Trisomy 13 (Patau Syndrome)

Condition & Description

Trisomy 13, also called Patau syndrome, was first described by Dr. Klaus Patau in 1960. A syndrome is a pattern or combination of physical, mental, and health characteristics that are usually seen together. Trisomy 13 is a condition that is associated with severe mental retardation and certain physical abnormalities due to an extra 13th chromosome. Trisomy 13 occurs in about 1 out of every 10,000 newborns. The rate of Trisomy 13 in California is 1.48 per 10,000 births.

Associated Conditions

All children with Trisomy 13 have developmental delays. Approximately 80% also have congenital heart disease. Additional conditions may include holoprosencephaly (the front part of the brain does not divide correctly) which occurs in 60% of infants and kidney defects which occur in 30% of infants.

Other complications may include difficulty in breathing or lack of breathing (apnea), deafness, feeding problems, seizures, and vision problems.

Causes & Risk Factors

The causes for chromosome abnormalities such as Trisomy 13 are unknown, although risk factors for Trisomies have been identified.

Mothers' age 35 and greater have been found to be at higher risk for having a baby with Trisomy 13. The chance of having another pregnancy that results in a Trisomy 13 child is about 1%.

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 13, during pregnancy through prenatal diagnostic procedures such as amniocentesis or chronic villus sampling. Ultrasound may be useful in screening for Trisomy 13. 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 13. For more information, please visit the California Prenatal Screening Program website.

Medical Care

Any course of treatment will be decided on an individual basis. Routine follow up care for a child with Trisomy 13 includes cardiac evaluations, eye evaluations, MRI of the head, abdominal ultrasound, hearing tests, referral for infant pre-school/early intervention programs, scoliosis checks through childhood and getting routine immunizations.

Family support and genetic counseling are recommended.

Long Term Outcomes

The syndrome involves multiple abnormalities. Of infants born with Trisomy 13, about 5% will survive the first six months. Those who do survive past one year will generally have severe developmental delays, growth problems, and seizures.
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
- Early Start: http://www.dds.ca.gov/EarlyStart/Home.cfm

**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

**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

**Other Information & Support Groups**

- Trisomy support group: http://www.trisomyonline.org/
- Family Resource Center: http://www.frncna.org/
- California Prenatal Screening Program: http://www.cdph.ca.gov/programs/pns/Pages/default.aspx

**References**