The Winner of

Best e-Poster on 20th Annual Scientific Meeting of AOMC 2022

CASE REPORT CATEGORY



20th Annual Scientific Meeting







Best e-Poster
Case Report Category

3rd Place

Pengfei Lin

Department of Neurology and Research Institute of Neuromuscular and Neurodegenerative Diseases, Qilu Hospital of Shandong University, Jinan, Shandong 250012, China

A female carrier of spinal and bulbar muscular atrophy diagnosed with DNAJB6-related distal myopathy









2nd Place

Hui Xiong

Department of Pediatrics, Peking University First Hospital, China

Uniparental disomy unmasks a homozygous mutation of POMGNT1 in a case of muscle-eye-brain disease









1st Place

Hui Xiong

Department of Pediatrics, Peking University First Hospital, China

PARS2 related developmental and epileptic encephalopathy: a case report





The Winner of

Best e-Poster on 20th Annual Scientific Meeting of AOMC 2022

RESEARCH ARTICLE CATEGORY



20th Annual Scientific Meeting





Best e-Poster Research Article Category

3rd Place

Anna Ka Yee Kwong

Paediatric Neurology Team, Department of Paediatrics and Adolescent Medicine, School of Clinical Medicine, LKS Faculty of Medicine, The University of Hong Kong, HKSAR, China

Genetic diagnosis of
Facioscapulohumeral Muscular Dystrophy
in Hong Kong Chinese patients using
Molecular Combing









Best e-Poster Research Article Category

2nd Place

Hui Xiong

Department of Pediatrics, Peking University First Hospital, China

Natural history of a novel mouse model for LAMA2-related congenital muscular dystrophy









Best e-Poster Research Article Category

1st Place

Yao Lei

Paediatric Neurology Division, Department of Paediatrics & Adolescent Medicine, School of Clinical Medicine, Li Ka Shing Faculty of Medicine, The University of Hong Kong

Exomiser accelerates the identification of second pathogenic variants by prioritizing the variants by pathogenicity













CONGRATULATIONS To All Winners!

See you on Next Meeting!



20th Annual Scientific Meeting