

SIMD2015 POSTER PRESENTATION (Numerical)

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4	Ah Mew, Nicholas	THE MISSED DIAGNOSIS OF ARGININOSUCCINATE LYASE DEFICIENCY IN THE OLDER SIBLINGS OF AFFECTED INFANTS
5	AlHashem, Amal	CLINICAL, BIOCHEMICAL, AND MOLECULAR STUDIES IN PYRIDOXINE-DEPENDENT EPILEPSY: REPORT OF 12 CASES
6	AlHashem, Amal	TREATMENT OF BIOTIN-RESPONSIVE BASAL GANGLIA DISEASE: OPEN COMPARATIVE STUDY BETWEEN THE COMBINATION OF BIOTIN PLUS THIAMINE VERSUS THIAMINE ALONE
7	Appadurai, Vivek	INCIDENCE ESTIMATE FOR CTX BASED ON 125,000 CHROMOSOMES REVEALS UNDER-DIAGNOSIS AND UNDERScores NEED FOR GREATER CLINICAL AND DIAGNOSTIC ATTENTION
8	Ayyub, Omar	ENGINEERING A COMPACT AND HIGH RESOLUTION BLOOD AMMONIA METER
9	Ayyub, Omar TRAVEL AWARD WINNER	DEVELOPMENT OF A RAPID POINT-OF-CARE BLOOD PHENYLALANINE METER FOR AT HOME AND BEDSIDE USE (See the abstract under Travel Award Abstracts)
10	Bellesheim, K.R.	ALTERED FUNCTIONAL BRAIN CONNECTIVITY IN PHENYLKETONURIA: EVIDENCE FROM GRAPH THEORY ANALYSIS
11	Boyer, Monica	ISOLATED SULFITE OXIDASE DEFICIENCY: NEONATAL PRESENTATION WITH ADDITIONAL BIOCHEMICAL FINDINGS AND DIET THERAPY
12	Plourde, F.	A PILOT OPEN LABEL TRIAL ASSESSING THE SAFETY AND EFFICACY OF BETAININE IN PATIENTS WITH A PEROXISOME BIOGENESIS DISORDER (PBD) AND PEX1-GLY843ASP (G843D) GENOTYPE
13	Gangoiti, Jon A.	TAMING MICROBIOTA. THE BALANCE OF CARNITINE SUPPLEMENTATION
14	Smith, Laurie D.	METHYLMALONIC ACIDEMIA OF UNCLEAR ETIOLOGY: IMPLICATION OF <i>ACSF3</i> BY WHOLE EXOME SEQUENCING
15	Byers, Stephanie	FAILED NEWBORN HEARING SCREEN AS FIRST MANIFESTATION OF MUCOPOLYSACCHARIDOSIS
16	Bieneck, Charlotte	ENERGY EXPENDITURE AND LIPID METABOLISM IN VERY LONG-CHAIN ACYL-COA DEHYDROGENASE (VLCAD) DEFICIENCY
17	Mokhtarani, M.	URINARY PHENYLACETYLGLUTAMINE (U-PAGN) CONCENTRATION AS AN ADHERENCE BIOMARKER FOR PATIENTS WITH UREA CYCLE DISORDERS (UCDS) TREATED WITH GLYCEROL PHENYLBUTYRATE (GPB)
18	Ganetzky, Rebecca D.	ECHS1 DEFICIENCY AS A CAUSE OF SEVERE NEONATAL LACTIC ACIDOSIS
19	Ganetzky, Rebecca D. TRAVEL AWARD WINNER	MUTATIONS IN <i>MTIF2</i> CAUSE A NOVEL DISORDER OF MITOCHONDRIAL TRANSLATION (See abstract under Travel Award Abstracts)
20	Chen, Bin	EVALUATION OF A CONTINUING EDUCATION ACTIVITY FOR QUALITY PRACTICES IN BIOCHEMICAL GENETIC TESTING AND NEWBORN SCREENING
21	Mei, Joanne	PROFICIENCY TESTING FOR LYSOSOMAL STORAGE DISORDERS IN IED BLOOD SPOTS TO DETECT KRABBE AND POMPE DISEASES
22	Ferreira, C.	THE NATURAL HISTORY PROTOCOL ON CONGENITAL DISORDERS OF GLYCOSYLATION
23	Ferreira, C.	CEREBROSPINAL FLUID FINDINGS IN A COHORT OF PATIENTS WITH <i>NGLY1</i> DEFICIENCY
24	Ferreira, C. (presented by Kristina Cusmano-Ozog)	HEREDITARY FRUCTOSE INTOLERANCE MIMICKING A BIOCHEMICAL PHENOTYPE OF MUCOLIPIDOSIS

25	Frigeni, Marta	DOMAIN-SWAPPING WITH OCTN2 SHOWS TOLERANCE TO SUBSTITUTIONS IN THE OCTN1 ERGOTHIONEINE TRANSPORTER
26	Reis, Claudia F.	IDENTIFICATION OF CITRIN DEFICIENCY BY NEWBORN SCREENING
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28	Buonuomo, Paola Sabrina	PEDIATRIC CHOLESTEROL SCREENING IN ITALY: THE SPIF PROJECT
29	Macchiaiolo, Marina	SUCCESSFUL PROSTHETIC EYE FITTING IN PATIENT WITH LIGNEOUS CONJUNCTIVITIS TREATED WITH TOPICAL PLASMINOGEN AND SURGERY
30	Bloom, Kaitlyn TRAVEL AWARD WINNER	IDENTIFICATION AND CHARACTERIZATION OF THE BIOCHEMICAL AND PHYSIOLOGICAL FUNCTIONS OF ACYL-CoA DEHYDROGENASE 10 (See the abstract under the Travel Award Abstracts)
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