digitalMLPA™ NXtec™

# Carrier Panels

MRC Holland

## Reliable analysis beyond routine sequencing

**Our digitalMLPA Carrier Panels are designed for carrier status determination of disorders in which CNVs and complex genomic regions play a key role. Combining the specificity of MLPA with the scale of NGS, these panels enable reliable analysis of challenging genomic targets.**

- ✓ Reliable analysis of challenging targets: SMN region, *CYP21A2*, *HBA1/HBA2*, *DMD* and more.
- ✓ Perfect complement to NGS; allows combination with other NGS libraries.
- ✓ Low costs: 10 to 26 genetic conditions for €35 - €40 per reaction.

- Robust detection of relevant CNVs and selected SNVs
- High specificity even in highly homologous regions
- Simple workflow with minimal hands-on time
- Easy data analysis with free software; no bioinformatics required

## Targeted content

### D038 CARRIER PLUS *Beyond the Essentials*

all D028 targets | 16 additional diseases

- ✓ Medium-chain aculcoenzyme A dehydrogenase deficiency (MCAD)
- ✓ Gaucher disease
- ✓ Fanconi anemia
- ✓ Familial dysautomia
- ✓ Niemann-Pick disease
- ✓ Smith-Lemli-Opitz syndrome
- ✓ Phenylketonuria
- ✓ Tay-Sachs disease
- ✓ Bloom syndrome
- ✓ Fragile X sydrome
- ✓ Hemophilia A

**... and more!**

### D028 CARRIER PANEL 1 *Core Insights*

10 common inherited conditions

- ✓ Spinal muscular atrophy (SMA)
- ✓ Duchenne and Becker muscular dystrophies (DMD/BMD)
- ✓ Alpha- and beta-thalassemia
- ✓ Cystric fibrosis (CF)
- ✓ Congenital adrenal hyperplasia (CAH)
- ✓ Cystinosis
- ✓ Hearing loss
- ✓ Juvenile neuronal ceroid lipofuscinosis

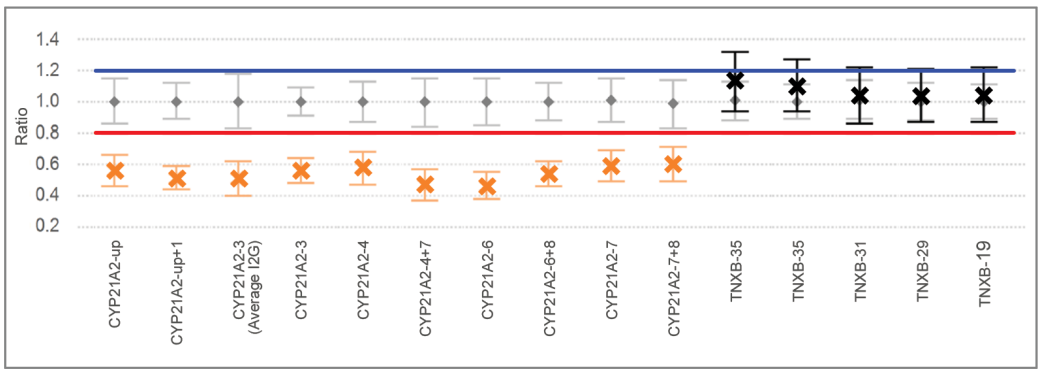
## Results

# Coffalyser digitalMLPA™

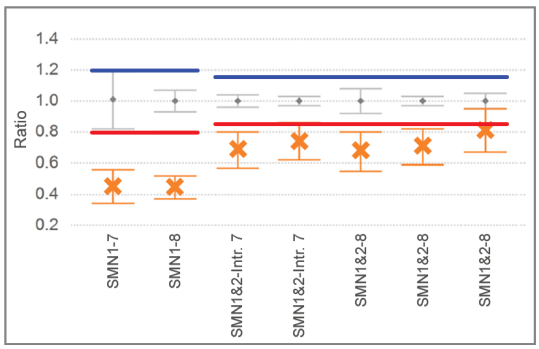
Free software that processes FASTQ files, runs QC, and delivers clear reports



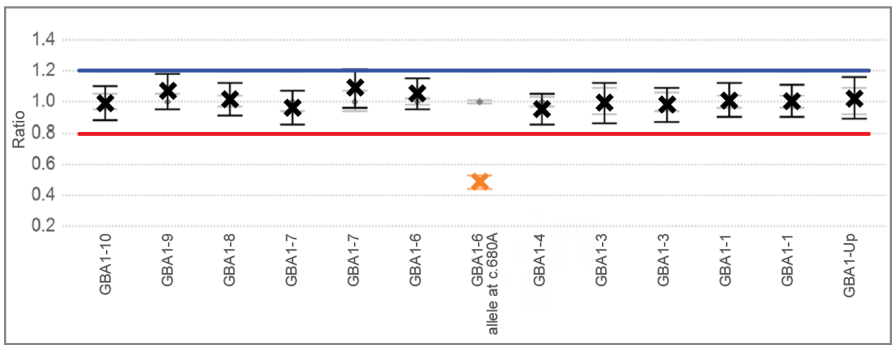

### 1. Heterozygous deletion of *CYP21A2*




### 2. Heterozygous deletion of *SMN1*




### 3. SNV detection in *GBA1* exon 6




Results obtained with NXtec D028 (1) and D038 (2, 3) Carrier Panels, demonstrating heterozygous deletions (1) of CYP21A2 region (normal copy number for *TNXB*); (2) of *SMN1* (5 probes detect sequences found in both *SMN1* and *SMN2*; a ratio of ~0.75 for those probes indicate a copy number of 3, suggesting a *SMN2* copy number of 2) and (3) loss of WT sequence allele c.680A>G in *GBA1* exon 6. A ratio of 1 indicates a normal copy number and ~0.5 indicates a heterozygous deletion. **Orange crosses:** aberrant ratios obtained in the test sample. **Black crosses:** normal range results obtained on the test sample. **Grey diamonds:** ratios obtained on healthy reference samples. The **blue** and **red** lines indicate the boundaries of the expected normal range.

## digitalMLPA Portfolio

**Where MLPA meets NGS.** digitalMLPA combines the precise probe design of MLPA with NGS-based detection, enabling the simultaneous analysis of up to 1600 genomic targets in a single workflow and delivering a powerful and scalable platform for genomic analysis

The digitalMLPA portfolio supports a broad range of genomic applications, from carrier status and hereditary cancer to genome-wide CNV detection and hematologic neoplasms, delivering targeted assays that meet the needs of today's genetic testing laboratories.

**See all our digitalMLPA assays!**

MRC Holland

## Confidence in Copy Number Determination

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