



Down Syndrome

Down Syndrome is a genetic disorder that often causes a variety of physical effects and developmental delays across the lifespan of an affected individual.

Common physical features include¹:

- a flattened face
- almond-shaped eyes that slant up
- a short neck
- small ears
- a tongue that tends to stick out of the mouth
- small hands and feet
- tiny white spots on the eye's iris
- a single line across the palm of the hand
- poor muscle tone or loose joints
- shorter height

Babies born with Down syndrome have a higher risk for¹:

- heart defects
- stomach and digestive problems
- eye problems
- hypothyroidism²
- ear infections
- hearing loss

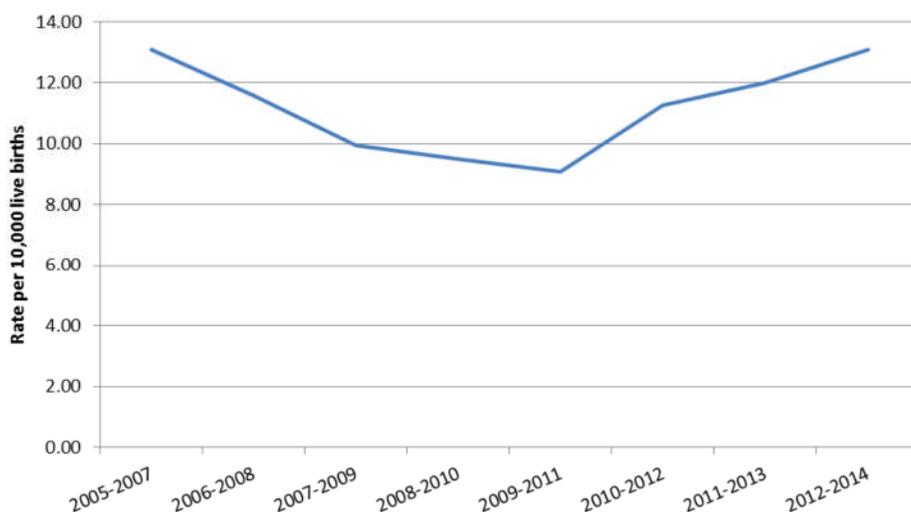
Causes

Chromosomes are the structures that carry all genetic information for an individual. An individual's chromosomes are in every cell of the body and instruct the body on growth and development. The typical individual has a total of 46 chromosomes (23 pairs of chromosomes). Individuals inherit one copy of each chromosome pair from their mother and the other copy from their father. Sometimes in development, an error occurs, and a baby has too much or too little chromosomal material. When an individual has three copies instead of two for a particular chromosome, it is called a trisomy. Down Syndrome is the most common of these conditions. It is sometimes called Trisomy 21 because there are three copies of Chromosome 21.

How Common is Down Syndrome?

- There are an average of 66 cases annually in Kentucky, which is about 11.6 cases per 10,000 live births.
- There are about 6,037 cases annually in the U.S., which is about 14.5 cases per 10,000 live births.³
- The rate in Kentucky in 2012-2014 is approximately the same as it was in 2005-2007.

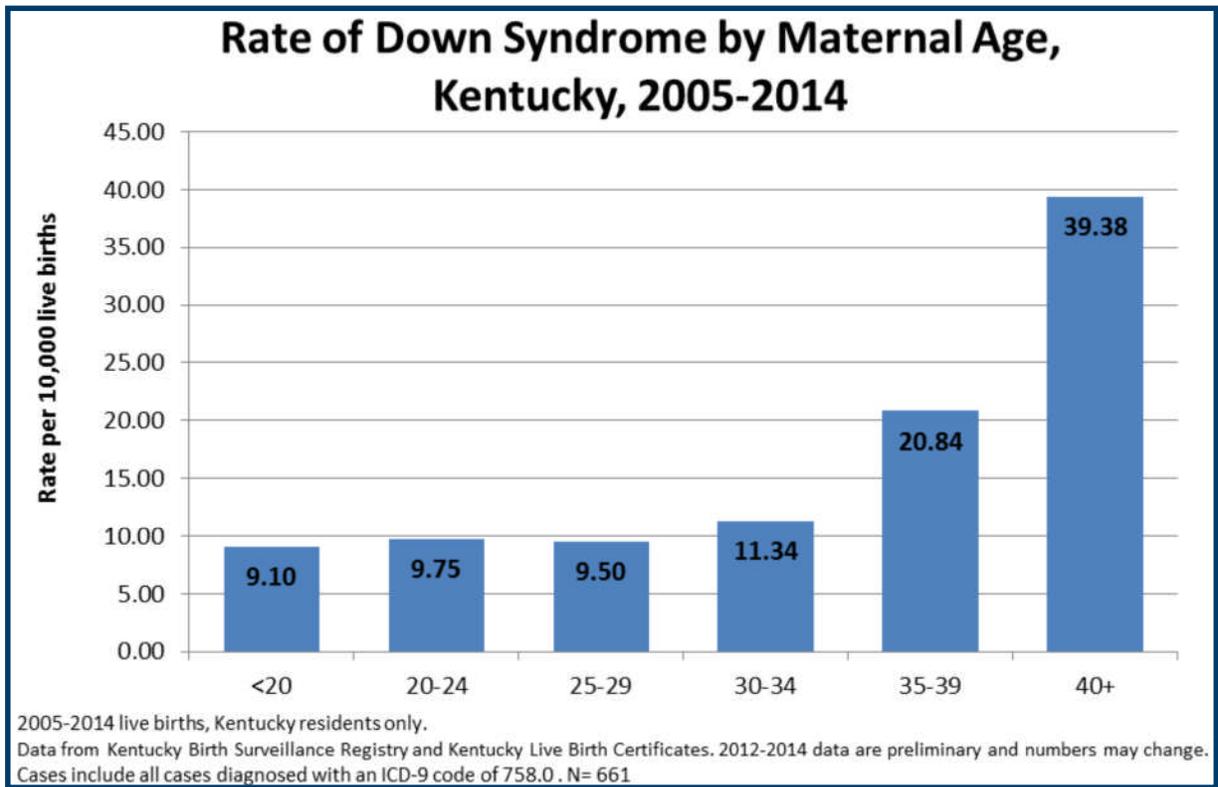
Rate of Down Syndrome, Kentucky, 2005-2014, 3-Year Rolling Averages



2005-2014 births, Kentucky residents only.
Data from Kentucky Birth Surveillance Registry and Kentucky Live Birth Certificates. 2012-2014 data are preliminary and numbers may change.
Cases include all cases diagnosed with an ICD-9 code of 758.0 (Down Syndrome). N (Down Syndrome)=661
Three Year Rolling Averages used to smooth rates and reduce instability due to small numbers.

Risk Factors

The risk of having a child with Down Syndrome increases with **maternal age**, with the increased risk becoming pronounced around age 35. However, most babies with Down Syndrome have mothers who are younger than 35, because there are many more births among younger women.¹ According to Kentucky Birth Surveillance Registry (KBSR) data from 2005-2014, mothers in the 35-39 year old age group have more than twice the risk of having a child with Down Syndrome versus mothers who are 25-29. Among mothers who are 40 years old or greater, the risk is magnified by four times compared to 25-29 year olds (see graph).



Prevention

Early and consistent prenatal care helps a woman monitor her health during pregnancy. Her provider can promote a healthy pregnancy by helping her manage health conditions, medications, and diet.

References

1. Centers for Disease Control and Prevention. Down Syndrome. National Center on Birth Defects and Developmental Disabilities. <http://www.cdc.gov/ncbddd/birthdefects/downsyndrome.html> Last updated November 12, 2015. Accessed February 16, 2016.
2. National Institutes of Health. Down Syndrome. Genetics Home Reference. <http://ghr.nlm.nih.gov/condition/down-syndrome>. Last updated June 2012. Accessed February 16, 2016..
3. Parker SE, Mai CT, Canfield MA, et al. Updated national birth prevalence estimates for selected birth defects in the United States, 2004-2006. National Center on Birth Defects and Developmental Disabilities. *Birth Defects Res A Clin Mol Teratol*. 2010 Dec;88(12):1008-16. doi: 10.1002/bdra.20735.