E-POSTER PRESENTATION PROGRAM

DAY 01 / 21 Sept.2024		SCREEN 01	
Abstract No.	**Topic**	**Presenter**	**Institute**
ABS002	BEYOND THE ORDINARY: A CASE REPORT ON MEPAN SYNDROME (By Dr. Sakshi Mathur, Dr. Priyanshu Mathur, Department of Paediatrics, SMS Medical College, Jaipur)	Dr. Sakshi Mathur	Jk Lon hospital, SMS medical college
ABS006	BIOTINIDASE DEFICIENCY: MIMIC OF SUBACUTE COMBINED DEGENERATION ON MRI SPINE	Kuldeep Nehra	SMS Hispital, Jaipur
ABS011	A RARE CASE OF SANFILIPPO SYNDROME WITH HOMOZYGOUS PATHOGENIC VARIANT OF HGSNAT GENE ON EXON 17	Dr. Oshima Goyal	SMS Medical College, Jaipur
ABS012	A case series on Morquio Syndrome in SPINPH and Attached group of hospitals	Urja Singhal	Sms hospital , Jaipur
ABS013	CREATINE DEFICIENCY DISORDER- A TREATABLE CAUSE OF AUTISM AND DEVELOPMENTAL DISORDER IN CHILDREN- A TERTIARY CARE CENTRE EXPERIENCE.	PRATHIBHA K	SRIHER
ABS015	RARE TRANSLOCATIONAL VARIANT OF DOWNS SYNDROME t(14;21) WITH CONGENITAL HYPOTHYROIDISM & CONGENITAL HEART DISEASE : A CASE REPORT	DR. SHREYANSH SHARMA	Jhalawar medical college ,jhalawar
ABS017	PYRUVATE DEHYDROGENASE COMPLEX (PDHC) DEFICIENCY DISEASE WITH UNUSUAL INITIAL PRESENTATION OF STROKE LIKE EPISODES AND RESPONSIVE TO THIAMINE	SONALI KACHRU GAUR	Sawai Man Singh Medical College, Jaipur
ABS018	A Case of Autosomal Dominate Lissencephaly type-1	Aditi yadav	SMS medical college and hospital
ABS022	A Sudden Turn: Navigating a diagnosis for Angiokeratomas	Dr Nehal Athreyi R	RNT medical college, Udaipur
ABS025	Sweetness can be Deadly: An Insight into Disorders of Fructose Metabolism	Soumi Das	PGIMER, Chandigarh
ABS026	THE X-FACTOR: MANIFESTATIONS OF X-LINKED METABOLIC DISORDERS IN FEMALES	Divya Thomas	Sir Ganga Ram Hospital
ABS027	Sandoff disease presenting as seizure	Dr Priyanka Kainthola	J K Lone Hospital

DAY 01 / 21 Sept.2024		SCREEN 02	
Abstract No.	**Topic**	**Presenter**	**Institute**
ABS028	UNMASKING NEURONAL CEROID LIPOFUSCINOSES 8: THE SILENT DRIVER OF PROGRESSIVE NEURODEGENERATION	SWATI GARG	
ABS030	UNUSUAL CASE OF MULTISUTURAL CRANIOSYNOSTOSIS IN ALPHA MANNOSIDOSIS	Lois Sara James	Christian Medical College
ABS033	LYSINURIC PROTEIN INTOLERANCE ??CLINICALLY HETEROGENOUS PRESENTATIONS	George Varghese Mani	Christian Medical College Vellore
ABS034	RARE CO-EXISTENCE OF MILLER DIEKER SYNDROME AND PHELAN-MCDERMID SYNDROME	Nitish Garg	SMS medical college
ABS035	HYALINE FIBROMATOSIS SYNDROME: A RARE INHERITED DISORDER	Aakash sharma	SMS medical college
ABS036	Understanding and Managing Phoshoglycerate Dehydrogenase Deficiency: A case Report and Literature Review	Mayank Nilay	Post Graduate Institute of Child Health
ABS037	RICKETS IN A CHILD WITH TYROSINEMIA TYPE 1 : A COMMON DISEASE WITH AN UNCOMMON ETIOLOGY	Dr Pradeep Kumar	AIIMS Patna
ABS039	ASPARAGINE SYNTHETASE DEFICIENCY: A RARE GENETIC DISORDER	Raveena Gehlot	Sawai Mansingh Hospital
ABS044	"Unraveling the Complexities of NBIA: A Neurological Perspective on Brain Iron Accumulation Disorders"	shubham satyanarayan agrawal	
ABS047	A RARE CASE REPORT WITH TWO GENE DEFCTS - EXPLORING THE WIDE VARIETY OF PHENOTYPICAL FEATURES ASSOCIATED WITH RASOPATHIES	Dr Saloni Sinha	SMS Medical Collage
ABS048	Case series of Metabolic cohort from a peripheral South Indian city	Kiruthika S	Salem Genetics Centre
ABS050	a case report on megalencephalic leucoencephalopathy: a rare genetic disease	Anjali gupta	Sms medical college, jaipur

DAY 01 / 21 Sept.2024		SCREEN 03	
Abstract No.	**Topic**	**Presenter**	**Institute**
ABS051	CARNITINE PALMITOYL TRANSFERASE ??1 DEFICIENCY ??CASE SERIES FROM A SINGLE CENTRE EXPERIENCE	Jagruthi Prathipati	
ABS052	X-LINKED HYPOPHOSPHATEMIC RICKETS: A NOVEL DE-NOVO MUTATION IN PHEX GENE	Dr. Rivneet Kaur	Sawai Maan Singh Medical College, Jaipur
ABS055	ISOLATED SULFITE OXIDASE DEFICIENCY CAUSING REFRACTORY SEIZURES IN A MODERATE PRETERM NEONATE-A RARE CASE STUDY	SHARANARAYAN M	SRI RAMACHANDRA INSTITUTE OF HIGER EDUCATION AND RESEARCH
ABS057	A rare cause of Progressive Myoclonic Epilepsy-Neuronal Ceroid Lipofuscinosis	KASAIAH SAIRAM	
ABS058	A rare cause of Progressive Myoclonic Epilepsy - Neuronal Ceroid Lipofuscinosis	KASAIAH SAIRAM	
ABS059	HOW RARE CAN BE RARE DISORDERS: UNFOLDING AN ACUTE ENCEPHALOPATHY	Dr M VIJAYALAKSHMI	Apollo women?? hospital
ABS060	NEWBORN SCREENING DETECTED NON CLASSICAL HYPERPHENYLALANIMEIA-Immediate management and follow up	Dr M VIJAYALAKSHMI	Apollo women?? hospital
ABS061	PROGRESSIVE NEURO-DEGENERATIVE DISORDER IN CHILDREN - never miss out lysosomal storage disorder	Dr Anjali Sharma	.
ABS065	Description of dramatic therapeutic responses in two rare adulthood onset metabolic diseases: glutaric acidemia-1 and glutaric acidemia-2	Dr Shruti Bajaj	..
ABS068	A case of Coarse facies, multiple skeletal abnormalities with persistent oxygen requirement	Abhinav Vishwakarma	INHS Asvini
ABS069	RARE CO OCCURRENCE OF CARNITINE DEFICIENCY WITH BETAKETOTHIOLASE DEFICIENCY: DIAGNOSTIC DILEMMA OF PRIMARY VS SECONDARY	Dr Ashka Prajapati	Neuberg Centre For Genomic Medicine
ABS072	Transient infantile hypertriglyceridemia- An unusual cause of hepatosplenomegaly with fibrosis	Tanuja Rajial	Aiims Jodhpur

DAY 01 / 21 Sept.2024		SCREEN 04	
Abstract No.	**Topic**	**Presenter**	**Institute**
ABS074	MEDNIK SYNDROME - A RARE DISORDER OF COPPER METABOLISM	Dr Sarah Bailur	Rainbow Childrens Hospital
ABS076	BIOTINIDASE DEFICIENCY : TREATABLE METABOLIC DISORDER	Dr Puneet Sharma	.
ABS077	SPINOCEREBELLAR ATAXIA 13 - A RARE CASE PRESENTATION	Dr Anmol Meena	.
ABS078	Biochemical Assays ??Important tools in diagnosis of Lysinuric Protein Intolerance	Dr. Papai Roy	Sir Ganga Ram Hospital
ABS079	ORGANIC ACIDEMIAS: EARLY DETECTION AND MANAGEMENT IN NEONATAL PERIOD?? NICU PERSPECTIVE	Dr.M.Arjun	Sriramachandra medical college and research institute
ABS081	Biotinidase Deficiency: A Case Series Highlighting the Importance of Early Diagnosis and Lifelong Management	Meghana S	Sri Ramachandra Institute of Higher Education and Research
ABS082	Profile of Metabolic Disorders at a Tertiary Care Centre in Central India- Need for increased awareness and Challenges faced.	Dr Jignesh Sharma	AIIMS BHOPAL
ABS084	SPECTRUM OF TREATABLE NEUROMETABOLIC DISEASES OF CHILDHOOD IN A TERTIARY CARE TEACHING HOSPITAL OF SOUTH INDIA- HOPE FOR FUTURE	INDRASISH RAY CHAUDHURI	Amrita Institute of Medical Sciences, Kochi
ABS085	Clinical, Biochemical, and Molecular Analyses of Medium-Chain Acyl-CoA Dehydrogenase Deficiency in Indian Patients	Dr Sarah Bailur	Rainbow Childrens Hospital
ABS088	Rare renal presentation of Gaucher disease: A case report	Adarsha	SGPGI, LUCKNOW
ABS089	PHENOTYPE AND GENOTYPIC CHARACTERIZATION OF SANFILIPPO SYNDROME: A CASE SERIES	Adarsha	SGPGI, LUCKNOW
ABS091	MENKES DISEASE: REFRACTORY EPILEPSY, NEUROREGRESSION AND UNIQUE HAIR CHARACTERISTICS	Dr Sarthak Khandelwal	J.K. Lone Hospital, Jaipur

DAY 01 / 21 Sept.2024		SCREEN 05	
Abstract No.	**Topic**	**Presenter**	**Institute**
ABS093	CITRULLINEMIA MASQUERADING AS FAILURE TO THRIVE	Niharika	SPINPH,SMS medical college,jaipur.
ABS094	Genetic Insight into Recurrent Fever-Unravelling the mystery	Shivangi	AIIMS Bhopal
ABS095	Liver Dysfunction following acitretin treatment in a case of Chanarin Dorfman Syndrome	Tanuja Rajial	Aiims Jodhpur
ABS097	PHENOTYPE-GENOTYPIC SPECTRUM OF A COHORT OF INDIAN PATIENTS WITH CONGENITAL DISORDERS OF GLYCOSYLATION	Devi Saranya S	AIIMS, New Delhi
ABS098	Succinic Semialdehyde Dehydrogenase (SSADH) deficiency-A case report.	DR VARSHA VIRANCHI VAIDYA	KPOND SUPERSPECILITY HOSPITAL
ABS101	GAIT ??A clue to a rare inherited disorder.	Dr Kandha Kumar U K	AIIMS Jodhpur
ABS105	ASAH1 ??elated disorders: Farber lipogranulomatosis to spinal muscular atrophy with progressive myoclonic epilepsy	Rahul Gupta	AIIMS Jodhpur
ABS109	The diverse clinical manifestations of homocystinuria: a series of 9 cases from a tertiary care hospital	Sahil Mathur	NIMHANS Bengaluru
ABS110	Fructose-1, 6-bisphosphatase deficiency ??a preventable metabolic disorder with distinctive biochemical features	Apoorva	
ABS112	Clinical, radiological and genetic characteristics of children with NDUFV1-related mitochondrial complex 1 deficiency	ARUSHI GAHLOT SAINI	PGIMER
ABS113	The spectrum of typical and atypical features in children with genetically confirmed Alexander disease with three novel variants	ARUSHI GAHLOT SAINI	PGIMER
ABS114	GAUCHER DISEASE: PHENOTYPE AND GENOTYPE FROM TERTIARY CARE CENTER, RAJASTHAN	Dr Toshika Agarwal	