MRC Holland

SALSA® digitalMLPA™ Probemix D007 Acute Lymphoblastic Leukemia

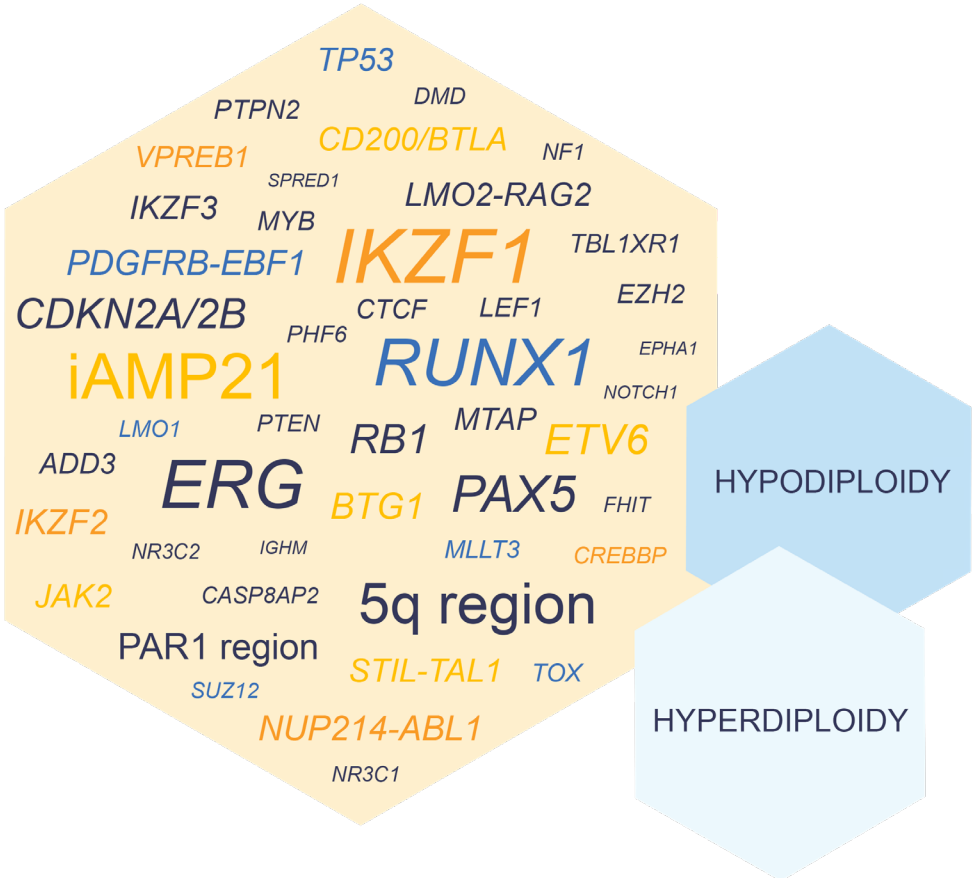
✓ **55 genes and regions** targeted
✓ **High dynamic range** for copy number alteration (CNA) detection
✓ **Quick turnaround** of 48-72 hours

Recurrent and clonal genetic alterations in different subtypes of Acute Lymphoblastic Leukemia (ALL) are well-characterized and known to be strong independent predictors of disease outcome. The conventional SALSA® MLPA® technology is one of the standard methods for the detection of *IKZF1* deletions, which are associated with unfavorable outcome in childhood ALL. To increase genomic coverage, the newer SALSA® digitalMLPA™ technology was employed to create a novel, all-encompassing ALL panel: **SALSA® digitalMLPA™ Probemix D007 Acute Lymphoblastic Leukemia**.

D007 Acute Lymphoblastic Leukemia is the perfect time-saving complement to next generation sequencing (NGS). digitalMLPA ensures high level of confidence in CNA calling and a wide range of ALL-related CNAs can be detected in one single reaction:

- Intragenic CNAs
- Partial chromosome gains, losses and high-level amplifications
- Hyper-/hypodiploidy
- Intrachromosomal gene fusions

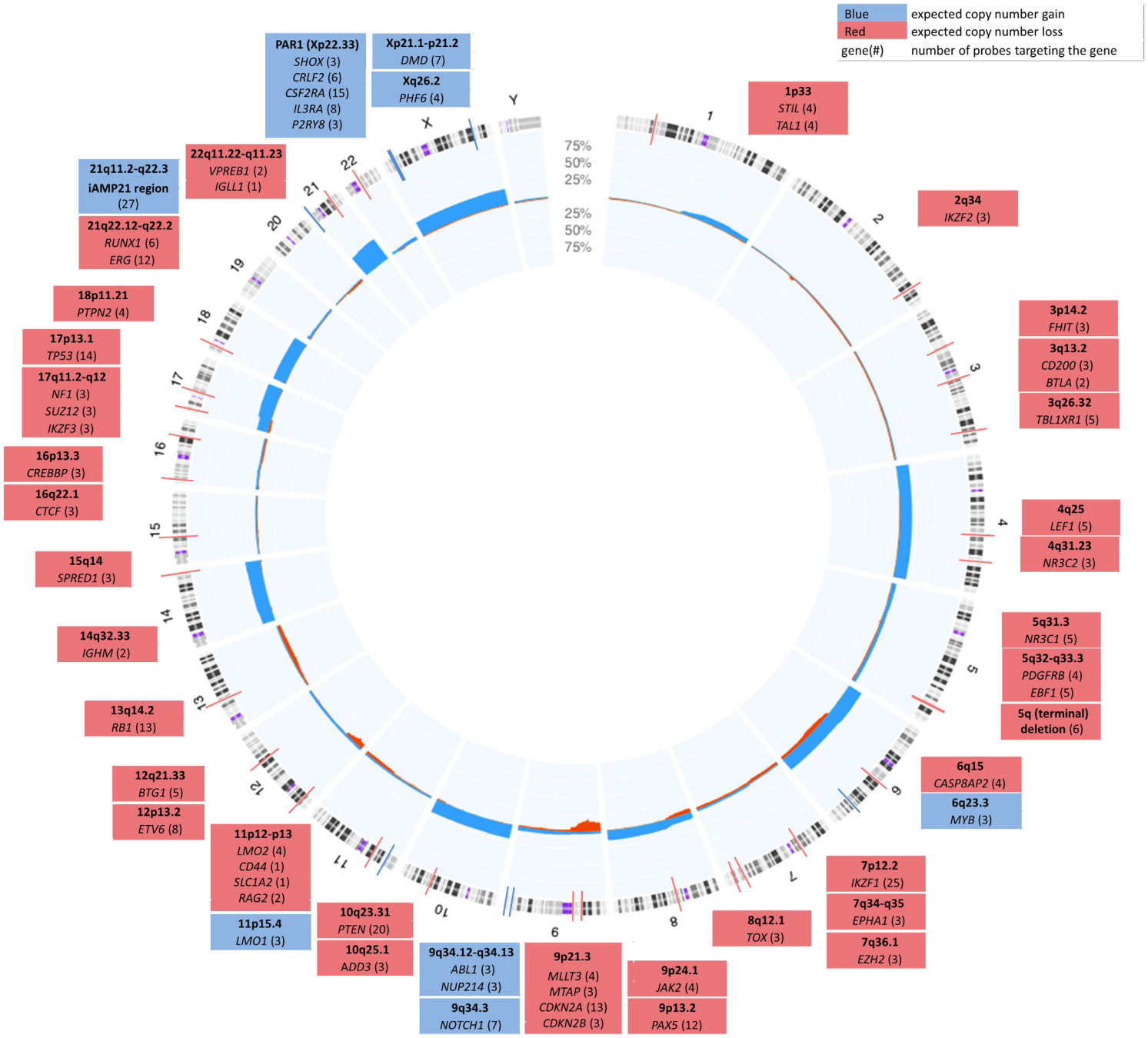
In addition, digitalMLPA can be combined with NGS libraries on the same flow cell, and is analysed using our free, easy-to-use Coffalyser digitalMLPA™ software - no bioinformatic skills are needed.



D007 Acute Lymphoblastic Leukemia contains probes targeting:

1. Genes and regions included in the well-established SALSA® MLPA® probemixes for ALL: P335 ALL-IKZF1, P202 IKZF1-ERG, P327 iAMP21-ERG, P329 CRLF2-CSF2RA-IL3RA and P383 T-ALL.
2. B-cell differentiation and cell cycle control genes, T-ALL-associated aberrations, iAMP21, 5q terminal region and copy number aberrations of PAR1 region.
3. Karyotyping probes for subtelomeric, pericentromeric and middle regions of chromosomal arms to detect larger copy number alterations and hyper-/hypodiploidy.

D007 Acute Lymphoblastic Leukemia: target genes and regions



Target genes and regions included in D007 Acute Lymphoblastic Leukemia. Circos plot shows CNA frequencies reported in the ALL patient population according to the Progenetix database. Inner circle: losses (red) and gains (blue). Outer circle: chromosomal locations. Red and blue boxes: deletions and respectively gains detected by ≥ 2 digitalMLPA probes. (n): number of probes per targeted region.

Required materials

- 20 ng of sample DNA derived from peripheral blood or bone marrow
- Thermocycler with heated lid
- Illumina sequencing platform (all devices), flow cell and reagents
- SALSA® digitalMLPA™ probemix, reagents and barcode plates

References

Benard-Slagter A et al. (2017). *J Mol Diagn*. 19:659-72.

Kiss R et al. (2020). *Mod Pathol*. 33:812-24.

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Confidence in Copy Number Determination

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