricket and Travelling