» DR. FRENNY JAYESH SHETH

Founding member and Director at Foundation for Research In Genetics and Endocrinology [FRIGE] and Institute of Human Genetics

Founder Trustee: Foundation For Research in Genetics and Endocrinology. Registered charitable Trust: Registration No: E/13237 (80G IT exemption)

Managing Trustee: Sheth Charitable Trust. Registration No: E 12862(80G IT)

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SCIENTIFIC REVIEWER

Editorial Board Member	Molecular Cytogenetics SM Journal of Gynecology and Obstetrics EC Pediatrics
Section Editor	The Indian Practitioner
Reviewer	Molecular Cytogenetics Journal of Translational Medicine Genetics in Medicine European Journal of Medical Genetics Gene Indian Pediatrics Indian Journal of Medical Research Indian Journal of Cancer Research and Therapeutics Indian Journal of Cancer Mutation Research Pediatric OnCall International Journal of Medicine and Medical Sciences International Journal of General Medicine Clinical Medicine Insights: Case Reports

MEMBERSHIP: PROFESSIONAL ORGANIZATIONS

» American Society of Human Genetics
» European Cytogenetic Association
» Indian Society of Human Genetics
» Indian Society for Prenatal Diagnosis and Therapy
» Indian Association of Cancer Research
» Indian Society of Cell Biology
» Indian Society of clinical, Pharmaceutical and Therapeutics
» Association of Genetic Technologist

PROJECTS

CONSULTANCY

» The Genetics of Consanguinity in Children with Multiple Disabilities with NIEPMD – National Institute for Empowerment of Persons with Multiple Disabilities

» Cell line authentication services – Zydus Biologics

RESEARCH PROJECTS
ON GOING

» Detection of copy number variants and Mendelian disorders in children with malformations and intellectual disabilities of unknown aetiology using various molecular modalities. Gujarat State Biotech Mission (GSBTM), PI: Dr. Frenny Sheth, GSBTM/MD/PROJECTS/SSA/4865/2016-2017 (2016 – 2019).

» The Clinical, Enzymological and Molecular Study of Children with Gaucher disease (GD): Identification of Demographical loci on GD phenotype DBT, Co-I: Dr. Frenny Sheth, BT/PR4112/MED/12/654/2014 (2016 – 2019).

CONCLUDED
NATIONAL

- Mutation study of the prevalent lysosomal storage disorders in India and extension of lysosomal enzyme study ICMR, Co-I: Dr. Frenny Sheth (2010 – 2013)
- Preparation and standardization of FISH probes for various genetic disorders and extension of services in Gujarat DBT, PI: Dr. Frenny Sheth (2008 – 2011)
- Study of LSDs in children with regression of milestone ICMR, Co-I: Frenny Sheth (2006 – 2009)
- Herbal Based preparations for degenerative disorders: Type II [NIDDM] diabetes Mellitus with emphasis on Insulin Sensitization and Herbo print-are tool for standardization for herbal medicine CSIR-NMITLI, Co-coordinated Genotoxicity studies (2004 – 2009)

INTERNATIONAL COLLABORATIVE PROJECTS:

- Re-examination of Individuals with balanced chromosomal breakpoints: International Breakpoint Mapping Consortium (IBMC) with University of Copenhagen, Denmark
- The genetics of primary microcephaly with Institut für Medizinische Genetik, Universität Zürich, Switzerland
- Gene Polymorphism and Folate Metabolism in mothers with Down syndrome child with Toxicological Research centre, Arkansas, USA
- Study of LAMIN gene mutation in large Indian Family with Familial Partial Lipodystrophy with Prof Robert Hegele, Canada
- Revaluation of the traditional production of the hybrid of the Indian wild ass (Equus hemionus khur) and the female donkey (E. asinus) in Gujarat, India. The study of Karyotype in hybrid of the Indian wild ass with Aquine Musium, Japan

EDUCATION

2002	**Advance Post Graduate course in Cytogenetic and Molecular Cytogenetics,** *University of Montpellier, Nimes, France*
1990	**Ph.D. in Cell Biology,** *Zoology Department, Gujarat University, Ahmedabad-380 009, India*
1983	**M.Sc. Applied Biology (by Dissertation)**, *Sir H. N. Hospital, Mumbai University, Mumbai-400 004, India.*
1980	**B.Sc. in Microbiology,** *M.. G. Science College, Ahmedabad-380 009, India.*

SPECIAL TRAININGS

2008	Comparative Genomic Hybridization array study in leukemia under the guidance of Dr. Joris Andrieux, Lille, France (8 weeks)
2006	Multipotent Adult Progenitor Stem cell training at University of Minnesota, Minneapolis, USA with Dr. Catherine Verfaillie (2 months)
2006	Stem cell Training at Coriell Institute of Medical Research, Camden, New Jersey, USA (2 weeks)
2003	Vysis sponsored hands-on FISH training using Path Vysion, Aneu Vysion and Uro Vysion FISH probes for the diagnosis of various genetic disorders at Bangkok (1 week)
2002	Visited Institute di Genetica, Bari – Italy and worked with Prof. Mariano Rocchi group for learning FISH techniques for the diagnosis of various Genetic disorders (10 weeks)
2002	European Advance Post Graduate Diploma in classical and Molecular Cytogenetics, University of Montpellier, Nimes, FRANCE (2 weeks)
1999	Visited Microbiology and Biotechnology Division, Univ. of Geneva Medical School, Cantonal Hosp. Geneva, Switzerland. Worked with Prof. S.E. Antonarakis and Prof. Potis Beris group for learning recent Cytogenetic and Molecular techniques for the diagnosis of Thalassemia and other Genetic diseases (8 weeks)
1998	Attended 4th National Congress and workshop on Prenatal Diagnosis and Therapy organized by ISPAT.
1995-96	Visited Microbiology and Biotechnology Division, Uni. Of Geneva to Medical School,

	Cantonal Hosp. Geneva,
	Switzerland. Worked with Prof. S. E. Antonarakis group for learning recent Cytogenetic and Molecular techniques for the diagnosis of Genetic diseases (8 weeks)
1995	Visited Institute Jules Bordet, Bruxelles. : Worked with Prof. Alian Verhest Group on Solid tumor and FISH (2 weeks)
1994	Attended IIIrd National workshop on Prenatal Diagnosis and Therapy organized by ISPAT
1993	Pre-Conference Workshop on DNA methodology at International Conference on Human Genetics and Family Welfare, B.J.Medical College, Pune.
1981	Training in Vaginal Cytology, Cama & Albless Hospital, Bombay (6 months)
1981	Training course on peptide hormones and RIA. At Institute for Research in Reproduction, Bombay (2 weeks)

ORGANIZATION OF SCIENTIFIC EVENTS

Jan. 2014	Organized International Conference on Human Genetics and 39th Annual meeting of ISHG Held at AMA during 22nd to 25th, 2014
Jan. 2014	Organized "Indo-German Workshop on Molecular Cytogenetics / Fluorescence In Situ Hybridization (FISH): Utility in clinical setting.21st-22nd. 2014
Jan. 2014	PCR based strategies for genetic testing. 23rd, 2014
Jan. 2014	NASI Student - Teacher interaction program 24th, 2014
Dec. 2010	Organized symposium on "Genetics in Clinical practice: Diagnosis to Therapeutics" jointly with IAP on 12th, 2010
Dec. 2009	Organized symposium on "Genetics in Clinical Practice" on 20th, 2009
Dec. 2008	Organized Winter Workshop on "Cytogenetics and Molecular Cytogenetics" at FRIGE, Ahmedabad Dec. 2008
Dec. 2007 – Jan. 2008	Organized workshop "GeneDioT-2007" at FRIGE jointly with GCRI and Gujarat University, Ahmedabad
Nov. 2008	Organized Winter workshop on "Molecular Diagnostics" at FRIGE, Ahmedabad No. 2008
Dec. 2005	Organized one-day scientific Symposium on "Obesity and Related Disorders", in collaboration with IMS, Ahmedabad
Jan. 2004	Organized 7th National conference on "Prenatal diagnosis and Therapy" during 23rd to 25th, Ahmedabad, Gujarat.
Jan. 2004	Organized two workshops "Molecular Cytogenetic Techniques" and "Antenatal Diagnostic Techniques", 20th To 23rd Jan. 2004
Dec. 2000	Organized one-day scientific symposium on "Genetic Disorders", Ahmedabad Dec. 2000
Dec. 2000	Organized Workshop on "Antenatal diagnosis of Beta- Thalassemia" at FRIGE, Genetics Centre, Ahmedabad Dec. 2000

AWARDS AND HONOURS

Dec. 2016	LIFETIME ACHIEVEMENT AWARD for Initiatives, Discoveries, and Developments in the discipline of Cytogenetics and Molecular Cytogenetics by Venus International Foundation. Chennai.
Oct. 2010	Travel award by ICMR to present the research paper at 60th Annual Meeting of ASHG, 2nd to 6th Nov. Washington DC. USA.
Sept. – Oct. 2008	UICC – ICRETT fellowship for learning Comparative Genomic Hybridization array study in leukemia at Institut de Génétique Médicale, Hôpital Jeanne de Flandre, Lille, France and worked with Dr. Joris Andrieux group
2006-07	Biography included and published by Marqui's Who is Who in the Medicine and HealthCare

Oct. 2004	Travel award by Fanconi Anemia Research Fund to present the research paper at 16th Annual International Fanconi Anemia Scientific Symposium, 14th to 17th Oct. Boston, USA.
Mar. 2003	European Cytogenetic Association fellowship for 'Candidate of Excellence'
Aug.2002	Fellow - UICC
May - Jul 2002	UICC – ICRETT fellowship for learning FISH techniques for the diagnosis of Leukemias at Institute di Genetica, Bari – Italy and worked with Prof. Mariano Rocchi group
Jan 1992 - Sept 1994	Research Associate, CSIR at Depart. of Zoology, Gujarat University, Ahmedabad.
Mar 1988 – Feb 1990	Senior Research Fellow, CSIR at Depart. of Zoology Gujarat University, Ahmedabad.
Apr 1987 - Feb 1988	Junior Research Fellow, UGC- DSA program at Depart. of Zoology, Gujarat University, Ahmedabad.
Sept 1985 - Mar 1987	Senior Research Fellow, UGC- DSA program at Depart. of Zoology, Gujarat University, Ahmedabad.
Sept 1983 - Aug 1985	Junior Research Fellow, UGC- DSA program at Depart. of Zoology, Gujarat University, Ahmedabad.

INVITED TALKS & LECTURES

- Characterising complex chromosomal re-arrangements at Global Trends in Genetic Diagnostic and Therapeutics, Hyderabad, 22nd – 23rd February, 2017
- Keynote speech on "The pleasure of finding things out" at Annual Research Meet – ARM 2016, Chennai, 3rd December, 2016
- Cytogenetics Revisited at workshop on Recent Techniques in Genetics. Gujarat University, 30th September, 2016
- Prenatal Genetic Testing - When is it "Toxic Knowledge" at BOGS and ISOPARB. Vadodara, 18th September, 2016
- Genodermatosis on Rare Disease Day at AMA on 28th February, 2016.
- A Journey from chromosome to aCGH at CME on Genetic Counseling Current Trends on 3rd January, 2016 at Sangli.
- Chromosomal Aberrations at Workshop on Perinatology on 28th June, 2015 at AMA, Ahmedabad.
- A Journey from Chromosomal Aneupolidy to CNVs - A New Paradigm Shift at National Seminar entitled "Recent Advances in Human Genetics: A Series of Lectures" on 23-24 March, 2015 at Patiala. Punjab.
- Understanding of chromosomal rearrangements - A challenge to Geneticist at 40th Annual meeting of ISHG 28-30th January, 2015
- The Joy of exploring new paradigms: Nirma University, 3rd July, 2014
- Detection and Inheritance Pattern of CNVs in MCA at Int. Con. On Hum Genet and 39th Annual Meeting of ISHG, Ahmedabad. 22nd-25th January, 2014
- Basics of FISH – Indo-German Preconference workshop at FRIGE's Institute of Human Genetics, Ahmedabad. 21st - 22nd January, 2014
- Important cases studied by FISH – Indo-German Preconference workshop at FRIGE's Institute of Human Genetics, Ahmedabad. 21st - 22nd January, 2014
- Genetic counseling for women and children. at ILS-AU-NASI Program Women in Science and Technology: The path to an empowered India. Ahmedabad 8th and 9th March 2013
- Techniques of Genetic Counseling April at Jodhpur Medical College by Rajasthan Chapter of ISPAT. Jodhpur on 21st April, 2012
- What is Karyotyping, FISH, CGH-array & SNIP–Basic "When they are indicated at Jodhpur Medical College Rajasthan Chapter of ISPAT. 22st April, 2012
- Applications of Array Comparative Genomic Hybridization in Today's Pediatric Practice at PediGen, Pune, 11th – 12th February, 2012
- Investigation of Index case at Fetal Medicine conference, Ahmedabad, 2011
- Basics of Genetics and Important cases in RFL at IMA, Godhara 18th June, 2011.
- Array-CGH – A newer tool of identifying genomic imbalance in children with MCA at Genetics in Clinical Practice. 12th December 2010.
- Role of Molecular Cytogenetics and CGH array in children with multiple congenital anomalies at Genetics in Clinical Practice. 20th December 2009.
- Cytogenetic and Molecular cytogenetic study in clinical practice. CME, Shimla. 15th July, 2009
- Scope of cytogenetics in cancer diagnosis and treatment. Muni Sevashram Cancer hospital, Goraj, Gujarat.21st March 2009

- Cytogenetic study in clinical practice. Symposium on Genetic Disorders. Dept of Pharmaceuticals, Saurastra University, Rajkot. 26th February, 2009 .
- Molecular cytogenetics in Leukemias. Symposium on Genetic Disorders. Dept of Pharmaceuticals, Saurastra University, Rajkot. 26th February, 2009
- Triple Marker followed by either AF, CVS – what are the lessons? CME, Gynac. Association. Baroda. 8thSeptember, 2008
- Clinical Application of Cytogenetics, CME of Hematological malignancies, AMA, Ahmedabad. February 2007
- Cytogenetics aspects in clinical practice. Baroda. January 2006
- Decision-making in pregnancy – Cytogenetic aspects. CME program at Palanpur. November 2005
- Basic of Genetics. Iladevi Cataract and amp IOL Research Centre, Ahmedabad. 1st April, 2004
- When and why to ask for genetic study and Triple Marker Study-Its role in Prenatal Diagnosis. Jiwaji University, Gwalior. May 2004
- Usefulness of cytogenetic study in day-to-day medical practice. CME Programme of Memnagar-Ghatlodia Medical Association. Ahmedabad. November 2002
- Usefulness of Cytogenetics in Medical Practice. At CME Programme of Obstetrics and Gynecology Society Bharuch wing. Gujarat. July 2002 .
- Recent advances in Genetics at Daman Medical Association, Daman. Gujarat. April 2001
- Molecular Biology and Its Clinical application: 6th Annual Conference of Association of Chest Physicians of Gujarat, Mt. Abu, Rajasthan. December 1999
- Trends in Genetics: The Ahmedabad GP Society CME. June 1997
- Genetic study in Genodermatosis: at XXI Annual State Conference of IADV&L, GSB. December 1996
- Laboratory investigations for metabolic disorders and chromosomal analysis: Post Graduate Revision course for Pediatrics. August 1996
- Laboratory diagnosis in spontaneous abortion: The Ahmedabad Obstetrics and Gynecological society. June 1995

PUBLICATIONS

1. Depienne C, Nava C, Keren B, Heide S, Rastetter A, Passemard S, Chantot-Bastaraud S, Moutard M, Agrawal P, VanNoy G, Stoler JM, Amor D, Billette T, Doummar D, Alby C, Cormier-Dair V, Garel C, Marzin P, Scheidecker S, Saint-Martin A, Hirsch E, Korff C, Bottani A, Faivre L, Verloes A, Orzechowski C, Burglen L, Leheup B, Roume J, Andrieux J, Sheth FJ, Datar C, Parker MJ, Pasquier L, Odent S, Naudion S, Delrue M, Caignec C, Vincent M, Isidor Bertrand, Renaldo F, Stewart F, Toutain A, Koehler U, Häckl B, Stülpnagel C, Kluger G, Moller R, Pal D, Jonson T, Soller M, Verbeek NE, van Haelst M, de Kovel C, Koeleman B, Monroe G, van Haaften G (2017). Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in *ZBTB18* and *HNRNPU*. Hum Genet. 136(4):463-479. doi: 10.1007/s00439-017-1772-0
2. Sheth JJ, Ranjan G, Shah K, Bhavsar R, Sheth FJ (2017). Novel LINS1 Missense Mutation in a Family with Non-Syndromic Intellectual Disability. Am J Med Genet A. 173(4):1041-1046. doi: 10.1002/ajmg.a.38089
3. Sheth JJ, Mistri M, Shah K, Chaudhary M, Godbole K, Sheth FJ (2016). Lysosomal storage disorders in non-immune hydrops fetalis (NIHF) - An Indian Experience. JIMD reports. DOI 10.1007/8904_2016_24 [Epub ahead of print]
4. Sheth JJ, Jijo J, Shah K, Muranjan M, Mistri M, Sheth FJ (2017). Pulmonary manifestations in Niemann-Pick type C disease with mutations in NPC2 gene: Case report and review of literature. BMC Med Genet. 18:5 DOI: 10.1186/s12881-017-0367-x
5. Solanki A, Selvaa CK, Sheth FJ, Radhakrishnan N, Kalra M, Vundinti BR (2017). Characterization of two novel *FANCG* mutations in Indian Fanconi anemia patients. Leukemia Res. 53:50-56. DOI: 10.1016/j.leukres.2016.11.013
6. Thuresson AC, Buggenhout GV, Sheth F, Kamate M, Andrieux J, Smith JC, Zander CS (2017). Whole gene duplication of *SCN2A* and *SCN3A* is compatible with normal intellectual development. Clin Genet. 91: 106–110. doi: 10.1111/cge.12797
7. Sheth FJ, Naznin L, Liehr T, Sheth JJ (2016). FISH – The Best Technique in Characterization of Prenatally Detected Small Supernumerary Marker Chromosomes (sSMC). Int J Preg Childbirth. 1(1): 00005. DOI: 10.15406/ipcb.2016.01.00005
8. Sheth JJ, Datar C, Mistri M, Bhavsar R, Sheth FJ, Shah K (2016). GM2 gangliosidosis AB variant: Novel mutation from India - a case report with a review. BMC Pediatr. 16:88. DOI 10.1186/s12887-016-0626-6
9. Sheth FJ (2015). What investigations would you suggest? Answer to MediQuiz. Indian Practitioner. 68: 37-38
10. Sheth FJ, Trivedi S, Andrieux J, Blouin JL, Sheth JJ (2015). Pure interstitial dup(6)(q22.31q22.31) - a case report. Ital J Pediatr. 41(1):5. doi:10.1186/s13052-015-0113-y
11. Sheth JJ, Shah A, Sheth FJ, Trivedi S, Lele M, Shah N, Thakor P, Vaidya R (2015). Does vitamin D play a significant role in type 2 diabetes? BMC Endocr Disord. 15:5. DOI 10.1186/s12902-015-0003-8
12. Vanlerberghe C, Petit F, Malan V, Vincent-Delorme C, Bouquillon S, Boute O, Holder-Espinasse M, Delobel B, Duban B, Vallee L, Cuisset JM, Lemaitre MP, Vantyghem MC, Pigeyre M, Lanco-Dosen S, Plessis G, Gerard M, Decamp M, Mathieu M, Morin G, Jedraszak G, Bilan F, Gilbert-Dussardier B, Fauvert D, Roume J, Cormier-Daire V,

Caumes R, Puechberty J, Genevieve D, Sarda P, Pinson L, Blanchet P, Lemeur N, Sheth FJ, Manouvrier-Hanu S, Andrieux J (2015). 15q11.2 microdeletion (BP1-BP2) and developmental delay, behaviour issues, epilepsy and congenital heart disease: a series of 52 patients. Eur J Med Genet. 58(3): 140-147.

13. Sheth JJ, Shah S, Patel H, Bhavsar R, Bhatt K, Sheth FJ (2015). A Study on Triplet Repeat Expansion Disorders in Western Indian Population. Hereditary Genet: Current Research. 4:1. http://dx.doi.org/10.4172/2161-1041.1000141
14. Sheth FJ, Rahman M, Liehr T, Desai M, Patel B, Modi C, Trived S, Sheth JJ (2015). Prenatal screening of cytogenetic anomalies - a Western Indian experience. BMC Pregnancy and Childbirth. 15:90. doi:10.1186/s12884-015-0519-y
15. Sheth H, Jackson MS, Santibanez-Koref M, Parikh K, Sheth JJ, Sheth FJ, Tyson J, Daly AK, Burn J (2015). Relevance of genetic factors to warfarin dosing in India. Blood. [E-letters]. 126:539-545. DOI: http://dx.doi.org/10.1182/blood-2015-02-627042
16. Sheth FJ, Liehr T, Shah K, Sheth JJ (2015). Prader-Willi syndrome - type 1 deletion, a consequence of an unbalanced translocation of chromosomes 13 and 15, easily to be mixed up with a Robertsonian translocation. Molecular Cytogenetics 8:52. DOI 10.1186/s13039-015-0163-2
17. Sheth H, Sheth JJ, Sheth FJ, Burn J (2015). The poor patient with diabetes 'should live like a saint'. Diabet Med.. 33(1):134-5.
18. Sheth JJ, Shah A, Sheth FJ, Trivedi SN, Nabar N, Shah N, Thakore P, Vaidya R (2015). The association of dyslipidemia and obesity with glycated hemoglobin. Clinal Diabet Endocrinol. 1:6. DOI 10.1186/s40842-015-0004-6
19. Sheth JJ, Mistri M, Bhavsar R, Patel H, Sheth FJ (2015). Novel mutation in the XPC gene: a case report of a patient with xeroderma pigmentosum. Int J Dematol. 54(11):e487-91. DOI:10.1111/ijd.13022
20. Sheth JJ, Mistri M, Bhavsar R, Sheth FJ, Kamate M, Shah H, Datar C (2015). Lysosomal Storage Disorders in Indian children with neuroregression attending genetic center. Indian Pediatr. 52(12):1029-33.
21. Sheth FJ, Sheth JJ, Shah K (2015). A Journey from Chromosomal Aneuplody to CNVs – A New Paradigm Shift. Pathol Lab Medicine. 7(2):89-91.
22. Sheth JJ, Sheth FJ, Shah K (2015). Next Generation Sequencing in Genetic Diagnosis. Pathol Lab Medicine. 7(2):86-88
23. Sheth JJ, Mistri M, Sheth FJ, Shah R, Bavdekar A, Godbole K, Nanavati N, Datar C, Kamate M, Oza N, Ankleshwaria C, Mehta S, Jackson M (2014). Burden of lysosomal storage disorders in India: experience of 387 affected children from a single diagnostic facility. JIMD reports. 12:51-63. DOI 10.1007/8904,2013_2014
24. Sheth FJ, Sheth H, Kumari P, Tewari S, Desai M, Pate B, Sheth JJ (2014). Evolution of Cytogenetics in Disease Diagnosis. J Translational Toxicol. 1(1): 3-9. DOI: http://dx.doi.org/10.1166/jtt.2014.1008
25. Ankleshwaria C, Mistri M, Bavdekar A, Muranjan M, Dave U, Tamhankar P, Khanna V, Jasinge E, Nampoothiri S, Kumar S, Sheth FJ, Gupta S, Sheth JJ (2014). Novel mutations in the glucocerebrosidase gene of Indian patients with Gaucher disease. J Hum Genet. 59(4): 223-228 doi:10.1038/jhg.2014.5
26. Sheth JJ, Mistri M, Datar C, Kalane U, Patil S, Kamate M, Shah H, Nampoothiri S, Gupta S, Sheth FJ (2014). Expanding the spectrum of HEXA mutations in Indian patients with Tay–Sachs disease. Mol Genet Metabol Rep. 1:425–430. http://dx.doi.org/10.1016/j.ymgmr.2014.09.004
27. Sheth JJ, Mistri M, Sheth FJ, Datar C, Godbole K, Kamate M, Patil K (2014). Prenatal diagnosis of Lysosomal Storage disorders by enzymes study using chorionic villus and amniotic fluid. J Fetal Med. 1: 17. doi:10.1007/s40556-014-0001-3
28. Sheth FJ, Datar C, Andrieux J, Pandit A, Nayak D, Rahman M, Sheth JJ (2014). Distal Deletion of Chromosome 11q Encompassing Jacobsen Syndrome without Platelet Abnormality. Clin Med Insight – Pediatr 2014;8:45-49. doi: 10.4137/CMPed.S18121
29. Shah H, Sheth FJ, Pandit VS, Langanecha B (2013). Bloom Syndrome: report of two cases in siblings. Int Dematol. 52(8):990–992.
30. Sheth FJ, Kaul M (2013). Diagnostic dilemma in overlapping congenital syndromes. Indian Pediatr. 50(1):157-158.
31. Asadollahi R, Oneda B, Sheth FJ, Azzarello-Burri S, Baldinger R, Joset P, Latal B, Knirsch W, Desai S, Baumer A, Houge G, Andrieux J, Rauch A (2013). Dosage changes of *MED13L* further delineate its role in congenital heart defects and intellectual disability. Euro J Hum Genet. 21(10):1100-1104.
32. Mampilly K, mampilly T, Chandramohan N, Velayutham M, Sheth JJ, Sheth FJ, Janaki V (2013). Prenatal dolichocephaly: sign of trouble? -A variant of Miller-Dieker syndrome. Fetal and Pediatric Pathology, 32(4):308-311.
33. Sheth JJ, Patel H, Mehta S, Tewari S, Sheth FJ (2013). Clinical and Molecular characterization of patients with gross hypotonia and impaired lower motor neuron function. Indian Pediatr. 50:591-593.
34. Sheth FJ, Andrieux J, Tewari S, Sheth H, Desai M, Kumari P, Nanavati N, Sheth JJ (2013). Chromosomal imbalance letter: Phenotypic consequences of combined deletion 8pter and duplication 15qter. Mol Cytogenet. 6(1):24. doi:10.1186/1755-8166-6-24
35. Sheth FJ, Liehr T, Kumari P, Akinde R, Sheth HJ, Sheth JJ (2013). Chromosomal Abnormalities In Couples With Repeated Fetal Loss: An Indian Retrospective Study. Indian J Hum Genet. 19(4); 415-422.
36. Sheth J, Mistri M, Kamate M, Vaja S, Sheth FJ (2012). Diagnostic strategy for Mucolipidosis II/III. Indian Pediatr. 49(12):975-977.
37. Mistri M, Tamhankar PM, Sheth FJ, Sanghavi D, Kondurkar P, Patil S, Thomas S; Gupta S; Sheth JJ (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
38. Sheth JJ, Ankleshwaria C, Pawar R, Sheth FJ (2012). Identification of Novel Mutations in FAH Gene and Prenatal Diagnosis of Tyrosinemia in Indian Family. Case Rep Genet. Article ID 428075, 4 pages. doi:10.1155/2012/428075
39. Othman M, Lier A, Junker S, Kempf P, Dorka F, Gebhart E, Sheth FJ, Grygalewicz B, Bhatt S, Weise A. Mrasek K,

Liehr T, Manvelyan M (2012). Does positioning of chromosomes 8 and 21 in interphase drive t(8;21) in acute myelogenous leukemia? BioDiscovery. 4:2; DOI: 10.7750/BioDiscovery.2012.4.2

40. Sheth FJ, Akinde OR, Datar C, Adeteye OV, Sheth JJ (2012).Genotype-phenotype characterization of wolf-Hirschhorn syndrome confirmed by FISH - Case Reports. Case Rep Genet. 878796. doi: 10.1155/2012/878796.
41. Sheth FJ, Gohel N, Liehr T, Akinde O, Desai M, Adeteye O, Sheth JJ (2012). Gain of chromosome 4qter and loss of 5pter – an unusual case with features of Cri du chat syndrome. Case Rep Genet. vol. 2012, Article ID 153405, 4 pages, doi:10.1155/2012/153405
42. Sheth JJ, Shah H, Sheth FJ (2011). Infantile Glaucoma with coarse facial features as an early complication of Hurler-Scheie. Pediatric oncall. Pediatric Oncall 8(1):22-23.
43. Sheth FJ, Andrieux J, Ewers E, Kosyakova N, Weise A, Sheth H, Romana S, Lorch M, Deloben B, Theisen O, Liehr T, Numpoothiri S, Sheth JJ (2011). Characterization of sSMC by FISH and molecular techniques. Euro J Med Genet. 54(3):247-255.
44. Sheth H, Blouin JL, Sheth JJ, Sheth FJ (2011). Triple-X syndrome in a trisomic Down syndrome child: Both aneuploidies originated from the mother. Int J Hum Genet. 11(1):51-53
45. Sheth JJ, Shah U, Sheth FJ, Shah N, Vaidya R, Vaidya A (2011). Genoprotective Effect of Indian Gentian in Type 2 Diabetes Mellitus (T2DM): Comet Assay, Sister Chromatid Exchanges and Protein Oxidation studies. Int J Hum Genet. 11(2):83-88.
46. Sheth FJ, Pani J, Desai M, Mehta S, Sheth JJ (2011). Single Cell Abnormality in couples with Bad Obstetric History and Repeated Fetal Loss: Occurrence and Clinical outcome. Int J Hum Genet. 11(4):271-276.
47. Sheth FJ, Shah U, Desai M, Sheth JJ (2011). Clinical Profile of Inversion Y in People of Gujarat, West India. Int J Hum Genet. 11(4):245-248.
48. Sheth JJ, Sheth FJ, Oza NJ, Gambhir PS, Dave UP, Shah RC (2010). Plasma Chitotriosidase activity in children with lysosomal storage disorders. Indian J Pediatr. 77(2):203-205.
49. Sheth F, Andrieux J, Sheth J (2010). Supernumerary marker chromosome in a child with microcephaly and mental retardation. Indian Pediatr. 47(3):277-279.
50. Sheth JJ, Mistri M, Godbole K, Sheth FJ (2010). Predominance of Morquio-B (Mucopolysaccharidosis IV-B) in children with skeletal dysplasia. Pathol Lab Medicine. 2:29-36.
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