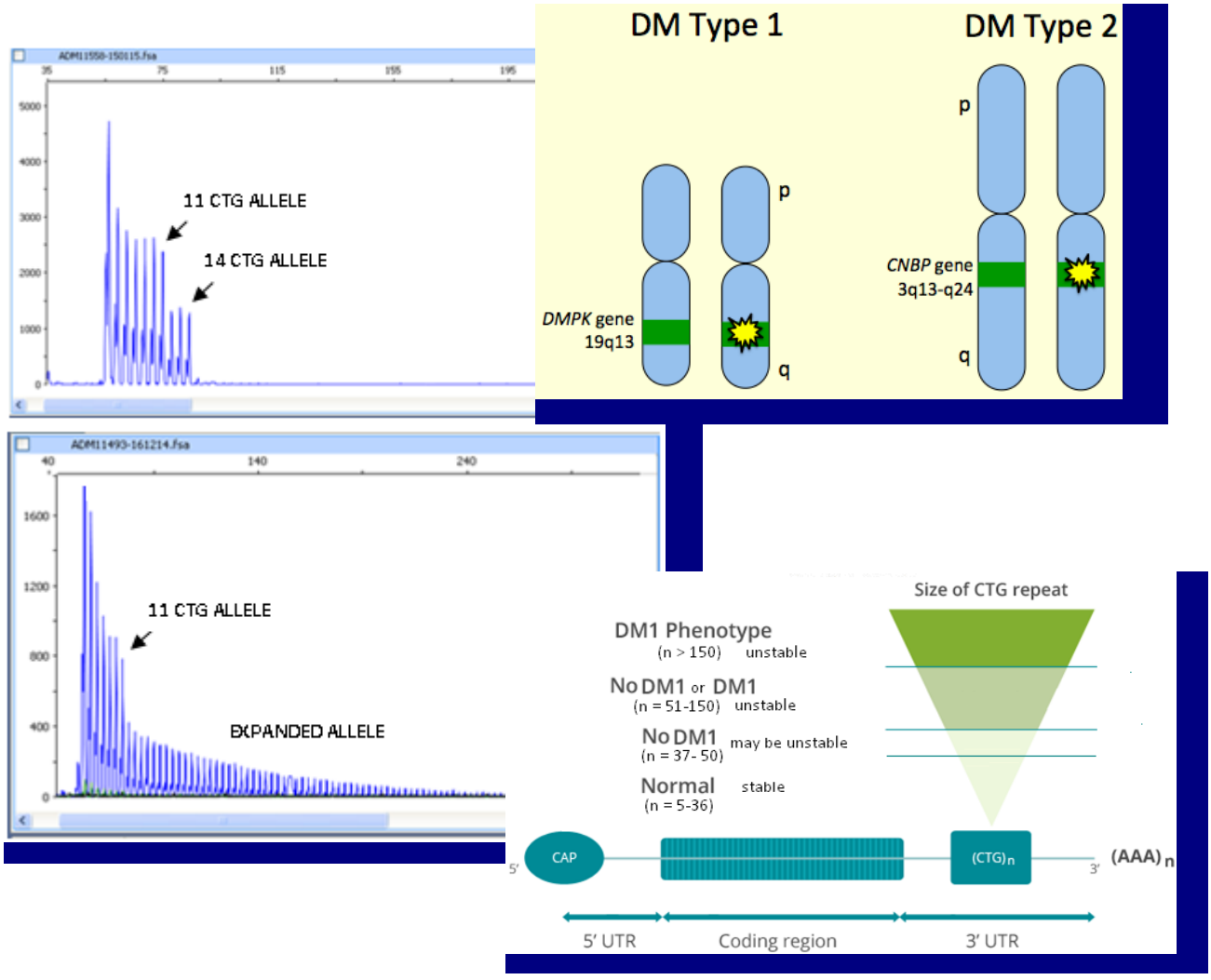
SISTEM for molecular testing of ***MYOTONIC DYSTROPHY type 1*** *by* ***Triplet Repeat Primed PCR*** *and*

CAPILLARY ELECTROPHORESIS

MYOTONIC DYSTROPHY type 1 GC KIT – FL

cod. DM.04FL



EXPERTEAM
via della Libertà 12-30175 Marghera (VE)
tel.: +39 041 5093101
fax: +39 041 5093102
e-mail: expertm@vegapark.ve.it
www.experteam.it

CE
IVD

Myotonic dystrophy (DM) is an autosomal dominant disorder characterized by myotonia, muscular dystrophy, catarats, testicular atrophy, frontal balding and cardiac conduction defects.
DM is clinically heterogeneous and at molecular level at least two types can be distinguished: DM type 1 (Steinert disease) and DM type 2 (proximal myotonic myopathy PROMM or Ricker syndrome). DM1 is the most common form of muscular dystrophy in adults with an estimated incidence of 1:8000.
It is caused by a[CTG]n repeat expansion in the 3'-untranslated region of the dystrophia myotonica-protein kinase gene **(DMPK)** on chromosome 19.
The $[CTG]_n$ repeat is polymorphic in the normal range, with repeat numbers ranging from **5** and **36**; alleles containing over 36 CTG-repeat demonstrate a lenght-dependent risk of instability on transmission. Alleles containing a CTG-repeat with a lenght of **51-150** may be either asymptomatic or may give rise to minimal or classical DM1. A more severe DM1 phenotype is associated with DMPK alleles with sized **>150** CTG.
Molecular testing for DM1 is extremely important as it can confirm the diagnosis in many cases where the symptoms are doubtful.
*The **Myotonic Dystrophy type 1 GC kit-FL** is based on* Triplet Repeat Primed PCR (TP-PCR) and allows to identify **all the DM1 alleles,** including those with very long expansion. Moreover, the kit eliminates the Southern blotting, reducing execution times (from a week to one day) and costs too.

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How do the kit work?

The ***Myotonic Dystrophy type 1 GC kit-FL*** is a system for molecular analysis of **Myotonic dystrophy type 1** by **TP-PCR** with three primers: a primer, 5' marked with a fluorophore, is located upstream of the repeated sequence, a second primer complementary to the repeated sequence (chimeric primer) and a reverse primer.
The chimeric primer hybridizes in multiple sites within the repeated region $CTG_{(n)}$, giving PCR products of different sizes.
These products are separated by capillary electrophoresis and form a characteristic fragment ladder of increasing lenght: the fragments differ from each other by 3 bp and allow rapid identification of pathogenic long CTG expansions which can not be amplified by primers flanking the repeated sequence.
The kit is based on a bidirectional TP-PCR to avoid false negatives, due to sequence interruptions by *CCG* and *GGC* triplets that may be present at 3' region of the repeated sequence.

Starting samples: peripheral blood

DNA isolation method: QIAamp DNA blood mini kit, QIAcube, QIAsymphony (Qiagen), High Pure PCR template preparation kit (Roche).

DNA Sequencers: 310, 3100, 3130, 3730, 3500 Genetic Analyzers (Applied Biosystmes),

Kit contents

Etichetta	Contenuto
DM1 TPF MASTER MIX	Mix for the amplification of DMPK gene
DM1 TPR MASTER MIX	
DM1 TP DNA polymerase	DNA polymerase for difficult amplification
Controllo WT	DNA with DM1 wild-type alleles 5- 28 CTG