

Table S1. Description of the children from groups E1, E2, E3 and C

No.	No. of patient	Age (years)	Sex	Presence of 22q11.2 microdeletion	CHM	Facial dysmorphism	Hypocalcemia	Thymic hypoplasia	Cleft palate	Reference
E1										
1	1	6.5	M	+	+	+	+	-	-	[21,22]
2	3	6.5	M	+	+	+	-	-	-	[21,22]
3	4	9	M	+	+	-	+	-	-	[21,22]
4	6	5.5	M	+	+	-	+	-	-	[21,22]
5	7	9	M	+	+	+	-	-	-	[21,22]
6	11	6.5	F	+	+	+	+	+	-	[21,22]
7	12	5.5	M	+	+	-	-	+	-	[21,22]
8	13	6	F	+	+	-	-	+	-	[21,22]
9	14	9.5	M	+	+	+	+	-	-	[21,22]
10	15	8	F	+	+	+	-	-	-	[18,22]
11	31	7.5	M	+	+	+	+	-	-	[18,22]
12	33	9.5	F	+	+	+	+	+	-	[21]
13	35	6	F	+	+	-	-	+	-	[22]
14	40	7.5	F	+	+	+	+	-	-	this study
15	60	6	F	+	+	+	-	-	-	this study
E2										
1	2	6	M	-	+	+	-	-	-	[21,22]
2	16	10	M	-	+	+	-	+	-	[21,22]
3	17	5.5	M	-	+	+	-	-	-	[21,22]
4	19	10	M	-	+	+	-	-	-	[21,22]
5	21	6	F	-	+	+	-	+	-	[21]
6	22	9	F	-	+	+	-	-	-	[21,22]
7	23	8	M	-	+	+	-	+	-	[22]
8	25	6	M	-	+	+	not available	+	-	[21]
9	26	6	M	-	+	+	-	-	-	[21,22]
10	27	7	M	-	+	+	+	-	-	[21]
11	28	6.5	M	-	+	+	-	-	-	[21]
12	32	5.5	F	-	+	+	+	-	-	[21]
13	41	9	F	-	+	+	-	-	-	[22]

