

Supplementary Table 3: Characteristics of RCDII patients according to *DDX3X* and *TNFAIP3/TNIP3* status

	Total (N=50)	<i>DDX3X</i>			<i>TNFAIP3/TNIP3</i>		
		Mutated (N=10)	Not mutated (N=40)	Stat	Mutated (N=10)	Not mutated (N=35)	Stat
Clinical features							
Age, mean in years (range)	57 (29-77)	56 (29-71)	57 (30-77)	ns	53 (37-77)	59 (29-77)	ns
Gender (F, %)	32 (64%)	5 (50%)	27 (68%)	ns	6 (60%)	26 (74%)	ns
Time from CeD diagnosis to molecular evaluation, mean in years (range)	5.4 (0.1-21.9)	5.8 (2.7-10.9)	5.3 (0.1-21.9)	ns	5.3 (0.2-21.7)	5.5 (0.1-21.9)	ns
Develops EATL (%)	16 (32%)	2 (22%)	14 (35%)	ns	3 (27%)	9 (26%)	ns
Biological features							
Flow cytometry (N=), small bowel biopsies	33	7	26		8	24	
% aberrant population in IEL (/CD45 ⁺)	72% (23-98)	79% (40-95)	70% (23-98)	ns	84% (40-98)	70% (23-98)	ns
% aberrant population in LPL (/CD45 ⁺)	32% (5-70)	36% (8-70)	31% (5-94)	ns	49% (8-94)	28% (5-70)	p=0.02
% aberrant population in PBL (/CD45 ^{hi})	7% (0-80)	6% (0-11)	7% (0-80)	ns	19% (0-80)	2% (0-16)	p=0.006
TCR status (frozen biopsies)							
Gamma clonal	48/50 (96%) [#]	10/10 (100%)	38/40 (95%) [#]	ns	10/10 (100%)	33/35 (94%) [#]	ns
VDJ Beta clonal	15/49 (31%)	3/10 (30%)	12/39 (31%)	ns	0/9 (0%)	15/35 (43%)	p=0.02
Mutational status (frozen biopsies)							
<i>JAK1/STAT3</i> mutations	38 (74%)	7 (70%)	31 (78%)	ns	9 (90%)	26 (74%)	ns
<i>JAK1</i>	24 (48%)	4 (40%)	20 (50%)	ns	4 (40%)	17 (49%)	ns
<i>STAT3</i>	19 (38%)	4 (40%)	15 (38%)	ns	7 (70%)	12 (34%)	ns
<i>TNFAIP3/TNIP3</i> mutations	10/47 (21%)	2/9 (22%)	8/38 (21%)	ns	-	-	-
<i>DDX3X</i> mutations	10/50 (20%)	-	-	-	2 (20%)	7 (20%)	ns

[#] two cases with polyclonal TCR γ and clonal for TCR δ