

**Office for Genetics
and
People with Special Health Care Needs**

Down's Syndrome (Trisomy 21)

Fact Sheet

What Is Down's Syndrome?

- A genetic disorder in which an infant has 47 chromosomes instead of the usual 46, due to an extra chromosome 21

Signs and Symptoms

- Similar facial appearance with eyes that slant downward, small ears and a small mouth, making the tongue appear to be large
- A short neck
- Heart and stomach/intestine abnormalities are more common than in children without this syndrome
- Low muscle tone
- Short stature
- Intellectual disability

What is the cause?

- Down's Syndrome is caused by an extra copy of chromosome 21. It is also called Trisomy 21 because there are three copies of this chromosome instead of two (chromosomes are the structures in cells that contains the genes)

Is there any treatment?

- Currently, there is no cure for Down's Syndrome, however, research has shown that women who get 0.4 mg of folic acid per day before they become pregnant and through early pregnancy have a lower risk of having a baby born with certain birth defects of the brain and spinal cord.
- Children with Down's Syndrome need to be followed by their pediatrician regularly since there are several medical concerns that they may have. These include: heart problems, abnormal thyroid function, neck instability, ear infections and hearing problems
- Children with Down's Syndrome benefit from early intervention services to help with motor and learning skills. You can contact the Maryland Infants and Toddlers program listed on the next page

Support Groups/Resources

- Kennedy Krieger Institute
<http://www.kennedykrieger.org>
443-923-9200
- March of Dimes
www.marchofdimes.com
1-410-546-2241
- Maryland State Department Of Education
www.marylandpublicschools.org/MSDE
410 767-0261
1-800-535-0182
- National Down Syndrome Society
<http://www.ndss.org>
1-800-221-4602
- Office for Genetics and Children with Special Health Care Needs
<http://fha.maryland.gov/genetics/bdris.cfm>
1 -800-638-8864
- The Parents' Place Of Maryland
www.ppmmd.org
410- 768-9100
- Maryland Infants and Toddlers Program
http://www.marylandpublicschools.org/MSDE/divisions/earlyinterv/infant_toddlers/
410-767-0261 or 1-800-535-0182
- NIH Health information:
<http://health.nih.gov/topic/BirthDefects>
- CDC National Center for Birth Defects and Developmental Disabilities
<http://www.cdc.gov/ncbddd/birthdefects/index.html>
- National Organization of Rare Disorders (NORD)
<http://www.rarediseases.org/>
- Birth Defects Research for Children, Inc
<http://www.birthdefects.org/>