



Down syndrome, also known as trisomy 21, is a genetic condition caused by an extra copy of the 21st chromosome. Each individual with Down syndrome is unique. Individuals with Down syndrome have variable health issues, including developmental delays and some degree of intellectual disability.

What is life like for people with Down syndrome?

Individuals with Down syndrome can have loving relationships with friends and family, and can learn and make progress in their social skills and communication at their own pace. All individuals with Down syndrome have some degree of intellectual delay, although it is quite variable. Some people with Down syndrome are able to live semi-independently lives, holding meaningful jobs and being active in their community. Others may have more significant delays, and can require continued care throughout their lifetime.

What causes Down syndrome?

Down syndrome, also known as trisomy 21, is caused by having an extra copy of the 21st chromosome. Chromosomes are structures that hold our genes, which is the information that tells our bodies how to grow and function. As humans, we typically have a total of 46 chromosomes, or 23 pairs, one set inherited from our mother through the egg and one from our father through the sperm.

Chromosome conditions are caused when a baby has extra or missing chromosomes. These conditions usually do not run in families. There is nothing you can do to cause or prevent a chromosome condition. For many of these conditions, the chance increases with the age of the mother, but there are babies born with chromosome conditions to mothers of all ages. Pregnancies where the baby has a chromosome condition have a higher risk for miscarriage. In rare cases, Down syndrome occurs due to a chromosome translocation (when two pieces of chromosomes break off and switch places), which can increase the risk to have another pregnancy with Down syndrome. A chromosome translocation related to Down syndrome may also increase the chance for family members to have a pregnancy with Down syndrome.

What are the health and developmental concerns associated with Down syndrome?

The health concerns associated with Down syndrome vary from person to person but may include:

- Heart defects, present at birth in approximately half of all babies with Down syndrome
- Low muscle tone (hypotonia), which affects approximately 80% of people with Down



syndrome

- Blockage in part of the intestine or functional problem with part of the intestine (gastrointestinal defect), which affects approximately 12% of people with Down syndrome
- Hypothyroidism
- Vision problems



- Hearing problems

How common is Down syndrome?

The incidence of Down syndrome is approximately 1/700 births. The chance of having a baby with Down syndrome due to trisomy 21 increases as the mother's age increases. Most cases of Down syndrome are sporadic, meaning they do not run in families. There is nothing we know of that can be done to cause or prevent Down syndrome from occurring.

If I have a baby with Down syndrome syndrome, what is the chance I will have another baby with this condition?



In cases where Down syndrome is due to trisomy 21 and not a chromosome translocation, the chance to have another baby with Down syndrome is approximately 1%. If your age-related risk at the time of the birth of the baby is higher than 1%, then that age-related risk would be the chance for the baby to have Down syndrome. There may be a much higher chance for recurrence of Down syndrome in families when it is



caused by a balanced translocation carried by one of the parents. Keep in mind, it is quite rare for Down syndrome to be caused by a translocation. A picture of the baby's chromosomes (karyotype) can determine if the Down syndrome is caused by trisomy 21 or the type that can run in the family due to a translocation.

What is the treatment for Down syndrome?

Sometimes surgery is needed to treat heart defects or abnormalities of the digestive tract present at birth. Other health conditions are treated as needed, such as hearing and vision concerns, and thyroid disorders.

Early intervention programs can help all individuals with Down syndrome reach their full potential. Intervention programs may include speech, physical, occupational, and/or educational therapy.

Federal law requires that children with Down syndrome receive an appropriate education in the public school system, and they may receive support services within the regular classroom. Many individuals with Down syndrome finish high school, and to date there are over 250 colleges offering programs for individuals with intellectual disabilities.

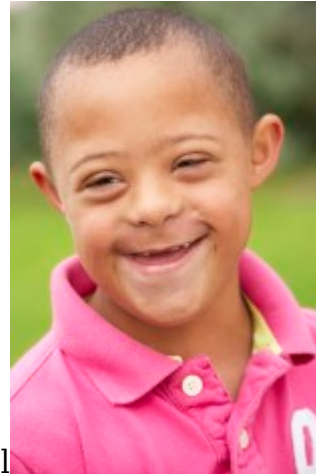
What would having a baby with Down syndrome mean for my family?

Raising a child with special needs, including Down syndrome, can result in more cost and may require more time and patience. However, research has shown that having a child with Down syndrome can have a positive effect on the family.

Outlook

Each individual is unique, however, this is a list of some things that are possible for individuals with Down syndrome:

- Participate in community sports and activities



- Can be in mainstream/'regular' classes at school
- Also may have special education classes
- May complete high school
- Can be employed competitively
- Independent living
- Living in a group home
- Have friends
- Have intimate relationships

Today, the life expectancy for someone with Down syndrome is approximately age 60.

Click [here](#) to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.

Click [here](#) to learn more about scheduling a genetic counseling appointment for infertility or preconception questions.

Click [here](#) to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.

Resources

[Understanding a Down Syndrome Diagnosis](#): A booklet for patients first learning about a prenatal diagnosis of Down syndrome.

[National Center for Prenatal and Postnatal Down Syndrome Resources](#)

[Down Syndrome Pregnancy](#): A book and website with detailed information for new and expectant parents anticipating the birth of a child with Down syndrome.

[Brighter Tomorrows](#): A website for both new and expectant parents.

[National Down Syndrome Congress](#)

[National Down Syndrome Society](#)

[International Mosaic Down Syndrome Association](#)

Related Articles

- [Trisomy 18 \(Edwards Syndrome\)](#)

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- [Open Neural Tube Defects](#)

Open neural tube defects (ONTDs), which includes spina bifida and anencephaly, is a range of birth defects that can include the brain and/or spinal cord. This is generally something that happens very early in pregnancy, and can vary widely in severity and expected outcome. Although the cause of most cases...

- [Sex Chromosome Variations](#)

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- [Microdeletion Syndromes](#)

There are many different kinds of genetic conditions. Some are caused by whole extra or missing chromosomes (such as Down syndrome), while others are caused by changes in a specific gene (such as sickle cell anemia). There is a third group of genetic conditions called microdeletion syndromes. A microdeletion is...