

# Down Syndrome

## *Making a Difference Today*

For some new parents, life can take a harrowing turn. Approximately one out of every 800 to 1,000 babies is born with Down syndrome, a disorder that includes a combination of birth defects such as mental retardation; certain physical distinctions, like a slightly flattened facial profile; and an increased risk of several medical conditions, including heart problems, intestinal malformations, and visual or hearing impairments. These children also tend to be plagued with ear infections and colds.

Down syndrome, or DS, can take its toll on families too. Having a child with DS can limit parents' ability to work, put a strain on their marriage, and lessen parental attention to other children.

The financial impact is also large. DS can result in high medical costs and nonmedical costs for special education, rehabilitation, and other services. In the United States the total lifetime cost of DS is estimated to be \$1.8 billion.

### Research Eases the Blow

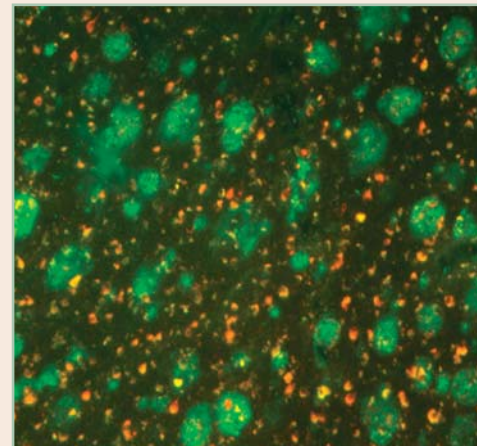
Although individuals with DS and their families currently still face a spectrum of obstacles, a growing body of research is lightening their burden. For example, research has led to the development of several medical tests that help identify whether a pregnant woman is carrying a baby with DS. These tests allow parents to prepare themselves mentally and financially for the birth and give them time to secure intervention programs that can aid their child's development. Scientists believe that the sooner after birth interventions are started, the better.

Today's most commonly used screening test, the Triple Screen, measures certain blood substances that, together with a pregnant woman's age, estimate her risk of having a child with DS. Screening tests accurately detect about 60 percent of fetuses with DS. Available prenatal diagnostic procedures, such as chorionic villus sampling and amniocentesis, can determine whether an unborn baby actually has DS. These procedures involve tissue or cell extraction and carry a small risk of miscarriage, but are about 98 to 99 percent accurate in detecting DS.

Once a child is born, research finds that early intervention programs that focus on the stimulation of developmental skills can benefit those with DS. For example, adolescents with DS who received intervention programs early in life had significantly higher scores on measures of intellectual functioning than a comparison group, according to several studies. Such improvements might help those with DS live more independently and maintain a job.

### Research Leads to Better Treatment Regimens

Research supported by the National Institutes of Health may further lighten the load of DS. Already, recent studies that found a link between DS and the brain disorder Alzheimer's disease, as well as genetic discoveries, hold promise for helping scientists determine better treatment regimens. But such progress will happen only if research receives continued financial support.



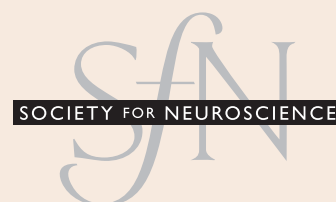
This section of brain tissue, taken from an individual with Down syndrome (DS), is packed with abnormal clumps (see green coloring) common to Alzheimer's disease, which links DS with the memory-robbing brain disorder. Based on this connection, also reported in other studies, scientists have started testing therapies in people with DS that have benefited patients with Alzheimer's disease.

### Continued funding for research could lead to:

- A better understanding of the molecular processes that create havoc in the DS brain.
- The introduction of new treatment regimens that counter various biological imbalances and help treat the condition.
- A decrease in the emotional suffering caused by DS.
- A reduced financial burden for families and society.

For more information please email [brss@sfn.org](mailto:brss@sfn.org).

© 2005 Society for Neuroscience. Image courtesy of Jorge Busciglio, PhD, and Pablo Helguera, PhD, University of California, Irvine. V.20.0.05



[www.sfn.org](http://www.sfn.org)

# Down Syndrome

## Making a Difference Tomorrow

Despite progress, Down syndrome (DS) continues to create an enormous burden for families and society.

Did you know that:

- DS, known for causing delays in intellectual development, affects more than 350,000 people in the United States alone.
- DS can result in high costs in human suffering, medical costs, and nonmedical costs for special education, rehabilitation, and other services.
- In the United States, the total lifetime cost of DS is estimated to be \$1.8 billion.

## Research Equals Hope for the Future

Fortunately, funding for research by the National Institutes of Health (NIH) could help scientists introduce new treatment regimens that significantly improve the lives and finances of numerous Americans.

One area of advance is being propelled by studies that appear to find a connection between DS and the memory-robbing ailment Alzheimer's disease, which normally afflicts the elderly. For example, brains from individuals with DS are often strewn with clumps, a tell-tale sign of Alzheimer's disease. Technically termed plaques, researchers believe these clumps injure or kill brain cells and help cause mental deficits.

Scientists also have long held that a cell system that uses the chemical acetylcholine to regulate memory is damaged in the Alzheimer's-afflicted brain. Evidence indicates that this same system is sometimes damaged in DS.

It is now thought that many people with DS develop Alzheimer's disease at an accelerated rate as they age. Some researchers also speculate that the same biological processes that underlie Alzheimer's disease may be associated with the childhood development of the intellectual impairments that characterize DS.

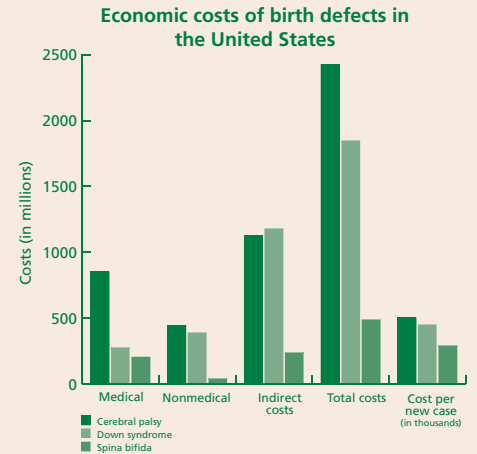
The findings recently led researchers to test therapies used for Alzheimer's disease in people with DS. Research partly funded by NIH found evidence that a medication that attempts to normalize the acetylcholine system may improve the thinking abilities of patients with DS and Alzheimer's disease.

Another study is testing the use of vitamin E, which has properties that may protect cells from dying. Past NIH-supported research has suggested that supplements of the vitamin may slow the progression of Alzheimer's disease.

Methods that target genes also hold promise for treating DS. Our genes guide the production of proteins, which direct the development and function of our body and brain. In DS, cells contain an excess of certain genes that, in turn, can abnormally boost protein production. Researchers, with the aid of NIH support, recently started to decipher these genes and unravel their specific roles in DS. One gene being studied, termed APP, is tied to plaque development. Methods developed to specifically counter or reduce the gene activity could potentially translate into treatments.

Ultimately, such therapies might help normalize brain function and further ease the burden of a DS diagnosis.

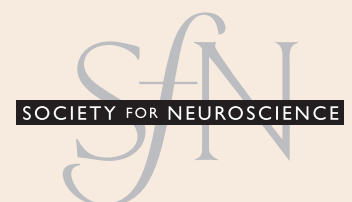
For more information please email [brss@sfn.org](mailto:brss@sfn.org).



The most clinically important birth defects in the United States, including Down syndrome, cause substantial economic burden, according to the most recent data (1992) released by the Centers for Disease Control and Prevention. Conditions with the highest cost per new case that year included cerebral palsy (\$503,000), Down syndrome (\$451,000), and spina bifida (\$294,000).

## Already research has led to:

- The introduction of several medical tests that help identify whether a pregnant woman is carrying a baby with DS, which provides parents time to secure intervention programs that can aid their child's development.
- Evidence that those with DS can benefit from early intervention programs that focus on the stimulation of developmental skills.
- Findings that hold promise in helping scientists create better treatment regimens, including studies that determined a link between DS and the brain disorder Alzheimer's disease, as well as genetic discoveries.



[www.sfn.org](http://www.sfn.org)