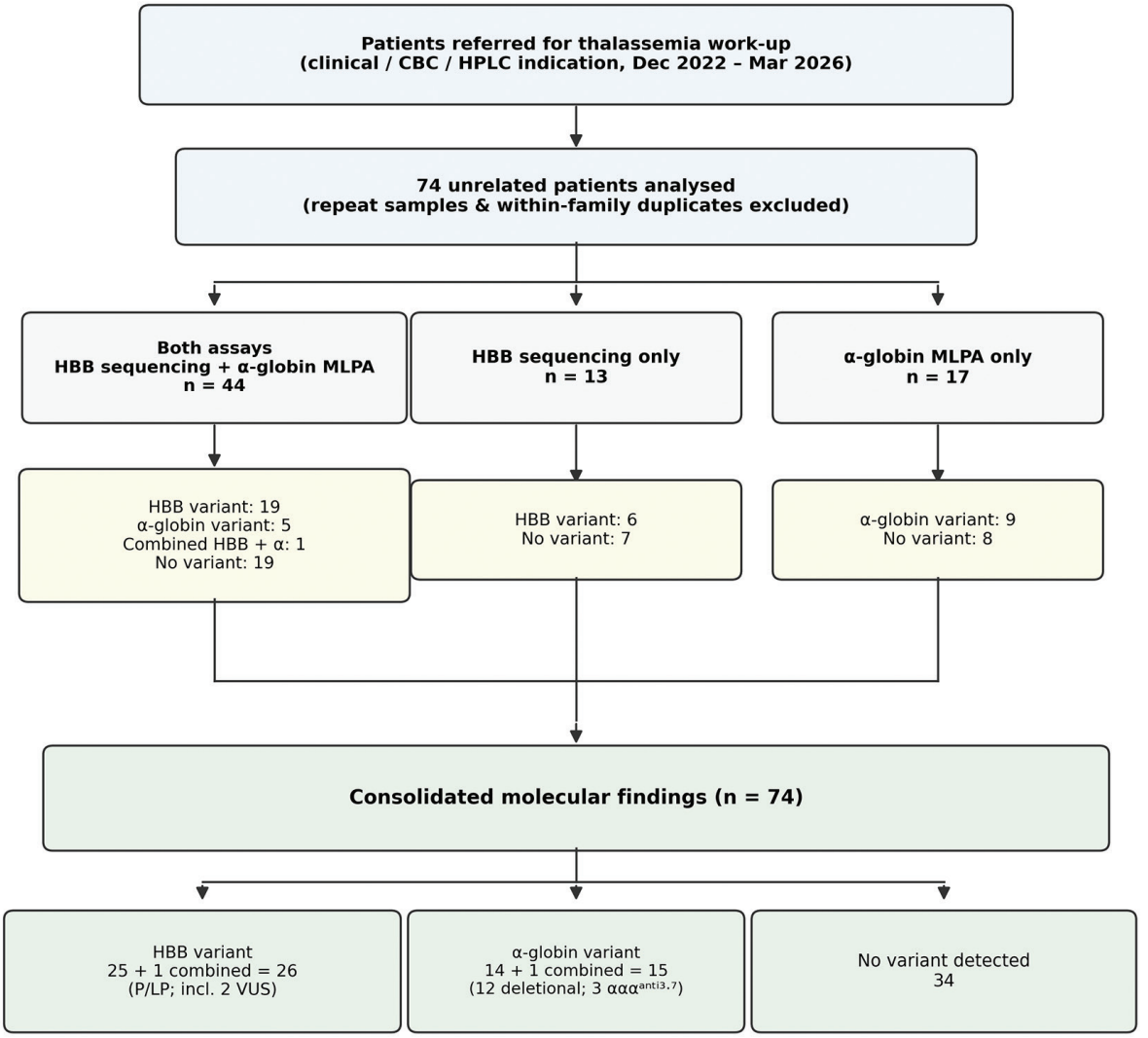


MLPA, multiplex ligation-dependent probe amplification; P/LP, pathogenic/likely pathogenic; VUS, variant of uncertain significance. The combined case carried an HBB codon 39 variant and a homozygous ααα$^{anti3.7}$ triplication and is counted once in each gene column. One variant-negative patient (P74) had a thalassemia-trait phenotype without a detectable variant on either assay.

Supplementary Figure 1. STARD-style flow of testing and molecular findings