MRC Holland

SALSA® MLPA®

The Gold Standard for DNA Copy Number Determination

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- ✓ **Reliable:** the best method for detecting gene copy number variations
- ✓ **Straightforward:** easy to perform, simple analysis & clear results
- ✓ **Cost-efficient:** run on standard lab equipment & low assay costs
- ✓ **Versatile:** from whole genes to single exons, from methylation patterns to complex regions - MLPA offers the best solution for each application

MLPA. The number one in CNV detection.

MLPA is the go-to technique for studying gene copy number variations (CNVs) associated with disease. With MLPA's optimized multiplex PCR-based method, it is possible to detect deletions and duplications in up to 60 DNA sequences in one easy reaction, without PCR primer bias. In addition, MLPA's detection range stretches from complete chromosomes down to single exons, and the method is so sensitive that it can reliably discriminate genes from highly similar pseudogenes. This is why laboratories worldwide rely on MLPA for studying genetic disorders and tumours.

Thousands of labs around the world rely on MLPA for the diagnosis and research of genetic disorders and tumours.

MLPA Application Highlight

Top 10 selling MLPA applications

Predisposition to Cancer	
BRCA1	**P002** BRCA1; **P087** BRCA1 confirmation
BRCA2	**P045** BRCA2/CHEK2; **P090** BRCA2; **P077** BRCA2 confirmation
Lynch Syndrome	**P008** PMS2; **P003** MLH1/MSH2; **ME011** Mismatch Repair Genes; **P072** MSH6-MUTYH
Congenital Disorders & Carrier Testing	
Alpha Thalassemia	**P140** HBA
Congenital Adrenal Hypoplasia	**P050** CAH
Cystic Fibrosis	**P091** CFTR
Duchenne Muscular Dystrophy	**P034** DMD-1; **P035** DMD-2
Spinal Muscular Atrophy	**P021** SMA; **P060** SMA Carrier; **P460** SMA (Silent) Carrier
Imprinting Disorders	
Prader-Willi/Angelman Syndrome	**ME028** Prader-Willi/Angelman
Beckwith-Wiedemann / Russell-Silver Syndrome	**ME030** BWS/RSS

MLPA Application Selection

Over 350 MLPA assays are available, covering hundreds of disorders and thousands of genes. A small selection of our panels and genes:

Predisposition to Cancer

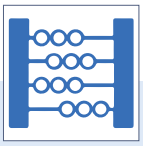
Breast Cancer (*BRCA1/2, CHEK1/2, TP53*) Lynch Syndrome (*MLH1**, *MSH2/6**, *PMS2**) Neurofibromatosis (*NF1/2*) *PTEN STK11 CDH1 PALB2 ATM*

Tumour Profiling

Tumour suppressors (*IKZF1, TP53, RB1**) Blood cancers (ALL, MDS, CLL, MM) Breast (BRCA1ness, *ERBB2, CCNE1*) Glioma (1p, 19q, *IDH1, IDH2, MGMT**)

Sensory Disorders

Retinoblastoma Uveal Melanoma Optic Atrophy type 1 Stargardt Disease Macular Degeneration Usher Syndrome Cone-rod Dystrophy 3



Intellectual Disability

Prader-Willi/Angelman Syndrome* Subtelomeres Microdeletion Syndromes Tuberous Sclerosis Rett Syndrome DiGeorge Syndrome UPD7/14*

Neurological Disorders

Parkinson's Disease Hereditary Spastic Paraplegia Epilepsy (*KCNQ2/3, SCN1A*) Dopa-responsive Dystonia

Neuromuscular Disorders

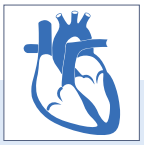
Spinal Muscular Atrophy (*SMN1, SMN2*) Duchenne Muscular Dystrophy (*DMD*) Charcot-Marie-Tooth Disease Limb Girdle Muscular Dystrophy

Immunological Disorders

Ataxia-Telangiectasia (AT) Acute Lymphoblastic Leukemia (ALL) Common Variable Immunodeficiency (CVID) Hereditary Angioedema (HAE)

Hereditary Blood Disorders

Thalassemia (Alpha, Beta) Fanconi Anemia Clotting Factor Deficiencies (V, IX, X, XI) Von Willebrand Disease



Cardiovascular Disorders

Marfan Syndrome *HHT/HPAH* Loeys-Dietz Syndrome Familial Hypertrophic Cardiomyopathy

Lung Disorders

Cystic Fibrosis Primary Ciliary Dyskinesia Alveolar Capillary Dysplasia AAT-deficiency

Kidney Disorders

Polycystic Kidney Disease Hemolytic Uremic Syndrome (HUS) Birt-Hogg-Dube Syndrome Alport Syndrome Nephronophthisis 1

Skeletal & Connective Tissue

Ehlers-Danlos (*PLOD1, COL3A1/5A1*) Marfan Syndrome Osteogenesis Imperfecta (*COL1A1/2*) *SHOX*

Endocrinological Disorders

Congenital Adrenal Hyperplasia MODY Multiple Endocrine Neoplasia (*MEN1*) Albright Hereditary Osteodystrophy (*GNAS*)*

Metabolic & Mitochondrial Disorders

Congenital Adrenal Hyperplasia (CAH) Hypercholesteremia Cytochrome *P450* Fabry Disease Phenylketonuria (PKU)

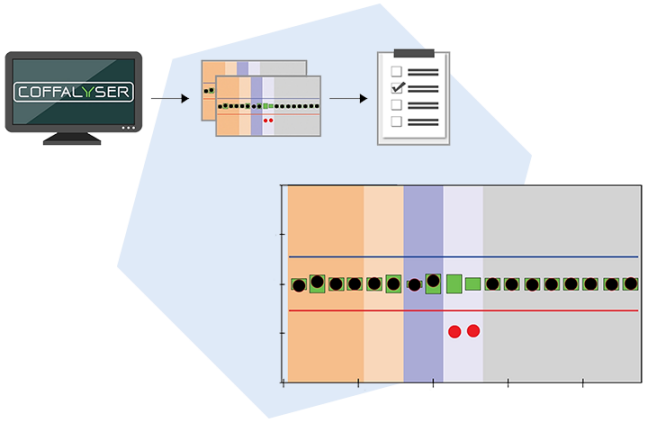
...and many more!

Scan the code or go to mrcholland.com/products

* For this gene/application, both copy number and DNA methylation can be determined.

DIY Analysis. No bioinformatic skills needed.
Coffalyser.Net™. For Results that Count.

✓ **Available free of charge**
✓ **Seamlessly Integrated**
✓ **Always up-to-date**



Coffalyser.Net is *free* MLPA analysis software made and supported by MRC Holland. Coffalyser.Net directly imports raw data files, performs advanced quality checks and data analysis to increase the robustness of your results. Analysis sheets for each product can be retrieved directly from our servers, ensuring you always have access to the latest versions.

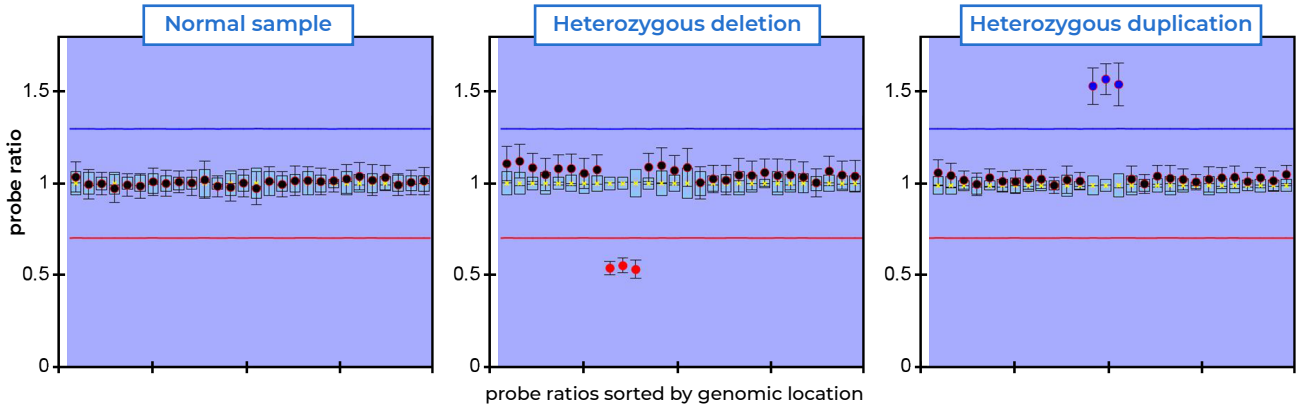


Figure 1. Determining *BRCA1* copy number using SALSA® MLPA® Probemix P002 BRCA1 and Coffalyser.Net.
Probe ratios sorted by genomic location. Only excerpt shown, from *BRCA1* exon 24 (left) to exon 7 (right).

Ratios indicate number of probe targets found in tested sample compared to reference genomes.

Ratio	Copy Number	Genotype
1	2	Normal
0.5	1	Heterozygous deletion
1.5	3	Heterozygous duplication

Left: *normal sample, no aberrations found.*

Middle: *sample showing a heterozygous deletion of exon 15 (1 probe) and exon 16 (2 probes).*

Right: *sample showing a heterozygous duplication of exon 13 (3 probes). This* BRCA1 *duplication is relatively common in the UK due to a founder mutation.*

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Confidence in Copy Number Determination

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