

Supplementary material

Complement Activation in 22q11.2 Deletion Syndrome

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Table S1 Immunological profile of patients with 22q11.2del

Parameters	Age groups							
	2-5 mo	5-9 mo	15-24 mo	2-5 yr	5-10 yr	10-16 yr	Adults	
Lymphocyte subpopulations								
CD3 ⁺ ^b	22q11.2 del ^a	1.6	2.2	1.8	1.4	1.3	1.1	1.1
	Ref.range ^h	3.4-12.2	1.8-18.7	1.4-12.1	1.4-5.5	1.2-4.7	1.4-4.2	1.2-4.1
CD4 ⁺ ^b	22q11.2 del ^a	1.2	1.4	1.0	0.8	0.7	0.5	0.6
	Ref.range ^h	1.6-6.5	1.0-7.2	0.4-7.2	0.5-2.7	0.4-2.5	0.4-2.1	0.5-2.0
CD8 ⁺ T ^b	22q11.2 del ^a	0.3	0.7	0.5	0.5	0.4	0.5	0.4
	Ref.range ^h	0.3-3.4	0.2-5.4	0.2-2.8	0.2-1.8	0.2-1.7	0.3-1.3	0.2-1.2
CD19 ⁺ ^b	22q11.2 del ^a	1.13	1.45	2.18	1.05	0.45	0.31	0.19

	Ref.range ^h	0.52-2.3	0.13-6.3	0.16-3.7	0.18-1.3	0.10-0.80	0.12-0.74	0.064-0.82
CD16+/CD56+/CD3- NK ^b	22q11.2 del ^a	0.976	0.775	0.412	0.410	0.040	0.363	0.249
	Ref.range ^h	0.097-1.99	0.068-3.9	0.055-4.0	0.061-0.51	0.070-0.59	0.092-1.2	0.10-1.2
CD3+/CD4-/CD8-/TCR alfa/beta + double negative ^c	22q11.2 del ^a	1.4	0.6	1.4	1.4	1.5	1.2	0.5
	Ref.range ^h	0.18-3	0.39-4	0.55-4	1-7	0.77-6	0.54-6	0.57-5
CD4+/CD45RO+ memory ^d	22q11.2 del ^a	25	29	33	38	45	63	50
	Ref.range ^h	2-51	2-59	0.09-40	15-56	0.035-100	13-76	18-95
CD4+/CD45RO+/CXCR5+ follicular- like ^d	22q11.2 del ^a	2	3	8	10	11	11	11
	Ref.range ^h	7-33	9-47	8-51	6-72	7-85	7-47	5-56
CD4+/CD45RA+ naïve ^d	22q11.2 del ^a	76	75	75	71	66	51	48
	Ref.range ^h	73 – 100	77 – 97	56 – 100	52 – 92	46 – 99	37-97	16-100
CD4+/CD45RA+/CD31+ recent thymic	22q11.2 del ^a	69	74	68	78	73	71	65

emigrants ^e	Ref.range ^h	64-94	65-90	40-100	37-100	41-81	31-81	7-100
CD4+/CD45RO+/CD127low/CD25+	22q11.2 del ^a	6	4	7	6	6	6	5
regulatory ^d	Ref.range ^h	6-11	4-18	6-13	3-17	4-14	4-20	4-17
CD8+/CD27+/CD28+ naïve ^f	22q11.2 del ^a	95	43	84	72	66	54	58
	Ref.range ^h	47-100	31-100	10-100	19-100	16-100	20-95	6-100
CD8+/CD27+/CD28+ early	22q11.2 del ^a	5	15	12	19	12	9	10
effector/memory ^f	Ref.range ^h	0.08-24	0.1-13	1-8	1-9	1-6	0.42-18	1-20
CD8+/CD27-/CD28- late	22q11.2 del ^a	0.20	16	0.48	4	15	35	25
effector/memory ^f	Ref.range ^h	0.47-70	1-100	2-100	10-55	5-100	4-100	14-98

mo, month; yr, years

^a Values are expressed as median

^b Absolute counts ($\times 10^9/L$)

^c % of CD3+ T lymphocytes

^d % of CD4+ T lymphocytes

^e % of CD4+/CD45RA+ T lymphocytes

^f % of CD8+ T lymphocytes

^g g/L

^h Reference range as reported by Schatorjé et al [1]

Table S2 Correlations coefficients between C3bc, TCC and lymphocyte subpopulations in 22q11.2 deletion syndrome

Parameters	C3bc ^a		TCC ^a	
	r	p-value	r	p-value
Lymphocyte subpopulations (n=52)				
CD3+ T lymphocytes	-0.173	0.220	-0.059	0.677
CD4+ T lymphocytes	-0.184	0.192	-0.086	0.546
CD8+ T lymphocytes	-0.086	0.543	0.046	0.747
CD19+ B lymphocytes	0.029	0.838	-0.020	0.890
CD16+/CD56+/CD3- NK cells	0.009	0.950	-0.015	0.914
T lymphocytes subpopulations (n=39)				
CD3+/CD4-/CD8-/TCR alfa/beta + double negative	0.111	0.500	-0.098	0.553

CD4+/CD45RO+ memory	0.207	0.205	0.189	0.250
CD4+/CD45RO+/CXCR5+ follicular-like	0.146	0.374	-0.036	0.828
CD4+/CD45RA+ naïve	-0.350	0.029*	-0.071	0.668
CD4+/CD45RA+/CD31+ recent thymic emigrants	-0.098	0.552	-0.017	0.918
CD4+/CD45RO+/CD127 ^{low} /CD25+ regulatory	-0.054	0.742	0.111	0.501
CD4+ naïve/memory ratio	-0.116	0.481	-0.094	0.569
CD8+/CD27+/CD28+ naïve	-0.087	0.601	-0.018	0.914
CD8+/CD27+/CD28+ early effector/memory	-0.243	0.141	-0.215	0.196
CD8+/CD27-/CD28- late effector/memory	0.180	0.280	0.105	0.530

^a Data are present as Spearman's rho correlation coefficients with corresponding p-values

* p < 0.05

References

1. Schatorje EJ, Gemen EF, Driessen GJ, Leuvenink J, van Hout RW, de Vries E. Paediatric reference values for the peripheral T cell compartment. *Scand J Immunol.* 2012;75(4):436-44. doi:10.1111/j.1365-3083.2012.02671.x.