Abstract


[Application of targeted capture technology and next generation sequencing in molecular diagnosis of inherited myopathy].

[Article in Chinese]

Objective: To elucidate the usefulness of next generation sequencing for diagnosis of inherited myopathy, and to analyze the relevance between clinical phenotype and genotype in inherited myopathy.

Method: Related genes were selected for SureSelect target enrichment system kit (Panel Version 1 and Panel Version 2). A total of 134 patients who were diagnosed as inherited myopathy clinically underwent next generation sequencing in Department of Pediatrics, Peking University First Hospital from January 2013 to June 2014. Clinical information and gene detection result of the patients were collected and analyzed.

Result: Seventy-seven of 134 patients (89 males and 45 females, visiting ages from 6-month-old to 26-year-old, average visiting age was 6 years and 1 month) underwent next generation sequencing by Panel Version 1 in 2013, and 57 patients underwent next generation sequencing by Panel Version 2 in 2014. The gene detection revealed that 74 patients had pathogenic gene mutations, and the positive rate of genetic diagnosis was 55.22%. One patient was diagnosed as metabolic myopathy. Five patients were diagnosed as congenital myopathy; 68 were diagnosed as muscular dystrophy, including 22 with congenital muscular dystrophy 1A (MDC1A), 11 with Ullrich congenital muscular dystrophy (UCMD), 6 with Bethlem myopathy (BM), 12 with Duchenne muscular dystrophy (DMD) caused by point mutations in DMD gene, 5 with LMNA-related congenital muscular dystrophy (L-CMD), 1 with Emery-Dreifuss muscular dystrophy (EDMD), 7 with alpha-dystroglycanopathy (α-DG) patients, and 4 with limb-girdle muscular dystrophy (LGMD) patients.

Conclusion: Next generation sequencing plays an important role in diagnosis of inherited myopathy. Clinical and biological information analysis was essential for screening pathogenic gene of inherited myopathy.

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