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Author(s)	Chan, SHS; Lo, IFM; Cherk, S; Cheng, WWS; Fung, E; Yeung, WL; Lee, WC; Wong, S; Ma, CKL; Tai, SM; Yau, MM; Ng, G; Wu, SP; Wong, VCN
Citation	The 18th International Congress of The World Muscle Society, Asilomar, CA., 1-5 October 2013. In Neuromuscular Disorders, 2013, v. 23 n. 9-10, p. 776, abstract no. P.7.10
Issued Date	2013
URL	http://hdl.handle.net/10722/197707
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Territory wide study of patients with dystrophinopathy in Hong Kong

S.H.S. Chan¹, I.F.M. Lo², S. Cherk³, W.W.S. Cheng⁴, E. Fung⁵, W.L. Yeung⁶, W.C. Lee⁷, S. Wong⁸, C.K.L. Ma⁸, S.M. Tai⁹, M.M. Yau¹⁰, G. Ng¹¹, S.P. Wu¹², C.V.N. Wong¹

¹ Dept of Paediatrics and Adolescent Medicine, Queen Mary Hospital, Hong Kong; ² Clinical Genetic Service, Dept of Health, Hong Kong; ³ Dept of Paediatrics and Adolescent Medicine, Kwong Wah Hospital, Hong Kong; ⁴ Dept of Paediatrics and Adolescent Medicine, Caritas Medical Centre, Hong Kong; ⁵ Dept of Paediatrics and Adolescent Medicine, Prince of Wales Hospital, Hong Kong; ⁶ Dept of Paediatrics and Adolescent Medicine, Alice Ho Mei Ling Nethesole Hospital, Hong Kong; ⁷ Dept of Paediatrics and Adolescent Medicine, Tuen Mun Hospital, Hong Kong; ⁸ Dept of Paediatrics and Adolescent Medicine, United Christian Hospital, Hong Kong; ⁹ Dept of Paediatrics and Adolescent Medicine, Pamela Youde Nethesole Eastern Hospital, Hong Kong; ¹⁰ Dept of Paediatrics and Adolescent Medicine, Tseung Kwan O Hospital, Hong Kong; ¹¹ Dept of Paediatrics and Adolescent Medicine, Princess Margaret Hospital, Hong Kong; ¹² Dept of Paediatrics and Adolescent Medicine, Queen Elizabeth Hospital, Hong Kong

This is a first territory wide study in Hong Kong on Chinese patients with dystrophinopathy on their genetic mutation, motor performance, use of steroid, and the interventions they received. This study is participated by all the paediatric departments in the eleven hospitals in Hong Kong. Clinical data was systemically collected from medical records of all patients diagnosed dystrophinopathy and were actively followed-up between May 2006 and April 2010. We found 91 individuals with dystrophinopathy, 82% have Duchenne Muscular Dystrophy (DMD) and 18% have Becker Muscular Dystrophy (BMD). The overall prevalence of dystrophinopathy, for 2010, is 1.03 per 10,000 males aged 0–24 years old. Mutation consists of 47% of large deletion and 10% of large duplication, and 43% of small rearrangement or point mutation, of which 59.4% are nonsense mutations. Exon deletion in the distal hot spot (exons 45–55) represent up to 54% of the identified deletion, while deletion starting in the proximal hot spot (exon 2–20) account for 37%. Only 23% of children had been on steroid and all stopped before or when they lost ambulation. For DMD individuals, the age ranges from 0.5 to 34 years old, with 48% are older than 13 years old, and the mean age of loss of ambulation is 10.5 years old. For those older than 13 years old, 30% have cardiomyopathy, 19% required noninvasive ventilation, 15% had scoliosis surgery and only 5% had gastrostomy. This first territory wide study for individuals with dystrophinopathy in Hong Kong confirms a similar prevalence of such condition with the western countries, but our population has a much higher percentage of point mutation. The findings of infrequent steroid use and the low percentage of gastrostomy among patients with non-invasive ventilation, allows us to compare our current approaches in Hong Kong with the DMD care guidelines to drive improvements in our health care delivery.