



LENA Q51: A retrospective analysis of fusion gene expression characteristics in acute myeloid leukemia (AML) and acute lymphoblastic leukemia (ALL)

- **Cohort:** 6,040 adults and children with newly diagnosed acute leukemia from 7 different sites (September 2016 - January 2020)
- **Study design:** Retrospective, multicenter analysis to evaluate fusion gene expression patterns and characteristics in acute leukemia patients using multiplex PCR screening.
- **Study objectives:** Analyze the expression of fusion genes in patients with newly diagnosed AML and ALL, explore characteristics of immunophenotypes and gene mutations in patients with different fusion gene families, and improve diagnostic accuracy.
- **Key outcomes:**
 - **AML Analysis (4,192 patients):** 46.47% positive for fusion genes with 29 different types detected. Expression showed "power law distribution" with PML-RAR α (18.96%), RUNX1-RUNX1T1 (14.98%), and CBF β -MYH11 (5.46%) being most common. Pediatric patients had significantly higher fusion gene positivity than adults (69.18% vs. 44.76%). MLL-FG and NUP98-FG positive patients were associated with FLT3 and RAS signaling pathway mutations.
 - **ALL Analysis (1,994 patients):** 44.3% positive for fusion genes with 15 different types detected. B-ALL had significantly higher fusion gene positivity than T-ALL (48.48% vs. 18.71%). Peak expression occurred at <1 year, 3-5 years, and 35-44 years. MLL-FG, TEL-AML1, and BCR-ABL1 were most common in infants, children, and adults respectively.
 - **Clinical Validation (1,750 samples, 2022):** 33.89% positive rate with 32 fusion gene types detected, confirming assay performance in routine clinical use.

The results from this large-scale retrospective study demonstrate that LENA Q51 multiplex PCR screening can accurately detect and classify fusion genes in acute leukemia, supporting its feasibility for broad clinical implementation in diagnosis, risk stratification, and treatment planning.