Screening of histone modifier gene mutations in peripheral T-cell lymphomas, not otherwise specified (PTCL-NOS)



Tumor samples of 125 patients with PTCL-NOS



Targeted sequencing of histone modifier genes



91 somatic mutations identified in 48% (60/125) of patients with PTCL-NOS

| Types of mutations | Missense mutations Nonsense mutations Frameshift | n=72 n=10 n=9 | Single-n variation | | de C>T/G>A | |
|--------------------|--|---------------------|----------------------------------|-----------------------|--|-------------------------------|
| Categories | Gories Histone methylation genes Histone acetylation genes | | KMT2D SETD2 KMT2A KDM6A | → → → → | H3K4 methyltransferase H3K36 methyltransferase H3K4 methyltransferase H3K27 demethylase | 20.0% 4.8% 2.4% 0.8% |
| | | | EP300 CREBBP | → | H3K18 acetyltransferase H3K18 acetyltransferase | 8.0% 4.0% |
| | DNA methylation genes | | TET2 TET1 DNMT3A | 12.0% 3.2% 3.2% | | |
| | IV Chromatin remodeler genes | | ARID1B ARID2 | | 4% 1.6% | |

Histone modifier gene mutations, particularly those involved in histone methylation and acetylation, are significantly associated with tumor chemoresistance and disease progression of PTCL-NOS