



# Population prevalence of Down's syndrome and cardiac complications in South Korea: Based on National Health Insurance Service (NHIS)

Na Yeong Lee, Won Kyoung Cho, Moonbae Ahn, Min Ho Jung, and Byung-Kyu Suh

Department of Pediatrics, College of Medicine, The Catholic University of Korea, Seoul, Republic of Korea

**Background:** There is a no reliable population-based prevalence data of Down's syndrome (DS) in South Korea. In the present study, we try to estimate the incidence and prevalence of DS and cardiac complications in South Korea using data of National Health Insurance Service (NHIS) data and Rare Diseases Registry.

**Methods:** We collected the data on DS patients who registered in the Rare Diseases Registry (RDR) between 2010 and December 2015. During this period, the total number of registered DS cases and the number of new registrations each year were identified. To estimate the prevalence of DS, the size of the Korean population in 2015 was ascertained from resident registration data with respect to population data gathered by the Korean Ministry of Security and Public.

## RESULTS

The prevalence of Down syndrome (per 100,000 was 2.75 in 2010, 2.90 in 2011, 3.04 in 2012, 3.25 in 2013, 3.88 in 2014, 4.03 in 2015) has been increased for 5 years. In 2015, the number of DS patients was 2,077 out of the total population of 51,574,044 South Koreans. When population prevalence was divided by sex and 10-year age group, in children under the age of 10, prevalence was 28.4 per 100,000 in males and 23.4 per 100,000 in females. Prevalence then decreased rapidly to 8.67 (per 100,000) in teenagers, 3.09 (per 100,000) in those in their 20s, 0.98 (per 100,000) in the 30s, 0.55 (per 100,000) in the 40s, and 0.26 (per 100,000) in the 50s.

After adjusting for age and sex, Hazard ratios with 95% CI that DS patients had statistically significant higher incidence rate (per 1000) of ASD (13.61 vs. 0.15, HR=87.8; 95% CI, 35.6 - 216.6), VSD (6.38 vs. 0.06, HR=107.0; 95% CI, 25.9 - 441.8), PDA (5.10 vs. 0.03, HR=178.9; 95% CI, 24.5 - 1304.8) or AVSD (3.30 vs. 0.02, HR=137.2; 95% CI, 18.6 - 1009.6) compared with matched control subjects in this study. In patients with DS, death rate per 1000 (11.7 vs. 0.4, HR=29.0; 95% CI, 13.0 - 64.9) was statistically significantly higher than that in matched control subjects.

Table 1. Prevalence of Down's syndrome in South Korea

	Population	Total Number of cases	Prevalence per 100,000 persons	Age standardization prevalence	Population	Total Number of cases	Incidence per 100,000 persons	Age standardization incidence
2010	50166793	1378	2.7468	2.7067	50163858	214	0.4266	0.4203
2011	50445164	1461	2.8962	2.9398	50441931	184	0.3648	0.3673
2012	50763154	1542	3.0376	3.1547	50759654	201	0.396	0.4068
2013	51013675	1657	3.2481	3.4273	51009857	146	0.2862	0.2991
2014	51281917	1992	3.8844	4.1837	51277830	155	0.3023	0.3214
2015	51574044	2077	4.0272	4.4105	51569629	173	0.3355	0.3582

## CONCLUSION

DS, trisomy 21 is the most common birth defects and are more likely to accompany cardiac complications such as ASD, VSD, and PDA..

Table 2. Population prevalence of Down's syndrome in South Korea according to age and sex at 2015

	Total Population	Total Number of cases	Prevalence Per 100,000 persons	Male Population	Total Number of cases	Prevalence Per 100,000 persons	Female Population	Total Number of cases	Prevalence Per 100,000 persons
0-9	4605930	1196	25.96653	2370685	674	28.4306	2235245	522	23.35314
10-19	5980471	519	8.678246	3126473	275	8.795854	2853998	244	8.54941
20-29	6821889	211	3.092985	3602702	118	3.275319	3219187	93	2.888928
30-39	7945906	78	0.981638	4074241	48	1.178134	3871665	30	0.77486
40-49	8934467	49	0.548438	4535856	27	0.595257	4398611	22	0.500158
50-59	8210181	21	0.25578	4121285	15	0.363964	4088896	6	0.146739
60-69	4691566	1	0.021315	2276167	0	0	2415399	1	0.041401
70-79	3124667	2	0.064007	1327943	0	0	1796724	2	0.111314
80-89	1110631	0	0	345069	0	0	765562	0	0

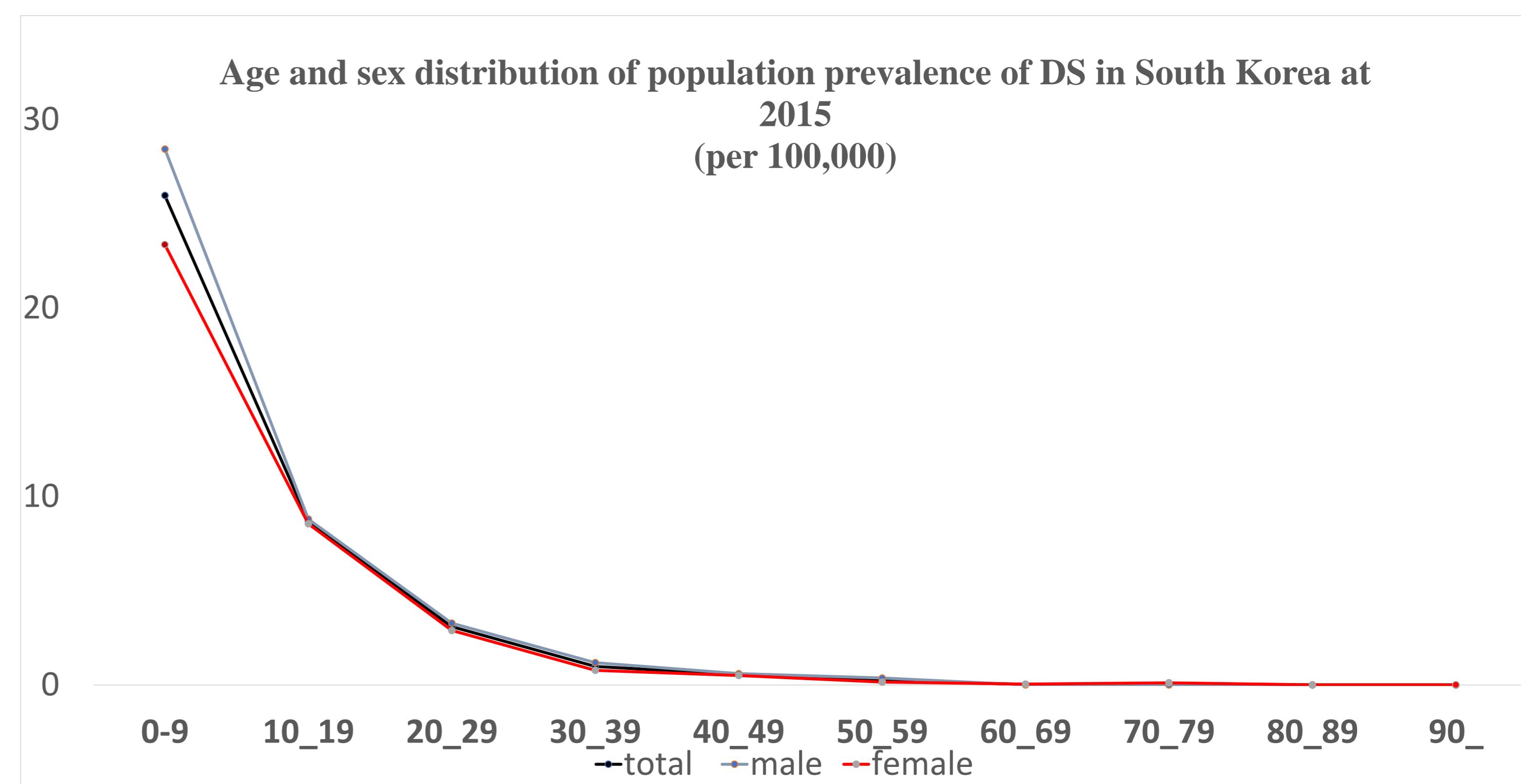


Figure 1. Population prevalence of Down's syndrome in South Korea according to age and sex at 2015

Table 3. Hazard ratios of congenital heart diseases and death in Down's syndrome in South Korea from 2010 to 2014 compared with matched controls adjusted for age, sex.

		N	event	Duration	IR (per 1000)	HR (95% CI)
ASD	Control	3855	4	14787.27	0.2705	1
	Case	771	58	2697.84	21.4986	80.1 (29.1-220.6)
VSD	Control	3865	3	14847.76	0.20205	1
	Case	773	27	2797.7	9.65077	46.3 (14.0-152.5)
PDA	Control	4000	1	15321.38	0.06527	1
	Case	800	26	2897.07	8.97458	135.3 (18.4-996.8)
PV_AS	Control	4470	1	17065.72	0.0586	1
	Case	894	1	3309.24	0.30218	5.1 (0.3-80.9)
TOF	Control	4410	0	16920.25	0	1
	Case	882	6	3265.3	1.83751	NC*
AVSD	Control	4265	0	16378.84	0	1
	Case	853	9	3152.61	2.85478	NC*
COA	Control	4490	0	17175.3	0	1
	Case	898	1	3326.34	0.30063	NC*
DORV	Control	4475	0	17118.93	0	1
	Case	895	2	3310.1	0.60421	NC*
Death	Control	4490	7	17168.63	0.4077	1
	Case	898	39	3329.49	11.7135	29.0 (13.0-64.9)

Abbreviations; ASD, Atrial Septal Defect; VSD, Ventricular septal defect, PDA, Patent ductus arteriosus; PV\_AS, PV atresia/stenosis; TOF, Tetralogy of Fallot; AVSD, Atrioventricular septal defect; COA, Coarctation of the Aorta; DORV, Double outlet right ventricle; NC, not calculated by zero counting.

