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Conradi Hünermann Syndrome

NORD gratefully acknowledges Nancy Braverman, MS, MD, Associate Professor, McGill University and the Montreal Children's Hospital Research Institute, for assistance in the preparation of this report.

Synonyms of Conradi Hünermann Syndrome

- CDPXD2
- chondrodysplasia punctata, X-linked dominant
- Conradi-Hunermann-Happle syndrome
- Happle syndrome

General Discussion

Conradi-Hünermann syndrome is a rare genetic disorder characterized by skeletal malformations, skin abnormalities, cataracts and short stature. The specific symptoms and severity of the disorder may vary greatly from one individual to another. Conradi-Hünermann syndrome is classified as a form of chondrodysplasia punctata, a group of disorders characterized by the formation of small, hardened spots of calcium on the "growing portion" or heads of the long bones (stippled epiphyses) or inside other areas of cartilage in the body. Conradi-Hünermann syndrome is commonly associated with disproportionate and assymetric shortening of long bones, particularly those of the upper arms (humeri) and the thigh bones (femora), curvature of the spine and mild to moderate growth deficiency, resulting in short stature. Many affected individuals also have a prominent forehead; unusually flattened midfacial regions (midfacial hypoplasia), with a low nasal bridge; loss of transparency of the lenses of the eyes (cataracts); sparse, coarse scalp hair; and/or abnormal thickening, dryness, and scaling of the skin. Conradi-Hünermann syndrome is inherited as an X-linked dominant trait that occurs almost exclusively in females.

Signs & Symptoms

The symptoms, progression and severity of Conradi-Hünermann syndrome can vary dramatically, even among members of the same family. The disorder can cause serious complications at birth or be so mild that individuals may not be identified until adulthood (usually after having an affected child). It is important to note that affected individuals may not have all of the symptoms discussed below. Affected individuals should talk to their physician and medical team about their specific case, associated symptoms and overall prognosis. The classic symptoms of Conradi-Hünermann syndrome involved the skeleton, skin and eyes. Intelligence is usually unaffected.

Affected infants may fail to grow and gain weight at the rate expected for age and gender (failure to thrive). Growth deficiencies may ultimately result in a final adult height that is below normal (short stature). Some affected infants are prone to developing repeated infections.

During infancy, individuals with Conradi-Hünermann syndrome have small, hardened spots of calcium on the “growing portion” or heads of the long bones (stippled epiphyses) as well as other areas of the cartilaginous skeleton. Cartilage is a tough, elastic type of connective tissue that provides cushion and structure within the body. When the skeleton begins to develop, it predominately consists of cartilage, which is gradually replaced by bone. The development of these abnormal calcified spots may also be known as chondrodysplasia punctata. In infants with Conradi-Hünermann syndrome, punctate calcifications may develop throughout the spinal column, the pelvis, the front ends of the ribs (costal cartilages), the breastbone, the shoulder blades, the collarbones, and, in rare cases, the larynx. Chondrodysplasia punctata tends to resolve on its own within the first few years of life.

Individuals with Conradi-Hünermann syndrome typically have additional musculoskeletal abnormalities. Such features commonly include asymmetric shortening of long bones of the limbs, particularly those of the upper arms (humeri) and the thigh bones (femora), causing disproportionate length of the arms and legs with one side typically more affected than the other. Affected individuals also frequently have abnormal sideways and, in some cases, front-to-back curvature of the spine (scoliosis or kyphoscoliosis). Abnormal stiffness of the joints or joints that are fixed or locked in a bent position (flexion contractures) may also occur. In some cases, additional musculoskeletal abnormalities have also been reported including malformation of the hips (hip dysplasia), webbing of the fingers (syndactyly) and/or extra fingers (polydactyly), defects of the spinal column, and deformities in which the feet are abnormally twisted out of shape or position (clubbed feet).

Some individuals with Conradi-Hünermann syndrome may have clouding of the lenses of the eye (cataracts). Cataracts may be present at birth (congenital) or may develop during infancy. Cataracts may affect one or both eyes. Cataracts can caused blurred vision or decreased clarity of vision. In rare cases, additional eye (ocular) abnormalities include abnormally small eyes (microphthalmos), abnormally small corneas (microcornea), down-slanting eyelid folds (palpebral fissures), rapid, involuntary eye movements (nystagmus), and degeneration of the main nerve that transmits nerve impulses from the retina to the brain (optic atrophy). In some cases, eye abnormalities can significantly reduce vision.

Additional distinctive facial features may occur in some cases including an unusually prominent forehead (frontal bossing), flattened cheekbones (malar hypoplasia), a flattened bridge of the nose, upturned nostrils (anteverted nares) and malformed (dysplastic) ears. Hearing loss has been reported in some cases.

In the newborn period, many affected infants also have redness (erythema) and unusual thickening, dryness, and scaling of the skin (ichthyosiform erythroderma) distributed in a linear, blotchy pattern over the body. Although the eruption usually resolves during infancy, older children may subsequently develop inflammation and wasting (atrophy) of follicles (follicular atrophoderma), causing pores to appear unusually large. In some cases,

affected areas of the skin may be darker or lighter than surrounding areas (hyper- and hypopigmentation). Patchy areas of hair loss and scarring may develop on the scalp (cicatricial alopecia). The sparse scalp hair may also be unusually coarse and lusterless.

In some instances, additional abnormalities have also been reported in association with Conradi-Hünermann syndrome. Such features have included abnormal calcifications and potential narrowing (stenosis) of the windpipe (trachea) and/or the larynx, which connects the throat and the trachea; an unusually short neck; abnormalities of the nails; various structural malformations of the heart (congenital heart defects); and/or other physical findings.

Causes

Conradi-Hünermann syndrome is caused by mutations of the emopamil-binding protein (EBP) gene. In many cases, this mutation occurs randomly, for no apparent reason (i.e., new mutation). The gene mutation is inherited as an X-linked dominant trait. Genetic diseases are determined by the combination of genes for a particular trait that are on the chromosomes received from the father and the mother.

X-linked dominant disorders are conditions caused by an abnormal gene on the X chromosome. Females have two X chromosomes, whereas males have one X chromosome and one Y chromosome. In females, certain disease traits on the X chromosome may in some cases be "masked" by the normal gene on the other X chromosome (random X-chromosome inactivation). However, since males have only one X chromosome, if they inherit a gene for a disease present on the X, it is more likely to be fully expressed. According to researchers, in males who inherit a disease gene for an X-linked dominant disorder (hemizygotes), it is suspected that full expression of the disorder may be associated with loss of life before birth. In rare cases, males have been reported with Conradi-Hünermann syndrome, most likely as a postzygotic mutation (acquired during embryonic life and not inherited from the parents) or in individuals with an abnormal XXY chromosomal makeup. Men with a disease gene for an X-linked disorder transmit the gene to their daughters but not to their sons. Women with a copy of the disease gene have a 50 percent risk of transmitting the gene to their daughters and their sons.

Investigators have determined that the EBP gene is located on the short arm (p) of the X chromosome (Xp11.23-p11.22). Chromosomes, which are present in the nucleus of human cells, carry the genetic information for each individual. Human body cells normally have 46 chromosomes. Pairs of human chromosomes are numbered from 1 through 22 and the sex chromosomes are designated X and Y. Males have one X and one Y chromosome and females have two X chromosomes. Each chromosome has a short arm designated "p" and a long arm designated "q". Chromosomes are further sub-divided into many bands that are numbered. For example, "chromosome Xp11.23-p11.22" refers to bands 11.23-11.22 on the short arm of the X chromosome. The numbered bands specify the location of the thousands of genes that are present on each chromosome.

The EBP gene creates a protein known as emopamil-binding protein. Mutations of the EBP gene result in deficient levels of functional copies of this protein. Emopamil-binding protein functions as a sterol-8-isomerase enzyme and plays a role in cholesterol biosynthesis. Biosynthesis is the building or conversion of complex chemical compounds from simple substances in the body. The clinical picture of Conradi-Hünermann syndrome is thought to be related to the accumulation of cholesterol precursor compounds (sterols) that are toxic.

In some cases of Conradi-Hünermann syndrome, where there is no apparent family history, the disorder may occur due to gonadal mosaicism. Such cases include rare instances in which more than one child of apparently unaffected parents have the disorder. In gonadal mosaicism, some of a parent's reproductive cells (germ cells) may carry the gene mutation while others contain a normal cell line ("mosaicism"). As a result, one or more of the parent's children may inherit the gene mutation, potentially leading to manifestation of the disorder, while the parent may have no apparent symptoms (asymptomatic carrier). Gonadal mosaicism may be suspected when apparently unaffected parents have more than one child with the same genetic abnormality. Within families, there is variation in the severity of the clinical picture between affected females, and this is largely secondary to differences in X-inactivation.

Affected Populations

Conradi-Hünermann syndrome is a rare disorder that almost exclusively affects women. The exact incidence of the disorder in the general population is unknown, although one estimate places it at 1 in 100,000 individuals. The disorder is often apparent at birth (congenital), but some individuals with mild cases may not be indentified until adulthood.

Chondrodysplasia punctata was first described in the medical literature by Drs. Conradi and Hünermann in 1931. Although the term Conradi-Hünermann was once used to described chondrodysplasia punctata in general, the term names is specifically used to denote X-linked dominant chondrodysplasia punctata, which as fully described in the medical literature by Happle in 1970s.

Related Disorders

Symptoms of the following disorders can be similar to those of Conradi-Hünermann syndrome. Comparisons may be useful for a differential diagnosis.

Rhizomelic chondrodysplasia punctata (RCDP) spectrum are a group of rare disorders that are also classified as peroxisomal biogenesis disorders. RCDP is characterized by bilateral and symmetric shortening of the upper long bone of the arms (humerus) and legs (femur), a condition known as rhizomelia. Additional findings include distinctive facial features, the formation of small, hardened spots of calcium (stippling) on the knee cap (patella) and long bones of the arms and legs (chondrodysplasia punctata), cataracts that are present at birth or shortly thereafter, profound growth deficiency after birth, mental retardation, and seizures. RCDP causes life-threatening complications during the first decade of life and in

some cases during the newborn (neonatal) period. Milder forms of RCDP have been identified in which affected individuals have less severe mental deficits and growth deficiency and often no rhizomelia. Many of these disorders are caused by mutations in the PEX7 gene and are inherited as an autosomal recessive trait.

Chondrodysplasia punctata, X-linked recessive type, is a form of chondrodysplasia punctata characterized by abnormal, symmetric, dotlike (punctate) calcifications within the growing ends of certain long bones (i.e., stippled epiphyses) and other regions; short stature; and underdevelopment (hypoplasia) of the bones at the ends of the fingers (distal phalanges). Additional characteristic findings may include sparse, unruly hair; ichthyosis, primarily over the neck, on the chest, under the arms, and on the backs of the legs; and underdevelopment of the nose (nasal hypoplasia). The severity of the disorder can vary greatly from one person to another. Some affected individuals may also have additional features, including a relatively small head (microcephaly), bilateral cataracts, breathing (respiratory) difficulties, narrowing of the cervical spine, hearing loss and/or cognitive impairment. Because the disorder is inherited as an X-linked recessive trait, it is typically fully expressed in males only. The disorder is caused by deletions or chromosomal rearrangements (translocations) involving the end of the short arm (p) of chromosome X (Xp22.3). A gene mapped to this chromosomal region regulates production of an enzyme known as arylsulfatase E (ARSE). (Enzymes are proteins that increase the rate of certain chemical reactions in the body.) Mutations of this gene have been identified in several individuals with the disorder, suggesting that altered ARSE activity plays a causative role in the development of X-linked recessive chondrodysplasia punctata.

CHILD syndrome, a rare genetic disorder that is apparent at birth (congenital), is characterized by distinctive skin abnormalities and limb defects affecting one side of the body (hemidysplasia). The term “CHILD” is an acronym that stands for (C)ongenital (H)emidysplasia with (I)chthyosiform erythroderma and (L)imb defects. The disorder is associated with mild growth deficiency before birth; stippled epiphyses; limb malformations on one side of the body (unilateral), which may range from underdevelopment (hypoplasia) of finger bones to absence of a limb; unilateral redness (erythema) and unusual thickening, dryness, and scaling of the skin (ichthyosiform erythroderma); and/or unilateral hair loss (alopecia). CHILD syndrome may also be characterized by hypoplasia of other tissues or organs on the affected side, such as other skeletal regions, the brain, spinal cord, thyroid, adrenal gland, and/or lung. Additional features may include an abnormal opening in the partition that separates the upper or lower chambers of the heart (cardiac septal defects); mental retardation; and/or other abnormalities. CHILD syndrome is inherited as an X-linked dominant trait that appears to primarily affect females. According to investigators, some cases of the disorder appear to result from different mutations of the same gene (i.e., EBP gene) responsible for Conradi-Hünermann syndrome (see “Causes” above), indicating that the disorders may sometimes be “allelic.” (An allele is one of two or more alternative forms of a gene that may occupy a particular chromosomal location.) However, the majority of cases are due to a mutation in

the NSDHL gene on Xq28. The gene encodes a steroid dehydrogenase enzyme that also plays a role in cholesterol metabolism. (For more information on this disorder, choose "CHILD" as your search term in the Rare Disease Database.)

Fetal warfarin syndrome, which may also be referred to as coumarin embryopathy, is a characteristic pattern of birth defects in a newborn resulting from exposure to certain anticlotting drugs (i.e., coumarin anticoagulants [vitamin K antagonists]), such as warfarin, during pregnancy. Evidence suggests that the greatest period of risk occurs from approximately six to nine weeks following conception. The most consistent feature is midfacial hypoplasia, with an unusually small, flattened nose; a deep groove between the "wings" of the nose (alae) and the tip; and abnormally small nostrils. Additional characteristic features may include stippled epiphyses, disproportionate short stature, mental retardation, eye abnormalities, hearing loss, sudden episodes of uncontrolled electrical activity in the brain (seizures), and/or other abnormalities. Experts indicate that Conradi-Hünermann syndrome and other forms of chondrodysplasia punctata, especially CDPX1, must be differentiated from the fetal effects of warfarin or other coumarin anticoagulants.

There are additional disorders that are associated with chondrodysplasia punctata or other skeletal symptoms similar to those found in Conradi-Hünermann syndrome. These disorders include Zellweger spectrum disorders, Smith-Lemli-Optiz syndrome, fetal alcohol syndrome, trisomies 18 and 12, Greenberg dysplasia, and chondrodysplasia punctata, tibia-metacarpal type. There are additional disorders that have skin abnormalities that are similar to those found in Conradi-Hünermann syndrome. These disorders include other forms of ichthyosis. (For more information on these disorders, choose the specific disorder name as your search term in the Rare Disease Database.)

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Diagnosis

A diagnosis of Conradi-Hünermann syndrome is made based upon identification of characteristic symptoms, a detailed patient history, a thorough clinical evaluation and a variety of specialized tests. X-ray (radiographic), eye (ophthalmologic), skin (dermatological), and biochemical examinations may all be performed to help make a diagnosis of Conradi-Hünermann syndrome. X-ray evaluation may reveal characteristic stippling of epiphyses and other regions of the cartilaginous skeleton. However, as noted above, there is loss of distinctive epiphyseal stippling over time, potentially making diagnosis difficult. In addition, there have been instances in which individuals with only mild manifestations have not been identified until adulthood.

An important test to confirm a diagnosis of Conradi-Hünermann syndrome is evaluating the plasma for elevated levels of a substance known as sterols. Mutations of the EBP gene result in the accumulation of sterols in the plasma and certain tissues of the body. Sterol levels are measured by gas chromatography-mass spectrometry.

A diagnosis of Conradi-Hünermann syndrome can be confirmed through molecular genetic testing, which can identify the characteristic genetic mutation that causes the disorder.

Standard Therapies

Treatment

The treatment of Conradi-Hünermann syndrome is directed toward the specific symptoms that are apparent in each individual. Such treatment may require the coordinated efforts of a team of medical professionals, such as pediatricians; physicians who diagnose and treat disorders of the skeleton, joints, muscles, and related tissues (orthopedists); skin specialists (dermatologists); eye specialists; and/or other health care professionals.

Various orthopedic measures, including surgery, may be recommended to help prevent, treat, or correct certain skeletal abnormalities associated with the disorder. Surgery may also be advised for certain craniofacial malformations, scoliosis or other physical abnormalities. The surgical procedures performed will depend on the nature, severity, and combination of anatomical abnormalities, their associated symptoms, and other factors.

Recommended treatment for congenital cataracts may include early surgical removal of the cataracts (when they interfere with vision); implantation of artificial lenses in some cases; and/or certain measures following surgery, such as the use of corrective lenses, to help achieve good vision.

For those affected by ichthyosis and skin abnormalities, supportive measures may be recommended, such as bathing with bath oil and/or applying appropriate skin ointments and lubricants that soften and soothe the skin (emollients).

Genetic counseling may be of benefit for affected individuals and their families. Other treatment is symptomatic and supportive.

Investigational Therapies

Information on current clinical trials is posted on the Internet at www.clinicaltrials.gov. All studies receiving U.S. government funding, and some supported by private industry, are posted on this government web site.

For information about clinical trials being conducted at the NIH Clinical Center in Bethesda, MD, contact the NIH Patient Recruitment Office:

Tollfree: (800) 411-1222

TTY: (866) 411-1010

Email: prpl@cc.nih.gov

For information about clinical trials sponsored by private sources, contact:

www.centerwatch.com

Contact for additional information about Conradi-Hünermann syndrome:

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Other Organizations

Genetic and Rare Diseases (GARD) Information Center (https://rarediseases.org/organizations/genetic-and-rare-diseases-gard-information-center/)

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Restricted Growth Association (https://rarediseases.org/organizations/restricted-growth-association/)

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