BIODATA

1) Name: Dr. A. Haseena

2) Email and contact number: hasi.flower@gmail.com

3) Institution: SGPGI, Lucknow

4) Date of Birth: 14th March 1991

6) Professional details:

S.No	Degree	Date of Passing	Attempts	Grade	Institution	University	Merit/Prizes/Medals
1	DM Medical Genetics	11/08/2022	First	-	SGPGI, Lucknow		
2	MD Pediatrics	08/05/2018	First	-	Maulana Azad Medical College, New Delhi	Delhi University	
3	MBBS	02/2013	First	76%	Madras Medical College, Chennai	The Tamil Nadu Dr. M.G.R Medical University	Distinction in microbiology, pharmacology, ophthalmology, general surgery. Honors in Pathology, oto-rhino-laryngology.

7) Professional Recognition/ Award/ Prize/ Certificate, Fellowship

S. No	Name of Award	Awarding agency	Year
1	Winner in the case presentation series in 5th National Conference on Primary Immunodeficiency Diseases. March 6-7	ISPID	2021
2	Runner up in Platform quiz ISIEM -Pune	ISIEM	2019
3	11th IAP pediatric quiz for postgraduates- winner in college round	Indian Academy of Pediatrics	2017
4	Winner of Satya Gupta Award in research paper category on Social Pediatrics	Indian Academy of Pediatrics	2017
7	Mrs Visalam Vijayan award for highest mark in physics in higher secondary school	Good Shepherd HSS	2008

8) Professional courses/Seminars, Workshops/Conferences attended

S no	Name	Year
1	Basic and Advanced life support skills and communication skills MAMC post graduate advanced life support course	2016
2	4th DHR-ICMR-PGI workshop on Primary Immunodeficiency diseasesAdvanced Pediatrics Centre, PGIMER,Chandigarh	2016
3	Badic Pediatric Cardiology Refresher Course Lady hardinge Medical College,New Delhi	2016
4	Provider Course of Advanced Life support MAMC, New Delhi	2017
5	Annual CME in Pediatrics Lady hardinge Medical College,New Delhi	2017
6	Pre conference workshop “At risk adolescent: Challenges and oppurtunities” , MAMC Delhi	2017
7	12th Asia Pacific Congress on Human Genetics, Thailand	2017
8	10th ISNS -Asian Pacific Region, Mongolia	2017
9	PIDCON, New Delhi	2017
10	Pediatric Conference of North India and National Conference of Research in Child Health Group	2017
11	IX Annual Training Course in Pediatric Nephrology	2017
12	Clinical Course for Postgraduates, UCMS, New Delhi	2017
13	2nd AOCN Masterclass in Pediatric Neurology for postgraduates	2018
14	Update on Prenatal and Newborn Screening MAMC, New Delhi	2019
15	Advanced Genomics in Health and Disease FRIGE’S institute of Human Genetics, Ahmedabad	2019
16	CME of genetics	2019
17	ISIEM 2019 national conference, Pune	2019
18	Sixth Annual National Conference of the Society for Indian Academy of Medical Genetics 2019, Hyderabad	2019
19	Post conference workshop on “Decoding Genetic Investigations: Interpretation of NGS and other Genetic Tests in clinical setting, NIMS and CDFD, Hyderabad	2019

20	Trainer for capacity building programme of SGPGI, Lucknow for pediatric COVID19 preparation	2019
21	25th International Conference on Prenatal Diagnosis and Therapy (Virtual)	2021
22	5th National Conference on Primary Immunodeficiency Diseases SGPGIMS, Lucknow	2021
23	6th ISIEM 2021 National Conference (Virtual)	2021
24	Faculty in Indo German Metabolic Meet 2021(IGMM)	2021
25	Next Generation Sequencing data analysis for clinical diagnostics CDFD, Hyderabad	2021
26	Workshop on interpretation of genomic sequence variations 7th Annual conference of Society for Indian Academy of Medical Genetics	2022
27	14th ISPAT Biennial Conference	2022
28	Demonstrator in 19th ICMR course on Medical Genetics and Genomic Counselling	2022
29	Demonstrator in 20th ICMR course on Medical Genetics and Genomic Counselling	2023
30	Faculty in genetic Counselling Workshop in 10th Annual Conference of Molecular Pathology Association of India, Dept of Hematology, SGPGIMS	2023

9) Presentations

S no	Title of presentation	Mode	Name of conference	Year
1	Correlation between maternal urinary iodine and neonatal TSH >5mIU/L level	Poster	12th Asia Pacific Congress on Human Genetics, Thailand	2017
2	Iodine excess and deficiency: Two sides of a coin	Oral	10th ISNS -Asian Pacific Region, Mongolia	2017
3	Iodine deficiency and iodine excess- A matter of public health importance	Oral	PIDCON, New Delhi	2017
4	Etiological and Molecular profile of Congenital Hypothyroidism: Insights From A Prospective Multi-centric Study On Newborn Screening In Delhi	Oral	ISIEM 2019 national conference, Pune	2019
5	Kallmann Syndrome Caused By Disruption of KAL1 Gene due to Translocation between Chromosomes X and Y	Poster	Sixth Annual National Conference of the Society for Indian Academy of Medical Genetics 2019, Hyderabad	2019

6	Prenatal presentation of a blended phenotype due to partial monosomy 9p and partial trisomy 2p	Poster	25th International Conference on Prenatal Diagnosis and Therapy (Virtual)	2021
7	Phenotypic and genotypic spectrum of CTSK variants in a cohort of twenty-five Indian patients with Pycnodysostosis	Poster	Research Day, SGPGIMS	2021
8	Inborn errors of ketogenesis: Novel variants, clinical presentation and follow up in a series of four patients	Poster	6th ISIEM 2021 National Conference (Virtual)	2021
9	Comprehensive study of prenatal diagnosis for monogenic disorders	Oral	14th ISPAT Biennial Conference	2022
10	Haemophilia management programme: Transformation during COVID-19	Poster	Research Day, SGPGIMS	2022
11	Genotypic and Phenotypic Landscape of Osteogenesis Imperfecta from India	Poster	7th Annual conference of Society for Indian Academy of Medical Genetics	2022

10) Membership of professional societies, bodies / associations

S No	Status	Name	Date of Membership
1	Trainee Membership	International Society for Prenatal Diagnosis	09/02/2022
2	Life Member	Society for Indian Academy Of Medical Genetics	08/11/2022
3	Life Member	AL/2017/H-72 Indian Academy of Pediatrics	2017
4	Life Member	The Indian society for Inborn Errors of Metabolism	2023

11) Papers published

S No	Authors	Title	Journal	Vol	Page	Yr
1	**Haseena Sait**, Seema Kapoor, Ankur Jindal, Ritika Garg, Ravi Shankar Belwal, Sangita Yadav, Sangeeta Gupta, Bk Thelma	Association Between Neonatal Thyroid Stimulating Hormone Status and Maternal Urinary Iodine Status	Indian Pediatrics	56	472-475	2019

2	**Haseena Sait**, Ashish Jain*1*, Seema Kapoor	Congenital Midline Cervical Cleft in a Neonate Born to Diabetic Mother – An Unusual Association	Journal of Clinical Neonatology	8	178-9	2019
3	Ritika Garg & **Haseena Sait** & Ankur Jindal& Monica Juneja & Sangeeta Gupta & BK Thelma & Seema Kapoor	Factors Associated with Transient Neonatal Hyperthyrotropinemia	Indian Journal of Pediatrics	6	482-3	2020
4	Priyanka Verma, Seema Kapoor1, **Haseena Sait**, Sujoy Ghosh, Sunita Gupta	Dental phenotype of multiple impacted supernumerary teeth in Wiedemann-Steiner syndrome	Journal of cleft lip palate and craniofacial anomalies	7	59-63	2020
5	**Haseena Sait**, Puneet Kaur Sahi And Seema Kapoor	Klippel Trenaunany syndrome- A rare cause for portal hypertension	Indian Pediatrics	57	754-755	2020
6	Francisca Díaz-González, Saruchi Wadhwa, Maria Rodriguez-Zabala,Somesh Kumar,Miriam Aza-Carmona,Lucia Sentchordi-Montané,Milagros Alonso-Blanco, Istaq Ahmad, Sana Zahra,Deepak Kumar, Neetu Kushwah, Uzma Shamim, **Haseena Sait**, Seema Kapoor, M Belen Rodan	Biallelic CGMP dependent type II protein kinase gene (PRKG2) variants cause a novel acromesomelic dysplasia	Journal of Medical Genetics	0	1-11	2020
7	**Haseena Sait** & Raghvendra Singh & Seema Kapoor	Bartter-Like Renal Phenotype in a Child with Donnai–Barrow Syndrome	Indian Journal of Pediatrics	88	390	2021
8	Somesh Kumar, **Haseena Sait**, Sunil K. Polipalli, Gaurav S. Pradhan ,Sumit Pruthi, Seema Kapoor	Loes Score: Clinical and Radiological profile of 22 patients of X-linked adrenoleukodystrophy: Case Series from a single centre	Indian Journal of Radiology and Imaging	31	383-390	2021
9	**Haseena Sait**, Raghvendra Singh, Seema Kapoor	An Unusual Occurrence of Protein C Deficiency and Cytomegalovirus Infection in a Case of Purpura Fulminans	Indian journal of dermatology	66	201-203	2021
10	**Haseena Sait**, Harikrishnan Gangadharan , Aviral Gupta , Amita Aggarwal , Manoj Jain Shubha R. Phadke	Monogenic Lupus with IgA Nephropathy Caused by Spondyloenchondrodysplasia with Immune Dysregulation	Indian journal of Pediatrics	88	819-823	2021

11	**Haseena Sait** , Priyanka Srivastava, Neerja Gupta , Madhulika Kabra , Seema Kapoor , Prajnya Ranganath , Ikrormi Rungsung , Kausik Mandal , Deepti Saxena , Ashwin Dalal , Ajitesh Roy , Jayalakshmi Pabbati , Shubha R Phadke	Phenotypic and genotypic spectrum of CTSK variants in a cohort of Twenty-Five Indian patients with Pycnodysostosis	European Journal of Medical Genetics	64	104235	2021
12	**Haseena Sait** , Priyanka Srivastava , Preeti Dabadghao, Shubha R Phadke	Kallmann Syndrome and X linked Ichthyosis due to Translocation between Chromosomes X and Y in Journal of Reproduction and Infertility	Journal of Reproduction & Infertility	22	302-306	2021
13	**Haseena Sait**, Lokesh Sharm, Preeti Dabadghao, Shubha R. Phadke	Congenital Hyperinsulinemia of Infancy: Role of Molecular Testing in Management and Genetic Counseling	Indian Journal of Pediatrics	89	395-398	2022
14	**Haseena Sait**, Amita Moirangthem, Vinita Agrawal, Shubha R. Phadke	Autosomal recessive spinocerebellar ataxia-20 due to a novel SNX14 variant in an Indian girl	American Journal of Medical Genetics	188	1909-1914	2022
15	**Haseena Sait,** Somya Srivastava, Deepti Saxena	Integrated Management Strategies for epidermolysis bullosa: current insights	International Journal of General Medicine	15	5807-5808	2022
16	**Haseena Sait,** Somya Srivastava, Somesh Kumar, Bijo Varughese, Manmohan Pandey, Manjunath Venkatramaiah, Parul Chaudhary, Amita Moirangthem, Kausik Mandal, Seema Kapoor	Inborn Errors of Ketogenesis: Novel Variants, Clinical Presentation, and Follow-Up in a Series of Four Patients	Journal of Paediatric Genetics	-	-	2022
17	Suzena Masih , Amita Moirangthem , Arya Shambhavi , Archana Rai , Kausik Mandal , Deepti Saxena , Mayank Nilay, Neha Agrawal , Somya Srivastava , **Haseena Sait** , Shubha R. Phadke	Deciphering the molecular landscape of microcephaly in 87 Indian families by exome sequencing	European Journal of Medical Genetics	65	104520	2022
18.	**Haseena Sait**, Shruti M. Sajjan & Shubha R. Phadke	Haemophilia management programme: Transformation during COVID-19	Indian Journal of Medical Research		472-474	2022

19.	**Haseena Sait,** Arya Shambhavi, Manmohan Pandey, Deepak Ravichandran, Shubha R. Phadke	T2 olivary nuclei hyperintensities: A characteristic neuroimaging finding in FIG4 related leukoencephalopathy	American Journal of Medical Genetics	191A	864-869	2023
20.	**Haseena Sait**, Somya Srivastava, Manmohan Pandey, Deepak Ravichandran, Anju Shukla, Kausik Mandal, Deepti Saxena, Arya Shambhavi, Purvi Majethia, Lakshmi Priya Rao, Suvasini Sharma, Shubha R. Phadke, Amita Moirangthem	Neurodegeneration with brain iron accumulation: a case series highlighting phenotypic and genotypic diversity in 20 Indian families	Neurogenetics	24	113-127	2023
21.	**Haseena Sait,** Manmohann Pandey, Shubha Phadke	COQ7 splice site variant causing a spastic paraparesis phenotype in siblings	Journal of Genetics	103	1-8	2024
22.	Deepashree K Rao, Ankur Jindal, Aashima Dabas, **Haseena Sait**, Sangeeta Yadav,Seema Kapoor	Effect of Maternal Iodine Excess during Pregnancy on Neonatal Thyroid Function and Neurodevelopmental Status at 12 Weeks	Journal of the ASEAN Federation of Endocrine Societies	-	-	2024
23.	Pooja Motwani, **Haseena Sait**	Immune dysregulation in a dysmorphic child with 6q23.3 deletion: a single case report	Clinical Dysmorphology	-	-	2024

12) Chapters contributed

1) Moderator for *NFIX* gene - Human diseases genes website series (https://humandiseasegenes.nl/moderators)
2) Moderator for *SNX14* gene - Human diseases genes website series (https://humandiseasegenes.nl/moderators)

13) Articles written in Genetic Clinics (First author)

Title	Volume	Year
Antisense Oligonucleotides: Adding Sense to Therapeutic Medicine	Jan – March, Vol 13, Issue 1	2020
Genetics of Diabetes Mellitus	July-Sept, Vol 13, Issue 3	2020
Genetic Counselling of Prenatally Detected Sex Chromosome Anomalies	July-Sept, Vol 14, Issue 3	2021
A New and Exciting Era of Genomics: No Region is Beyond Reach	January - March | Vol 15 | Issue 1	2022

Balancing the Sweetness of the Family Between Monogenic Hypoglycemia and Multifactorial Hyperglycemia	July - September | Vol 15 | Issue 3	2022
Validation of MLPA-detected Single Exon Deletion of the DMD Gene by Multiplex PCR	October-December | Vol 16| Issue 4	2023
Mosaicism in Clinical Genetics: Counselling Challenges and Diagnostic Dilemmas	July-September/Vol 17/Issue 3	2024