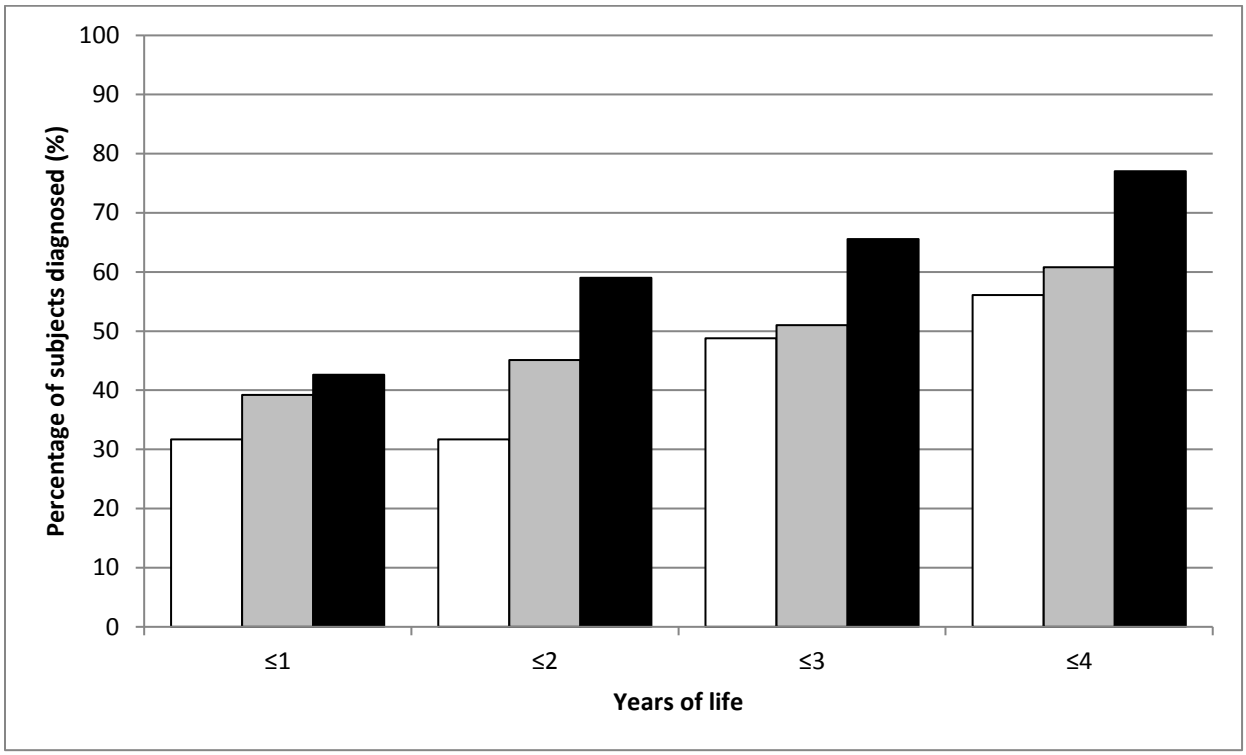
SUPPLMENTAL MATERIAL

Supplemental Figure 1. Cumulative percentage of subjects who were born in the molecular era (1994 and thereafter) and molecularly diagnosed with 22q11.2DS in the first 4 years of life



Bar graphs for the Canadian (n=41) and US (n=51) samples born 1994 to 1997 inclusive (group 3) are shown in white and in grey, respectively and for the pediatric US sample (n=61, group 4) born 1998 and thereafter, shown in black.

Supplemental Table 1. Medical/surgical specialty and allied health areas involved prior to molecular diagnosis in a subset of 100 Canadian patients with 22q11.2DS

Medical/surgical[a]	n	%	Allied health / other	n	%	Molecular testing ordered by	n	%
Genetics[b]	77	77%	Speech language pathology	44	44%	Genetic specialists	62	62%
Cardiology	70	70%	Psychology	29	29%	Cardiologists	15	15%
ENT/Otolaryngology[c]	45	45%	Dentistry[g]	24	24%			
Neurology	39	39%	Social services[h]	21	21%	Neurologist	1	1%
General surgery	33	33%	Audiology	19	19%	Immunologists	2	2%
Orthopedics	32	32%	Dietetics/nutrition	9	9%			
Psychiatry	27	27%	Physiotherapy/Occupational therapy	8	8%	Psychiatrists[i]	6	6%
Ophthalmology	22	22%						
Gastroenterology	16	16%						
Obstetrics and gynecology[d]	14	14%						
Plastic surgery[e]	13	13%				Plastic Surgeons	2	2%
Endocrinology	13	13%						
Dermatology	11	11%						
Immunology	11	11%						
Respirology	8	8%						
Hematology	7	7%						
Urology	7	7%						
Nephrology	5	5%						
Infectious disease[f]	2	2%						
						Pediatricians/family doctors	12	12%

[a](See text for details of Methods). Median of 5.5 (range 2-11) medical and a median of one (range 0-5) allied health who provided primary care.
[b]Including consults that may have involved other testing (i.e. karyotype, fragile X cytogenetic test, screening for inborn errors of metabolism and/or other genetic tests).
[c]Including ear nose and throat (ENT)/otolaryngology consults and myringotomies, tympanoplasties, otoplasties.
[d]Including consults for pregnancy, amenorrhea and endocervical cysts.
[e]Including cleft palate team surgical consultations, craniofacial surgeons, plastic surgeons performing palatal repairs, and rhinoplasty.
[f]Including consults for post-surgical and other infections (e.g., septicemia, bacterial endocarditis).
[g]Including special consults for very poor dentition, dental restorations and surgeries.
[h]Including social workers, family therapists and corrections officer.
[i]Limited to those outside of 22q11.2DS clinics.

Supplemental Table 2. Demographic and clinical factors, including number of specialty areas seen prior to molecular diagnosis, and their association with time to molecular diagnosis of 22q11.2 deletion syndrome (22q11.2DS) in a subset (n=100) of a Canadian cohort of adults with 22q11.2DS

Demographic and clinical factors	**n**	**%**		**Linear regression analysis[a]**			
				B	**95% CI**		***p***
Male sex	52	52.0		0.98	-0.94	2.91	0.3123
European ethnicity	72	72.0		-2.04	-4.28	0.20	0.0744
Born in era of molecular testing (Group 3)	21	21.0		-0.22	-1.81	1.37	0.7852
Palatal anomaly	45	45.0		-4.24	-6.25	-2.23	**<0.0001**
Cardiac anomaly	64	64.0		-1.64	-3.62	0.34	0.1036
Developmental delay/intellectual disability	56	56.0		-0.82	-2.79	1.16	0.4133
	Median	**Range**					
Number of specialty areas seen prior to molecular diagnosis of 22q11.2 deletion	7	2	15	0.46	0.12	0.80	**0.0088**

The regression model was significant for time to molecular diagnosis of 22q11.2DS: Adjusted $R^2 = 0.22$, df=7, $p < 0.0001$.
See text of manuscript and footnote to Table 1 for details about the regression model and the factors used.