# PEER REVIEW PUBLICATIONS & POSTER/ORAL PRESENTATIONS

**Related to Ph.D topic**

- 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: 17-24*. (DOI: 10.1007/s40556-014-0001-317).

- 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).

- **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).

**Other publications**

- 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*.

- 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).
- Jayesh Sheth, **Mehul Mistri**, Mahesh Kamate, SashiVaja, FrennySheth (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)

**Abstract published in different journals**

- **Mehul Mistri**, Parag M Tamhankar, Pratima Kondurkar, Frenny Sheth, Jayesh Sheth (2015). Biochemical study and molecular analysis identifying novel alleles in children affected with Sandhoff disease from India. *J Inherit Metab Dis 38 (1); P-580.*

- Jayesh Sheth, **Mehul Mistri**, Nrupesh Oza, Frenny Sheth (2011) Screening Strategy for Lysosomal storage disorders. *J Inherit Metab Dis 34 (2); P-15.*

- 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 01/2011; 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*

**Oral presentation**

- **Mehul Mistri**, Jayesh Sheth, Frenny Sheth, Sarita Gupta. Identification of Novel mutations in *HEXA* gene in children affected with Tay-Sachs disease from India. International Conference on Human Genetics and 39$^{th}$ Annual Meeting of the Indian Society of Human Genetics,

January 22-25, 2014, Ahmedabad, India. **(Full paper selected for ISHG young scientist award)**

- **Mehul Mistri,** Jayesh Sheth, Mahesh Kamate, Eresha Jasinge, Frenny Sheth. Identification of GSD type III by Debrancher Enzyme Activity form Leucocytes. International conference of inborn error of Metabolism (ICIEM), 19-21 September, 2014, Hyderabad, India.

**Poster Presentation**

- **Mehul Mistri**, Parag M Tamhankar, Pratima Kondurkar, Frenny Sheth, Jayesh Sheth. Biochemical study and molecular analysis identifying novel alleles in children affected with Sandhoff disease from India. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM 2015), September 1-4, 2015, Lyon, France (Received International travel scholarship from SSIEM).

- **Mehul Mistri**, Jayesh Sheth, Frenny Sheth, Sarita Gupta. Identification of Novel mutations in *HEXA* gene in children affected with Tay-Sachs disease from India. International Conference on Human Genetics and 39$^{th}$ Annual Meeting of the Indian Society of Human Genetics, January 22-25, 2014, Ahmedabad, India

- Jayesh Sheth, **Mehul Mistri**, Harsh Patel, Chitra Ankleshwaria, Aradhana Parikh. Autosomal Dominant Mutation in *COL7A1* Gene Causing Epidermolysis Bullosa Dystrophica. International Conference on Human Genetics and 39$^{th}$ Annual Meeting of the Indian Society of Human Genetics, January 22-25, 2014, Ahmedabad, India

- Jayesh Sheth, **Mehul Mistri**, Frenny Sheth, Sarita Gupta. Prenatal Diagnosis of Tay-Sachs disease: Our Experience in India. International Conference on Human Genetics and 39$^{th}$ Annual Meeting of the Indian Society of Human Genetics, January 22-25, 2014, Ahmedabad, India

- **Mehul Mistri**, Harsh Patel, Tanmay Tanna, Chitra Ankleshwari, Frenny Sheth. Prenatal Diagnosis of Autosomal Recessive Osteopetrosis: A Case Report, International Conference on Human Genetics and 39$^{th}$ Annual Meeting of the Indian Society of Human Genetics, January 22-25, 2014, Ahmedabad, India

- **Mehul Mistri**, Nrupesh Oza, Freeny Sheth, Jayesh Sheth. Prenatal Diagnosis of Lysosomal storage disorders: Our experience, International Conference on Human Genetics and 39$^{th}$ Annual Meeting of the Indian Society of Human Genetics, January 22-25, 2014, Ahmedabad, India

- Chitra Ankleshwaria, Jayesh Sheth, **Mehul Mistri**, Ashish Bavdekar, Sheela Nampoothiri, Sarita Gupta, Frenny Sheth. Identification of Novel Mutations in Glucocerebrosidase (GBA) Gene in Indian Patients with Gaucher Disease (GD), International Conference on Human Genetics and 39$^{th}$ Annual Meeting of the Indian Society of Human Genetics, January 22-25, 2014, Ahmedabad, India

- Harsh Patel, **Mehul Mistri**, Chitra Ankleshwaria, Frenny Sheth, Jayesh Sheth. Frequency analysis of Spinocerebellar ataxia types 1, 2, 3 & 6 in patients with ataxia from Gujarat,

International Conference on Human Genetics and 39th Annual Meeting of the Indian Society of Human Genetics, January 22-25, 2014, Ahmedabad, India

- **Mehul Mistri**, Chitra Ankleshwaria, Ashish Bavdekar, Mahesh Kamate, Chaitanya Datar, Frenny Sheth, Jayesh Sheth. Occurrence of Lysosomal storage disorders in children with Hepatosplenomegaly and Hepatomegaly: An Institutional experience. 62nd ASHG meeting, November 6-10, 2012 in San Francisco, California. (Abstract ID – 700T)

- Jayesh Sheth, **Mehul Mistri**, Chitra Ankleshwaria, Parag Tamhankar, Ashish Bavdekar, Chaitanya Datar, Mahesh Kamate, Sarita Gupta, Sanjeev Mehta, Frenny Sheth. Molecular analysis for Gaucher, Tay-Sach's and Sandhoff disease in Indian patients, 62nd Annual Meeting of The American Society of Human Genetics, November 6-10, 2012 in San Francisco, California. (Abstract ID – 2803W)

- Jayesh Sheth, Raju Shah, **Mehul Mistri**, Vishal Kuchhy, Freny Sheth. Spectrum of metabolic disorders in high risk group of children from western India (Gujarat), SSIEM, Annual Symposium 2012, 4-7th September, 2012, ICC, Birmingham, UK. (Abstract ID - ARROEP-132887-804978-SSIEM2012)

- Jayesh Sheth, **Mehul Mistri**, Frenny Sheth, Nrupesh Oza, Raju Shah, Mahesh Kamate, Koumudi Godbole, Ashish Bavdekar, Chaitanya Datar. Burden of Lysosomal storage disorders in India: Study of 946 children, SSIEM, Annual Symposium 2012, 4-7th September, 2012, ICC, Birmingham, UK. (Abstract ID - CEPUTO-931689-828742-SSIEM2012)

- **Mehul Mistri**, Parag Tamhankar, Susan Thomas, Parag Kondurkar, Sanjeev Mehta, Daksha Sanghavi, Jayesh Sheth. Identification of novel mutations in HEXA gene in children affected with Tay-sachs disease from India. 61st Annual meeting of American Society of Human Genetics and 12th International Congress of Human Genetics (ICHG) meeting, Montreal, Canada. 11th – 15th Oct., 2011. (Abstract ID-1322W) **(Received the developing country travel award from ICHG).**

- Chitra Ankleshwaria, **Mehul Mistri**, Ashish Bavdekar, Mamta Muranjan, Jayesh Sheth. Study of mutation spectrum in patients with Gaucher disease from India. 61st Annual meeting of American Society of Human Genetics and 12th International Congress of Human Genetics (ICHG) meeting, Montreal, Canada. 11th – 15th Oct., 2011. (Abstract ID: 1308T)

- Jayesh Sheth, **Mehul Mistri**, Nrupesh Oza, Frenny Sheth. Screening Strategy for Lysosomal storage disorders. The 7th ISNS European Neonatal Screening Regional Meeting, Geneva, Switzerland, 28 – 30 August, 2011.

- Jayesh Sheth, **Mehul Mistri**, Nrupesh Oza and Frenny Sheth. Occurrence and Screening Strategy of Lysosomal Storage Disorders In India: Our Experience. 7th World Symposium of Lysosomal Storage Diseases Network. Las Vegas, USA, February, 2011.(Abstract ID-Sheth12)

- **Mehul Mistri**, Nrupesh Oza, Frenny Sheth and JayeshSheth. Occurrence and Screening Strategy of Lysosomal Storage Disorders In India: Our Experience. 36th Annual conference of

Indian Society of Human Genetics and international conference of Genomics, Genetics disease and diagnostics, Manipal, Karnataka. February, 2011. Poster ID-215.

- Jayesh Sheth, **Mehul Mistri**, Nrupesh Oza, Usha Dave, Prakash Gambhir, Raju C. Shah, Frenny Sheth. Prevalence of Lysosomal storage disorders in India: Our experience. 60th Annual meeting of American Society of Human Genetics, Washington DC, USA. November, 2010. Poster ID-2052.

- Jayesh Sheth, **Mehul Mistri**, Nrupesh Oza and Frenny Sheth. The spectrum of Lysosomal Storage Disorders (LSDs) in children with neuroregression in India. 59th Annual meeting of American Society of Human Genetics (ASHG-2009). 20 – 24 Oct., 2009, Honolulu, Hawaii.

- Mehul Mistri, Jayesh Sheth, Nrupesh Oza, Frenny Sheth, Prakash Gambhir and Usha Dave. The Prevalence of Lysosomal storage disorders (LSDs) in western India. 4th International conference of Birth Defects and Disabilities in the developing world. 4 -7 Oct., 2009, New Delhi, India.

- **Mehul Mistri**, Jayesh Sheth. Prenatal diagnosis of lysosomal storage disorders in India. International Symposium on Advances in Molecular Medicine and Clinical Implication, Dhirubhai Ambani Life Sciences symposia series – IV. 24 – 25 Jan., 2009, Navi Mumbai, India.

- Jayesh Sheth, **Mehul Mistri**, Nrupesh Oza, Chitra Ankleshwaria, Ashish Bavdekar, Koumudi Godbole, Frenny Sheth. Occurrence of Lysosomal Storage Disorders in children with Hepatomegaly/ Hepatosplenomegaly in India. 37th Annual conference of Indian Society of Human Genetics, Chandigarh, India.

- Participate as delegate in the one day seminar one "HUMAN HEALTH CARE: REDEFINED – 2008" Organized by the department of Zoology, Biomedical technology and Human Genetics, University School of Sciences, Gujarat University, Ahmedabad on 17th March, 2008.

- Participate as delegate in the XXIV – Gujarat Science Congress 2010 Organized by the Gujarat University and Gujarat Science academy, University School of Sciences, Gujarat University, Ahmedabad on 21st March, 2010.

**Training**

- Actively participated for training of "Introduction to Real-Time PCR" under the guidance of Dr. Sangita Thatai, during June 2011 at Applied Biosystems, Gurgaon, Haryana.

- Actively participated for training of "Introduction and application of sequencer (ABI-3100)" under the guidance of Dr. Parg M. Tamhankar, during January 2011 at Genetic research centre-NIRRH-ICMR, Parel, Mumbai.

- Selected to attend ICMR workshop on "Basics of Bioinformatics and it Application" to be held at the National Institute for Research in Reproductive Health, J. M. Street, Parel, Mumbai – 400012 from June 5th -7th, 2012.

**Awards & Honors**

- International travel scholarship from Society for the Study of Inborn Errors of Metabolism (SSIEM), for poster presentation at Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM 2015), September 1-4, 2015, held at Lyon, France.

- The developing country travel award from International Congress of Human Genetics (ICHG), for poster presentation at 61$^{st}$ Annual meeting of American Society of Human Genetics (ASHG) and 12$^{th}$ International Congress of Human Genetics (ICHG) meeting, October 11-15, 2011, held at Montreal, Canada.