Background: Congenital myopathies (CMs) are a genetically and clinically heterogeneous group of neuromuscular disorders. Historically, the congenital myopathies are classified according to muscle biopsy findings – Rods (Nemaline myopathy) (NM), cores (central core disease and multinucleomeric disease) (CM), and selective hypotrophy of type 1 fibres (congenital fibre type disproportion) (CTFD). Over twenty genes have been implicated in CMs. The overlapping clinical presentations among different histopathological findings and different mutations poses major diagnostic challenge.

Objective: We investigated the characteristics of children with congenital myopathies in Hong Kong.

Patients and methods: We identified all patients with a confirmed diagnosis of CM between 2012-March 2015. Their clinical presentation, muscle biopsy, muscle MRI and genetic analysis results were evaluated.

Results: Patients: Total 15 patients have been diagnosed to have CM. Nine were males (60%), 6 were females (40%).

Genetic findings: (1) A genetic diagnosis could be established in 11 (73%) out of 15 patients. Nine were males (60%), 6 were females (40%).

Histopathological features: (1) Muscle biopsy evaluation were available in all 15 patients. Nemaline myopathy were the most frequent histopathological diagnosis, in 5 patients (33%), followed by core myopathy, in 4 patients (26%), centronuclear myopathy in 2 patients (13%), fibres type disproportion in 2 patients (13%), zebra bodies in 1 (6.7%) patient and type 1 predominance in 1 (6.7%) patient.

Clinical features: (1) Of the 15 patients, 9 (60%) had age of onset at birth or before one month, 3 (20%) between 1 and 12 months, and 3 (20%) between 1 and 5 years. Out of the 9 patients with early neonatal presentation, 3 (33%) patients died before 15 months.

Muscle imaging: Selective muscle involvement with Rectus Femoris sparing provides helpful clues to a possible underlying RYR1 mutation.