Introduction: A genotype-phenotype correlation was established by dystrophin gene deletion analysis in Duchenne and Becker muscular dystrophies patients (DMD/BMD). Objectives: To establish a correlation between molecular genotype and clinical phenotype in a Colombian population. Materials and methods: A PCR (Polymerase Chain Reaction) amplification of 18 exons (included in the two hot spots regions) was performed in 62 affected families. Results: Nineteen patients showed deletions in several exons of the dystrophin gene. This corresponds to 31% of analyzed males in the present population. Conclusions: For each DMD/DMB affected male with deletion in the dystrophin gene, a correlation between disease severity and extent of deletion was established. The data showed that most out-frame deletions cause DMD phenotype, while the in-frame deletions results in BMD phenotype. This correlation was described by Koenig in his open reading frame hypothesis. In the present study it was possible to establish a direct correlation between mutation state and clinical severity in 79% of patients. This may help clinical evaluation of DMD/DMB patients.

Keywords
Duchenne and Becker muscular dystrophy (DMD/DMB);
Multiplex polymerase chain reaction (PCR); Open reading frame hypothesis; X-linked diseases.

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