E-POSTER PRESENTATION PROGRAM

DAY 02 / 22 Sept.2024		SCREEN 01	
Abstract No.	**Topic**	**Presenter**	**Institute**
ABS115	Inborn Error of metabolism masquerading as sick infant at tertiary paediatric care referral hospital of North India.	Dr Diksha Asati	Hope Children hospital
ABS118	A NEW PERSPECTIVE IN DIAGNOSIS OF AUTOIMMUNE HEMOLYTIC ANEMIA	Dr Pokar Chand	
ABS119	Reporting first case of Osteo-oto-hepato-enteric syndrome (OOHE) in an Indian neonate presenting with intractable diarrhoea and cholestasis.	Dr Diksha Asati	Hope Children hospital
ABS120	Developmental and epileptic encephalopathy -5 (DEE5): A novel mutation in SPTAN1 gene, a Case report	DR DHAN RAJ BAGRI	
ABS122	A COMPREHENSIVE CASE SERIES ON HYPOMYELINATING LEUKODYSTROPHY: CLINICAL MANIFESTATIONS, IMAGING AND GENETIC ANALYSIS	Jigil Joy	NIMHANS
ABS124	A rollercoaster ride of blood glucose from HYPO to HYPER- a double whammy	Dr. Veena Bharathi Kota	.
ABS125	JUVENILE FORM OF METACHORMATIC LEUKODYSTROPHY, A RARE CASE REPORT	Dr SAKSHI SHARMA	.
ABS126	Prenatal diagnosis of inborn errors of metabolism(IEM) in tertiary care centre	Roja Ravali Boppana	Rainbow children?? hospital

DAY 02 / 22 Sept.2024		SCREEN 02	
Abstract No.	**Topic**	**Presenter**	**Institute**
ABS129	LATE FORM OF METACHORMATIC LEUKODYSTROPHY A RARE CASE REPORT	Dr SAKSHI SHARMA	.
ABS131	USE OF OCTREOTIDE SUBCUTANEOUS INFUSION IN CONGENITAL HYPERINSULINEMIC HYPOGLYCEMIA- FIRST TIME IN INDIA	Dr Neha Sharma	surya hospital, jaipur
ABS134	Xia-Gibbs syndrome- A Case Report	Dr Gaurav	SMSHospital , Jaipur
ABS005	Gaucher Disease: The Importance of Genetic Studies in Early Case Detection and Diagnosis in a Young Age.	Aditya Kanade	
ABS007	CLINICAL & GENOMIC SPECTRUM OF INHERITED LIVER DISEASE: MANIPAL EXPERIENCE	Dr. Priyanka Patra	Kasturba Medical college, Manipal, Manipal Academy of Higher Education, Manipal
ABS031	CASE REPORT OF FRUCTOSE 1,6 BISPHOSPHATASE DEFICIENCY	Utkarsha P. Kulkarni	AIIMS Jodhpur
ABS032	THIAMINE-RESPONSIVE MEGALOBLASTIC ANEMIA RARE CASE OF DIABETES: CASE REPORT	Anisha Tanwar	AIIMS Jodhpur
ABS042	The Diagnostic Dilemma of Cockayne syndrome :A Report of two cases	Dr Sivakumar C	.

DAY 02 / 22 Sept.2024		SCREEN 03	
Abstract No.	**Topic**	**Presenter**	**Institute**
ABS071	Reanalysis of Whole Exome Sequencing Facilitates Diagnosis of Caf? Au Lait Spots with Freckling due to KIT gene variants in a Child with Juvenile Xanthogranulomatosis and Glycogen Storage Disorder Type VI.	Aruna Gowdra	Indira Gandhi Institute of Child Health
ABS083	IMPACT OF GCH1 GENE VARIANT ON PHENOTYPE OF THIAMINE METABOLISM DYSFUNCTION SYNDROME 5	Shilpa Kamble	Apollo Genomics Institute, Apollo Hospitals Navi Mumbai
ABS090	NEWBORN SCREENING FOR BIOTIDINASE DEFICIENCY ??HYPE OR HOPE? EXPERIENCE FROM A TERTIARY CARE CENTRE	Aarti Javeri	
ABS099	Reanalysis of Whole Exome Sequencing Facilitated the Diagnosis of Caf? Au Lait like Macules with Freckling and Juvenile Xanthogranulomatosis due to KIT gene variant in a Child with Glycogen Storage Disorder Type VI.	Aruna Gowdra	Indira Gandhi Institute of Child Health
ABS130	Knowing the unknown- A rare case of Citrullinemia	Tanya Jain	
ABS008	INTEGRATING GENETIC AND BIOCHEMICAL ANALYSIS FOR EFFECTIVE MANAGEMENT OF CBLA TYPE METHYLMALONIC ACIDURIA: A CLINICAL CASE STUDY	Dr. Priyanka Patra	Kasturba Medical college, Manipal, Manipal Academy of Higher Education, Manipal
ABS009	MONITORING OF BIOCHEMICAL MARKERS IN HEREDITARY TYROSINEMIA TYPE 1 PATIENTS FOR EFFECTIVE TREATMENT	Dr. Papai Roy	Sir Ganga Ram Hospital
ABS038	3-HYDROXYISOBUTYRYL-COA HYDROLASE (HIBCH) DEFICIENCY PRESENTING AS SEVERE METABOLIC CRISIS IN AN INFANT	Dr Pradeep Kumar	AIIMS Patna

DAY 02 / 22 Sept.2024		SCREEN 04	
Abstract No.	**Topic**	**Presenter**	**Institute**
ABS053	OUTCOME OF HEMATOPOIETIC STEM CELL TRANSPLANT IN ATTENUATED MPS I	Dr vadde Yaswanth Reddy	.
ABS066	MANAGEMENT AND EXTENDED FOLLOW UP IN METHYLMALONIC AND PROPIONIC ACIDURIA FROM A TERTIARY CENTRE.	Dr. Ami Shah	B J Wadia hospital for Children
ABS073	Double Trouble - Coexisting metabolic diseases	Dr Sarah Bailur	Rainbow Childrens Hospital
ABS075	Isovaleric Acidemia: Clinical, Biochemical, Molecular and Therapeutic response - a 10-year follow-up study from South India	Dr Sarah Bailur	Rainbow Childrens Hospital
ABS086	ACRODERMATITIS DYSMETABOLICA AS A COMPLICATION OF METHYLMALONIC ACIDEMIA- A DOUBLE-EDGED SWORD	INDRASISH RAY CHAUDHURI	Amrita Institute of Medical Sciences, Kochi
ABS100	The impact of nutrient adequacy on nutritional status of children with Inborn errors of metabolism (IEM).	Anushia K	
ABS102	Tolvaptan as a Potential Rescue Therapy for SIADH with Refractory Hyponatremia in a child with Acute Intermittent Porphyria	Rahul Gupta	AIIMS Jodhpur
ABS104	Tolvaptan as a Potential Rescue Therapy for SIADH with Refractory Hyponatremia in a child with Acute Intermittent Porphyria	Rahul Gupta	AIIMS Jodhpur

DAY 02 / 22 Sept.2024		SCREEN 05	
Abstract No.	**Topic**	**Presenter**	**Institute**
ABS127	Effect of long-term treatment with enzyme replacement therapy in Indian patients with Gaucher disease	Dr Shashank S Koundinya	AIIMS New Delhi
ABS014	PRECISION IN RESEARCH: LEVERAGING EMTREE FOR STUDYING INBORN ERRORS OF METABOLISM	Harini Challapalli	Elsevier
ABS045	Newborn Screening (NBS) for inherited metabolic disorder in a Tertiary Care Newborn Unit: An 8-year journey		Sri Ramachandra Institute of Higher Education and Research
ABS117	CORRELATION BETWEEN TRANSVERSE CEREBELLAR DIAMETER MEASURED BY ULTRASOUND WITHIN 48 HOURS OF BIRTH AND GESTATIONAL AGE	Dr Pokar Chand	
ABS121	??ACTORS INFLUENCING COMPLEMENTARY FEEDING IN CHILDREN (AGE GROUP 6 MONTHS TO 2 YEARS) IN WESTERN RAJASTHAN - HOSPITAL BASED CROSS-SECTIONAL STUDY?? media /abstracts /c17f24697eff61932db1f1b27045649cAbstract for	Dr Mahendra	
ABS132	SERUM HOMOCYSTEINE IS AN UNDERUTILIZED METABOLIC BIOMARKER- A CASE CONTROL STUDY IN CHILDREN WITH GLOBAL DEVELOPMENTAL DELAY AND THOSE WITH NORMAL DEVELOPMENT	Dr Krishna Mehta	
ABS133	THE EXPANDING CLINICAL, RADIOLOGICAL AND GENETIC SPECTRUM OF MITOCHONDRIOPATHIES FROM A TERTIARY CARE CENTRE	Dr Mehak Malhotra	
ABS 135	Developmental delay, encephalopathy with seizures in an Infant: Glutaric acidemia type	Dr Sunil Gothwal	SMSHospital , Jaipur