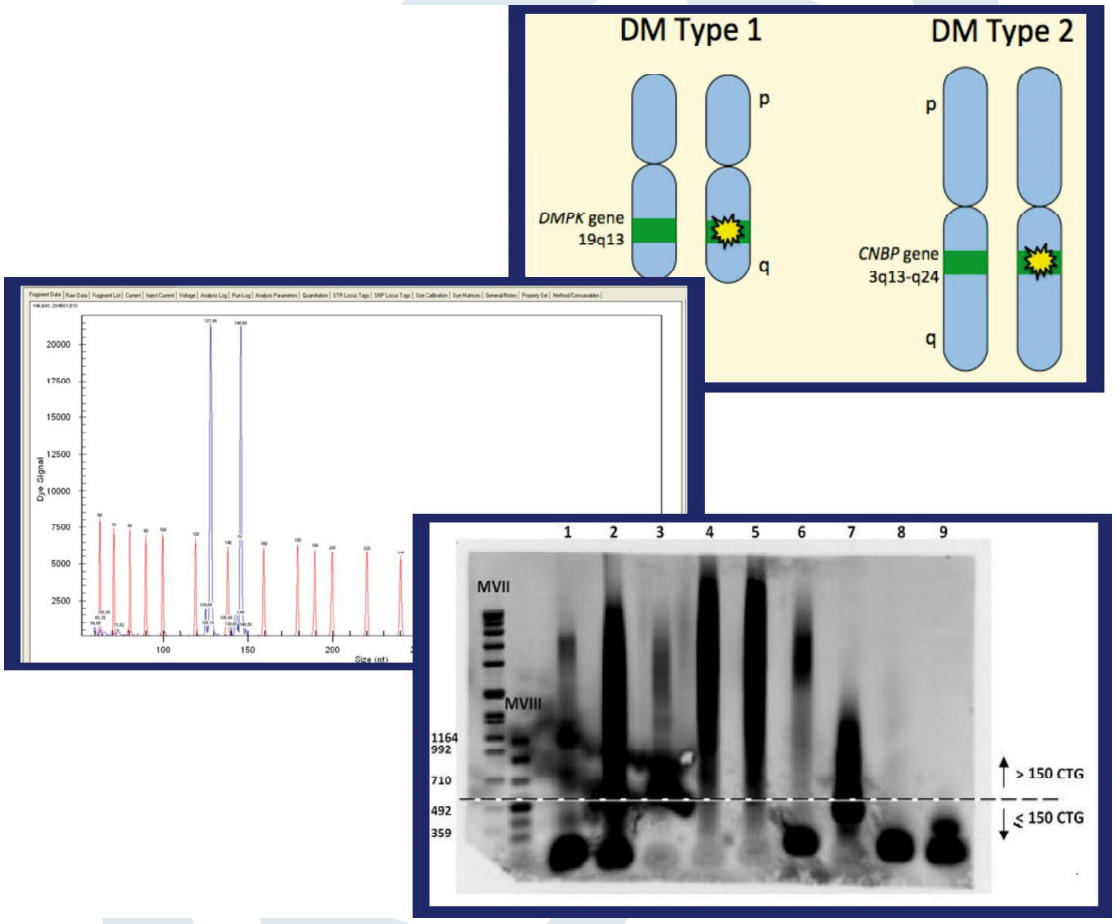
CYTOGEN

Produkte für Medizin und Forschung GmbH

MYOTONIC DYSTROPHY SB KITS

Systems for molecular testing of Myotonic Dystrophy type 1 (DM1) and Myotonic Dystrophy type 2 (DM2) by "long range-PCR" and Southern blotting



CE IVD

EXPERTEAM
l'esperienza in biologia molecolare

CytoGen GmbH
Langgasse 73
35576 Wetzlar, Germany

Tel. + 49 6441-6795588
Fax +49 6441-6795589

web: www.cytogen info
e-mail: cytogen@eurobiz.de

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) . 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 Contenuto del kit rise minimal or classical DM1. A more severe DM1 phenotype is associated with DMPK alleles with sized >**150** CTG. DM2 is caused by a [CCTG]n expansion in the first intron of the ZNF9 (zinc finger protein 9) gene on chromosome 3q21; this gene harbors a complex trait [TG]n[TCTG] n[CCTG]n. Non pathogenic alleles contain up to 26 CCTG repeat units; in DM2 patients, the range of repeat units is extremely wide, ranging from 75 to over 11.000, with a mean of 5.000.

Myotonic Dystrophy SB kits are systems for molecular diagnostic test of the DM1 and DM2 by "**long range-PCR**" and **Southern blotting** according to the **"EMQN Best Practice Guidelines and Recommendations on Myotonic Dystrophy type 1 and 2"**.

Containing of the kits

Label	Contents
DM1 /DM2 MASTER MIX	Mix for the amplification of DMPK or ZNF9 genes
DM1/DM2 DNA polymerase	DNA Polymerase for difficult amplification
DM1/DM2 DIG-probe	DIG-end labelled oligo (CTG)n

How does the kit work?

Myotonic Dystrophy SB kits are based on the **long range-PCR** method followed by **southern blotting** of the PCR products. This technique is able to detect all the alleles containing from 5 to **900/1000** repeat units; they use the **MD DNA polymerase** with proofreading 3´ to 5´ exonuclease activity and whose fidelity, accuracy, and specificity make it ideal for the amplification of GC-rich templates. The procedure requires a single reaction of long-range PCR followed by two step: the first step that is for the analysis of **normal alleles** and/or **small expansions**, consits in sizing of the amplified products by **capillary electrophoresis** (or high resolution agarose gel electrophoresis). The second step performs the characterisation of the **larger expansions** by **southern blotting** of the long range PCR fragments to a nylon membrane and subsequently **hybridization** with a **DIG**-end labelled **$(CTG)_n$ probe.** (For detecting expanded alleles > 900/1000 CTG in DM1 patients use the **Myotonic Dystrophy type 1 GC kit-FL** (cod. DM.04FL).

Starting samples: fresh or recently frozen peripheral blood. **DNA isolation method:** QIAamp DNA blood mini kit, High Pure PCR template preparation kit (Roche). **DNA Sequencer:** CEQ 8000/8800 Genetic Analysis System (Beckman Coulter); 310, 3100, 3130, 3730, 3500 Genetic Analyzers (Applied Biosytems).

Procedure: according to the "EMQN Best Practice Guidelines and Recommendations on Myotonic Dystrophy type 1 and 2" (2010).

Product	Unit	Cat.-No.
Myotonic Dystrophy type 1 SB Kit - FL	40 tests	DM.02FL
Myotonic Dystrophy type 2 SB Kit - FL	40 tests	DM.03FL

Distributor:
CytoGen GmbH
Langgasse 73
35576 Wetzlar, Germany
Tel. + 49 6441 6795588
Fax +49 6441 6795589
web: www.cytogen info
e-mail: cytogen@eurobiz.de

Producer:
EXPERTEAM S.r.l.
via della Libertà 12
30175 Marghera, Italy
tel.: +39 041 5093101
fax: +39 041 5093102
web: www.experteam.it
e-mail:expertm@vegapark.ve.it