

Transposition of the Great Arteries/DESCRIPTION

DESCRIPTION

6761 TRANSPOSITION OF THE GREAT ARTERIES

Synonym: Transposition of the great vessels

Involves the following Biologic System(s):

Cardiovascular Disorders

Transposition of the great arteries is a heart defect that is present at birth (congenital) in which the major blood vessels that transport blood away from the heart (aorta and pulmonary artery) are switched (transposed) from their normal position. The pulmonary artery normally arises from the base of the lower right-sided pumping chamber (right ventricle) of the heart and carries oxygen-poor blood to the lungs, where the exchange of oxygen and carbon dioxide occurs. The aorta, the main artery of the body, normally arises from the base of the left ventricle and carries oxygen-rich (oxygenated) blood to the body's tissues. However, in infants with transposition of the great arteries, the aorta arises from the right ventricle and the pulmonary artery arises from the left ventricle. As a result, oxygenated blood recirculates to the lungs, while the oxygen-poor blood recirculates throughout the body, and bodily tissues receive insufficient levels of oxygenated blood (hypoxia).

Transposition of the great arteries is not compatible with life unless there is some communication between the pulmonary and systemic circulation, thus allowing for some mixing of deoxygenated and oxygenated blood. Certain fetal shunts may provide such mixing. These include persistence of the fetal channel that joins the pulmonary artery and the aorta (ductus arteriosus), an opening in the fibrous partition (septum) between the upper chambers (atria) of the heart (patent foramen ovale). Some patients with transposition have mixing of blood through openings in the septum between the ventricles (ventricular septal defect, VSD) or atria (atrial septal defects, ASD)

In newborns with transposition of the great arteries, symptoms are primarily cyanosis (bluish discoloration of fingers and toes and mucous membranes). Shortly after birth, affected infants may experience abnormally rapid and deep breathing (tachypnea, hyperpnea) and cyanosis. Without treatment, life-threatening complications will result. Medical treatment includes a medication called prostaglandin E to open the ductus arteriosus and allow mixing. A cardiac catheterization to place a balloon catheter across the atrial septum (balloon septostomy) may be necessary to allow for mixing of blood. Permanent treatment of infants with transposition of the great arteries includes surgery to switch the aorta and coronary arteries and pulmonary artery back to their normal positions (arterial switch operation). This operation is done in the first weeks of life.

Transposition of the great arteries is more common in males than females and affects approximately one in 2,000 newborns. Infants are most often normal sized, full term, and otherwise healthy. The condition is thought to result from the interactions of several different genes, possibly in association

with the involvement of environmental factors (multifactorial inheritance). Although the exact underlying cause of this heart defect is unknown, rese archers suggest that it may result from an error during the development of an embryonic structure that later divides the aorta and pulmonary artery.

Government Agencies

6762 NIH/National Heart, Lung and Blood Institute

National Institute of Health
31 Center Dr MSC 2486, Bldg 31, Room 5A48
Bethesda, MD 20892
301-592-8573
Fax: 240-629-3246
TTY: 240-629-3255
e-mail: NHLBInfo@nhlbi.nih.gov
www.nhlbi.nih.gov

Primary responsibility of this organization is the scientific investigation of heart, blood vessel, lung and blood disorders. Oversees research, demonstration, prevention, education, control and training activities in these fields and emphasizes the prevention and control of heart diseases.

Elizabeth G Nabel, MD, Director
Susan Shurin, MD, Deputy Director

6763 NIH/National Heart, Lung, and Blood Institute

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Bethesda, MD 20824
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www.nhlbi.nih.gov

Provides leadership for a national program in diseases of the heart, blood vessels, lungs, and blood; blood resources; and sleep disorders.

Elizabeth G Nabel MD, Director

6764 NIH/National Institute of Child Health and Human Development

31 Center Drive, Building 31
Bethesda, MD 20892
301-496-1333
Fax: 301-496-1104
www.nichd.nih.gov

Established in 1962 by congress, today the institute conducts and supports research on topics related to the health of children, adults, families and populations. Some of these topics include: developmental disabilities, growth and development, infant death, reproductive health and birth defects.

Jay H Hoofnagle, Director
Lisa Kaeser, Program & Public Liaison

National Associations & Support Groups

6765 American Academy of Pediatrics

141 NW Point Boulevard
Elk Grove Village, IL 60007
847-228-0604
Fax: 847-434-8000
e-mail: kidsdocs@aap.org
www.aap.org

The American Academy of Pediatrics and its member pediatricians dedicate their efforts and resources to the health, safety and well-being of infants, children, adolescents and young adults.

Joann Barbour, Manager

6766 American Heart Association

7272 Greenville Avenue
Dallas, TX 75231
214-373-6300
800-242-8721
Fax: 214-706-1341
e-mail: inquire@amhrt.org
www.americanheart.org

Transposition of the Great Arteries/Web Sites

Supports research, education and community service programs with the objective of reducing premature death and disability from cardiovascular diseases and stroke; coordinates the efforts of health professionals, and others engaged in the fight against heart and circulatory disease.

M Cass Wheeler, CEO

6767 Congenital Heart Information Network

600 North 3rd Street, First Floor
Philadelphia, PA 19123

215-627-4034
Fax: 215-627-4036
e-mail: mb@tchin.org
www.tchin.org

CHIN is an international organization that provides reliable information, support services and resources to families of children with congenital heart defects and acquired heart disease.

Mona Barmash, President

6768 Genetic Alliance

4301 Connecticut Avenue NW
Washington, DC 20008

202-966-7955
800-336-4363
Fax: 202-966-8553
e-mail: info@geneticalliance.org
www.geneticalliance.org

A coalition of voluntary genetic support groups, consumers and professionals addressing the needs of individuals and families affected by genetic disorders from a national perspective.

Sharon Terry, President

6769 United Network for Organ Sharing

700 N 4th Street, PO Box 2484
Richmond, VA 23219

804-782-4800
Fax: 804-782-4816
www.unos.org

Our mission is to advance organ availability and transplantation by uniting and supporting our communities for the benefit of patients through education, technology and policy development.

Walter K Graham, CEO
Vicki F Sauer, Executive VP/COO

Web Sites

6770 American Academy of Pediatrics

The American Academy of Pediatrics and its member pediatricians are committed to the attainment of optimal physical, mental and social health and well-being for all infants, children, adolescents, and young adults.

6771 American Heart Association

www.amhrt.org

Supports research, education and community service programs with the objective of reducing premature death and disability from cardiovascular diseases and stroke; coordinates the efforts of health professionals, and others engaged in the fight against heart and circulatory disease.

6772 Congenital Heart Information Network

www.tchin.org

An international organization that provides reliable information, support services and resources to families of children with congenital heart defects and acquired heart disease and adults with congenital heart defects, and the professionals who work with them.

6773 NIH/National Heart, Lung and Blood Institute

www.nhlbi.nih.gov

Provides leadership for a national program in diseases of the heart, blood vessels, lungs, and blood; blood resources; and sleep disorders.

6774 Southern Illinois University School of Medicine

www.siumed.edu/peds/index.htm

The mission of SUI School of Medicine is to assist the people of Central and Southern Illinois in meeting their present and future health care needs through education, clinical service and research.

6775 United Network for Organ Sharing

www.unos.org

Our mission is to advance organ availability and transplantation by uniting and supporting our communities for the benefit of patients through education, technology and policy development.

6776 Yale University School of Medicine

www.info.med.yale.edu/intmed/cardio/chd

A site that offers information on Transposition of the Great Arteries and other congenital heart conditions.

DESCRIPTION

6777 TRISOMY 18 SYNDROME

Synonyms: Chromosome 18, trisomy 18, Edwards syndrome

Covers these related disorders: Trisomy 18 mosaicism

Involves the following Biologic System(s):

Genetic/Chromosomal/Syndrome/Metabolic Disorders

Trisomy 18 syndrome is a chromosomal disorder that affects about one in 300 newborns. With the exception of reproductive cells, cells of the body normally have 23 pairs of chromosomes that are numbered from 1 to 22 (with a 23rd pair consisting of one X chromosome from the mother and an X or a Y chromosome from the father). However, in infants with trisomy 18 syndrome, all or a portion of chromosome 18 is present three times (trisomy) rather than twice in cells of the body. In some affected infants, only a percentage of cells may contain the trisomy 18 chromosomal abnormality (mosaicism).

The symptoms and physical findings associated with trisomy 18 syndrome are variable and depend upon the exact location, and percentage, of body cells containing the additional chromosomal material from chromosome 18. However, infants with trisomy 18 syndrome experience development delays, usually severe mental retardation, low birth weight, difficulties feeding and breathing, and a failure to gain weight and grow at the expected rate (failure to thrive). In addition, almost all infants with trisomy 18 have complex structural heart defects, failure of one or both testes to descend into the scrotum (cryptorchidism) in affected males, malformations of the hands and feet, additional skeletal abnormalities, and characteristic malformations of the head and facial (craniofacial) area.

In infants with trisomy 18 syndrome, defects of the hands and feet of ten include closed fists with overlapping, abnormally bent fingers; underdeveloped or absent thumbs; and webbing between certain fingers or toes (syndactyly). Affected infants also often have additional skeletal abnormalities, such as a small pelvis, narrow hips with limited movements, fusion of certain bones of the spinal column (vertebrae), or sideways curvature of the spine (scoliosis). Characteristic craniofacial abnormalities associated with trisomy 18 syndrome typically include an abnormally small head (microcephaly); a prominent back portion of the head (occiput); a small mouth (microstomia) and a small jaw (micrognathia); malformed, low-set ears; and short, narrow eyelid folds (palpebral fissures). Additional craniofacial malformations may be present, such as incomplete closure of the roof of the mouth (cleft palate), an abnormal groove in the upper lip (cleft lip), and drooping of the upper eyelids (ptosis). Some infants may have kidney defects. The abnormalities of trisomy 18 are generally not compatible with more than a few months of life. Fifty percent of the affected infants do not survive beyond the first week of life. Although the exact cause of trisomy 18 syndrome is unknown, it is thought to result from errors during division of a parent's reproductive cells (meiosis) and, in some cases of mosaicism, errors during cellular division after

fertilization (e.g., postzygotic nondisjunction). Parents who have a child with translocational trisomy 18 and want additional children should have chromosome studies, because they are at increased risk to have another child with trisomy 18.

Government Agencies

6778 NIH/National Institute of Child Health and Human Development

31 Center Drive, Building 31
Bethesda, MD 20892

301-496-1333
Fax: 301-496-1104
www.nichd.nih.gov

Established in 1962 by congress, today the institute conducts and supports research on topics related to the health of children, adults, families and populations. Some of these topics include: developmental disabilities, growth and development, infant death, reproductive health and birth defects.

Jay H Hoofnagle, Director
Lisa Kaeser, Program & Public Liaison

National Associations & Support Groups

6779 Chromosome 18 Registry & Research Society

7155 Oakridge Drive
San Antonio, TX 78229

210-657-4968
Fax: 210-657-4968
e-mail: office@chromosome18.org
www.chromosome18.org

The purpose of the Chromosome 18 Registry & Research Society is to offer support to patients and families, to educate the public about different available treatments and to connect families and doctors to the research community.

500 Members

Jammine Cody, President
Ben Flowe Jr, VP Public Relations

6780 Congenital Heart Anomalies, Support, Education & Resources (CHASER)

2112 N Wilkins Road
Swanton, OH 43558

419-825-5575
Fax: 419-825-2880
e-mail: chaser@compuserve.com
www.csun.edu/~hcmth011/chaser/

National organization for support, education and resources for families and patients who deal with children born with congenital heart malformations.

Anita Myers, Executive Director

6781 Genetic Alliance

4301 Connecticut Avenue NW, Suite 404
Washington, DC 20008

202-966-7955
Fax: 202-966-8553
e-mail: info@geneticalliance.org
www.geneticalliance.org

The Genetic Alliance promotes healthy living by working to speed the translation of genetic advances into quality and affordable healthcare, public awareness and consumer-centered public policies.

Sharon Terry, President

6782 MUMS: National Parent to Parent Network

150 Custer Street
Green Bay, WI 54301

920-336-5333
877-336-5333
Fax: 920-339-0995
e-mail: mums@netnet.net
www.netnet.net/mums

A national parent-to-parent organization for parents or care providers of a child with any disability, rare or not so rare disorder, chromosomal abnormality or health condition.

Julie J Gordon, Director

6783 Support Organization for Trisomy 18, 13, and Related Disorders (SOFT)

2982 S Union Street
Rochester, NY 14624

585-594-4621
800-716-7638

e-mail: barbsoft@rochester.rr.com
www.trisomy.org

SOFT is a network of families and professional dedication to provide support and understanding to families involved in the issue and decision surrounding the diagnosis and care related to chromosome disorders. Support is provided throughout prenatal diagnosis, the child's life and after their passing. It is committed to the support of families and personal decisions in alliance with a parent-professional partnership. Includes listings of local chapters in 25 states.

Barb Vanherreweghe, Contact

6784 Trisomy 18 Foundation

4491 Cheshire Station Plaza, Suite 157
Dale City, VA 22193

810-867-4211

e-mail: t18info@trisomy18.org
www.trisomy18.org

The foundation's mission is to search for a cure and treatments; to educate and support medical professionals; and to create a worldwide caring community for those affected.

Victoria Miller, Executive Director
Mindy Wilsford, Operations Director

Research Centers

6785 Trisomy 18 Foundation

4491 Cheshire Station Plaza, Suite 157
Dale City, VA 22193

e-mail: t18info@trisomy18.org
www.trisomy18.org

The foundation's mission is to search for a cure and treatments; to educate and support medical professionals; and to create a worldwide caring community for those affected.

Victoria Miller, Executive Director
Mindy Wilsford, Operations Director

Book Publishers

6786 Introduction to Trisomy 18

SOFT
2982 S Union Street
Rochester, NY 14624

585-594-4621
800-716-7638

e-mail: barbsoft@rochester.rr.com
www.trisomy.org

Addresses parent question regarding the disorder as well as explains the chromosomes, diagnosis and characteristics.

Revised 1998

Barb Vanherreweghe, Contact

DESCRIPTION

6787 TRISOMY 13 SYNDROME

Synonyms: Chromosome 13, trisomy 13, D1 trisomy syndrome, Patau syndrome

Covers these related disorders: Trisomy 13 mosaicism

Involves the following Biologic System(s):

Genetic/Chromosomal/Syndrome/Metabolic Disorders

Trisomy 13 syndrome is a chromosomal disorder that is thought to affect approximately one in 5,000 newborns. With the exception of reproductive cells, cells of the body normally have 23 pairs of chromosomes that are numbered from 1 to 22. The 23rd pair includes one X chromosome from the mother and an X or a Y chromosome from the father. In infants with trisomy 13 syndrome, all or a portion of chromosome 13 is present three times (trisomy) rather than twice. In some affected infants, a certain percentage of cells contain the extra chromosome 13, whereas other cells have the normal two. This finding is known as chromosomal mosaicism.

In infants with trisomy 13 syndrome, associated symptoms and physical findings are pronounced and depend upon the specific length and location of the duplicated portion of chromosome 13 as well as the percentage of the body cells containing the defect.

Abnormalities associated with trisomy 13 syndrome include severe developmental delays, profound mental retardation, incomplete closure of the roof of the mouth (cleft palate), an abnormal groove in the upper lip (cleft lip), and unusually small eyes (microphthalmia). Additional characteristic symptoms and findings include abnormal bending of the fingers, the presence of extra fingers and toes (polydactyly), failure of the testes to descend into the scrotum (cryptorchidism in affected males, and malformation of the uterus in affected females, i.e., bicornuate uterus). Many infants have severe feeding difficulties, abnormally diminished muscle tone (hypotonia), and episodes of temporary cessation of breathing (apnea).

Defects in the brain can result in seizure activity and deafness. Most infants with trisomy 13 syndrome also have additional physical malformations, including an abnormally small head (microcephaly) with a sloping forehead; widely set eyes (ocular hypertelorism); a broad, flat nose; low-set, malformed ears; and a small jaw (micrognathia). Reddish, purplish benign growths (hemangiomas) may be present on the forehead or other areas due to an abnormal distribution of minute blood vessels (capillaries). Many affected infants may also have additional skeletal abnormalities, heart defects, and brain malformations. More than 80% of children with trisomy 13 die in the first month. Because of the severity of congenital defects, life-sustaining procedures are generally not attempted. Parents of infants with trisomy 13 caused by a translocation should have genetic testing and counseling, which may help them prevent recurrence. The exact cause of trisomy 13 syndrome is unknown.

Government Agencies

6788 NIH/National Institute of Child Health and Human Development

31 Center Drive, Building 31
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301-496-1333
Fax: 301-496-1104
www.nichd.nih.gov

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Jay H Hoofnagle, Director
Lisa Kaeser, Program & Public Liaison

National Associations & Support Groups

6789 Congenital Heart Anomalies, Support, Education & Resources (CHASER)

2112 N Wilkins Road
Swanton, OH 43558

419-825-5575
Fax: 419-825-2880
e-mail: chaser@compuserve.com
www.csun.edu/~hcmth011/chaser/

National organization for support, education and resources for families, patients and professionals who deal with children born with congenital heart malformations. Information on hospitals, medical assistance, and schooling. Offers Chaser News, an international newsletter and Chaser's Pediatric Heart Surgeons Facility Directory.

Anita Myers, Executive Director

6790 Genetic Alliance

4301 Connecticut Avenue NW
Washington, DC 20008

202-966-7955
800-336-4363
Fax: 202-966-8553
e-mail: info@geneticalliance.org
www.geneticalliance.org

A coalition of voluntary genetic support groups, consumers and professionals addressing the needs of individuals and families affected by genetic disorders from a national perspective.

Sharon Terry, President

6791 National Dissemination Center for Children with Disabilities

PO Box 1492
Washington, DC 20013

202-884-8200
800-695-0285
Fax: 202-884-8441
e-mail: nichcy@aed.org
www.nichcy.org

A national information and referral center for families, educators and other professionals on: disabilities in children and youth; programs and services; IDEA, the nation's special education law; and research-based information on effective practices.

Suzanne Ripley, Executive Director

6792 Support Organization for Trisomy 18, 13, and Related Disorders (SOFT)

2982 S Union Street
Rochester, NY 14624

585-594-4621
800-716-7638
e-mail: barbsoft@rochester.rr.com
www.trisomy.org

SOFT is a network of families and professional dedication to providing support and understanding to families involved in the issue and decision surrounding the diagnosis and care in related chromosome disorders. Support is provided throughout pre-natal diagnosis, the child's life and after their passing. It is committed to the support of families personal decision in alliance with a parent-professional partnership. Site includes a listing of local chapters in 25 states.