digitalMLPA™ NXtec™

# D028 Carrier Panel 1

MRC Holland

## Reliable carrier status analysis in complex genomic regions

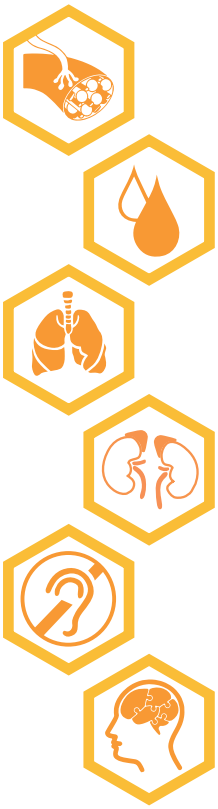
**D028 Carrier Panel 1** delivers an unparalleled detection of CNVs associated with 9 autosomal recessive or X-linked disorders, with additional coverage of 30+ relevant SNVs.

- ✓ **High-specificity:** distinguishes even highly homologous regions
- ✓ **High-throughput:** easy integration into existing NGS workflows
- ✓ **Low Cost:** <€35 (~US$40-45) reagent costs per sample*
- ✓ **Fast:** <24 h from sample to sequencer, with minimal hands-on time
- ✓ **Free software included:** no bioinformatics expertise required

*D028 Carrier Panel 1* brings an MLPA-grade copy number analysis into an NGS-based workflow - even for complex genomic regions.

Romy Kuilboer, MSc
Senior Product Developer MRC Holland

## Panel Coverage



**Spinal Muscular Atrophy (SMA)** - *SMN1-specific targets and silent carrier risk*
**Duchenne & Becker Muscular Dystrophy (DMD & BMD)** - *all 79 DMD exons*

**Alpha-thalassemias** - *HBA region, incl. HBA2 Constant Spring Mutation*
**Beta-thalassemias** - *HBB region, incl. WT sequence of c.316-3C>A mutation*

**Cystic Fibrosis (CF)** - *CFTR, incl. WT sequence of F508del mutation*

**Congenital Adrenal Hyperplasia (CAH)** - *CYP21A2, incl. I2G locus*
**Cystinosis** - *CTNS*

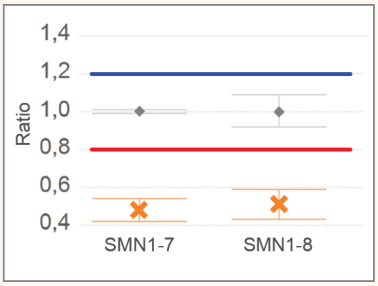
**Hearing loss** - *DFNB1 region, incl. WT sequence of GJB2 c.35delG mutation*

**Juvenile Neuronal Ceroid Lipofuscinosis** - *CLN3*

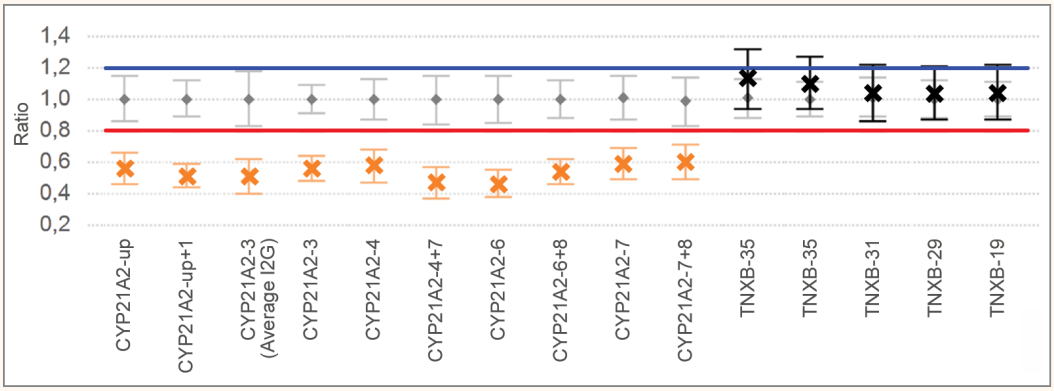
* Costs based on digitalMLPA reagent list prices; possible additional costs not included. Prices may differ when ordering through our sales partners.

## Results

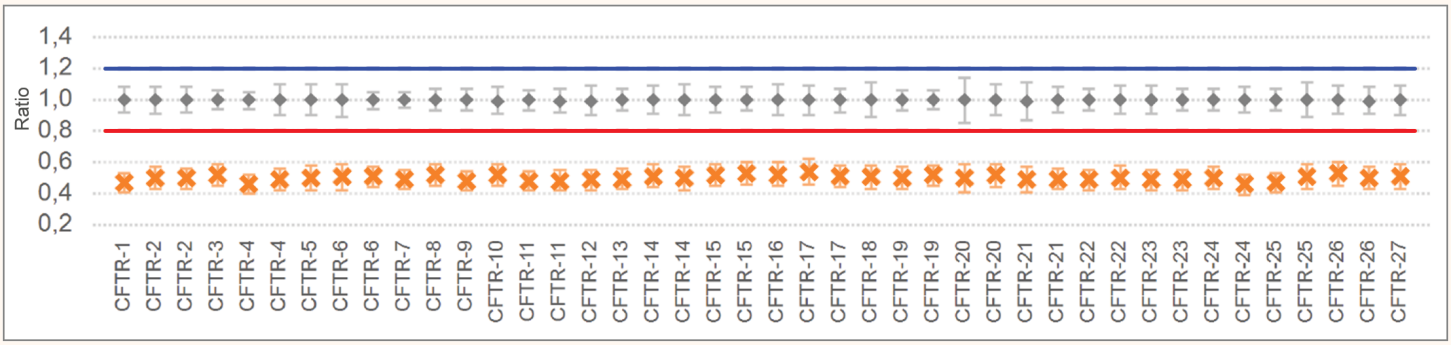
### 1. Heterozygous deletion of *SMN1*




### 2. Heterozygous deletion of *CYP21A2*




### 3. Heterozygous deletion of *CFTR*




Results obtained with NXtec D028 Carrier Panel 1, demonstrating heterozygous deletions in (1) *SMN1* – deletion of exons 7 and 8; (2) *CYP21A2* region (normal copy number for *TNXB*) and (3) *CFTR* – all exons. The **black** and **orange** crosses represent the ratios obtained in the test sample within and outside the borders respectively; a ratio of 1 indicates a normal copy number and ~0.5 indicates a heterozygous deletion. **Grey diamonds** represent ratios obtained in healthy reference samples. The **blue** and **red** lines indicate the upper and lower boundaries of the expected range.

## Software

# Coffalyser digitalMLPA™

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

Versions v2.5.1 and later are compatible with NXtec D028 Carrier Panel 1 and allow for optional masking of selected genes. This software is suitable for all digitalMLPA NXtec assays, including our panels for hereditary cancer, genome-wide CNV detection, acute lymphoblastic leukemia and more!


Scan to check our full NXtec portfolio!

MRC Holland

## Confidence in Copy Number Determination

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