TASMANIAN HEALTH SERVICE

Rapid Integrated DNA/RNA Panel Testing for Acute Leukaemias: A Regional Centre Experience

THS Pathology – Royal Hobart Hospital

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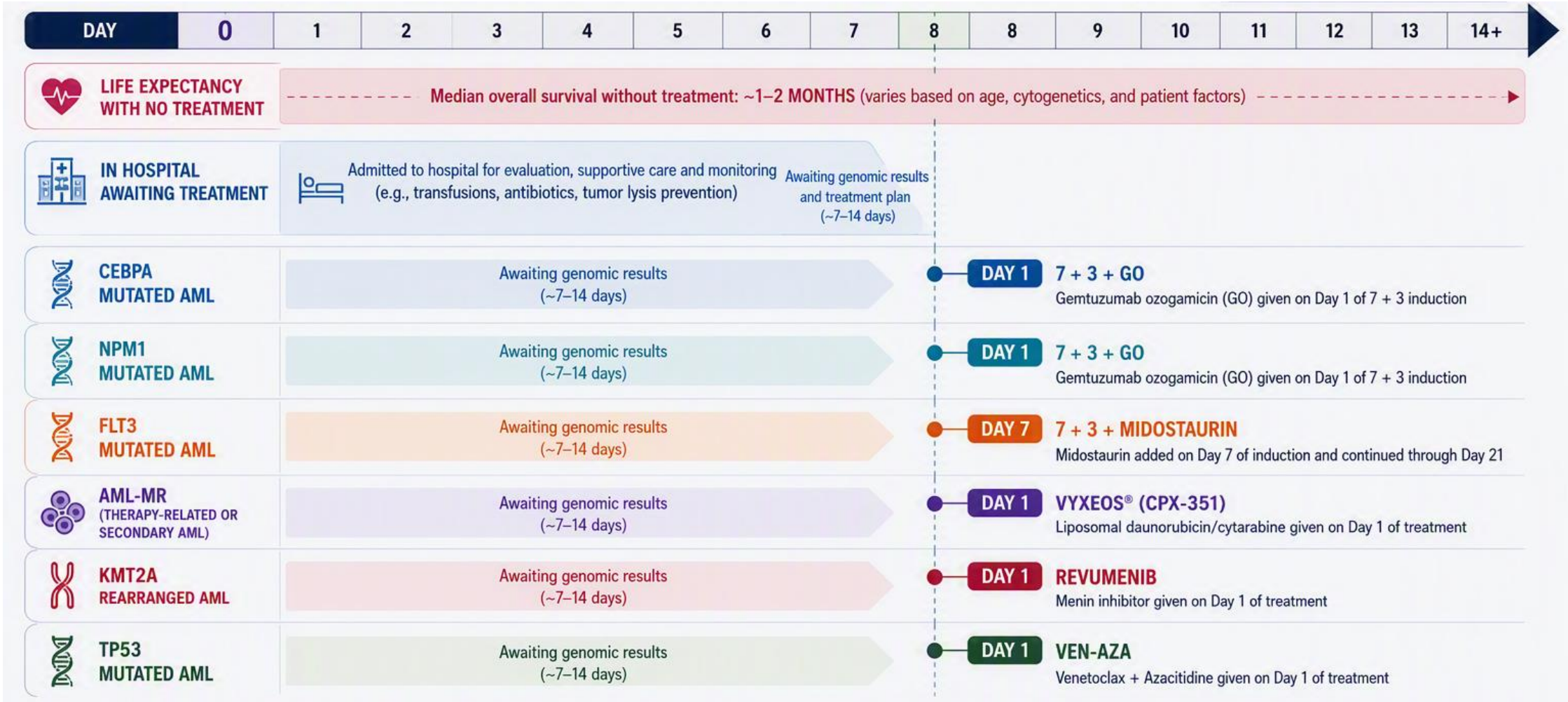
PPA NATIONAL FORUM, INNOVATIONS PLENARY 2026

Acute Myeloid Leukaemia (AML) treatment



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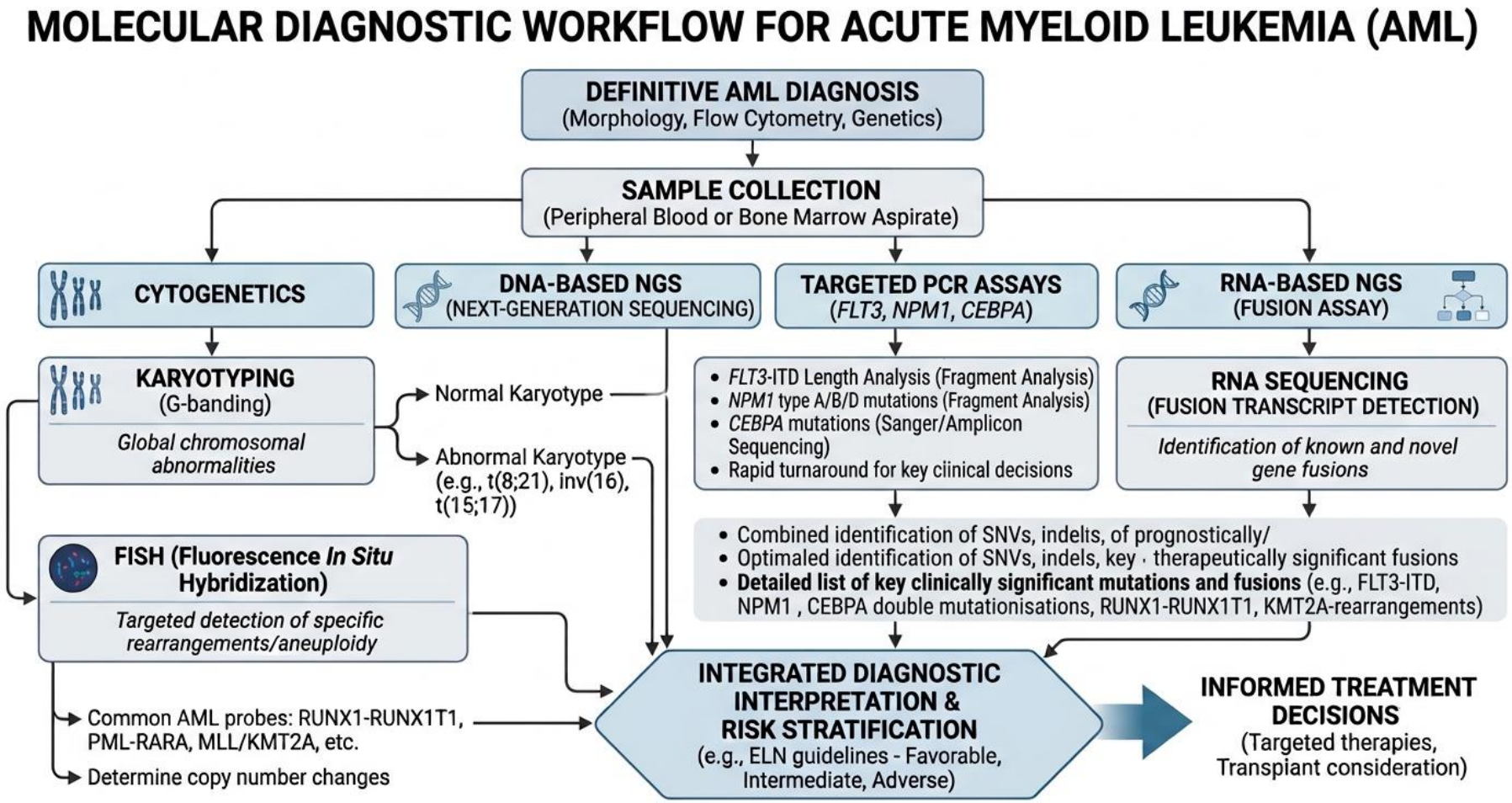
Conventional workflow



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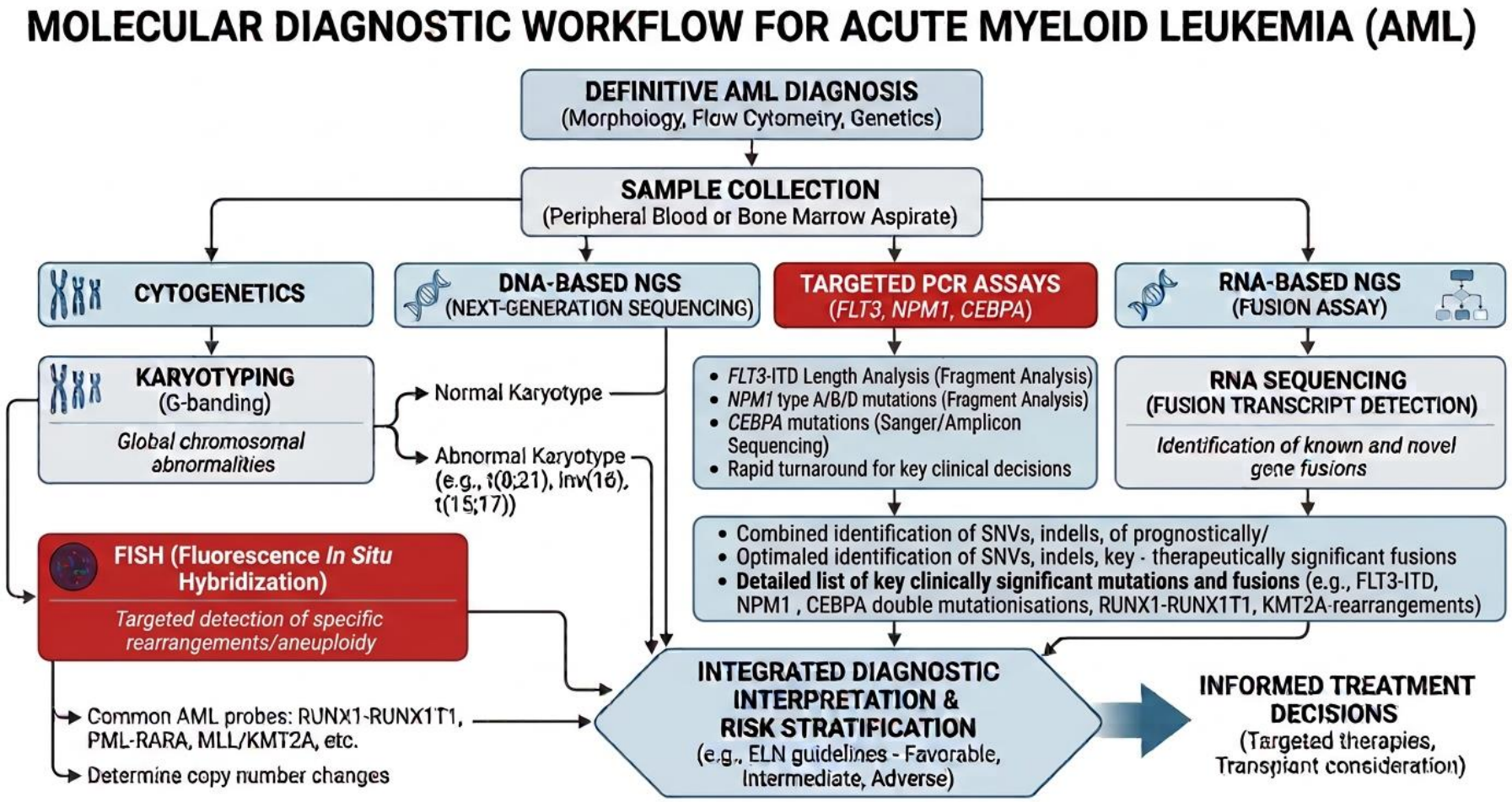
Conventional workflow



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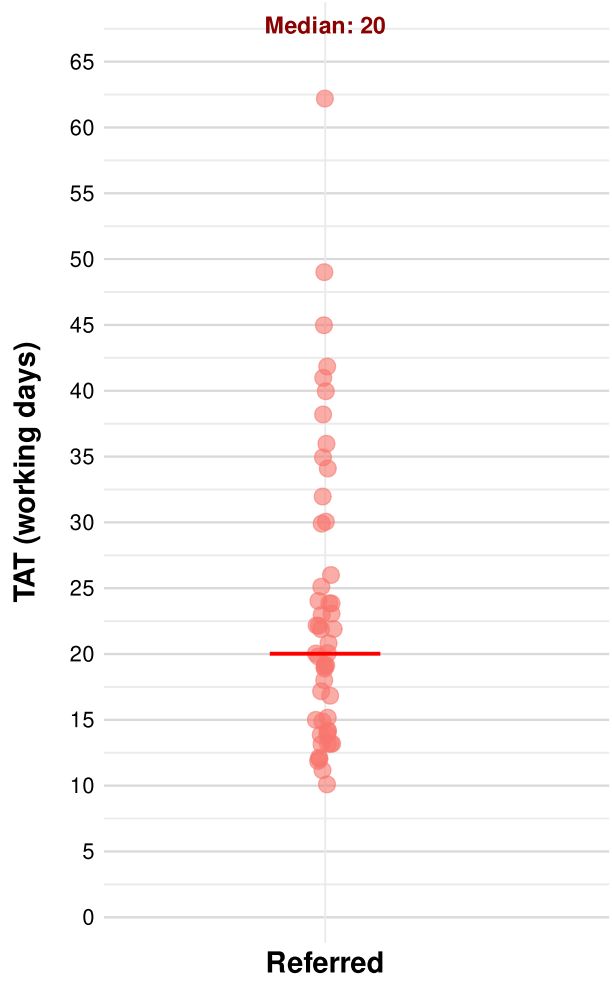
Risk and impact of slow turn around time

- Non-ideal empirical treatment e.g. 7 + 3 without targeted agent
- Missed clinical trial opportunities
- Patient deconditioning
- Futile treatment
- Increased length of hospital stay



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Aims

- Bring testing in-house.
- Reduce TAT to <5 working days.
- Do it with existing staff resource and while maintaining other services.
- Reduce costs.

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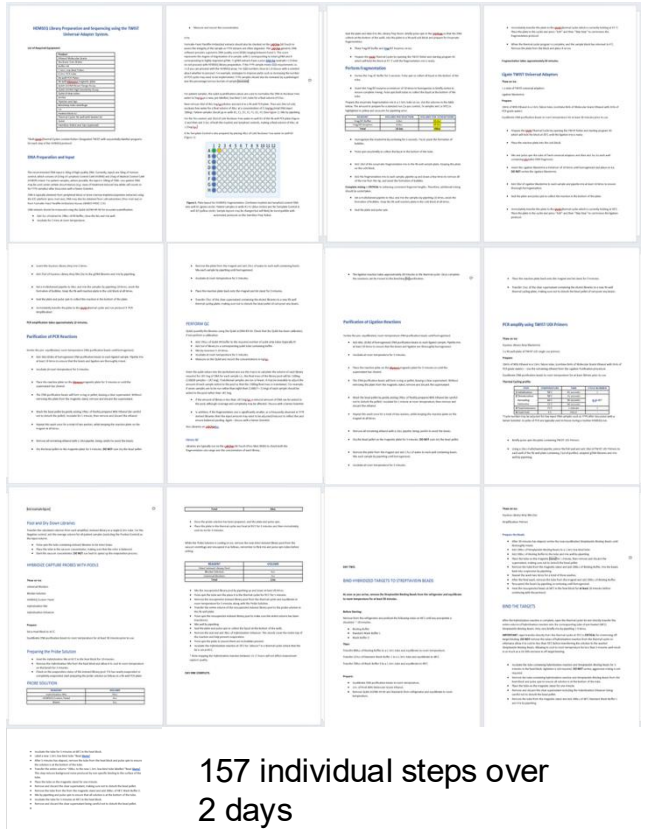
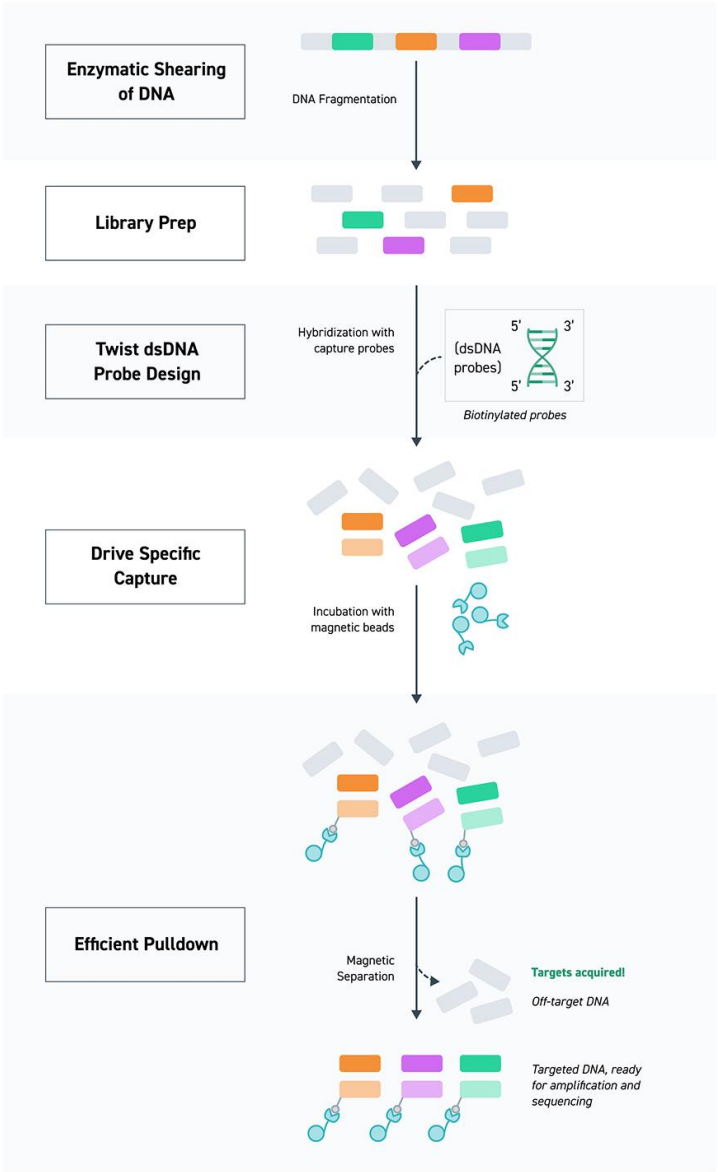
Twist assay

- Excellent quality results.
- Cost effective.
- DNA only.
- Time consuming manual process.
- Expensive and/or difficult to fully automate.



157 individual steps over 2 days



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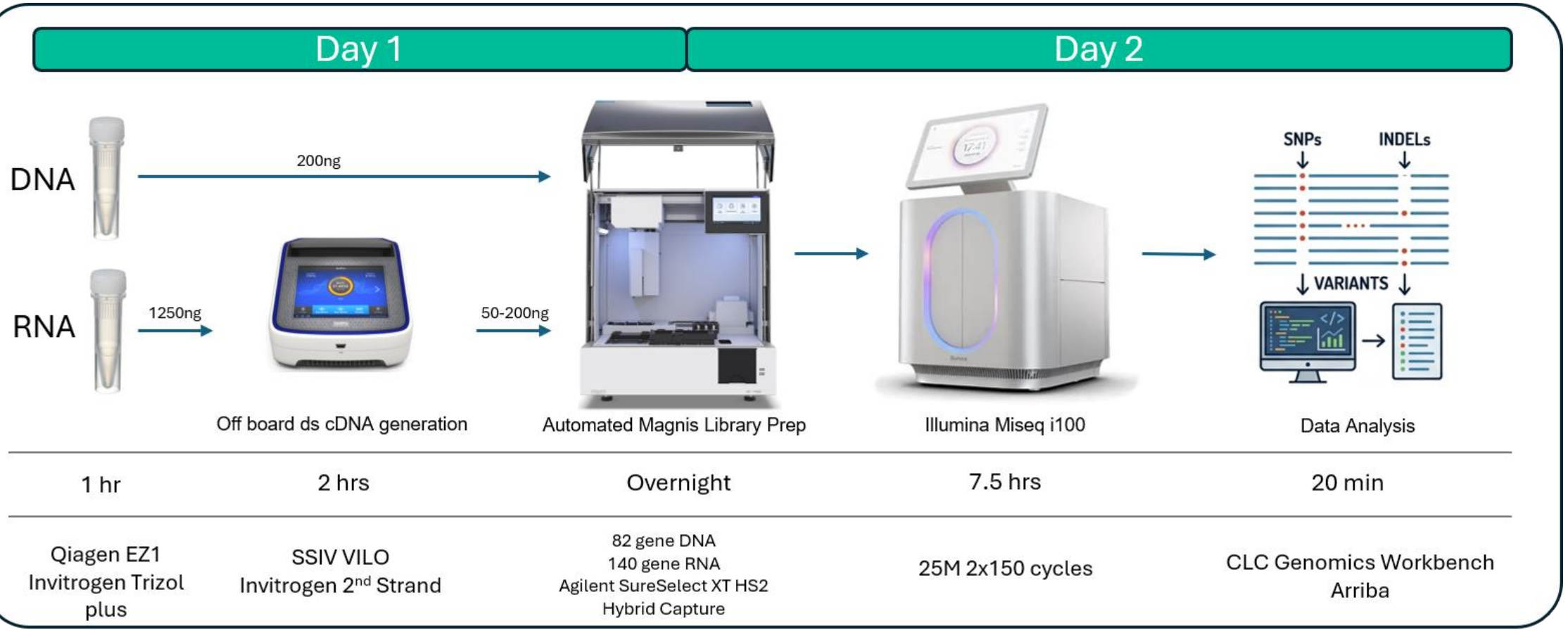
Magnis SureSelect assay



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HAEMATOLYMPHOID MALIGNANCY NGS PANEL

COMPREHENSIVE GENOMIC PROFILING FOR BLOOD CANCERS

82 DNA GENES + 19 RNA FUSION TARGETS
One comprehensive test

ACTIONABLE INSIGHTS
For diagnosis, prognosis and therapy selection

HIGH SENSITIVITY
Approximate detection to 2% VAF

BONE MARROW & BLOOD COMPATIBLE
Suitable for routine clinical use

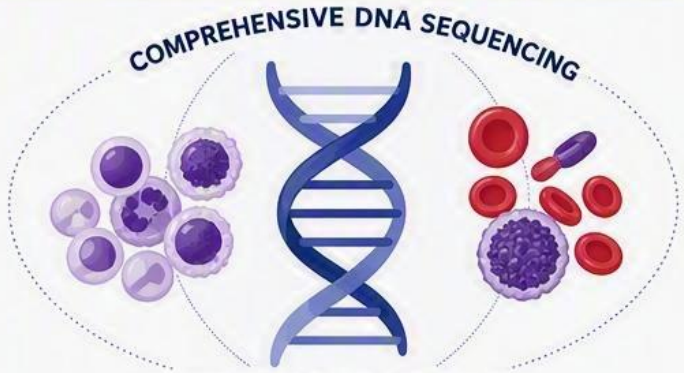
DISEASES COVERED

MYELOID NEOPLASMS

- Acute Myeloid Leukaemia (AML)
- Myelodysplastic Syndromes (MDS)
- Myeloproliferative Neoplasms (MPN)
- Chronic Myelomonocytic Leukaemia (CMML)
- Systemic Mastocytosis

LYMPHOID MALIGNANCIES

- Acute Lymphoblastic Leukaemia (ALL)
- Chronic Lymphocytic Leukaemia (CLL)
- B-cell Lymphomas
- T-cell Lymphomas
- Plasma Cell Neoplasms



DETECTING SOMATIC VARIANTS & GENE FUSIONS TO GUIDE PRECISION THERAPY

KEY DNA BIOMARKERS (Examples)

AML / MDS

- FLT3
- NPM1
- CEBPA
- TP53
- RUNX1
- ASXL1
- DNMT3A
- TET2
- IDH1 / IDH2

MPN

- JAK2
- CALR
- MPL
- CSF3R
- SH2B3

LYMPHOID

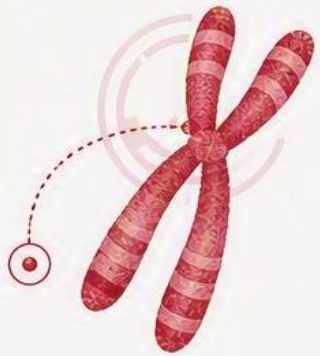
- MYD88
- NOTCH1
- SF3B1
- BTK
- PLCG2
- CARD11
- BCL2

RNA FUSION DETECTION

19 CLINICALLY RELEVANT FUSION TARGETS

- BCR::ABL1
- PML::RARA
- RUNX1 Rearrangements
- CBFB::MYH11
- KMT2A Rearrangements
- ETV6 Rearrangements
- MECOM Rearrangements
- JAK2 Fusions
- ZNF384 Rearrangements

... and more



ASSAY FEATURES

- Targeted sequencing of 82 DNA genes (SNVs & indels)
- RNA sequencing for 19 fusion genes
- FLT3-ITD and UBTF tandem duplication assessment
- Hybrid capture technology
- Illumina NGS platform
- High analytical specificity
- Rigorous bioinformatics and clinical interpretation

CLINICAL APPLICATIONS

- DIAGNOSIS & CLASSIFICATION
- RISK STRATIFICATION
- THERAPY SELECTION
- TARGETED TREATMENT ELIGIBILITY
- PROGNOSTIC ASSESSMENT
- DISEASE MONITORING SUPPORT

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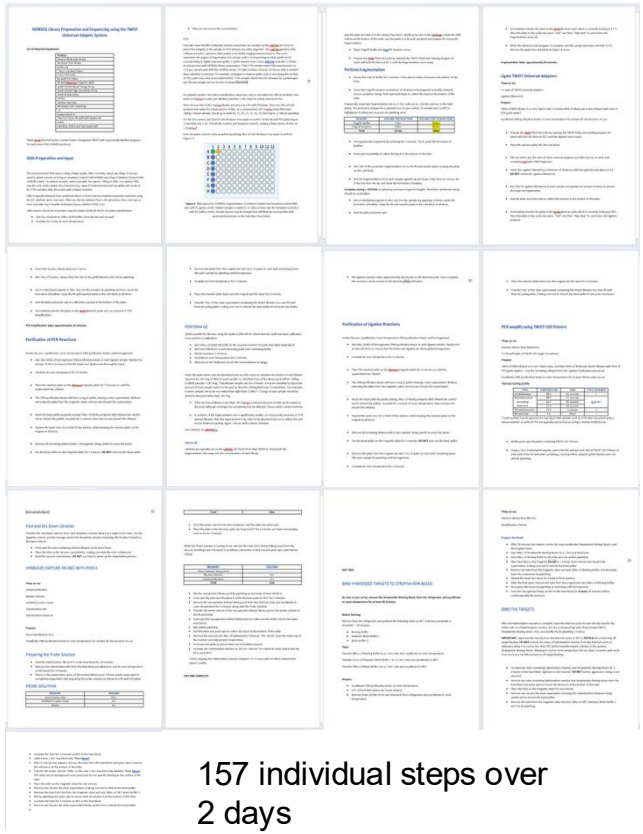
Key Changes Implemented

Referred

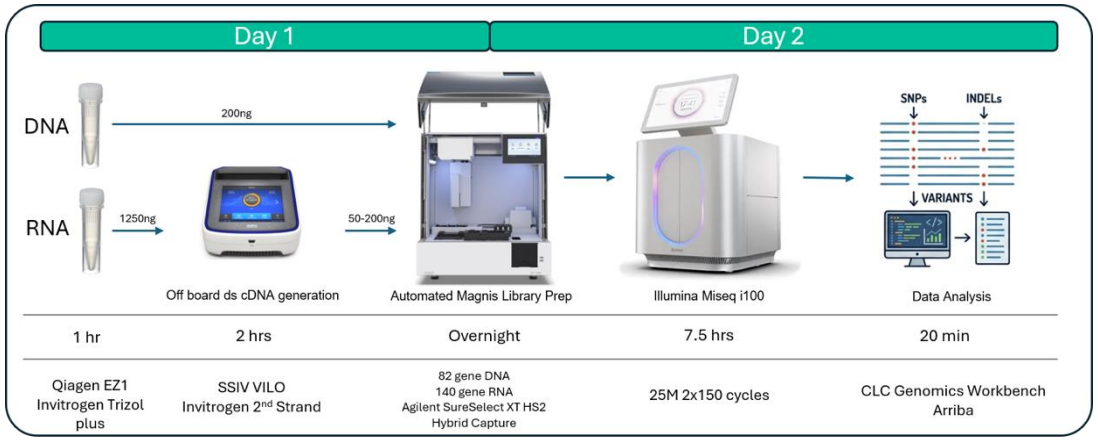
Manual Twist assay



157 individual steps over
2 days

1.5 days hands on
DNA only

Magnis Sureselect assay



0.5 – 2hrs hands on
DNA + RNA concurrently

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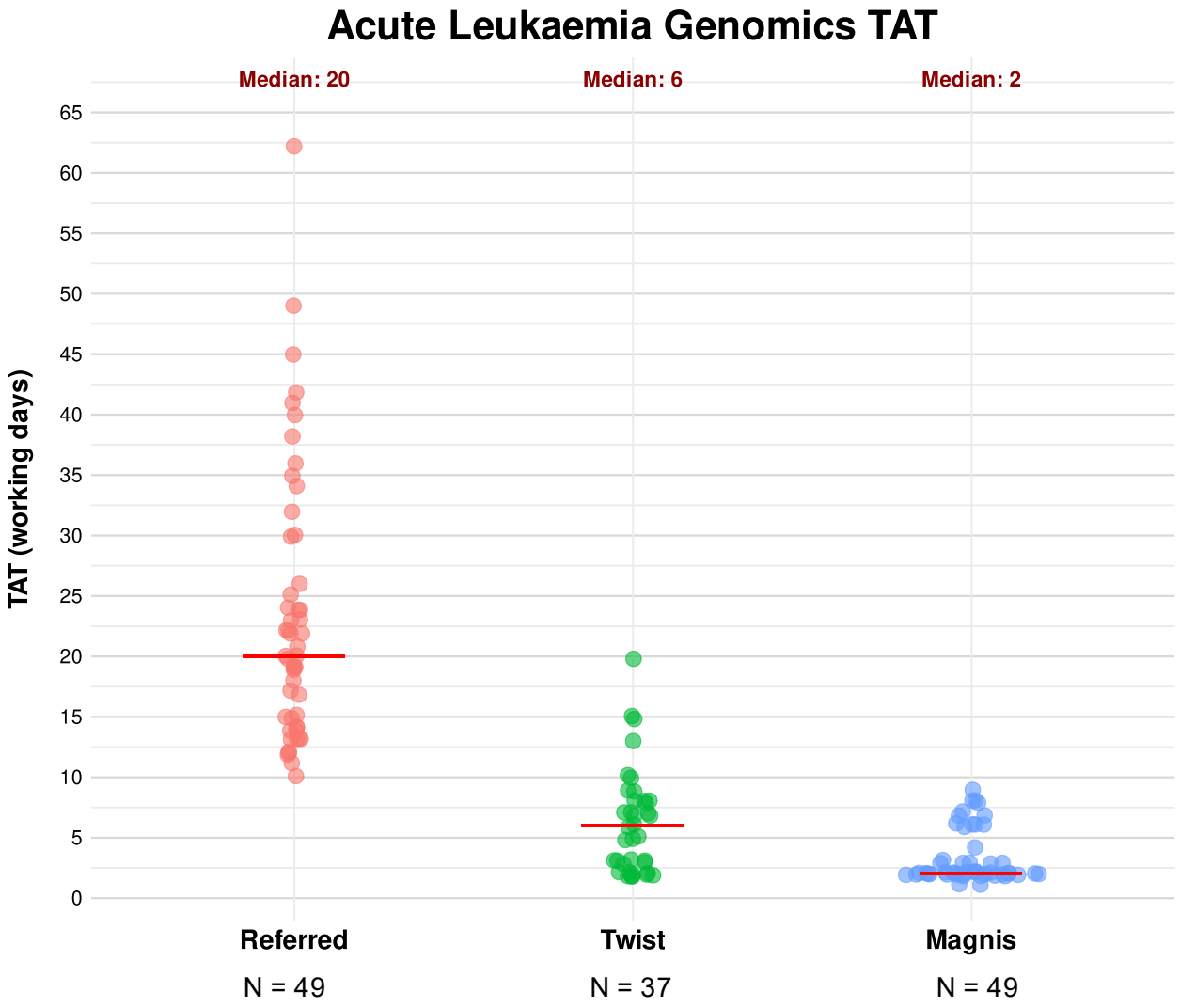
Outcomes so far



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Conclusion

- It is possible to offer complex molecular diagnostics for acute leukaemias in a small regional hospital and to do it cost effectively.
- Success relies on correct choice of equipment and a skilled/knowledgeable workforce willing to be flexible and push the envelope a little bit.

Rapid Integrated DNA/RNA Panel Testing for Acute Leukaemias: A Regional Centre Experience

THS Pathology

Problem: *Conventional molecular diagnostic workflows were too expensive and too slow for evolving needs in treating acute leukaemia. Patients were being disadvantaged.*

Solution: *Adopting the Magnis automated NGS platform and modifying the RNA assay to run concurrently with the DNA panel.*

Results: *Able to offer comprehensive molecular diagnostics in as little as 1 day. Reduced costs by avoiding most FISH and PCR testing. Reduced complexity for treating clinicians by reducing reports.*

Acknowledgements

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Thank you.

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