

Trisomy 18 (Edward's Syndrome)

Version: April 2009



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Condition & Description

Trisomy 18 is a serious chromosome abnormality caused by the presence of an extra #18 chromosome. It affects about 1 in every 3000 newborns¹. A syndrome is a pattern or combination of physical, mental, and health characteristics that are usually seen together. Approximately 80% of children born with this are female². It was first described by Dr Edwards in 1960³. There are more than 130 documented abnormalities that may be associated with Trisomy 18, although no one will have all of these features³. In California, the rate of Trisomy 18 is 3.27 per 10,000 births⁷.

Associated Conditions

All children with Trisomy 18 have developmental delays. Approximately 80-90% will have a congenital heart malformation¹, 65% will display central nervous system malformations⁴, more than 50% will experience hearing loss¹, and approximately 25% will display defects of the digestive system or abdominal wall⁴.

Other complications may include feeding difficulties, slow growth, apnea, seizures, kidney defects, urinary tract infections, and scoliosis¹.

Causes & Risk Factors

The causes for chromosome abnormalities such as Trisomy 18 are unknown, although several risk factors for Trisomys have been identified.

Mothers who are age 35 or greater have been found to be at higher risk for having a baby with Trisomy 18⁵. The chance of having another pregnancy with Trisomy 18 is about 1%⁵.

Prevention

There are a number of steps a woman can take that may reduce her risk of having a baby with a birth defect. Please see CBDMP's fact sheet on [Reducing the Risk of Birth Defects](#).

Preconception & Prenatal Care

It is possible to diagnose Trisomy 18 during pregnancy through prenatal diagnostic procedures such as amniocentesis or chorionic villus sampling. Ultrasound may be useful in screening for Trisomy 18. There are some signs that may be seen on a thorough ultrasound (sometimes called level II or targeted ultrasound) that may indicate that there is an increased chance for Trisomy 18⁶.

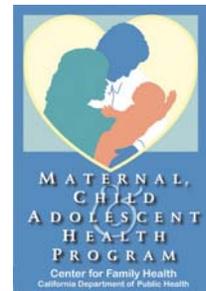
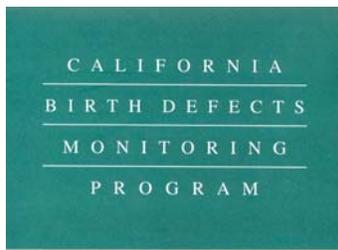
Studies suggest that first trimester screening using a combination of a blood test and ultrasound can detect up to 95% of pregnancies affected by Trisomy 18⁵. For more information, please visit the [California Prenatal Screening Program](#) website.

Medical Care

Any course of treatment will be decided on an individual basis. Follow-up care includes cardiac and eye evaluations, hearing tests, routine ultrasounds, scoliosis checks and routine immunizations. In addition, infant/pre school early intervention for the affected child and ongoing support for the family should occur¹.

Long Term Outcomes

Of infants born with Trisomy 18, 5% to 10% will survive beyond one year. All children with Trisomy 18 have severe developmental delays, but it has been noted that older children with Trisomy 18, interact, laugh and relate to their families³.



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Resources For Families

Regional Centers

Regional centers are nonprofit private corporations that contract with the Department of Developmental Services to provide or coordinate services and supports for individuals with developmental disabilities. They have offices throughout California to provide a local resource to help find and access the many services available to individuals and their families.

- Services Provided By Regional Centers
<http://www.dds.cahwnet.gov/RC/RCSvs.cfm>
- Who is Eligible for Services?
<http://www.dds.cahwnet.gov/General/Eligibility.cfm>
- Early Start
<http://www.dds.ca.gov/EarlyStart/Home.cfm>

California Healthy Families Program

Healthy Families is low cost insurance for children and teens. It provides health, dental and vision coverage to children who do not have insurance and do not qualify for free Medi-Cal.

- Phone: 1-800-880-5305 if you do not already have a child enrolled in Healthy Families.
- E-mail: HealthyFamilies@MAXIMUS.com
- Be sure to include your name and phone number.
- In person: Many community organizations have people who are trained to help you apply. Call to find a *Certified Application Assistants*, or CAAs in your area: 1-888-747-1222. The call is free.

Website: www.healthyfamilies.ca.gov

California Children's Services (CCS)

California Children's Services (CCS) is a state program for children with certain diseases or health problems. Through this program, children up to 21 years old can get the health care and services they need. CCS will connect you with doctors and trained health care people who know how to care for your child with special health care needs.

(Service eligibility is income-based, but you may be eligible for services if the health care costs exceed 20% of your income.)

Find your local CCS Office –

- In the phone book under *California Children's Services* or *County Health Department*
- Or online at: www.dhs.ca.gov/ccs

Birth Defects Research for Children, Inc.

Birth Defect Research for Children is a resource for free birth defect information, parent networking and birth defect research through the National Birth Defect Registry.

- Birth Defect Research for Children, Inc.
800 Celebration Avenue, Suite 225
Celebration, FL 34747
- Phone: 407-566-8304
- Fax: 407-566-8341
- Website: <http://www.birthdefects.org/>

Other Information & Support Groups

- S.O.F.T.: <http://www.trisomy.org/index.php>
- Trisomy 18 Support Group: <http://www.trisomy18support.org/>
- Medline: <http://www.nlm.nih.gov/MEDLINEPLUS/ency/article/001661.htm>
- Stanford: <http://www.lpch.org/DiseaseHealthInfo/HealthLibrary/genetics/trisomy.html>
- Family Resource Center: <http://www.frcnca.org/>
- California Prenatal Screening Program: <http://www.cdph.ca.gov/programs/pns/Pages/default.aspx>

References

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4. Goc, Z., Walencka, Z., Wojciechowska, E., Wiecek-Wlodarska, D., Krzystolik-Ladzinska, J. et al. (2006). Trisomy 18 in neonates: Prenatal diagnosis, clinical features, therapeutic dilemmas and outcomes. *Journal of Applied Genetics*, 47(2): 165-170.
5. March of Dimes (2009). Maternal Blood Screening for Birth Defects. Retrieved on October 27, 2008, from http://www.marchofdimes.com/professionals/14332_1166.asp.
6. Support Organization for Trisomy 18, 13, and Related Disorders (SOFT) (2009). How is Trisomy 18 Diagnosed? Retrieved on October 27, 2008, from http://www.trisomy18.org/site/PageServer?pagename=parents_diagnosed.
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