AOMC 2022 Accepted Abstracts

The following abstracts are grouped by AOMC 2022 abstract topic and not based on abstract ranking

Topic: Genetic

Paper ID	Author	Affiliation	Title		Manuscrypt Category	Type
CPF-164	Yao Lei	THE UNIVERSITY OF HONG KONG	Exomiser accelerates the identification of second pathogenic variants by prioritizing the variants by pathogenicity	**Genetic**	Research	ePoster

Topic: Muscle

Paper ID	Author	Affiliation	Title		Manuscrypt Category	Type
CPF-010	dr. Citra Mega Kharisma	UNIVERSITAS SUMATERA UTARA	Dermatomyositis	**Muscle**	Case Report	ePoster
CPF-030	Hui Xiong	PEKING UNIVERSITY FIRST HOSPITAL	PARS2 related developmental and epileptic encephalopathy: a case report	**Muscle**	Case Report	ePoster
CPF-031	Hui Xiong	PEKING UNIVERSITY FIRST HOSPITAL	Uniparental disomy unmasks a homozygous mutation of POMGNT1 in a case of muscle-eye-brain disease	**Muscle**	Case Report	ePoster
CPF-092	dr. Purwadhani Sophia Nur Handini	UNIVERSITAS GAJAH MADA	Diagnostic Approach of Polymyositis and Dermatomyositis : A Case Report	**Muscle**	Case Report	ePoster
CPF-126	dr. Fuji Restu Firma, M.M	UNIVERSITAS UDAYANA	Asymmetrical Triangular Shoulder pada Fascioascapulohumeral Muscular Dystropy (FSHD)	**Muscle**	Case Report	ePoster
CPF-134	dr. Muhammad Arif Budi Prakoso	UNIVERSITAS GAJAH MADA	Recurrent Hypokalemic Periodic Paralysis In COVID-19 and Hyperthyroid A Case Report	**Muscle**	Case Report	ePoster
CPF-177	Wonjae Sung	HANYANG UNIVERSITY HOSPITAL	Very long-chain Acyl-CoA Dehydrogenase Deficiency due to Compound Heterozygote Variants of ACADVL Gene	**Muscle**	Case Report	ePoster
CPF-181	dr. Septian Widyantoro	UNIVERSITAS GAJAH MADA	Steroid-Induced Myopathy in Nephrotic syndrome : A Case Report	**Muscle**	Case Report	ePoster
CPF-196	Sanggon Lee	HANYANG UNIVERSITY HOSPITAL	Myofibrillar myopathy presenting with muscle biopsy findings of Mitochondrial myopathy	**Muscle**	Case Report	ePoster
CPF-203	Pengfei Lin	DEPARTMENT OF NEUROLOGY AND RESEARCH INSTITUTE OF NEUROMUSCULAR AND NEURODEGENERATIVE DISEASES, QILU HOSPITAL, SHANDONG UNIVERSITY	Elongation and truncation variants in the TTN gene causing limb-girdle muscular dystrophy type 2J in a Han Chinese family	**Muscle**	Case Report	ePoster
CPF-224	Mr.Chayanont Netsawang	RAMATHIBODI HOSPITAL, MAHIDOL UNIVERSITY	Mitochondrial Myopathy in a 50-Year-Old Woman Mimicking Ocular Myasthenia	**Muscle**	Case Report	ePoster
CPF-229	dr. Luh Ari Indrawati, Sp.N(K)	UNIVERSITAS INDONESIA	A case of Seronegative Immune Mediated Necrotizing Myopathy with Nonspecific Interstitial Lung Disease	**Muscle**	Case Report	ePoster

CPF-230	Wan-Ling Hsiao	KAOHSIUNG MEDICAL UNIVERSITY HOSPITAL KAOHSIUNG MEDICAL UNIVERSITY KAOHSIUNG	Clinical, pathological and genetic diagnosis of multiminicore myopathy in three Taiwanese patients: SEPN1 myopathy and MEGF10 myopathy	**Muscle**	Case Report	ePoster
CPF-021	Dr. Dipti Baskar	NATIONAL INSTITUTE OF MENTAL HEALTH AND NEUROSCIENCES (NIMHANS)	ADSSL1 myopathy: Diagnostic challenges with variable phenotypes in three Indian patients	**Muscle**	Research	ePoster
CPF-022	Dr. Dipti Baskar	NATIONAL INSTITUTE OF MENTAL HEALTH AND NEUROSCIENCES (NIMHANS)	Clinico-pathological features of beta-sarcoglycanopathy with a highly prevalent mutation in an Indian cohort	**Muscle**	Research	ePoster
CPF-025	Hui Xiong	PEKING UNIVERSITY FIRST HOSPITAL	Natural history of a novel mouse model for LAMA2-related congenital muscular dystrophy	**Muscle**	Research	ePoster
CPF-026	Hui Xiong	PEKING UNIVERSITY FIRST HOSPITAL	Muscle transcriptomic study of a novel LAMA2-related congenital muscular dystrophy mouse model	**Muscle**	Research	ePoster
CPF-029	Hui Xiong	PEKING UNIVERSITY FIRST HOSPITAL	Clinical and genetic study of LAMA2-related muscular dystrophy patients with seizures	**Muscle**	Research	ePoster
CPF-067	SRIDHAR S	NATIONAL INSTITUTE OF MENTAL HEALTH AND NEUROSCIENCES (NIMHANS)	Magnetic Resonance Imaging of Muscles in different subtypes of Inflammatory Myositis	**Muscle**	Research	ePoster
CPF-114	Dr Anna Ka Yee Kwong	THE UNIVERSITY OF HONG KONG	Genetic diagnosis of Facioscapulohumeral Muscular Dystrophy in Hong Kong Chinese patients using Molecular Combing	**Muscle**	Research	ePoster
CPF-123	Dr. Abel Thomas Oommen	NATIONAL INSTITUTE OF MENTAL HEALTH AND NEUROSCIENCES (NIMHANS)	Myofibrillar Myopathy: Clinico-Genetic Spectrum from an Indian Neuromuscular Centre	**Muscle**	Research	ePoster
CPF-180	Pengfei Lin	DEPARTMENT OF NEUROLOGY AND RESEARCH INSTITUTE OF NEUROMUSCULAR AND NEURODEGENERATIVE DISEASES, QILU HOSPITAL, SHANDONG UNIVERSITY	A female carrier of spinal and bulbar muscular atrophy diagnosed with DNAJB6-related distal myopathy	**Muscle, motor neuron**	Case Report	ePoster
CPF-199	Sanggon Lee	HANYANG UNIVERSITY HOSPITAL	Hypokalemic periodic paralysis with myokymic movements due to secondary distal renal tubular acidosis : Initial manifestation in Sjogren syndrome without any sicca symptom	**Muscle; peripheral nerve**	Case Report	ePoster
CPF-212	dr. Luh Ari Indrawati, Sp.N(K)	UNIVERSITAS INDONESIA	A Case Report of Miyoshi Myopathy with a Novel Variant of DYSF	**Muscle**	Clinical Pathogi	Clinical Pathogical Conference
CPF-213	POH YEN YEONG	NATIONAL NEUROSCIENCE INSTITUTE	Late onset distal myopathy masquerading as right common peroneal nerve palsy	**Muscle**	Clinical Pathogi	Clinical Pathogical Conference
CPF-173	Pimchanok Kulsirichawaroj	SIRIRAJ GENOMICS CENTER, MAHIDOL UNIVERSITY	Utilisation of Whole-Exome Sequencing for Muscular Disorders in Thai Paediatric Patients: Diagnostic Yield and Mutational Spectrum	**Muscle**	Research	ePoster
CPF-191	Pengfei Lin	DEPARTMENT OF NEUROLOGY AND RESEARCH INSTITUTE OF NEUROMUSCULAR AND NEURODEGENERATIVE DISEASES, QILU HOSPITAL, SHANDONG UNIVERSITY	Clinical and pathological analysis of four Chinese patients with LGMD2G	**Muscle**	Research	ePoster

CPF-197	Pengfei Lin	DEPARTMENT OF NEUROLOGY AND RESEARCH INSTITUTE OF NEUROMUSCULAR AND NEURODEGENERATIVE DISEASES, QILU HOSPITAL, SHANDONG UNIVERSITY	Clinical, pathological, and molecular genetic analysis of 7 Chinese patients with hereditary myopathy with early respiratory failure	**Muscle**	Research	ePoster
CPF-200	Pengfei Lin	DEPARTMENT OF NEUROLOGY AND RESEARCH INSTITUTE OF NEUROMUSCULAR AND NEURODEGENERATIVE DISEASES, QILU HOSPITAL, SHANDONG UNIVERSITY	Tcap deficiency in zebrafish leads to ROS production and mitophagy, and idebenone improves its phenotypes	**muscle**	Research	ePoster
CPF-227	dr. Luh Ari Indrawati, Sp.N(K)	UNIVERSITAS INDONESIA	Performance of Diagnostic Studies of Muscle Diseases in Department of Neurology Dr. Cipto Mangunkusumo Hospital: Results of the Myopathy Registry	**Muscle**	Research	ePoster

Topic: **Peripheral Nerve**

Paper ID	Author	Affiliation	Title		Manuscrypt Category	Type
CPF-063	dr. Eka Mahendrayana	UNIVERSITAS SUMATERA UTARA	Charcot-Marie-Tooth disease (CMT)	**Peripheral nerve**	Case Report	ePoster
CPF-091	dr. Diah Ariesa	UNIVERSITAS GAJAH MADA	Guillain-Barre Syndrome Post Sinovac Vaccination: A Case Report	**Peripheral nerve**	Case Report	ePoster
CPF-095	dr. Eva Nur Fadila	UNIVERSITAS GAJAH MADA	Guillain-Barré Syndrome Following COVID-19 Vaccination : A Case Report	**Peripheral nerve**	Case Report	ePoster
CPF-132	dr. Luhur Budi Adhiapto	UNIVERSITAS GAJAH MADA	Guillain Barre Syndrome with Vitamin D Deficiency in Pregnancy Woman, A Case Report	**Peripheral nerve**	Case Report	ePoster
CPF-167	Trianggoro Budisulistyo, MD, Dipl of Pain-	UNIVERSITAS DIPONEGORO	PAIN AND MOTOR DISORDERS ACCOMPANIED OF CHRONIC CARPAL TUNNEL SYNDROME SHOWED IMPROVEMENT AFTER ULTRASOUND-GUIDED OF 10% DEXTROSE PERINEURAL DISSECTION INJECTION: A CASE REPORT	**Peripheral nerve**	Case Report	ePoster
CPF-068	LI WAI YIN VIVIEN	THE UNIVERSITY OF HONG KONG	Debilitating osteo-chondral manifestations in patients with Congenital Insensitivity to Pain with Anhidrosis (CIPA) due to NTRK1 mutations	**Peripheral nerve**	Research	ePoster
CPF-121	dr. Muchtar Leonardi	UNIVERSITAS SRIWIJAYA	Effectiveness of 5?xtrose Subcutaneous Injection in Painful Diabetic Neuropathy Patients in Mohammad Hoesin General Hospital Palembang	**Peripheral nerve**	Research	ePoster

Topic: **Motor Neuron**

Paper ID	Author	Affiliation	Title		Manuscrypt Category	Type
CPF-094	dr. Noor Alia Susianti	UNIVERSITAS GAJAH MADA	Spinal Muscular Atrophy-Type 3: Early Diagnostic And Management in Yogyakarta, Indonesia, A Case Report	**Motor Neuron**	Case Report	ePoster
CPF-151	dr. Aurelia Vania	UNIVERSITAS UDAYANA	A RARE CASE OF JUVENILE AMYOTROPHIC LATERAL SCLEROSIS	**Motor neuron**	Case Report	ePoster
CPF-023	Hui Xiong	PEKING UNIVERSITY FIRST HOSPITAL	Clinical and genetic study of a spinal muscular atrophy family with variable phenotypes	**Motor Neuron**	Research	ePoster
CPF-024	Hui Xiong	PEKING UNIVERSITY FIRST HOSPITAL	Baseline nutrition investigation in a Chinese cohort of pediatric patients with spinal muscular atrophy	**Motor Neuron**	Research	ePoster

CPF-115	M S Keerthipriya	NATIONAL INSTITUTE OF MENTAL HEALTH AND NEUROSCIENCES (NIMHANS)	RARE FORMS OF GENETICALLY MEDIATED FAMILIAL AND SPORADIC AMYOTROPHIC LATERAL SCLEROSIS FROM INDIA	**Motor Neuron**	Research	ePoster
CPF-117	Atchayaram Nalini	NATIONAL INSTITUTE OF MENTAL HEALTH AND NEUROSCIENCES (NIMHANS)	Hirayama Disease: Surgical Outcome In A Large Cohort Of Indian Patients	**Motor Neuron**	Research	ePoster
CPF-214	Sanggon Lee	HANYANG UNIVERSITY HOSPITAL	SOD1 variants in Korean patients with motor neuron disease.	**Motor Neuron**	Research	ePoster

Topic: **Multiple System**

Paper ID	Author	Affiliation	Title		Manuscrypt Category	Type
CPF-028	Hui Xiong	PEKING UNIVERSITY FIRST HOSPITAL	Clinical and genetic study of rare cases with coexistence of dual genetic diagnoses	**Multiple system**	Research	ePoster

Topic: **Neuromuscular Juncion Disease**

Paper ID	Author	Affiliation	Title		Manuscrypt Category	Type
CPF-099	dr. Mira Tamila Nurul Maulida Awaliyah	UNIVERSITAS GAJAH MADA	Myasthenic Crisis in Covid-19: A Challenging Case in Dr. Sardjito General Hospital Yogyakarta, Indonesia	**Neuromuscular Juncion Disease**	Case Report	ePoster
CPF-057	Dr Seena Vengalil	NATIONAL INSTITUTE OF MENTAL HEALTH AND NEUROSCIENCES (NIMHANS)	Clinical and genotypic characterization of glycosylation defects in Congenital Myasthenic Syndrome in Indian patients	**Neuromuscular Juncion Disease**	Research	ePoster
CPF-084	Dr Saraswati Nashi	NATIONAL INSTITUTE OF MENTAL HEALTH AND NEUROSCIENCES (NIMHANS)	Clinico-Genetic Profile Of Congenital Myasthenic Syndromes: Study of Mutations In Neuromuscular Junction Development And Maintenance Genes	**Neuromuscular Juncion Disease**	Research	ePoster
CPF-143	Widya Prawirani Siahaan	UNIVERSITAS SUMATERA UTARA	The Profile Quality of Life Myasthenia Gravis Patients in General Hospital Haji Adam Malik Medan	**Neuromuscular Juncion Disease**	Research	ePoster