digitalMLPA™ NXtec™

D008 DMD

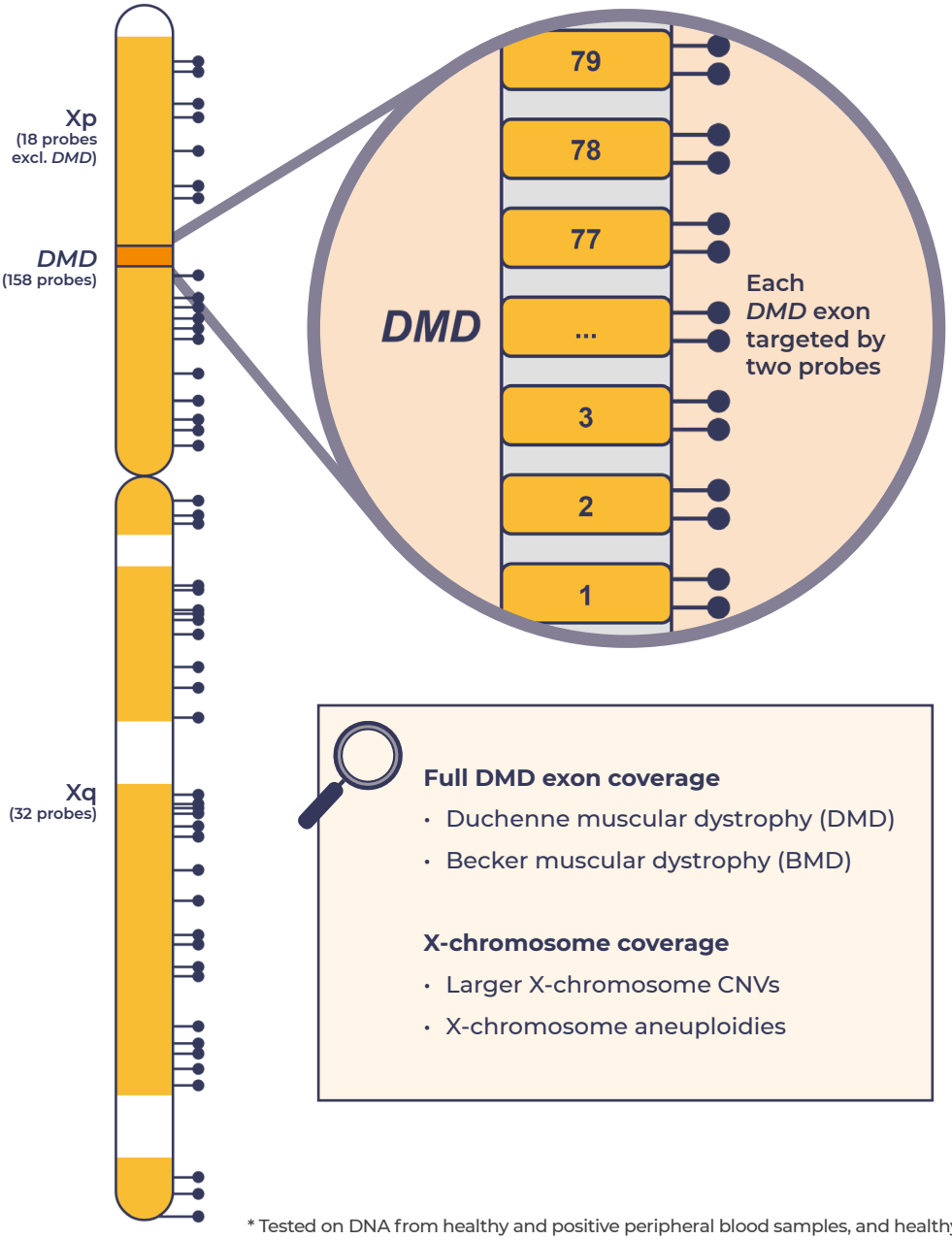
MRC Holland

Gold standard certainty for CNV detection in *DMD*

NXtec D008 DMD delivers highly reliable detection of CNVs linked to Duchenne and Becker muscular dystrophies. Covering the entire *DMD* gene at single-exon resolution, this digitalMLPA assay provides comprehensive results in just one reaction.

- ✓ **Comprehensive:** each *DMD* exon targeted by two probes
- ✓ **Specific:** high-confidence CNV detection from single exon to whole-gene changes, including larger X-chromosome CNVs, and whole X-chromosome aneuploidies
- ✓ **Versatile:** robust performance on DNA from different sample types*
- ✓ **Fast:** <24 h from sample to sequencer, with minimal hands-on time
- ✓ **Simple analysis:** free and easy-to-use software, no bioinformatics expertise required
- ✓ **Flexible:** combine digitalMLPA with other libraries to maximise sequencing capacity

Panel Coverage



Full DMD exon coverage

- Duchenne muscular dystrophy (DMD)
- Becker muscular dystrophy (BMD)

X-chromosome coverage

- Larger X-chromosome CNVs
- X-chromosome aneuploidies

Required materials

- As little as 20 ng of sample DNA
- Thermocycler with heated lid
- Compatible NGS instrument and reagents
- digitalMLPA NXtec probemix, reagents and barcode plates



Figure legend

- *DMD*: target gene
- Targeted X-chromosome regions
- Non-targeted regions
- Probe

* Tested on DNA from healthy and positive peripheral blood samples, and healthy amniotic fluid (23-30 weeks) and CVS samples (12-19 weeks).

Comparison

	NXtec D008 DMD	NXtec D028 Carrier Panel 1	SALSA MLPA probemixes P034 DMD Mix 1 & P035 DMD Mix 2
Technique	digitalMLPA	digitalMLPA	MLPA
Coverage (*DMD*)	All 79 exons covered with 2 probes.	All 79 exons covered with 1 probe.	All 79 exons covered with 1 probe; additional probes for exon 1.
Coverage (additional)	50 probes distributed across the X chromosome	131 probes targeting *SMN1, CYP21A2, CFTR, CLN3, CTNS* and the HBB, HBA and DFNB1 regions.	-
DNA input	20 ng	20 ng	50 ng
# of reactions for full *DMD* coverage	1	1	2
Costs** for full *DMD* coverage	~€30	~€35	~€30
Equipment	Thermocycler with heated lid + compatible NGS instrument	Thermocycler with heated lid + compatible NGS instrument	Thermocycler with heated lid + compatible capillary electrophoresis instrument
CE-marked	No	No	Yes (IVDR)

** Costs based on list prices. Prices may differ when ordering through our sales partners.

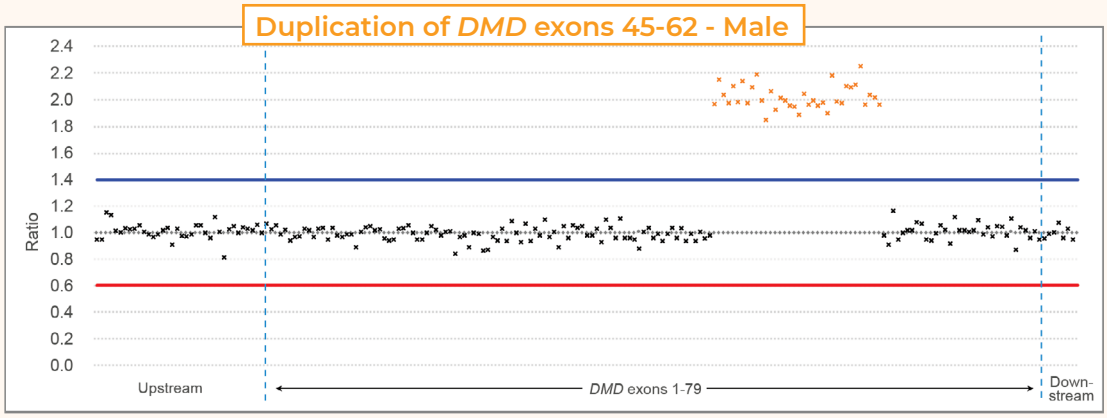
Results

Coffalyser digitalMLPA™ data analysis software

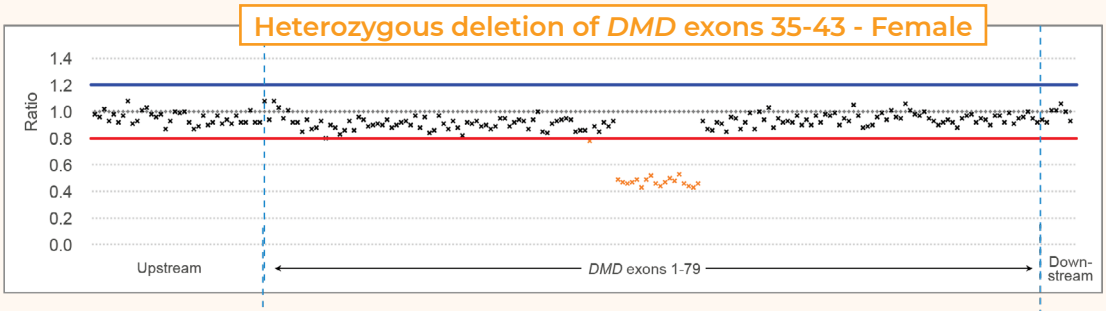
Free software that processes FASTQ files, runs QC, and delivers clear CNV reports

NXtec D008 DMD detects CNVs in *DMD* and whole X-chromosome aneuploidies

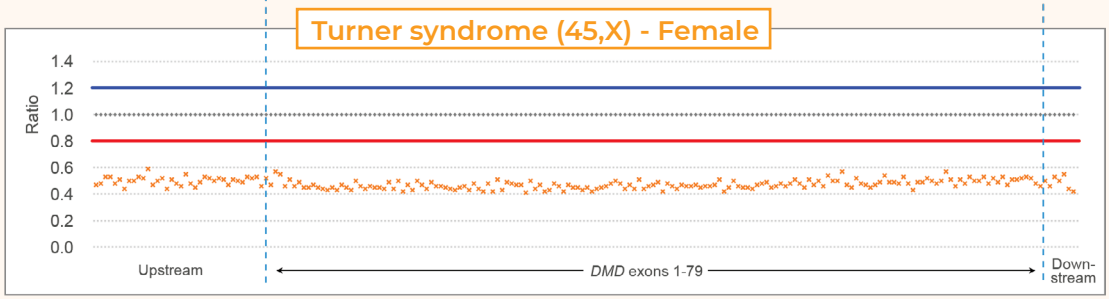
Duplication of *DMD* exons 45-62 - Male



Heterozygous deletion of *DMD* exons 35-43 - Female



Turner syndrome (45,X) - Female



Ratio 1.0 = normal copy number.

Values beyond **blue**/**red** thresholds indicate duplications/deletions.

Grey diamonds = probe ratios of reference samples.

black/**orange** crosses = probe ratios of test sample (normal/aberrant).

DNA samples used: Coriell NA05123, NA05169, NA20027 (top–bottom).

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Confidence in Copy Number Determination

Scan for full NXtec portfolio

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