

D028 Carrier Panel 1

Reliable carrier status analysis in complex genomic regions

D028 Carrier Panel 1 delivers an unparalleled detection of CNVs associated with 10 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.

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Panel Coverage



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



Alpha-thalassemia - *HBA region, incl. HBA2 Constant Spring Mutation*
Beta-thalassemia - *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. 12G locus*
Cystinosis - *CTNS*



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

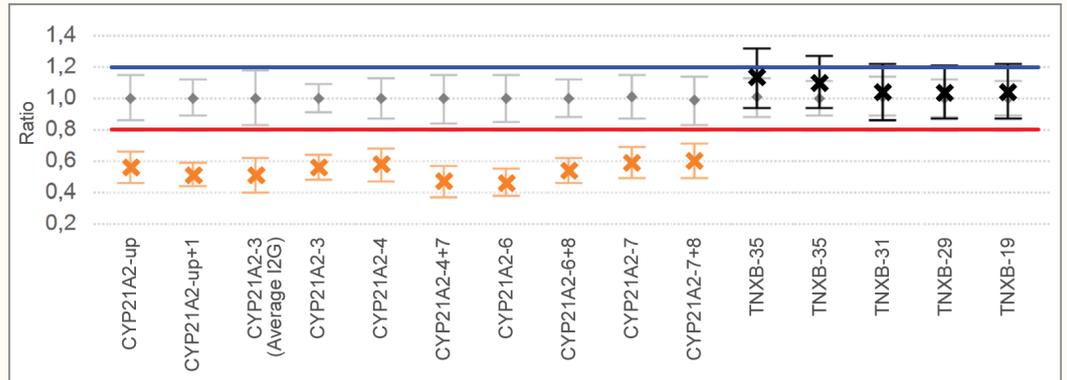


Juvenile Neuronal Ceroid Lipofuscinosis - *CLN3*

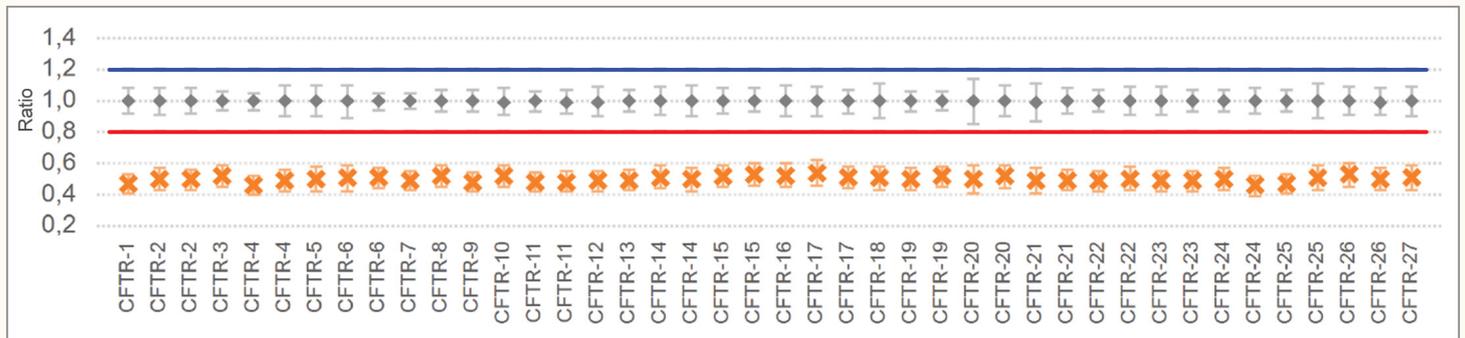
1. Heterozygous deletion of *SMN1*



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!

