香港神經肌肉疾病學會
The Hong Kong Society of Neuromuscular Diseases

Academic Meeting
Expanding Histopathologic and Clinical Spectrum of *RYR1* associated Myopathy

Speaker: Professor Mariarita Santi

Department of Pathology and Laboratory Medicine
The Children's Hospital of Philadelphia, Philadelphia, USA

RYR1 mutation are the most common overall cause of congenital myopathies. It can cause both autosomal dominant and autosomal recessive congenital myopathies. With the availability of genetic testing for the entire *RYR1* coding sequence, this has led to a dramatic expansion in the identification of more patients with a wide clinical spectrum of presentation as well as the histopathological findings. Comprehensive analysis of genotype-phenotype association in *RYR1*-myopathies will be discussed.

Date: 09 January 2017 (Monday)
Time: 18:30 - 20:30
Venue: Block A, Ground floor, Seminar Room, Queen Elizabeth Hospital, Kowloon
Moderator: Dr. Amanda Kan

All are welcome. No registration required.

CME: for paediatricians – 2 points; physicians (IM, NU), pathologists (AP, CP, PA), family physicians - pending

Registration fee: Free-of-charge

Enquiry: Dr. Sandy Cheng (Tel : 64600089/ Email : chengy@ha.org.hk)

Co-organized with Department of Medicine, Queen Elizabeth Hospital.