



Spectrum and Frequency of Cytogenetic and Molecular Abnormalities in Paediatric, Teenage and Young Adult Patients with Acute Myeloid Leukaemia

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BACKGROUND & OBJECTIVE

- Genetic lesions set the AML **risk band** and treatment intensity [1].
- Reported abnormality rates vary (23–64%), largely by **technique** [3,4].
- Objective:** define the spectrum and frequency of cytogenetic and molecular abnormalities in patients under 30 years.

METHODOLOGY

- Descriptive cross-sectional study; **89** newly diagnosed AML patients (<30 y), consecutive sampling.
- Diagnostic marrow: **G-banded karyotype + FISH; PCR** for FLT3-ITD, NPM1, CEBPA, RUNX1.
- SPSS 27; chi-square / Fisher's exact; stratified by age, gender, FAB.

RESULTS

89

patients; mean age 14.4 y; 61.8% paediatric

59.6%

cytogenetic abnormality (53/89)

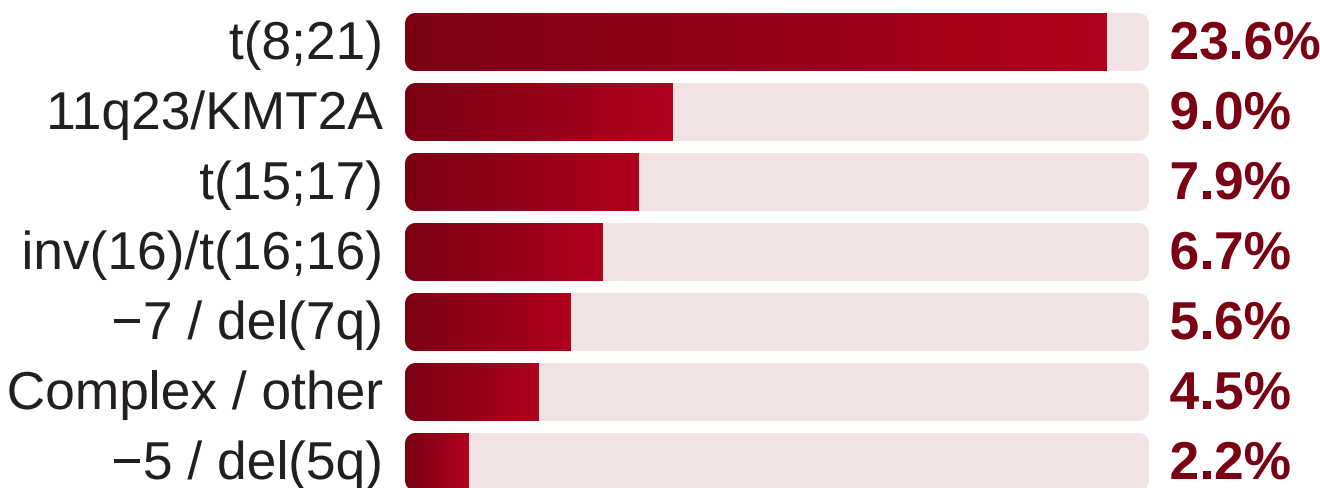
30.3%

core-binding factor AML (27/89)

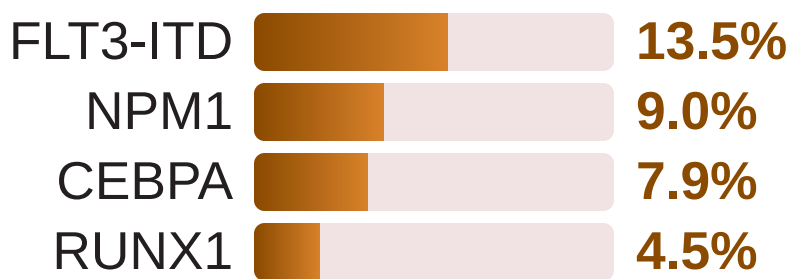
30.3%

≥1 molecular marker positive (27/89)

Cytogenetic lesions (% of 89)



Molecular markers (% of 89)



Techniques are complementary

63.3%

molecular positive if karyotype **normal** (19/30)

13.2%

if karyotype **abnormal** (7/53); p<0.001

- All 8 NPM1 cases had a normal karyotype; no association with age, gender or FAB group (p>0.15).
- Exploratory (small strata): abnormality commoner in teenage/young adult (TYA) males (81.0% vs 54.5%, p=0.048) and M0–M2 (80.0% vs 42.4%, p=0.015).

DISCUSSION

- Yield matches New Delhi (64.3%) and exceeds banding-alone series (23.3%): a **technical** difference [2,4].
- t(8;21) and CBF rates mirror Karachi (26.4%; 32.5%) [3].
- Limits: single centre, four markers only, small strata.

CONCLUSION

- Lesions in **~3 of 5** young AML patients; few translocations account for most.
- Molecular markers positive in many with a **normal karyotype**.
- A small combined panel can risk-stratify most at diagnosis.

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