

**Appendix 1: Genotype frequencies in controls and cases (persistent islet autoimmunity including T1D)**

SNP	Genotype	DR3/DR4 heterozygotes			Other HLA genotypes		
		Controls	Cases	OR (95% CI)	Controls	Cases	OR (95% CI)
CTLA4-318	CT/TT	12.0	22.5	2.13 (0.77-5.89)	18.7	17.7	0.94 (0.42-2.07)
CTLA4 ex.1	AG/GG	61.3	62.5	1.05 (0.48-2.32)	65.9	61.3	0.82 (0.44-1.54)
INS HphI	AT/TT	54.7	32.5	<b>0.40 (0.18-0.89)</b>	45.5	43.5	0.92 (0.50-1.71)
IL4R -3223	CT/TT	48.0	42.5	0.80 (0.37-1.73)	44.7	62.9	<b>2.10 (1.12-3.92)</b>
IL4R -1914	CT/TT	64.0	72.5	1.48 (0.64-3.43)	71.5	56.5	<b>0.52 (0.27-0.97)</b>
IL4R 50	AG/GG	73.3	65.0	0.68 (0.29-1.54)	67.5	82.3	<b>2.23 (1.05-4.74)</b>
IL4R 142	CT/TT	17.3	15.0	0.84 (0.29-2.41)	21.1	19.4	0.89 (0.42-1.92)
IL4R 375	AC/CC	22.7	22.5	0.99 (0.40-2.48)	21.1	27.4	1.41 (0.69-2.86)
IL4R 389	GT/TT	22.7	22.5	0.99 (0.40-2.48)	21.1	27.4	1.41 (0.69-2.86)
IL4R 406	TC/CC	21.3	22.5	1.07 (0.42-2.70)	21.1	24.2	1.19 (0.58-2.46)
IL4R 478	TC/CC	30.7	32.5	1.09 (0.48-2.48)	32.5	35.5	1.14 (0.60-2.17)
IL4R 551	AG/GG	34.7	40.0	1.25 (0.57-2.77)	43.9	45.2	1.05 (0.57-1.94)
IL4R 761	TC/CC	1.3	2.5	N/A	1.6	0.0	N/A
IL4 -524	CT/TT	37.3	22.5	0.49 (0.20-1.17)	30.1	40.3	1.57 (0.83-2.97)
IL13 -1512	AC/CC	29.3	40.0	1.61 (0.72-3.59)	30.9	32.3	1.06 (0.55-2.05)
IL13 -1112	CT/TT	33.3	40.0	1.33 (0.60-2.95)	31.7	32.3	1.02 (0.53-1.97)
IL13 intr.3	CT/TT	42.7	35.0	0.72 (0.33-1.60)	35.8	33.9	0.92 (0.48-1.75)
IL13 110	GA/AA	41.3	35.0	0.76 (0.34-1.69)	35.0	32.3	0.89 (0.46-1.69)

Genotype frequencies are shown after stratification for HLA-DR3/4 status. Since several cells had values < 5 in the homozygote variant, both heterozygotes and homozygotes variant have been combined for analysis. ORs refer to the combined variant group. Statistically significant OR values (P < 0.05) are indicated in bold.

**Appendix 2: Pairwise linkage disequilibrium between IL4 and IL13 SNPs**

	<b>IL4 -524</b>	<b>IL13 -1512</b>	<b>IL13 -1112</b>	<b>IL13 int3</b>	<b>IL13 110</b>
<b>IL4 -524</b>		0.15*	0.20*	0.53**	0.43**
<b>IL13 -1512</b>	0.02		0.96**	0.57**	0.58**
<b>IL13 -1112</b>	0.03	0.13		0.48**	0.47**
<b>IL13 int3</b>	0.07	0.07	0.07		1.00**
<b>IL13 110</b>	0.06	0.08	0.07	0.16	

Upper right triangle: absolute D' (standardized LD,  $D' = D/D_{max}$ )

Lower left triangle: D. Values refer to the reference allele.

Statistically significant LD values are indicated with \*  $P < 0.05$  and \*\*  $P < 0.0001$

**Appendix 3: Pairwise linkage disequilibrium between IL4R SNPs**

	<b>-3223</b>	<b>-1914</b>	<b>50</b>	<b>142</b>	<b>375</b>	<b>389</b>	<b>406</b>	<b>478</b>	<b>551</b>	<b>761</b>
<b>-3223</b>		0.04	0.90**	0.98**	0.29	0.29	0.27	0.48*	0.56*	0.45
<b>-1914</b>	- 0.005		0.09	0.04	0.12	0.12	0.15	0.16	0.24*	0.23
<b>50</b>	0.13	- 0.02		1.00**	0.12	0.12	0.10	0.35*	0.07	0.18
<b>142</b>	- 0.03	- 0.002	0.06		1.00*	1.00*	1.00*	0.40**	0.32*	0.33
<b>375</b>	- 0.01	0.01	0.01	- 0.01		1.00**	1.00**	0.98**	1.00**	1.00
<b>389</b>	- 0.01	0.01	0.01	- 0.01	0.10		1.00**	0.98**	1.00**	1.00
<b>406</b>	- 0.01	0.01	0.01	- 0.01	0.10	0.10		1.00**	1.00**	1.00
<b>478</b>	- 0.02	0.01	0.03	0.03	0.09	0.09	0.09		0.96**	0.86*
<b>551</b>	- 0.04	0.03	0.01	0.03	0.09	0.09	0.09	0.13		0.16
<b>761</b>	- 0.001	- 0.001	0.001	0.002	- 0.001	- 0.001	- 0.001	0.01	- 0.000	

Upper right triangle: absolute D' (standardized LD,  $D' = D/D_{max}$ )

Lower left triangle: D. Values refer to the reference allele.

Statistically significant LD values are indicated with \*  $P < 0.05$  and \*\*  $P < 0.0001$