

# 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.<sup>[1-3]</sup>

## Births

- **How many babies are born with Down syndrome each year?** As of 2012, we estimate that 1 in every 790 liveborn babies has Down syndrome (12.65 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)

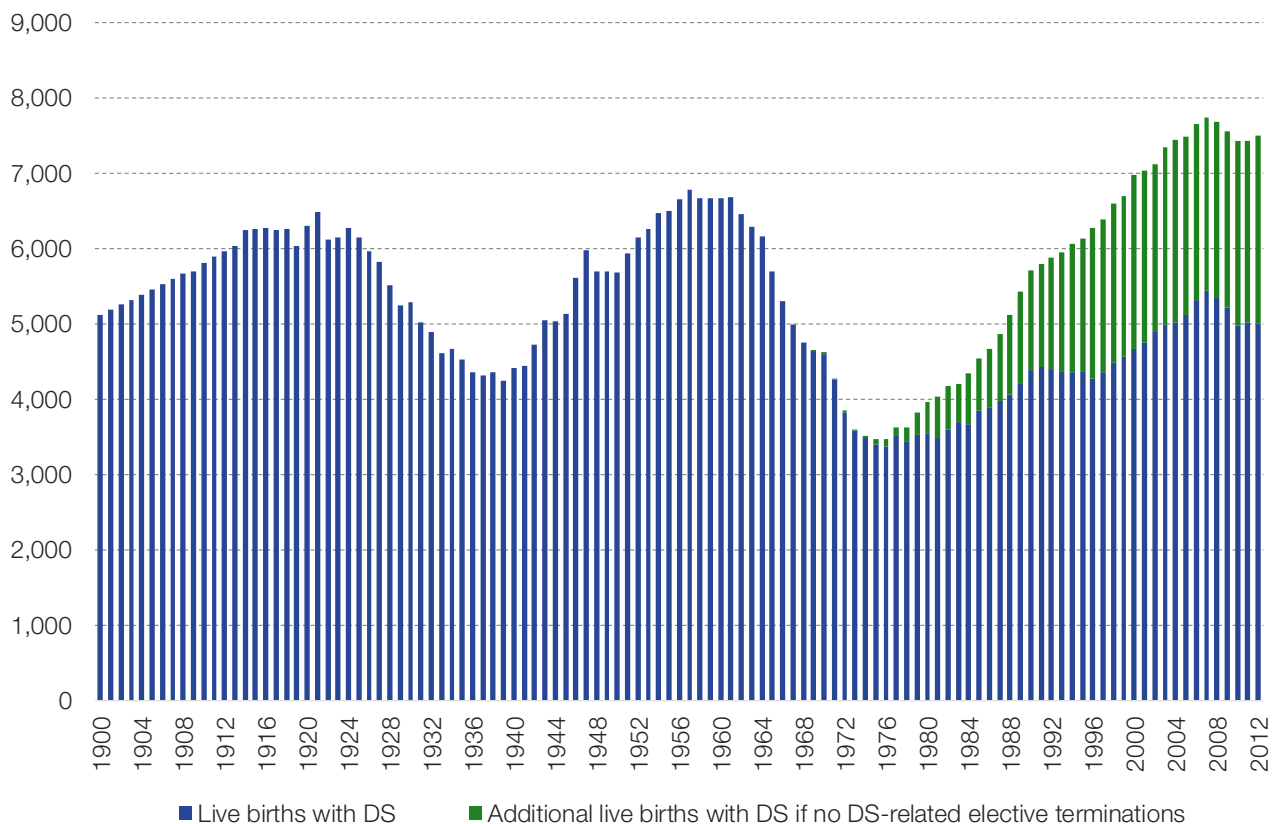
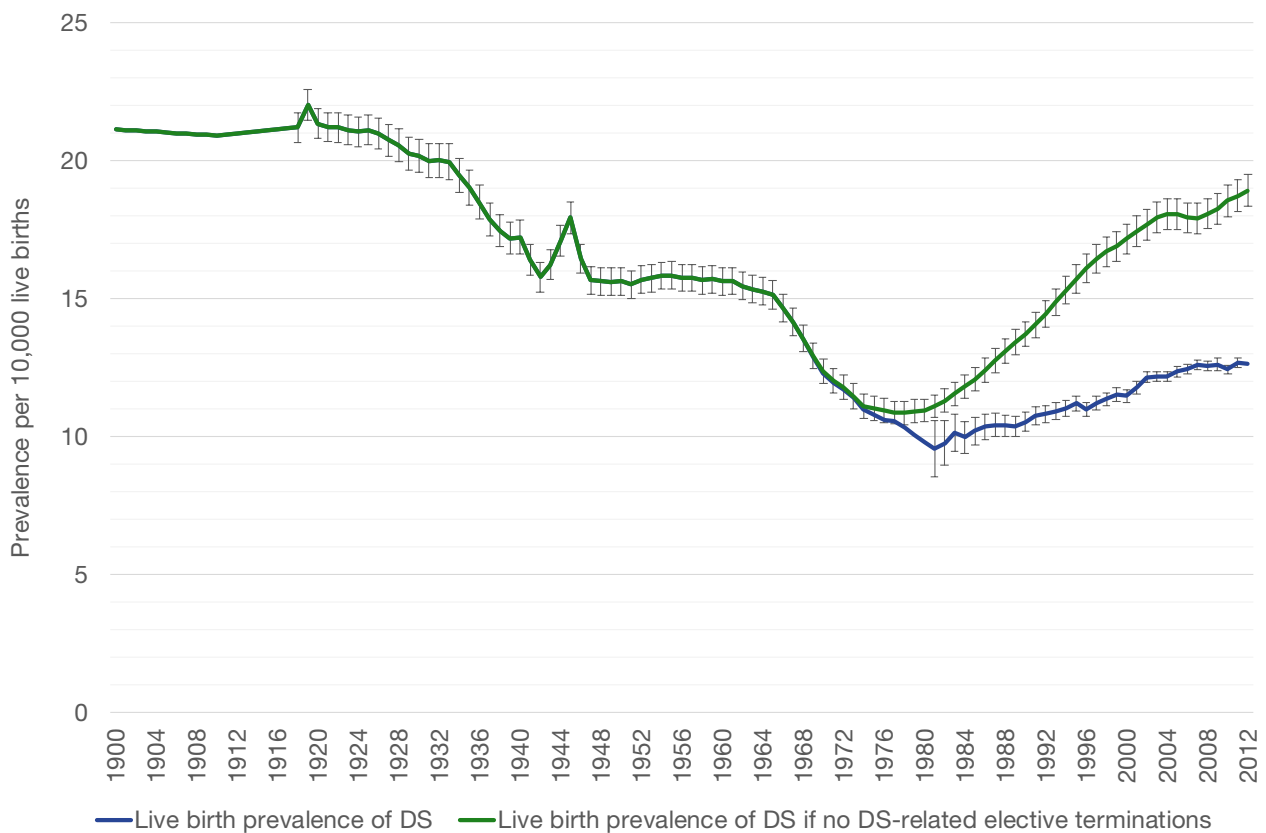


Figure 1. Births of babies with Down syndrome in the USA, 1900-2012

- **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, 3,400 Down syndrome-related elective pregnancy terminations were performed as of 2012 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.<sup>[4]</sup> 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 2012. 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, 1900-2012**

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

State	AC DS LBs	AC DS LB prev. per 10,000	AC DS LB prev. as 1 in X	LB reduction rate	prevented LBs	nonselective LB prev. per 10,000	nonselective DS LB prev. as 1 in X
MA	87	12.0	833	51%	91	24.6	407
NJ	121	11.3	882	52%	133	23.8	420
NY	287	11.8	844	49%	274	23.2	432
IL	199	12.0	832	38%	124	19.5	513
IN	97	11.5	871	26%	34	15.5	645
MI	148	13.0	770	26%	51	17.5	573
FL	294	13.7	729	27%	111	18.9	529
KY	44	8.0	1256	47%	39	15.0	666
AZ	109	12.5	797	27%	41	17.3	579
<b>All 9 states</b>	<b>1386</b>	<b>12.1</b>	<b>824</b>	<b>39%</b>	<b>898</b>	<b>20.0</b>	<b>500</b>

AC DS LBs: Actual number of livebirths of children with DS

AC DS LB prec. per 10,000: Actual livebirth prevalence per 10,000 livebirths

AC DS LB prev. as 1 in X: Actual live birth prevalence as 1 in X

LB reduction rate: Net effect of elective terminations, i.e.: prevented LBs divided by (AC DS LBs + prevented LBs) multiplied by 100%

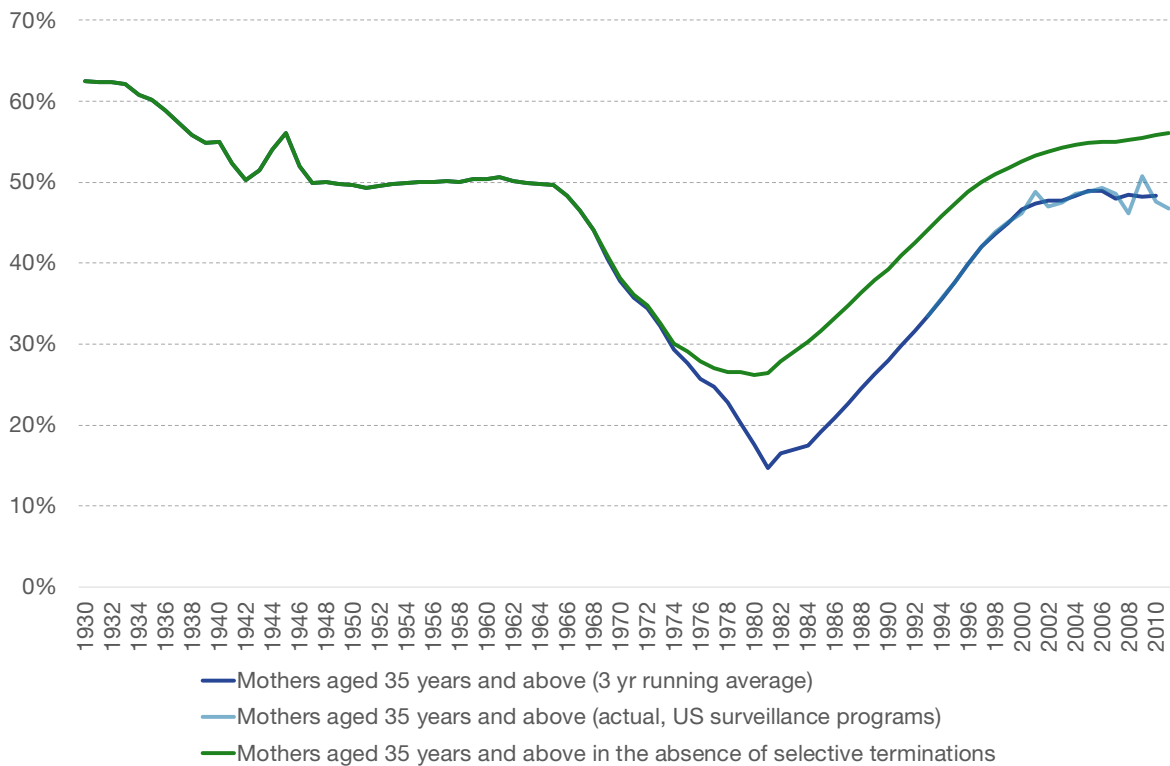
prevented LBs: Number of children with DS that would have been born extra, absent elective terminations

nonselective LB prev. per 10,000: Nonselective livebirth prevalence per 10,000 livebirths

nonselective LB prev. as 1 in X: Nonselective live birth prevalence as 1 in X

**Table 1. Estimates of live birth prevalence of children with DS in 9 U.S. states in 2010**

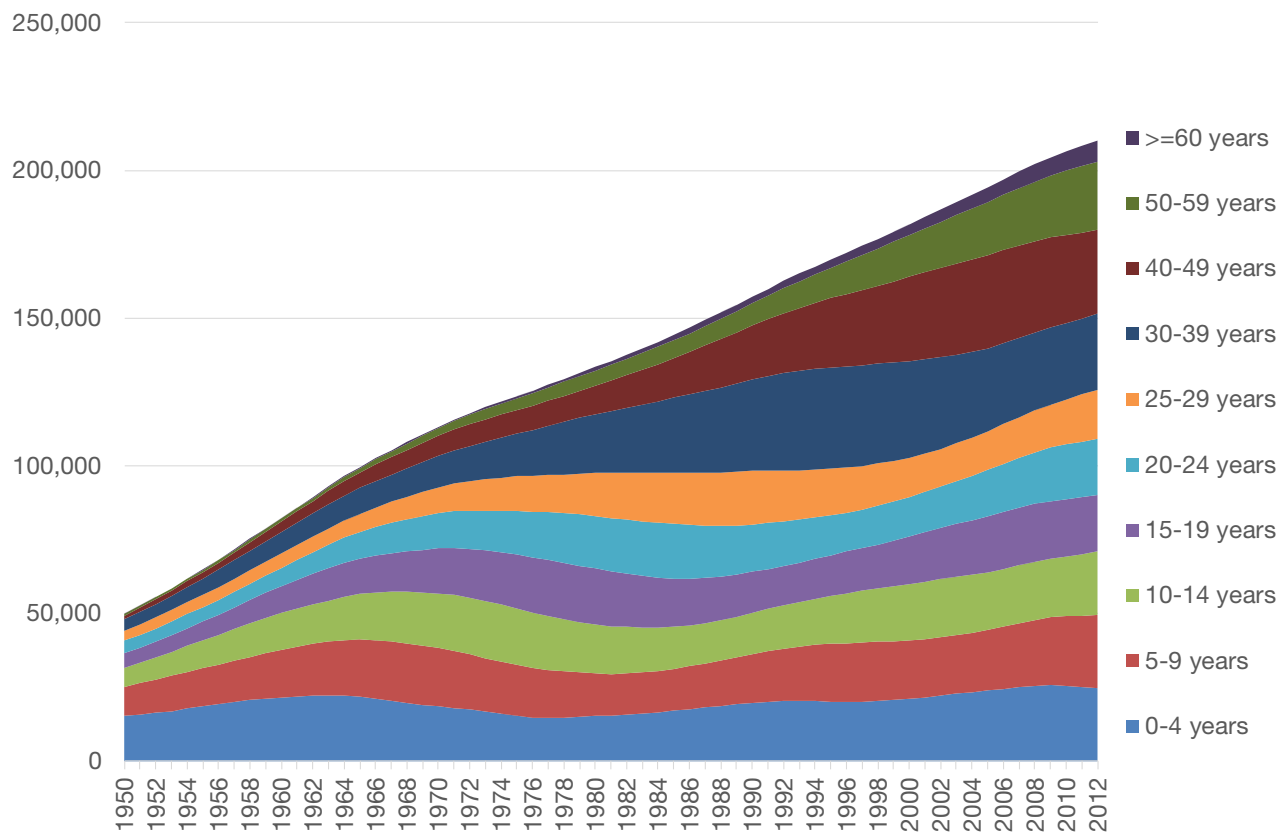
- **How are newer non-invasive screening technologies influencing birth rates?** Noninvasive prenatal screens (NIPS) were introduced in October of 2011 in the United States. No significant changes in birth rates occurred in 2012, the latest year of available data. Surveillance program in the U.S. report 5-year running averages; so data from 2012 represents the average from 2010-2014. New data is 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 56% in 2010. 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 48% around 2010 (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, 1930-2011**

## Population

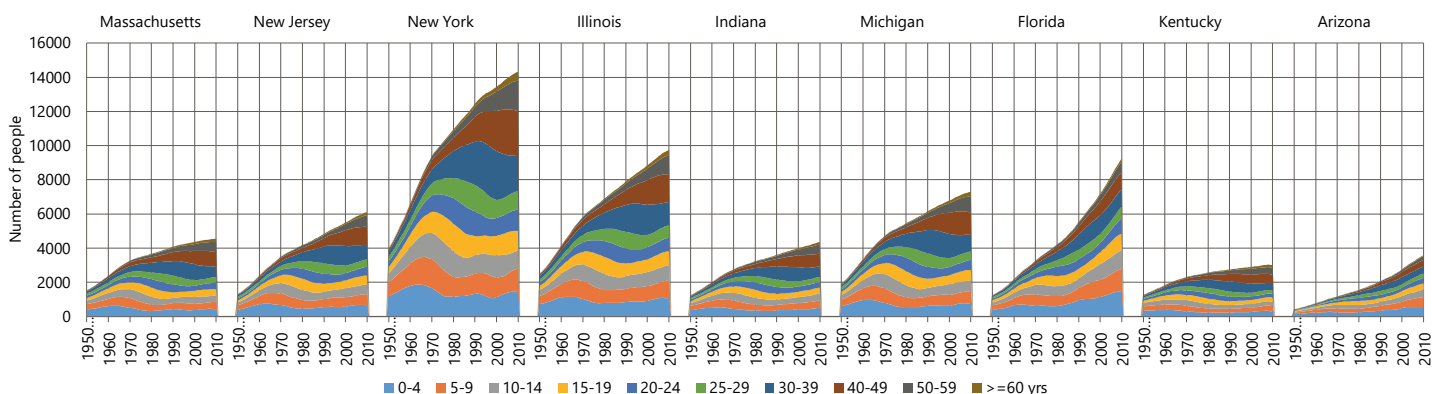
- **How many people with Down syndrome are living in the U.S. today?** Including people born outside of the U.S., we estimate that the number of people with Down syndrome living in the U.S. has grown from 49,923 in 1950 to 206,366 in 2010, and 210,168 in 2012 (Figure 4).
- **What proportion of the U.S. population are people with Down syndrome?** The population prevalence of Down syndrome, as of 2012, is estimated at 6.7 per 10,000 inhabitants (or 1 in 1,493).
- **What is the population of people with Down syndrome in individual U.S. states?** Nine U.S. states had sufficient publicly available data to estimate the populations of people with Down syndrome (Figure 5).<sup>[3]</sup>
- **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 4. Population of people with Down syndrome in the USA, 1950-2012**

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 2010.<sup>a</sup>



**Figure 5. Historical populations: Number of people with DS by age group and state (all ethnic groups combined), corrected for interstate migration, from 1950 to 2010.**

- **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.

## 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.

## 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 [<http://dx.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 [<http://dx.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 [<http://dx.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 [<http://dx.doi.org/10.1002/pd.2910>]