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ADELLGENE® MYOTONIC DYSTROPHY

References:

AD-MD-16
SCREENING

AD-MD-C-16
CONFIRMATORY

EMQN
The European Molecular Genetics Quality Network

External Quality Assessment (EQA) schemes passed (run by EMQN)

DETERMINATION OF THE NUMBER OF CTG TRIPLETS OF THE DMPK GENE

Kit for the determination, by fluorescent fragment analysis, of the number of repeats of CTG triplets of the DMPK gene

DETERMINACIÓN DEL NÚMERO DE TRIPLETES CTG DEL GEN DMPK

Kit para la determinación del número de repeticiones del triplete CTG del gen DMPK, mediante análisis de fragmentos fluorescentes

CE-IVD

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ADELLGENE® MYOTONIC DYSTROPHY

INTRODUCTION

Myotonic dystrophy type 1 or Steinert's disease is currently the most common form of muscular dystrophy in adults. Inheritance of this multisystem disease is autosomal dominant, and phenotypic expression is highly variable due to an unstable expansion CTG trinucleotide repeats dystrophia myotonica protein kinase gene (DMPK, MIM*605377). Depending on the number of repetitions of this triplet, three categories can be established:

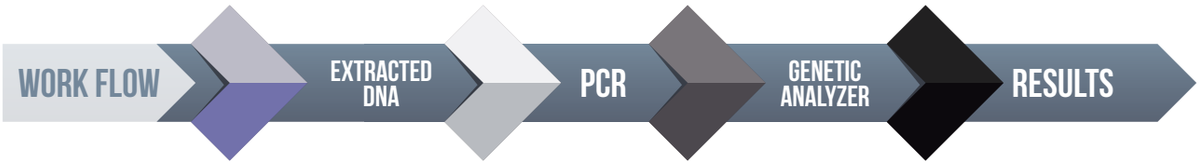
- From 5 to 34 repeats: individuals with healthy alleles.
- From 35 to 49 repeats: individuals with permutated alleles.
- Over 50 repeats: individuals with mutated alleles.

INTRODUCCIÓN

La Distrofia Miotónica tipo 1 o enfermedad de Steinert es actualmente la forma más común de la Distrofia Muscular en adultos.

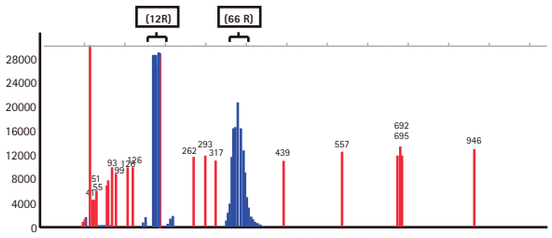
La herencia de esta enfermedad multisistémica es autosómica dominante, y la expresión fenotípica es muy variable debido a una expansión inestable de trinucleótidos CTG localizada en el gen de la proteína distrofia miotónica quinasa (DMPK, OMIM * 605377). Dependiendo del número de repeticiones de este triplete, se pueden establecer tres categorías:

- De 5 a 34 repeticiones: individuos con alelos sanos.
- De 35 a 49 repeticiones: individuos con alelos premutados.
- Más de 50 repeticiones: individuos con alelos mutados.

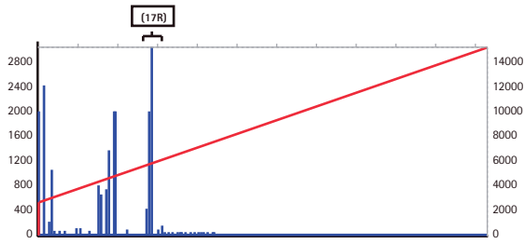


RESULTS / RESULTADOS

ADELLGENE® MYOTONIC DYSTROPHY SCREENING



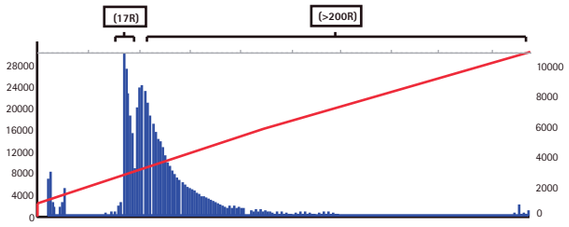
Heterozygous: 12, 66 repeats



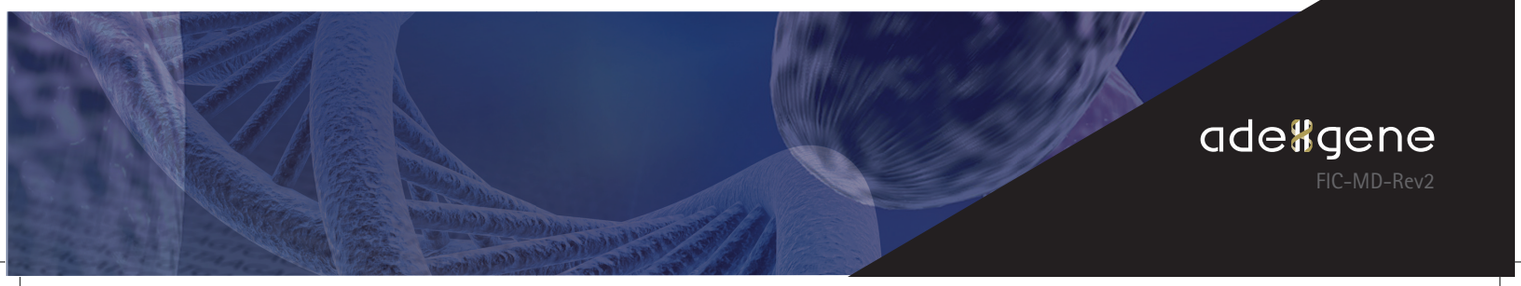
Sample with only one peak: 17 repeats*

* Samples with one peal must be confirmed with Adellegne Myotonic Dystriphy Confirmatory

ADELLGENE® MYOTONIC DYSTROPHY CONFIRMATORY



Sample with one peak: (17R) and one peak > 200



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FIC-MD-Rev2