

People living with Down syndrome in the USA: BIRTHS AND POPULATION

Gert de Graaf, PhD,ⁱ Frank Buckley^{ii,iii} and Brian Skotko, MD, MPP^{iv,v}

(i) Dutch Down Syndrome Foundation, Meppel, The Netherlands; (ii) Down Syndrome Education International, Cumbria, UK; (iii) Down Syndrome Education USA, Irvine, California, USA; (iv) Division of Medical Genetics, Department of Pediatrics, Massachusetts General Hospital, Boston, Massachusetts, USA; (v) Harvard Medical School, Boston, Massachusetts, USA.

This fact sheet summarizes recently published and updated estimates of the numbers of babies born and people living with Down syndrome in the USA.^[1-3]

Births

- **How many babies are born with Down syndrome each year?** As of 2014, we estimate that 1 in every 768 liveborn babies has Down syndrome (13.0 per 10,000). This means that there were about 5,200 babies with Down syndrome born annually in the U.S. in recent years. (Figure 1)

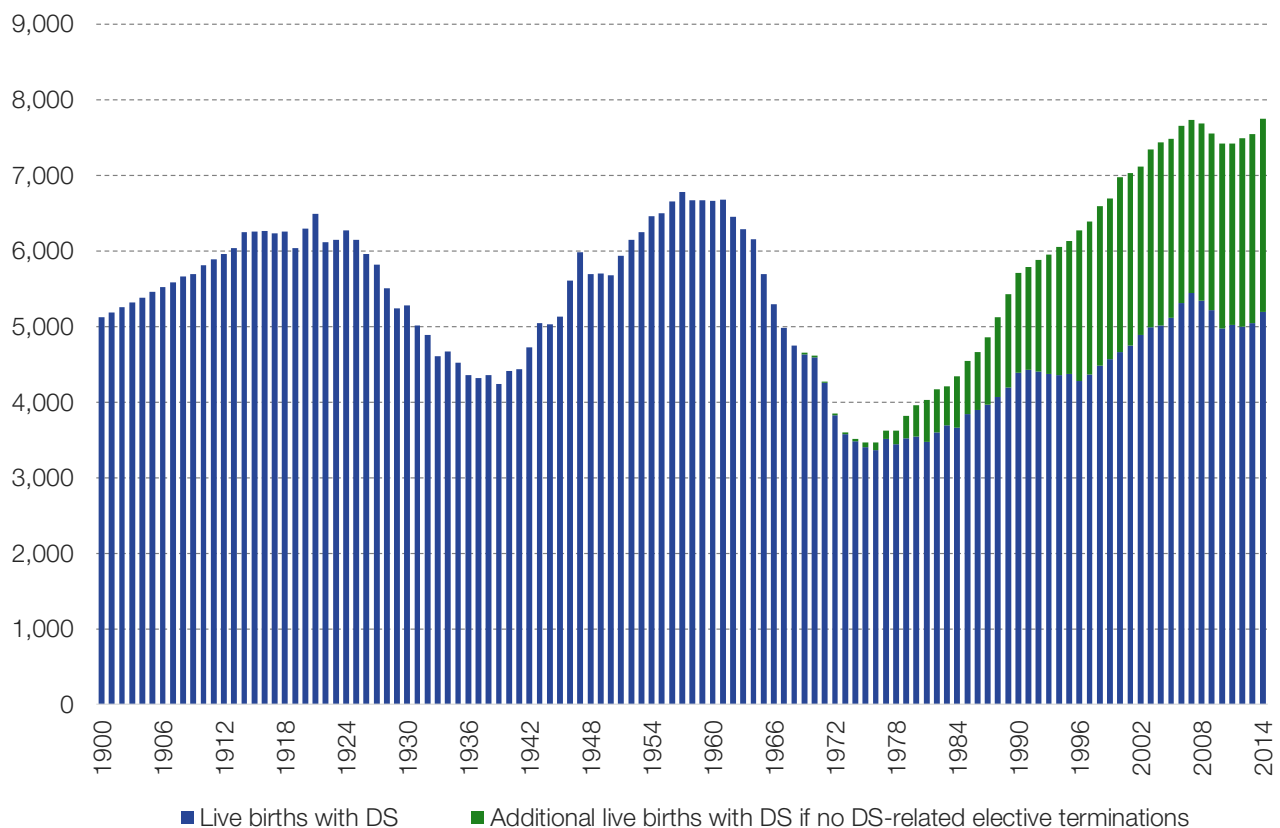


Figure 1. Births of babies with Down syndrome in the USA, 1900-2014^[d]

- **Are more pregnancies with Down syndrome being terminated than in the past?** In the few decades since prenatal screening was introduced, more pregnancies with Down syndrome have been diagnosed prenatally and terminated. However, not all children born with Down syndrome are

diagnosed prenatally, and many expectant parents do not choose screening. Therefore, reductions in live birth rates are influenced by the number of people choosing prenatal testing, the accuracy of the screening tests, and parents' decisions given a prenatal diagnosis. Approximately, 3,500 Down syndrome-related elective pregnancy terminations were performed as of 2014 in the U.S.

- Are most pregnancies with Down syndrome now terminated?** Previous studies have suggested that around 74% of expectant parents in the U.S. who learn of a Down syndrome diagnosis prenatally choose to terminate.^[4] However, many expectant couples choose not to pursue prenatal screening or diagnostic testing. Therefore, the 74% termination rate does not imply that 74% fewer babies were born; instead, it only reflects the decisions of couples who have already opted into screening. Actually, in the U.S., as a result of elective terminations, we estimate there was a 33% reduction in the numbers of babies with Down syndrome born in 2014. This means that in recent years there were 33% fewer babies with Down syndrome than could have been born, absent elective terminations. (Figure 1, green bars).
- What has happened to the overall birth rate?** Since the early 1980s, the effect of increasing maternal age has slightly outweighed the growth of prenatal screening followed by elective terminations, leading to an increase in the live birth prevalence of Down syndrome in the USA in recent decades - rising from around 10.1 per 10,000 livebirths (1 in 990) in the 1980s to around 12.3 per 10,000 livebirths in the 2000s (1 in 813). (Figure 2)

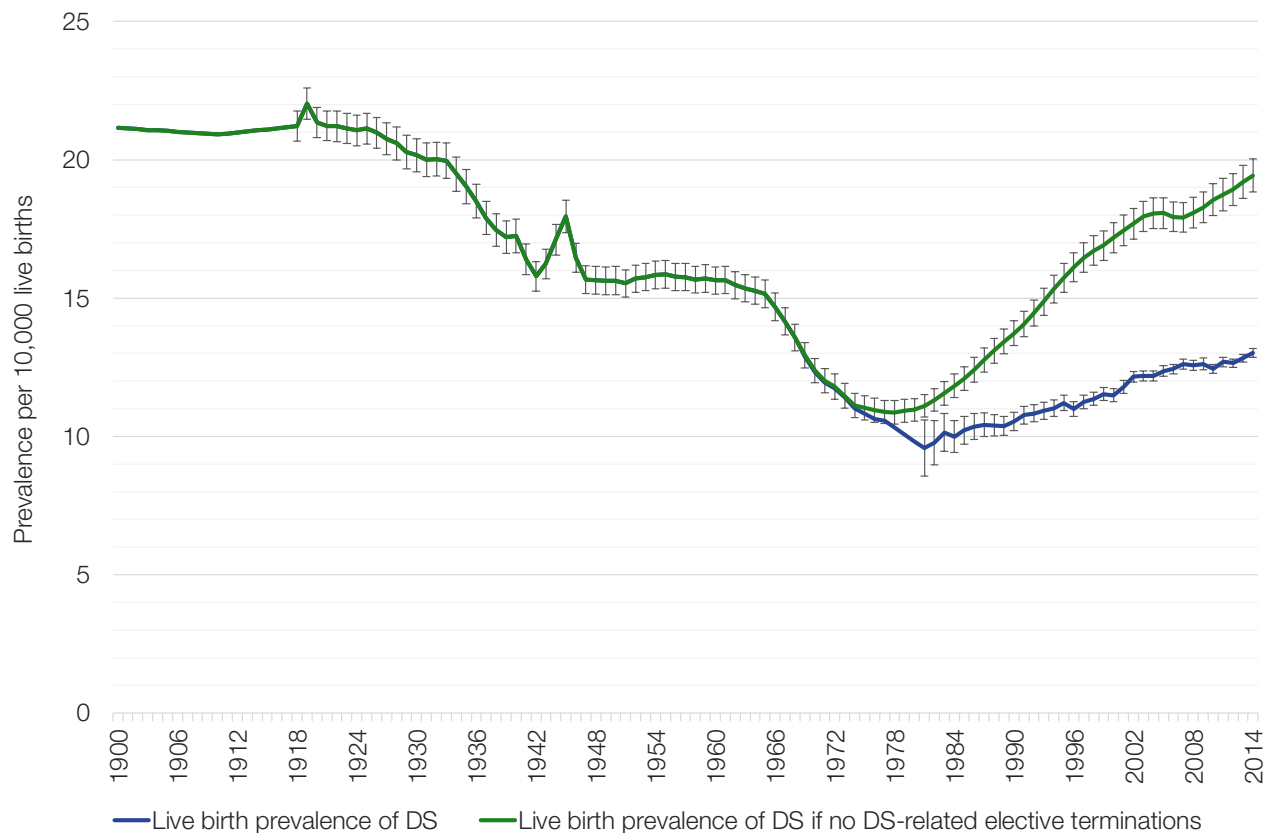


Figure 2. Live birth prevalence of Down syndrome in the USA, 1900-2014

- Are similar numbers of babies with Down syndrome born in all regions and all communities?** Previous research suggests that Down syndrome naturally occurs in all races and

ethnicities, and that only maternal age differences influence the number of births. Our research adds that there are cultural differences between regions of the US in regards to Down syndrome-related terminations. As of 2007, the reduction in babies born with Down syndrome was highest in the Northeast region and Hawaii. They were lowest in the South. There also appears to be racial/ethnic differences. From 2005-2009, the reduction of babies born with Down syndrome was highest among Asians/Pacific Islanders followed by non-Hispanic whites. The reduction was lowest among Hispanics and American Indians. However, higher reduction percentages tend to co-occur with higher maternal ages (and therefore with more pregnancies with a child with Down syndrome). As a result, the actual differences between regions and between ethnic groups in live birth prevalence are relatively small. There were 32 U.S. states that had sufficient publicly available data to estimate the birth prevalences of Down syndrome (Table 1).

- **How are newer non-invasive screening technologies influencing birth rates?** Noninvasive prenatal screens (NIPS) were introduced in October of 2011 in the United States. No significant changes in birth rates occurred in 2012, 2013, or in 2014, the latest year of available data. The surveillance program in the U.S. reports 5-year running averages; so data from 2014 represents the average from 2012-2016. New data are released around November of each year.
- **What percentage of children with Down syndrome are born to women older than 35 in the U.S.?** Without elective terminations, the percentage of mothers, 35 years of age or older, who had children with Down syndrome would have increased from 26% in 1980 to 57% in 2014. As a result of elective terminations, however, the actual percentage of mothers, 35 years of age or older, who had children with Down syndrome changed from around 18% in 1980, to 33% in 1993, to 42% in 1997, and to 47% around 2014 (Figure 3).

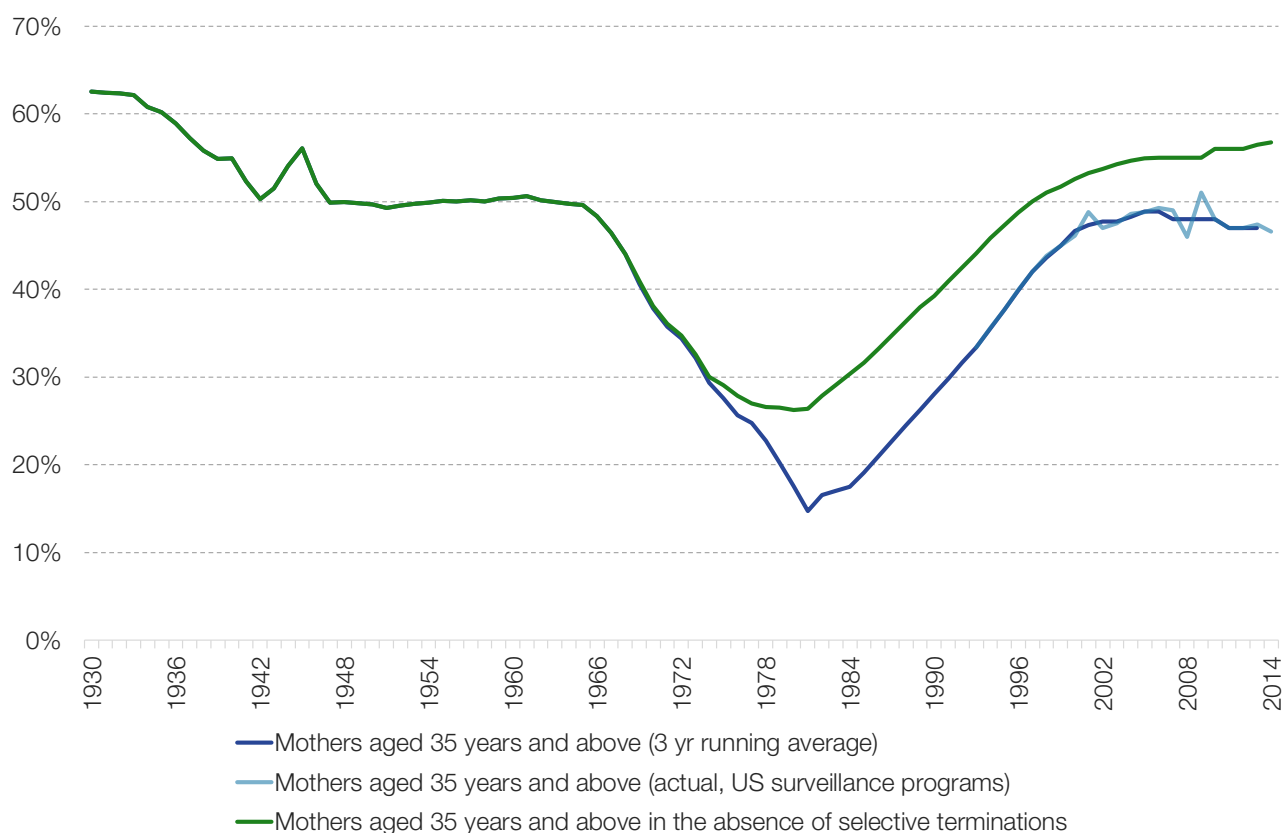


Figure 3. Percentage of mothers of children with Down syndrome aged 35 years or over in year of child’s birth in the USA, 1930-2014

Population

- **How many people with Down syndrome are living in the U.S. today?** Including people born outside of the U.S., we estimate that the number of people with Down syndrome living in the U.S. has grown from 49,923 in 1950 to 213,841 in 2014 (Figure 4).

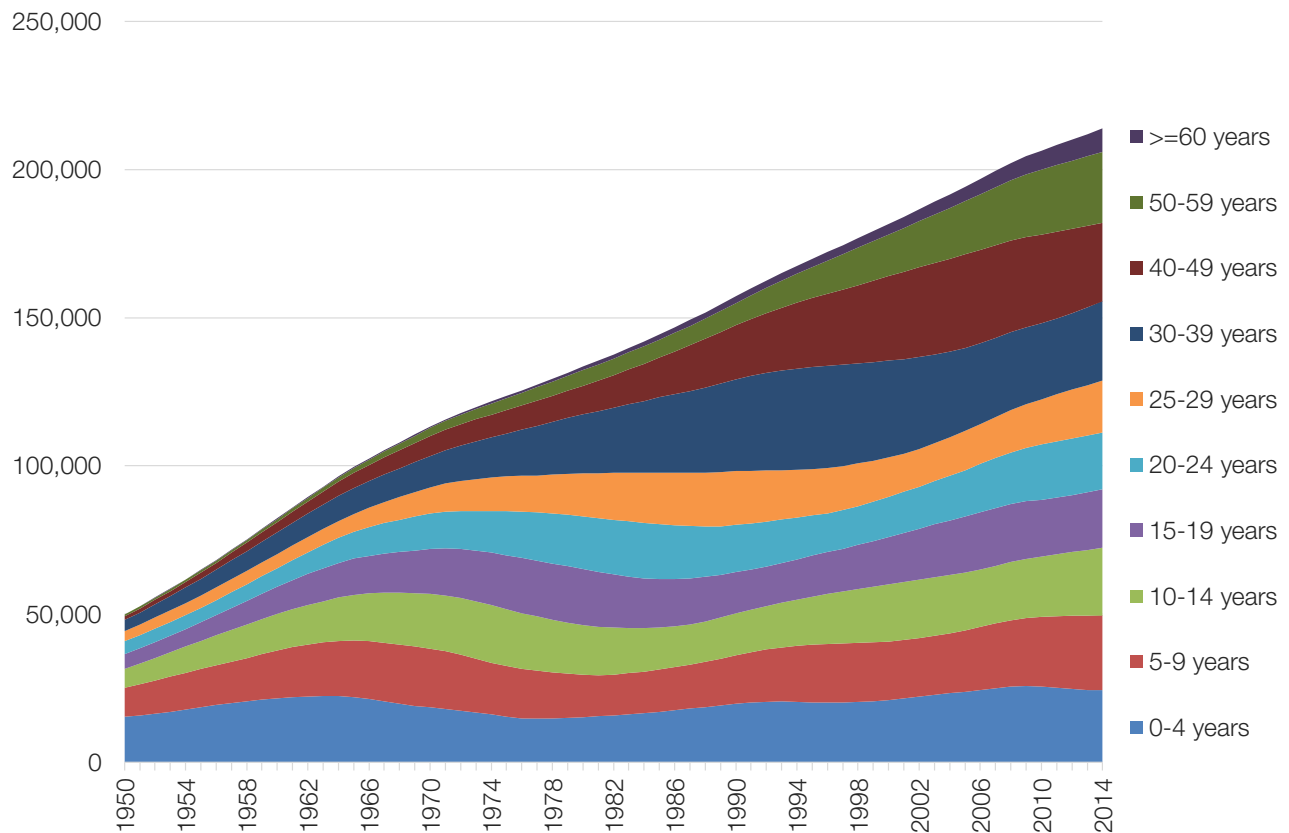


Figure 4. Population of people with Down syndrome in the USA, 1950-2014

- **What proportion of the U.S. population are people with Down syndrome?** The population prevalence of Down syndrome, as of 2014, is estimated at 6.7 per 10,000 inhabitants (or 1 in 1,490).
- **What is the population of people with Down syndrome in individual U.S. states?** Nine U.S. states had sufficient publicly available data, for a substantial number of years, to estimate the populations of people with Down syndrome (Figure 5).^[3]
- **Is Down syndrome a “rare disease”?** Our estimates indicate that until 2008, Down syndrome was a rare disease, typically defined in the U.S. as a population of fewer than 200,000 persons.
- **What is the ethnic composition of today’s population of people with Down syndrome?** We estimated the population of people with Down syndrome living in the U.S. in 2010 to include 138,019 non-Hispanic whites, 27,141 non-Hispanic blacks, 32,933 Hispanics, 6,747 Asians/Pacific Islanders, and 1,527 American Indians/American Natives.
- **Are the populations of people with Down syndrome growing at similar rates in all ethnic groups?** In more recent decades, the population growth of people with Down syndrome has leveled off for non-Hispanic whites, a consequence of elective terminations. The growth in the population

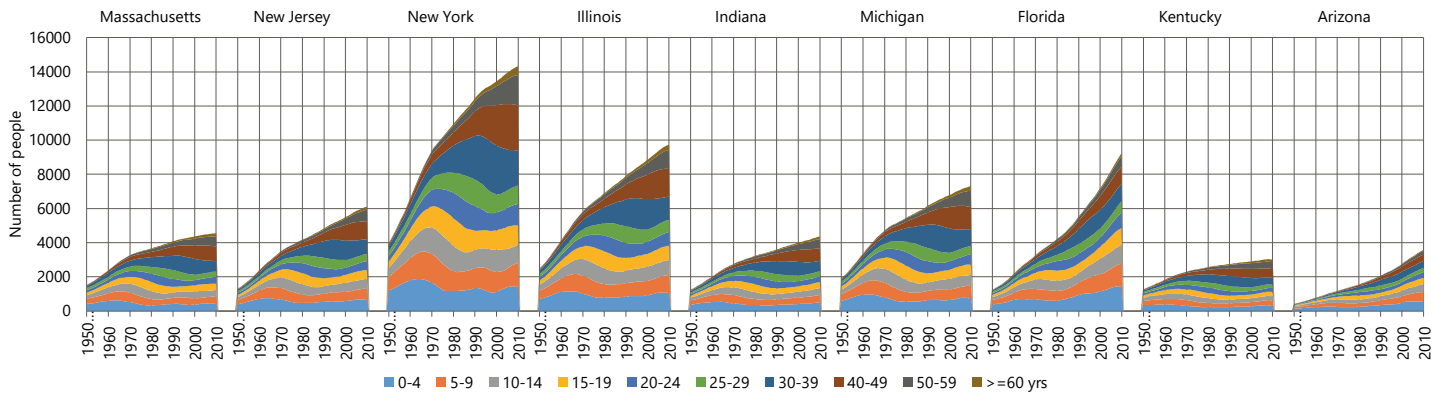


Figure 5. Number of people with Down syndrome by age group and state in 9 U.S. States, corrected for interstate migration, from 1950 to 2010.

of people with Down syndrome is strongest in the more recent immigration groups, Asians/Pacific Islanders and Hispanics. This is a result of these ethnic groups consisting of relatively many young people starting families and having children.

- **How has life expectancy changed for people with Down syndrome?** There has been a rising mean and median life expectancy, growing from an estimated 26 years (mean) and 4 year (median) in 1950 to 53 years (mean) and 58 years (median) in the 2010s.^a
- **What is the life expectancy for people with Down syndrome of different racial and ethnic groups?** As a result of ethnic differences in childhood survival, there also are some ethnic differences in life expectancy. For people with Down syndrome who are non-Hispanic blacks, or American Indians/Alaska Natives, our estimates of mean and median life expectancy were 22 years (mean) and 2 (median) years, respectively, in 1950 and 50 (mean) and 57 (median) years in 2010. For people with DS who are non-Hispanic whites, Asian/Pacific Islanders or Hispanics, we estimate that mean and median life expectancy rates were 26 years (mean) and 4 years (median) in 1950 to 54 years (mean) and 58 years (median) as of 2010. As such, ethnic differences in life expectancy appear to be decreasing.

Table 1. Estimates of live births of children with Down syndrome, live birth prevalence (per 10,000 live births), reduction rates, by US state, 2014

Region	State	Live births of babies with DS	Live birth prevalence of DS	Live birth reduction rate	Live births prevented by selective terminations	Non-selective live birth prevalence of DS	
NE	Connecticut					23.8	
	Maine ^[b]	15	11.8	37%	9	18.7	
	Massachusetts ^[b]	83	11.5	54%	98	25.0	
	New Hampshire					19.9	
	New Jersey	115	11.2	54%	137	24.4	
	New York	299	12.5	47%	269	23.8	
	Pennsylvania					19.2	
	Rhode Island					20.5	
	Vermont	5	8.7	58%	7	20.6	
	MW	Illinois	216	13.6	33%	109	20.5
Indiana		108	12.8	20%	28	16.1	
Iowa						16.3	
Kansas		40	10.1	38%	24	16.3	
Michigan		152	13.3	25%	51	17.7	
Minnesota ^[c]		116	16.6	15%	21	19.6	
Missouri		107	14.2	13%	16	16.4	
Nebraska		47	17.4	0%		17.3	
North Dakota ^[b]		11	10.7	31%	5	15.6	
Ohio		194	13.9	18%	44	17.0	
South Dakota						15.9	
Wisconsin		69	10.3	43%	52	18.0	
S		Alabama					15.1
		Arkansas					14.6
	Delaware ^[b]	16	15.0	22%	4	19.2	
	District of Columbia					25.6	
	Florida	279	12.7	36%	156	19.8	
	Georgia ^[b,c]	169	12.9	31%	76	18.7	
	Kentucky	79	14.0	7%	6	15.2	
	Louisiana ^[c]	75	11.6	22%	22	15.0	
	Maryland	65	8.8	61%	101	22.5	
	Mississippi	50	13.0	7%	4	14.0	
	North Carolina					18.3	
	Oklahoma					14.7	
	South Carolina					16.7	
	Tennessee	116	14.2	12%	16	16.2	
	Texas ^[b]	533	13.8	22%	154	17.7	
Virginia	123	12.0	43%	91	21.0		

Region	State	Live births of babies with DS	Live birth prevalence of DS	Live birth reduction rate	Live births prevented by selective terminations	Non-selective live birth prevalence of DS
	West Virginia ^[b]	15	7.4	50%	15	14.8
W	Alaska ^[b]	17	14.5	15%	3	17.1
	Arizona	111	13.0	28%	43	18.1
	California					23.1
	Colorado					20.6
	Hawaii					22.4
	Idaho					16.6
	Montana					17.2
	Nevada	41	11.6	40%	27	19.2
	New Mexico					16.1
	Oregon	83	18.2	10%	9	20.3
	Utah ^[b]	61	12.1	30%	26	17.3
	Washington	124	14.1	31%	56	20.4
	Wyoming					15.2
DoD	Department of Defense	165	14.0	11%	20	15.7

Notes

- a. Importantly, there is a difference between “life expectancy” and “mean age of death”. “Life expectancy” is a prediction of how many years a person born in a specific year of birth probably will live, whereas “mean age of death” tells us what is the average age of death in the calendar year under observation. Mean age of death is strongly influenced by the age distribution of people living in the specific population, which is a result of the relative sizes of birth cohorts and of historical childhood survival rates within these cohorts. According to our model, mean and median age of death increased, too, and even more rapidly from respectively an estimated 3 years (mean) and 0 years (median) in 1950, 12 years (mean) and 2 years (median) in 1970, 35 years (mean) and 38 years (median) in 1990, to 48 (mean) years and 54 years (median) in 2010. There are some small differences in life expectancy for people with Down syndrome between ethnic groups. However, there are pronounced differences between ethnic groups in age of death. In particular, more recent immigrant groups have lower ages of death, not because of less favorable survival rates, but because these groups include relatively more children and fewer older people.
- b. For estimating actual LB prevalence for 2014, data were used from the period 2012-2016 for most states. For some states we present older data (most recent that were available): Massachusetts 2013-14; Georgia 2011; Texas 2011; Utah 2012; Alaska 2012-15; Maine 2009-2013; North Dakota, Delaware, West Virginia 2010-2014; Arizona, Nevada, and Virginia 2011-2015.
- c. Most surveillance programs cover the whole state. However, actual LB prevalence for Georgia is based on data from Atlanta only (38% of the general births in Georgia), for Minnesota on Hennepin and Ramsey Counties only (34%), for Louisiana on 78% of total births (not all hospitals were included)
- d. The information that we had published in 2019, included an error in the estimates of live births, terminations and population for 2013. The corrected numbers for 2013 are: 5,045 live births of children with DS, 3,429 terminations, a reduction rate of 33%, and DS population size of 211,960.

References

1. de Graaf G., Buckley F., Skotko B. G. (2015). Estimates of the live births, natural losses, and elective terminations with Down syndrome in the United States. *American Journal of Medical Genetics Part A*, 167A, 756-76. [doi:10.1002/ajmg.a.37001](https://doi.org/10.1002/ajmg.a.37001)
2. de Graaf G., Buckley F., Skotko B. G. (2017). Estimation of the number of people with Down syndrome in the United States. *Genetics in Medicine*, 19, 439-447. [doi:10.1038/gim.2016.127](https://doi.org/10.1038/gim.2016.127)
3. de Graaf G., Buckley F., Dever J., Skotko B. G. (2017). Estimation of live birth and population prevalence of Down syndrome in nine U.S. states. *Genetics in Medicine*, advance online publication. [doi:10.1002/ajmg.a.38402](https://doi.org/10.1002/ajmg.a.38402)
4. Natoli, J. L., Ackerman, D. L., McDermott, S. and Edwards, J. G. (2012), Prenatal diagnosis of Down syndrome: a systematic review of termination rates (1995–2011). *Prenatal Diagnosis*, 32: 142–153. [doi:10.1002/pd.2910](https://doi.org/10.1002/pd.2910)