Association Between Clonal Hematopoiesis of Indeterminate Potential and Brain β-Amyloid Deposition in Korean Patients With Cognitive Impairment

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Supplemental methods

CHIP variant calling strategy

Single-nucleotide variants and small insertion-deletion variants were retained based on the following criteria: Phred quality score ≥20, altered read numbers ≥10, variant allele frequency (VAF) 2%–30%, non-strand-biased variants (forward- and reverse-stranded alternative counts have a ratio between 3:7 and 7:3), coding region ±10 base pairs, and non-synonymous variants. Variants with a minor allele frequency ≥1% in the Genome Aggregation Database version 2.1 or the Korean Reference Genome Database were excluded. Variants included in the "Panel of Normals" using a sample from a healthy person were excluded [1]. To remove false-positive variants, single-nucleotide deletions or insertions within a homopolymer stretch and trinucleotide deletions or insertions within the trinucleotide repeat-rich region, which were repeatedly detected with a low VAF in our study cohort, were excluded. Finally, variants reported in ClinVar as benign or likely benign (last accessed on August 16, 2022) were filtered. The Catalogue of Somatic Mutations in Cancer version 94 was used to annotate the final variant list.

Reference

1. Caetano-Anolles D. Panel of Normals (PON). https://gatk.broadinstitute.org/hc/en-us/articles/360035890631-Panel-of-Normals-PON- (Updated on November 17, 2022).

Supplemental Data Table S1. Genes included in the 61-gene panel for detecting CHIP

ABL1	*ANKRD26*	*ASXL1*	*ATRX*	*BCOR*	*BCORL1*	*BRAF*	*CALR*
CBL	*CBLB*	*CBLC*	*CEBPA*	*CSF3R*	*DDX41*	*DNMT3A*	*ETV6*
EZH2	*FLT3*	*GATA1*	*GATA2*	*GNAS*	*GNB1*	*HRAS*	*IDH1*
IDH2	*IKZF1*	*JAK2*	*JAK3*	*KDM6A*	*KIT*	*KMT2A*	*KRAS*
MPL	*MYD88*	*NF1*	*NOTCH1*	*NPM1*	*NRAS*	*PDGFRA*	*PHF6*
PPM1D	*PRPF8*	*PTPN11*	*RAD21*	*RB1*	*RUNX1*	*SETBP1*	*SF3B1*
SH2B3	*SMC1A*	*SMC3*	*SRSF2*	*STAG1*	*STAG2*	*STAG3*	*STAT3*
TET2	*TP53*	*U2AF1*	*WT1*	*ZRSR2*			