# PEER REVIEW PUBLICATIONS

## Ph.D related topics:

- **Chitra Ankleshwaria**, Mehul Mistri, Ashish Bavdekar, Mamta Muranjan, Usha Dave, Parag Tamhankar, Varun Khanna, Eresha Jasinge, Sheela Nampoothiri, Suresh Kumar EK, FrennySheth, Sarita Gupta, Jayesh Sheth (2014). Novel mutations in the glucocerebrosidase gene of Indian patients with Gaucher Disease. J of Hum Genet. 59: 223-228.

- 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 Rep12: 51-63.

- Jayesh Sheth, **Chitra Ankleshwaria**, Mehul Mistri, Nidhish Nanavaty, Sanjiv Mehta (2011). Splenomegaly, Cardiomegaly and Osteoporosis in a Child with Gaucher Disease. Case Rep in Pediatr: 1-4.

**Other topics:**

- Jayesh Sheth, **Chitra Ankleshwaria**, Rajeshwari Pawar, Frenny Sheth(2012). Identification of Novel mutations in FAH gene and Prenatal diagnosis of Tyrosinemia in Indian Family. Case Rep in Genet doi:10.1155/2012/428075.

## ABSTRACT PUBLISHED IN DIFFERENT JOURNALS

- **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). Mol. Cytogenet 7: P52 doi:10.1186/1755-8166-7-S1-P52.

- Jayesh Sheth, Mehul Mistri, Harsh Patel, **Chitra Ankleshwaria**, Aradhana Parikh(2014). Autosomal Dominant Mutation in *COL7A1* Gene Causing Epidermolysis Bullosa Dystrophica. Mol. Cytogenet 7: P58 doi:10.1186/1755-8166-7-S1-P58.

- Mehul Mistri, Harsh Patel, Tanmay Tanna, **Chitra Ankleshwaria**, Frenny Sheth(2014). Prenatal Diagnosis of Autosomal Recessive Osteopetrosis: A Case Report. Mol. Cytogenet 7: P125 doi:10.1186/1755-8166-7-S1-P125.

- 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. Mol. Cytogenet 7: P64 doi:10.1186/1755-8166-7-S1-P64.

## ABSTRACT PRESENTED AND PUBLISHED

### Poster Presentation

- **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.

- 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.

- Mehul Mistri, Harsh Patel, Tanmay Tanna, **Chitra Ankleshwaria**, 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.

- 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.

- **Chitra Ankleshwaria**, Jayesh Sheth, Harsh Patel, Jyothi Lekshami, Frenny Sheth.Single gene disorders in Western India, presented at 62nd ASHG Annual Meeting held at San Francisco, 2012.

- 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**, ParagTamhankar, AshishBavdekar, ChaitanyaDatar, Mahesh Kamate, Sarita Gupta, Sanjeev Mehta, FrennySheth. 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, Mehul Mistri, Nrupesh Oza, **Chitra Ankleshwaria**, Ashish Bavdekar, Koumudi Godbole, Frenny Sheth. "Occurrence of Lysosomal Storage Disorders in children with Hepatomegaly or Hepatosplenomegaly in India" was presented at XXXVII Annual Conference of the Indian society of Human Genetics at Punjab University, Chandigarh, 2012.

- **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 12thInternational Congress of Human Genetics (ICHG) meeting, Montreal, Canada. 11th – 15th Oct. 2011. (Abstract ID: 1308T).

- Toral Shah, Anamika Bansal, Frenny Sheth, **Chitra Ankleshwaria,** Jayesh Sheth. **"**DMD gene deletions study in children with muscular dystrophy in Gujarat" at International Conference on Genomics, Genetic Diseases and diagnostics and XXXVI Annual Conference of the Indian society of Human Genetics at Manipal University, Manipal, 2011.

## AWARDS

- **Travel award**

  Received DBT travel Grant for presenting poster and attending 12$^{th}$ ICHG and 61$^{st}$ ASHG meeting held at Montreal, Canada during 11$^{th}$ - 15$^{th}$ October 2011.

  **Chitra Ankleshwaria**, Mehul Mistri, Ashish Bavdekar, Mamta Muranjan, Jayesh Sheth. "Study of mutation spectrum in patients with Gaucher disease from India" was presented at 12th International Congress of Human Genetics (ICHG) and the 61st ASHG Annual Meeting held at Montreal, Canada in 2011.

- **Paper selected for young scientist award**

  Paper selected for the young scientist award by Indian society of Human Genetics (ISHG), for oral presentation at International Conference on Human Genetics and 39$^{th}$ Annual Meeting of the Indian Society of Human Genetics, 22-25$^{th}$January, 2014, Ahmedabad, India

**Chitra Ankleshwaria**. Identification of novel mutation and high prevalence of L444P mutation in glucocerebrosidase (GBA) gene with severe type-1 phenotype: Our experience of 32 gaucher disease patients from India.