Abstract

Duchenne and Becker muscular dystrophies (DMD and BMD) represent the most frequent neuromuscular diseases in humans (1/3,500-6,000 live male births), characterized by an X-linked recessive pattern of inheritance and therefore affecting mainly male individuals. DMD and BMD are allelic disorders resulting from genetic defects, mostly intragenic deletions, in the dystrophin gene. Using multiplex polymerase chain reaction (PCR), we have analyzed 170 male patients from unrelated families originating from Algeria, showing that 68 % of them harbored deletion events affecting the known 5′ or 3′ hot spot regions. The distal portion was predominantly involved (85 %), whereas 37 distinctive patterns of deletion were identified in our panel. The extent of deletion varied from 1 to 32 exons, although the average number was about four exons. The lack of seven exons (45, 46, 47, 48, 50, 51 and 52), each alone or in combination, represented about 78 % of the alterations encountered, while exon 48 was most frequently involved (50 %). The effect of the deletions showed that the reading frame rule proved mostly true, correlating with the clinical diagnosis suggested. Moreover, the c.525delT mutation in the γ-sarcoglycan gene was present in non-deleted patients (7 %), suggesting that clinical features can still be misleading. Finally, multiplex PCR proved to be a simple, fast and low-cost approach for the molecular diagnosis of dystrophinopathies in Algeria, whereas our data could contribute to the creation of a national registry of DMD/BMD patients in our country, which would give them hope to an access to already available genotype-based therapies