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

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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 2018, we estimate that 1 in every 775 liveborn babies has Down syndrome (13.2 per 10,000). This means that there were about 5,000 babies with Down syndrome born annually in the U.S. in recent years. (Figure 1)

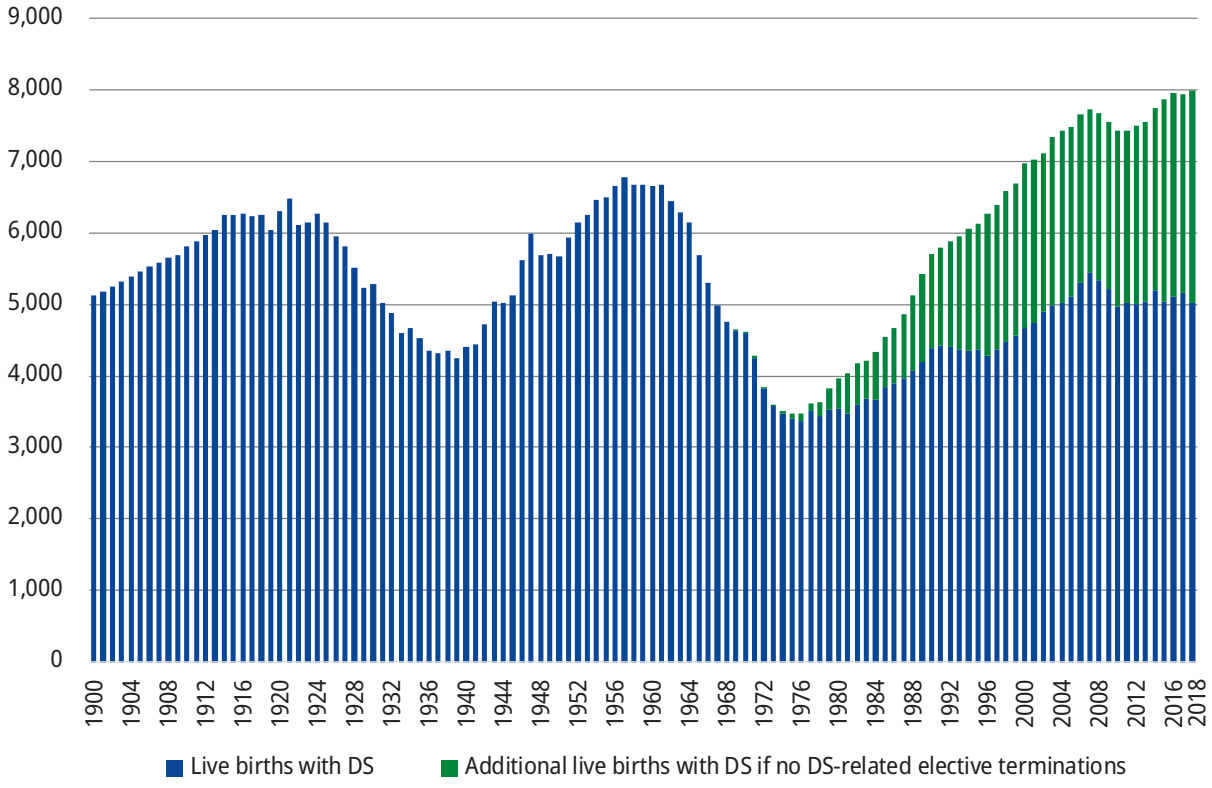

Figure 1. Births of babies with Down syndrome in the USA, 1900-2018[a]

- **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, 4,100 Down syndrome-related elective pregnancy terminations were performed as of 2018 in the U.S. (Figure 2).

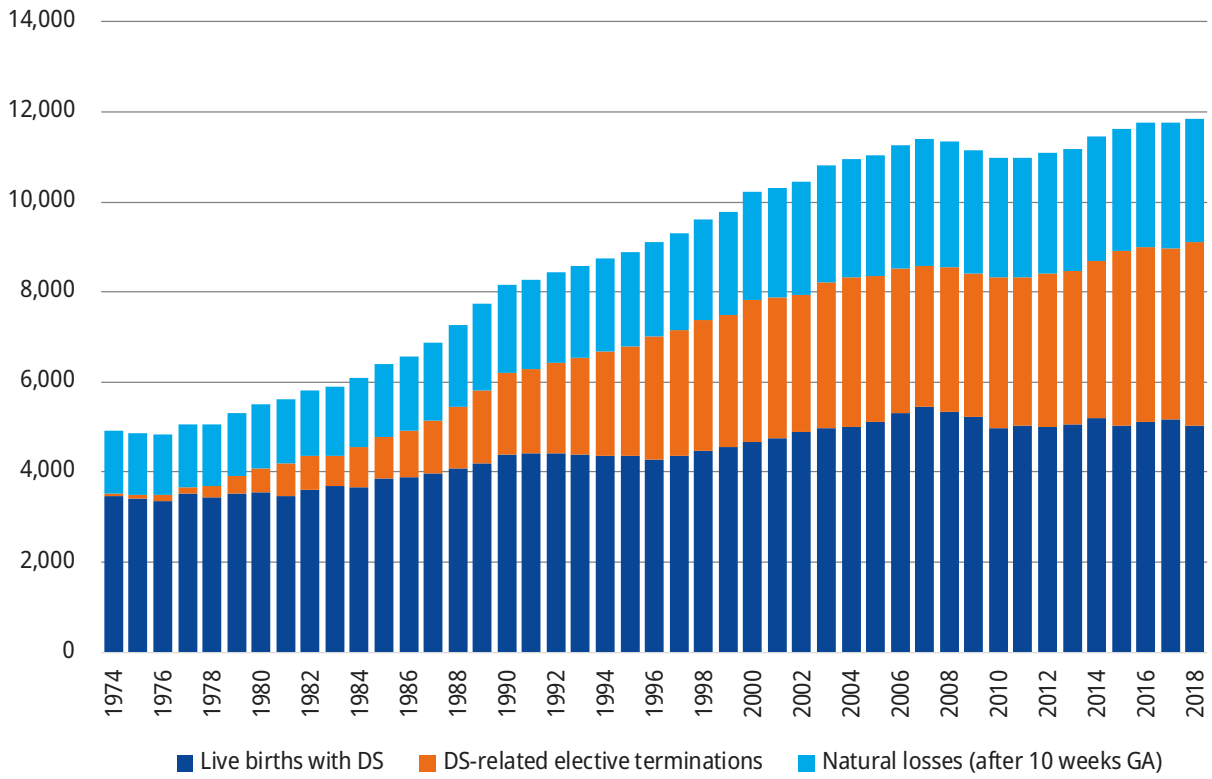

Figure 2. Live births, elective terminations and natural losses in the USA, 1974-2018

- **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 prenatal diagnosis of Down syndrome choose to terminate.[4] However, many parents never pursue prenatal testing to begin with, either by choice or because of limitations with insurance coverage. As such, the 74% termination rate does not imply that 74% fewer babies were born; instead, this number only reflects the decisions of couples who have received a definitive prenatal diagnosis of Down syndrome. Our analyses show that as a result of these elective terminations in the U.S., there was a 37% reduction in the numbers of babies with Down syndrome born in 2018. This means that in recent years there were 37% 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 3)

- **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. 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 34 U.S. states that had sufficient publicly available data to estimate the birth prevalences of Down syndrome (Table 1).

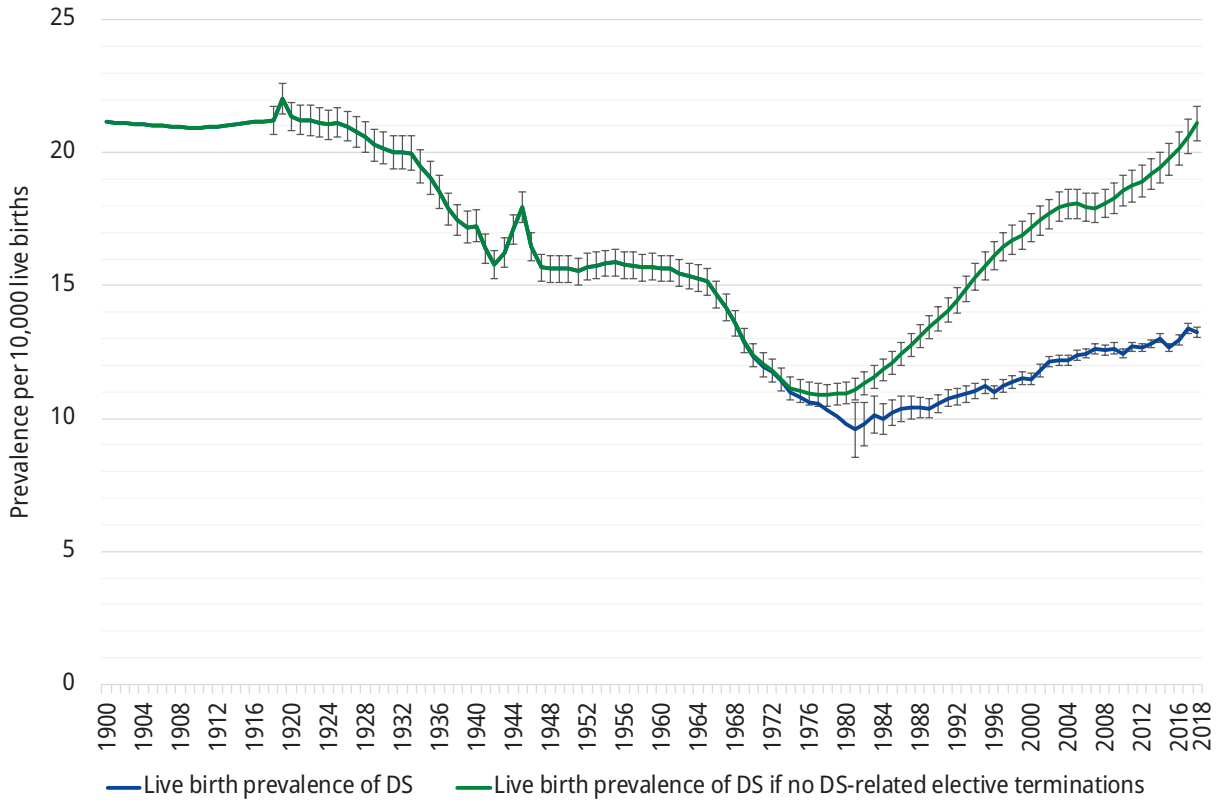

Figure 3. Live birth prevalence of Down syndrome in the USA, 1900-2018

- **How are newer screening technologies influencing birth rates?** Prenatal cell-free DNA (cfDNA) screening (also referred to as non-invasive prenatal screening [NIPS] or testing [NIPT]) was introduced in October of 2011 in the United States. No significant changes in birth rates occurred immediately after 2011. However, the reduction percentage in 2018 was estimated at 37%, which is higher than in past years (from 1996 to 2014, the average value was 32%). Since the introduction of cfDNA screening, it has taken time for medical societies to recommend universal expansion and for some third-party insurers to begin covering the costs. Surveillance programs in the U.S. also report their results as 5-year running averages. So, our estimates of the number of live births of children with DS in 2018, for example, represent the average for the period 2016-2020. If cfDNA screening took several years to become more widely adopted in the U.S., then the possible effects might be more pronounced in future years. We will continue to monitor possible changes.

- **What proportion of children with Down syndrome are born to women older than 35 in the U.S.?** Without elective terminations, the proportion of mothers, 35 years of age or older, who had children with Down syndrome would have increased from 26% in 1980 to 60% in 2018. As a result of elective terminations, however, the actual proportion 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 51% around 2018 (Figure 4).

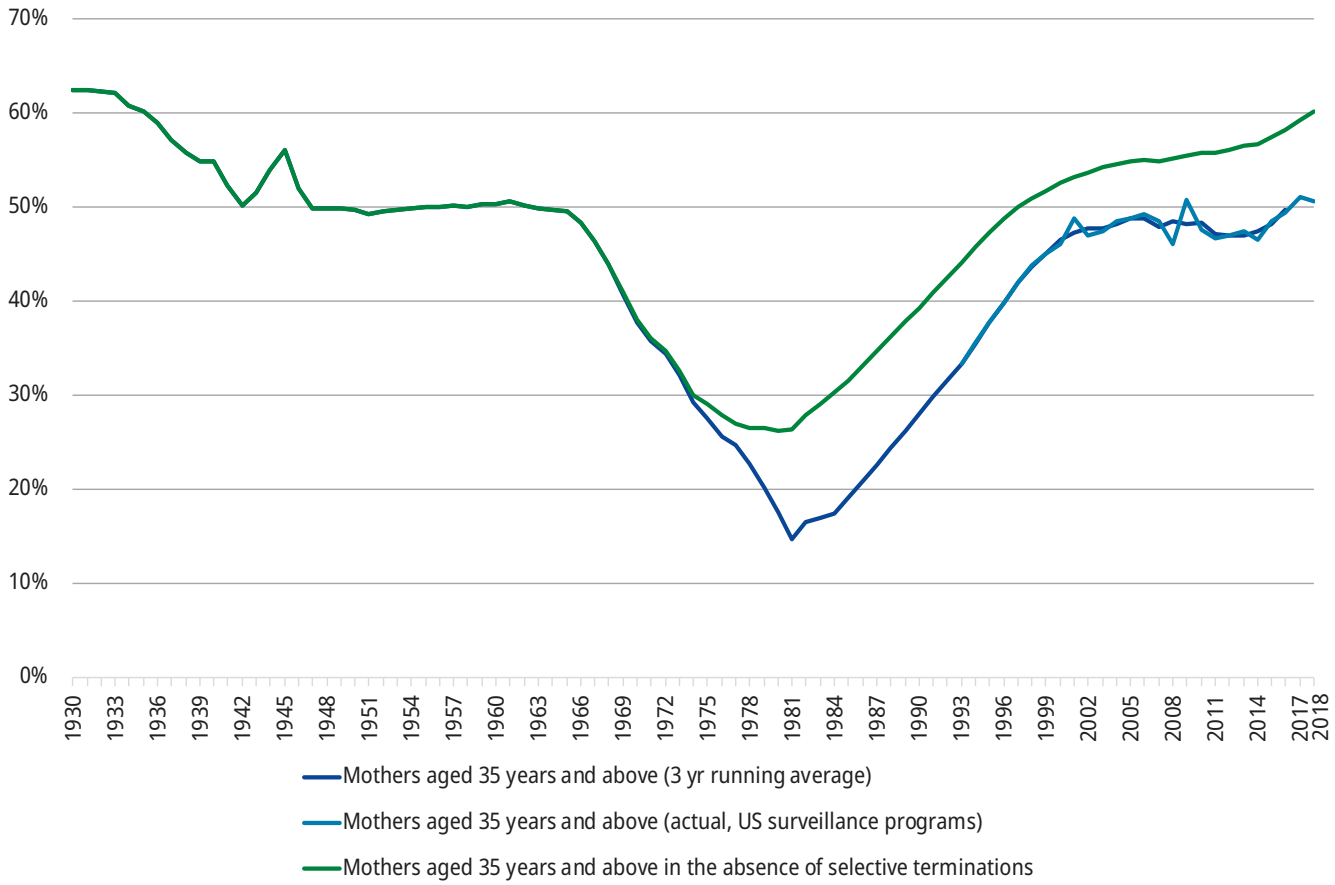

Figure 4. Proportion of mothers of children with Down syndrome aged 35 years or over in year of child's birth in the USA, 1930-2018

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 220,186 in 2018 (Figure 5).

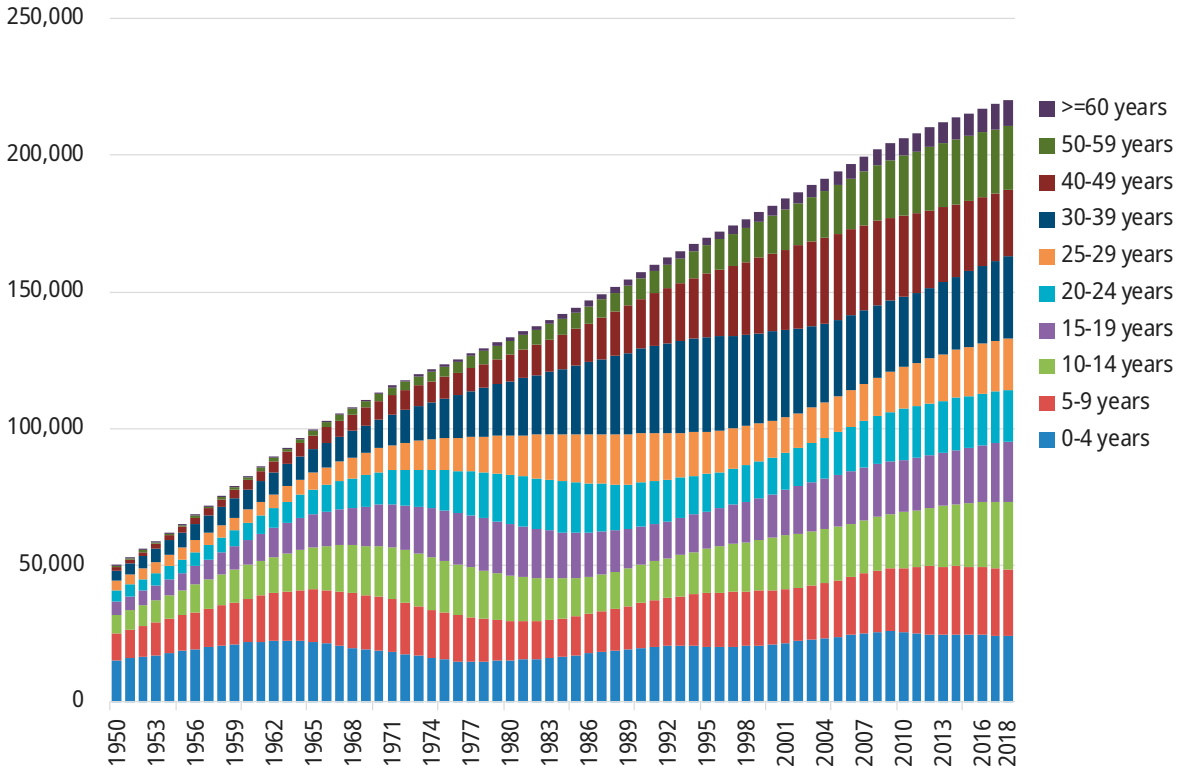

Figure 5. Population of people with Down syndrome in the USA, 1950-2018

- **What proportion of the U.S. population are people with Down syndrome?** The population prevalence of Down syndrome, as of 2018, is estimated at 6.7 per 10,000 inhabitants (or 1 in 1,484).
- **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 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 years (median) in 1950 to 53 years (mean) and 58 years (median) in the 2010s.[b]

- **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, 2018

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					25.4
	Maine [c]	15	11.8	34%	8	17.8
	Massachusetts [c]	81	11.7	53%	92	25.0
	New Hampshire					21.9
	New Jersey	105	10.4	61%	163	26.5
	New York	279	12.3	53%	313	26.2
	Pennsylvania					20.5
	Rhode Island					22.8
	Vermont	6	11.6	47%	6	21.9
MW	Illinois	209	14.4	35%	112	22.2
	Indiana	109	13.4	23%	32	17.3
	Iowa					17.6
	Kansas	44	12.2	32%	20	17.8
	Michigan [c]	198	18.0	5%	10	18.9
	Minnesota	105	15.6	27%	39	21.4
	Missouri [c]	101	13.7	22%	29	17.7
	Nebraska [c,e]	47	17.4	0%		17.3
	North Dakota [c]	11	10.7	29%	5	15.1
	Ohio [e]	251	18.2	0%		17.5
	South Dakota					17.1
	Wisconsin	108	16.3	12%	15	18.5
S	Alabama					16.2
	Arkansas					15.5
	Delaware [c]	16	15.0	21%	4	19.0
	District of Columbia					29.2
	Florida	289	13.0	40%	192	21.7
	Georgia [d]	154	11.9	38%	93	19.0
	Kentucky	61	11.4	30%	26	16.3
	Louisiana [d]	72	12.0	28%	28	16.8
	Maryland	60	8.4	66%	114	24.4
	Mississippi	30	8.0	45%	25	14.5
	North Carolina					19.5
	Oklahoma	58	11.7	27%	22	16.1
	South Carolina					18.1
	Tennessee	113	14.1	19%	27	17.4
	Texas [c]	533	13.8	20%	129	17.1
	Virginia	122	12.3	46%	103	22.6

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	16	9.0	37%	10	14.3
W	Alaska [c]	17	14.5	15%	3	17.1
	Arizona [c]	111	13.0	27%	41	17.8
	California					25.4
	Colorado	115	18.3	17%	24	22.1
	Hawaii					24.2
	Idaho					17.7
	Montana					18.3
	Nevada [c]	41	11.6	39%	26	19.1
	New Mexico					17.7
	Oregon [c]	65	15.3	31%	29	22.2
	Utah [c]	61	12.1	25%	21	16.2
	Washington [c]	105	12.2	46%	90	22.7
	Wyoming					17.8
DoD	Department of Defense	151	14.0	18%	34	17.2

Notes

a. Data on actual DS live birth prevalence for 2015 (period 2013-2017) were based on Heinke et al. (2021). For other recent periods, we made use of the annual reports by the National Birth Defects Prevention Network (NBDPN), adapted as explained in de Graaf G., Buckley F., Skotko B. G. (2015).

b. 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.

c. For estimating actual LB prevalence for 2018, data were used from the period 2016-2010 for most states. For some states we present older data (most recent that were available): Georgia, Mississippi, Ohio, Wisconsin 2014-18; Massachusetts 2015-18; Nebraska 2012-2016; Alaska 2012-15; Arizona and Nevada 2011-2015; Utah 2012; Delaware and North Dakota 2010-2014; Texas 2011; Maine 2009-2013. For these states, we have used the nonselective prevalence of the corresponding year too.

d. Most surveillance programs cover the whole state. However, actual LB prevalence for Georgia is based on data from Atlanta only (27% of births in Georgia), for Louisiana on average 95% of total births (not all hospitals were included).

e. The reduction rates of 0 in Nebraska and Ohio are an artefact of the estimation method. It means that actual prevalence was higher than expected on the basis of maternal ages. However, especially in small populations, as result of chance the “real” number of DS LBs in the absence of selective terminations can be somewhat higher or lower than what is estimated on the basis of maternal ages. If there are relatively few terminations, the reduction can then turn out negative. These negative values were set at zero. It means that reduction will have been low.

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