MOLECULAR PROFILING OF CHRONIC LYMPHOPROLIFERATIVE DISEASES IN BRAZILIAN PATIENTS: A REAL-WORLD NGS PANEL EXPERIENCE

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BACKGROUND

Next-generation sequencing (NGS) panels identify clinically relevant mutations in lymphoproliferative diseases, enabling accurate diagnosis and personalized therapy.

However, these panels are rarely available in low- and middle-income countries, where molecular data remain scarce.

AIM

To describe the molecular landscape and the clinical utility of a targeted NGS panel in Brazilian patients with mature lymphoproliferative diseases.

METHODS

Retrospective analysis of Brazilian patients with mature lymphoproliferative disorders who underwent molecular testing with a targeted NGS panel.

Tumor mutational burden (TMB) and microsatellite instability (MSI) were assessed, and mutated genes were grouped by functional pathway.

Variants were reviewed for diagnostic contribution, therapeutic actionability and resistance-predictive value.

CONCLUSIONS

NGS panels provide significant clinical value in Brazilian patients with lymphoproliferative diseases, through diagnostic clarification and identification of actionable biomarkers.

These findings support broader implementation of NGS panels in Latin American oncology practice, to improve diagnostic accuracy and enable precision medicine approaches.

RESULTS

61
patients profiled with a targeted NGS panel

95%
with at least one mutation identified

46%
with **potentially** actionable biomarkers

Diagnostic categories

Category	n	%
Aggressive B-cell lymphomas	28	46
T-cell lymphoproliferative diseases	13	21
CLL / SLL	6	10
Other B-cell lymphomas	14	23

Median TMB 4.7 mut/Mb (range 0.0–192.7); MSI detected in 1 patient (1.6%), with PTLD.

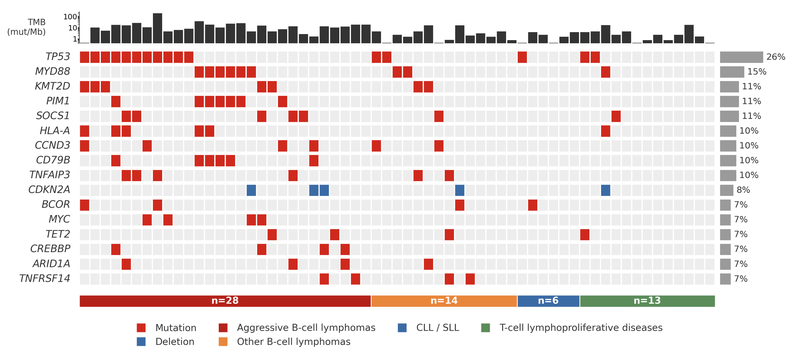

Figure 1. Mutational landscape of the cohort (genes altered in at least 4 patients (≥ 7%)).

Panel contribution to diagnosis

Diagnosis	Key alterations	Panel contribution
cHL	*CCND3*, *SOCS1*, *IGH* rearrangement	Excluded ALCL and corroborated cHL
PMBCL	*SOCS1*, *TP53*, *TNFAIP3*, *XPO1*	Supported PMBCL rather than DLBCL
Nodal marginal zone lymphoma	*TNFAIP3*, *KMT2D*, *ANKRD11*, *BARD1*	Profile favoured MZL with plasmacytic differentiation (differentials: LPL, atypical SLL)
T-LGL leukaemia	*STAT3*, *ASXL1*	Supported the diagnosis
B-cell LPD, unclassified	*TP53*, *NCOR1*	Excluded mantle cell lymphoma
B-cell LPD, unclassified	*TNFRSF14*, *B2M*, *CD58*	Excluded plasmablastic lymphoma
Splenic diffuse red pulp small B-cell lymphoma	*CCND3*, *TP53*	Supported the diagnosis alongside clinical and immunophenotypic features
Extranodal NK/T-cell lymphoma	*TP53*, *STAT3*, *TET2*	Supported the diagnosis

LIMITATIONS

- Retrospective design, restricted to patients referred for clinical molecular testing;
- Heterogeneous diagnostic categories limit subgroup analysis;
- Actionability was inferred from molecular findings; treatment impact was not assessed.

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