Genetic and phenotypic characterization of indolent T-cell lymphoproliferative disorders of the GI tract

10 indolent T-cell lymphoproliferative disorders of the gastrointestinal tract

- CD4+ (n=4)
- CD8+ (n=4)
- CD4+/CD8+ (n=1)
- CD4-/CD8- (n=1)

Targeted next-generation sequencing
Comprehensive immunophenotypic analysis

80% had genetic alterations (recurrent mutations and novel rearrangements)

- CD4+ (n=4)
- CD4+/CD8+ (n=1)
- CD4-/CD8- (n=1)

- 82% Alterations of the JAK-STAT pathway genes
- 50% STAT3 mutations
- 17% SOCS1 deletion
- 17% STAT3-JAK2 rearrangement
- 67% Concomitant mutations in epigenetic modifier genes (TET2, DNMT3A, KMT2D)
- CD8+ (n=4)
- 50% Structural alterations involving the 3’ untranslated region of the IL2 gene

Immunophenotypic features

- CD4+ (n=4)
- CD4+/CD8+ (n=1)

Heterogeneous Th1 (T-bet+), Th2 (GATA3+) or hybrid Th1/Th2 (T-bet+/GATA3+) profiles

- Majority of CD8+ (n=4)
- CD4-/CD8- (n=1)

Type-2 polarized (GATA3+) effector T-cell (Tc2) phenotype

- CD8+ (n=4)
- 50% CD103 expression

Soderquist et al., Haematologica, 2020