ACHONDROGENESIS

A 3-in-1 Medical Reference

A Bibliography and Dictionary
for Physicians, Patients,
and Genome Researchers

TO INTERNET REFERENCES

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ACHONDROGENESIS

A BIBLIOGRAPHY AND DICTIONARY FOR PHYSICIANS, PATIENTS, AND GENOME RESEARCHERS

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CHAPTER 1. INTRODUCTION

In March 2001, the National Institutes of Health issued the following warning: "The number of Web sites offering health-related resources grows every day. Many sites provide valuable information, while others may have information that is unreliable or misleading."[1] Furthermore, because of the rapid increase in Internet-based information, many hours can be wasted searching, selecting, and printing. Since only the smallest fraction of information dealing with achondrogenesis is indexed in search engines, such as **www.google.com** or others, a non-systematic approach to Internet research can be not only time consuming, but also incomplete. This book was created for medical professionals, students, and members of the general public who want to know as much as possible about achondrogenesis, using the most advanced research tools available and spending the least amount of time doing so.

In addition to offering a structured and comprehensive bibliography, the pages that follow will tell you where and how to find reliable information covering virtually all topics related to achondrogenesis, from the essentials to the most advanced areas of research. Special attention has been paid to present the genetic basis and pattern of inheritance of achondrogenesis. Public, academic, government, and peer-reviewed research studies are emphasized. Various abstracts are reproduced to give you some of the latest official information available to date on achondrogenesis. Abundant guidance is given on how to obtain free-of-charge primary research results via the Internet. **While this book focuses on the field of medicine, when some sources provide access to non-medical information relating to achondrogenesis, these are noted in the text.**

E-book and electronic versions of this book are fully interactive with each of the Internet sites mentioned (clicking on a hyperlink automatically opens your browser to the site indicated). If you are using the hard copy version of this book, you can access a cited Web site by typing the provided Web address directly into your Internet browser. You may find it useful to refer to synonyms or related terms when accessing these Internet databases. **NOTE:** At the time of publication, the Web addresses were functional. However, some links may fail due to URL address changes, which is a common occurrence on the Internet.

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[1] From the NIH, National Cancer Institute (NCI): **http://www.cancer.gov/**.

For readers unfamiliar with the Internet, detailed instructions are offered on how to access electronic resources. For readers unfamiliar with medical terminology, a comprehensive glossary is provided. We hope these resources will prove useful to the widest possible audience seeking information on achondrogenesis.

The Editors

CHAPTER 2. STUDIES ON ACHONDROGENESIS

Overview

In this chapter, we will show you how to locate peer-reviewed references and studies on achondrogenesis. For those interested in basic information about achondrogenesis, we begin with a condition summary published by the National Library of Medicine.

Genetics Home Reference

Genetics Home Reference (GHR) is the National Library of Medicine's Web site for consumer information about genetic conditions and the genes or chromosomes responsible for those conditions. Here you can find a condition summary on achondrogenesis that describes the major features of the condition, provides information about the condition's genetic basis, and explains its pattern of inheritance. In addition, a summary of the gene or chromosome related to achondrogenesis is provided.[2]

The Genetics Home Reference has recently published the following summary for achondrogenesis:

What Is Achondrogenesis?[3]

Achondrogenesis is a group of severe disorders that affect cartilage and bone development. These conditions are characterized by a small body, short limbs, and other skeletal abnormalities. As a result of their serious health problems, infants with achondrogenesis are usually born prematurely, are stillborn, or die shortly after birth from respiratory failure. Some infants, however, have lived for a while with intensive medical support.

Researchers have described at least two forms of achondrogenesis, designated as type 1B and type 2. These types are distinguished by their signs and symptoms, inheritance pattern,

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[2] This section has been adapted from the National Library of Medicine: **http://ghr.nlm.nih.gov/**.

[3] Adapted from the Genetics Home Reference of the National Library of Medicine: **http://ghr.nlm.nih.gov/condition=achondrogenesis**.

and genetic cause. Other types of achondrogenesis may exist, but they have not been characterized or their cause is unknown.

Achondrogenesis, type 1B is characterized by extremely short limbs, a narrow chest, and a prominent, rounded abdomen. The fingers and toes are short and the feet may be rotated inward. Affected infants frequently have a soft out-pouching around the belly-button (an umbilical hernia) or near the groin (an inguinal hernia).

Infants with achondrogenesis, type 2 have short arms and legs, a small chest with short ribs, and underdeveloped lungs. This condition is also associated with a lack of bone formation (ossification) in the spine and pelvis. Typical facial features include a prominent forehead, a small chin, and, in some cases, an opening in the roof of the mouth (a cleft palate). The abdomen is enlarged, and affected infants often have a condition called hydrops fetalis in which excess fluid builds up in the body before birth.

How Common Is Achondrogenesis?

Achondrogenesis, type 1B is a rare genetic disorder; its incidence is unknown. Achondrogenesis, type 2 and hypochondrogenesis (a similar skeletal disorder) together affect 1 in 40,000 to 60,000 births.

What Genes Are Related to Achondrogenesis?

Mutations in the **COL2A1** (**http://ghr.nlm.nih.gov/gene=col2a1**) and **SLC26A2** (**http://ghr.nlm.nih.gov/gene=slc26a2**) genes cause achondrogenesis.

Achondrogenesis, type 1B is the most severe condition in a spectrum of skeletal disorders caused by mutations in the SLC26A2 gene. This gene provides instructions for making a protein that is essential for the normal development of cartilage and for its conversion to bone. Cartilage is a tough, flexible tissue that makes up much of the skeleton during early development. Most cartilage is later converted to bone, except for the cartilage that continues to cover and protect the ends of bones and is present in the nose and external ears. Mutations in the SLC26A2 gene disrupt the structure of developing cartilage, preventing bones from forming properly and resulting in the skeletal problems characteristic of achondrogenesis, type 1B.

Achondrogenesis, type 2 is one of several skeletal disorders caused by mutations in the COL2A1 gene. This gene provides instructions for making a protein that forms type II collagen. This type of collagen is found mostly in cartilage and in the clear gel that fills the eyeball (the vitreous). It is essential for the normal development of bones and other tissues that form the body's supportive framework (connective tissues). Mutations in the COL2A1 gene interfere with the assembly of type II collagen molecules, which prevents bones and other connective tissues from developing properly.

How Do People Inherit Achondrogenesis?

Achondrogenesis, type 1B is inherited in an autosomal recessive pattern, which means two copies of the gene in each cell are altered. Most often, the parents of an individual with an autosomal recessive disorder are carriers of one copy of the altered gene but do not show signs and symptoms of the disorder.

Achondrogenesis, type 2 is considered an autosomal dominant disorder because one copy of the altered gene in each cell is sufficient to cause the condition. It is almost always caused by new mutations in the COL2A1 gene and typically occurs in people with no history of the disorder in their family. This condition is not passed on to the next generation because affected individuals do not live long enough to have children.

Where Can I Find Additional Information about Achondrogenesis?

You may find the following resources about achondrogenesis helpful. These materials are written for the general public.

NIH Publications - National Institutes of Health

- **http://www.niams.nih.gov/hi/topics/connective/connective.htm**

MedlinePlus - Health Information

- Encyclopedia: Achondrogenesis:
  http://www.nlm.nih.gov/medlineplus/ency/article/001247.htm
- Health Topic: Bone Diseases:
  http://www.nlm.nih.gov/medlineplus/bonediseases.html
- Health Topic: Connective Tissue Disorders:
  http://www.nlm.nih.gov/medlineplus/connectivetissuedisorders.html
- Health Topic: Dwarfism:
  http://www.nlm.nih.gov/medlineplus/dwarfism.html

Educational Resources - Information Pages

- Madisons Foundation:
  http://www.madisonsfoundation.org/content/3/1/display.asp?did=380
- Orphanet:
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=932

Patient Support - for Patients and Families

- Human Growth Foundation:
  http://www.hgfound.org/

- International Skeletal Dysplasia Registry (Cedars-Sinai Medical Center): **http://www.csmc.edu/3805.html**
- Little People of America: **http://www.lpaonline.org/**
- National Organization for Rare Disorders: **http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=Achondrogenesis**
- Resource list from the University of Kansas Medical Center: **http://www.kumc.edu/gec/support/dwarfism.html**
- The MAGIC Foundation: **http://www.magicfoundation.org/**

Professional Resources

You may also be interested in these resources, which are designed for healthcare professionals and researchers.

- Gene Reviews - Clinical summary: **http://www.genetests.org/query?dz=achon1b**
- Gene Tests - DNA tests ordered by healthcare professionals: **http://ghr.nlm.nih.gov/condition=achondrogenesis/show/Gene+Tests;jsessionid=41AB3EDA3A440943CA3C1FFA20AECFDA**
- PubMed - Recent literature: **http://ghr.nlm.nih.gov/condition=achondrogenesis/show/PubMed;jsessionid=41AB3EDA3A440943CA3C1FFA20AECFDA**
- OMIM - Genetic disorder catalog: **http://ghr.nlm.nih.gov/condition=achondrogenesis/show/OMIM;jsessionid=41AB3EDA3A440943CA3C1FFA20AECFDA**

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A summary of the genes related to achondrogenesis is provided below:

What Is the Official Name of the COL2A1 Gene?[4]

The official name of this gene is "collagen, type II, alpha 1 (primary osteoarthritis, spondyloepiphyseal dysplasia, congenital)."

COL2A1 is the gene's official symbol. The COL2A1 gene is also known by other names, listed below.

What Is the Normal Function of the COL2A1 Gene?

The COL2A1 gene provides instructions for the production of the pro-alpha1(II) chain of type II collagen. Type II collagen adds structure and strength to the tissues that support the body's muscles, joints, organs, and skin (the connective tissue). Type II collagen is found primarily in cartilage, the tissue that cushions bones and joints and makes up the flexible portions of the nose and ears. It is also part of the jelly-like substance that fills the eyeball (the vitreous), the inner ear, and the center portion of the discs between the vertebrae in the spine (nucleus pulposus).

Three pro-alpha1(II) chains twist together to form a triple-stranded, ropelike procollagen molecule. These procollagen molecules must be processed by enzymes in the cell. Once these molecules are processed, they leave the cell and arrange themselves into long, thin fibrils that cross-link to one another in the spaces around cells. The cross-linkages result in the formation of very strong, mature type II collagen fibers.

[4] Adapted from the Genetics Home Reference of the National Library of Medicine: **http://ghr.nlm.nih.gov/gene=col2a1;jsessionid=41AB3EDA3A440943CA3C1FFA20AECFDA**.

What Conditions Are Related to the COL2A1 Gene?

Achondrogenesis - Caused by Mutations in the COL2A1 Gene

Mutations in the COL2A1 gene cause a form of achondrogenesis known as type 2. Some mutations delete part of the COL2A1 gene or lead to pro-alpha1(II) chains that are missing critical segments. Other mutations change one of the protein building blocks (amino acids) used to make the pro-alpha1(II) chain. Specifically, the amino acid glycine is replaced by a different amino acid at various places in this collagen chain. All of these mutations prevent the normal production of mature type II collagen, which results in the severe skeletal abnormalities seen in this disorder.

Hypochondrogenesis - Caused by Mutations in the COL2A1 Gene

Mutations in the COL2A1 gene cause a form of achondrogenesis known as type 2. Some mutations delete part of the COL2A1 gene or lead to pro-alpha1(II) chains that are missing critical segments. Other mutations change one of the protein building blocks (amino acids) used to make the pro-alpha1(II) chain. Specifically, the amino acid glycine is replaced by a different amino acid at various places in this collagen chain. All of these mutations prevent the normal production of mature type II collagen, which results in the severe skeletal abnormalities seen in this disorder.

Kniest Dysplasia - Caused by Mutations in the COL2A1 Gene

Several different types of mutations in the COL2A1 gene are responsible for hypochondrogenesis. Some mutations delete part of the COL2A1 gene or lead to pro-alpha1(II) chains that are missing critical segments. Other mutations change one of the protein building blocks (amino acids) used to make the pro-alpha1(II) chain. Specifically, the amino acid glycine is replaced by a different amino acid at various places in this collagen chain. All of these mutations interfere with the formation of mature triple-stranded type II collagen molecules, which results in this type of hypochondrogenesis by affecting tissues that are rich in type II collagen.

Platyspondylic Lethal Skeletal Dysplasia, Torrance Type - Caused by Mutations in the COL2A1 Gene

Most of the mutations responsible for Kniest dysplasia cause abnormally short pro-alpha1(II) collagen chains to be produced in the cell. These short chains join with longer, normal-length collagen chains. The resulting abnormal type II collagen molecules are shorter than normal, causing the signs and symptoms of Kniest dysplasia, a disorder that prevents normal bone growth and development.

Spondyloepimetaphyseal Dysplasia, Strudwick Type - Caused by Mutations in the COL2A1 Gene

Fewer than 10 mutations in the COL2A1 gene have been identified in people with platyspondylic lethal skeletal dysplasia, Torrance type. Most of these mutations change a single protein building block (amino acid) in the pro-alpha1(II) chain. These COL2A1 mutations lead to the production of an abnormal version of the pro-alpha1(II) chain that cannot be incorporated into type II collagen fibers. As a result, cells make a reduced amount of type II collagen. Instead of forming collagen molecules, the abnormal pro-alpha1(II) chains build up in cartilage cells (chondrocytes). These changes disrupt normal bone development, resulting in skeletal abnormalities such as short arms and legs, a small chest, flattened vertebrae, and short fingers and toes.

Spondyloepiphyseal Dysplasia Congenita - Caused by Mutations in the COL2A1 Gene

All reported COL2A1 mutations change one of the protein building blocks (amino acids) used to make the pro-alpha1(II) chain of type II collagen. Specifically, the amino acid glycine is replaced by a different amino acid at various places in this collagen chain. The substitution of another amino acid for glycine in this chain inhibits the formation of stable, triple-stranded, ropelike collagen molecules. This alteration results in spondyloepimetaphyseal dysplasia, Strudwick type by affecting tissues that are rich in type II collagen. This disorder is characterized by abnormal bone growth and vision problems.

Spondyloperipheral Dysplasia - Caused by Mutations in the COL2A1 Gene

Spondyloepimetaphyseal dysplasia congenita can be caused by several types of mutations in the COL2A1 gene. These mutations may result in an incorrect amino acid in the pro-alpha1(II) chain. Specifically, the amino acid glycine is replaced by a different amino acid at various places in this collagen chain. Mutations may also result in the production of an abnormally short pro-alpha1(II) chain. All of these changes interfere with the formation of mature triple-stranded type II collagen molecules, which result in spondyloepimetaphyseal dysplasia congenita by affecting tissues that are rich in type II collagen. Skeletal abnormalities, vision problems, and hearing loss are the most common problems seen in this disorder.

Stickler Syndrome - Caused by Mutations in the COL2A1 Gene

Mutations that cause spondyloperipheral dysplasia lead to the production of an abnormally short pro-alpha1(II) chain that cannot be incorporated into type II collagen fibers. As a result, cells make a reduced amount of type II collagen. Instead of forming collagen molecules, the abnormal pro-alpha1(II) chains build up in cartilage cells (chondrocytes). These changes disrupt normal bone development, resulting in flattened vertebrae, short fingers and toes, and the other features of spondyloperipheral dysplasia.

Other Disorders - Associated with the COL2A1 Gene

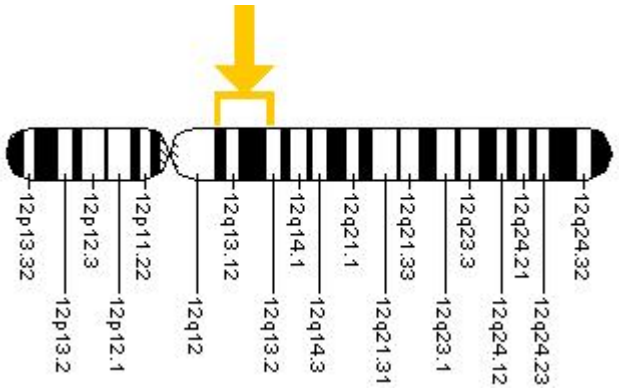
Several of the mutations in the COL2A1 gene result in the production of an abnormally short protein that cannot be incorporated into a type II collagen fiber. Other COL2A1 mutations create a premature stop signal in the instructions for making the pro-alpha1(II) chain. As a result, cells produce only half the normal amount of this collagen chain. This shortage results in an underproduction of type II collagen in cartilage and the signs and

symptoms of Stickler syndrome, namely, overly flexible joints, distinctive facial features, hearing loss, and severe nearsightedness with associated eye problems.

Where Is the COL2A1 Gene Located?

Cytogenetic Location: 12q13.11-q13.2

Molecular Location on chromosome 12: base pairs 46,653,017 to 46,684,527



The COL2A1 gene is located on the long (q) arm of chromosome 12 between positions 13.11 and 13.2.

More precisely, the COL2A1 gene is located from base pair 46,653,017 to base pair 46,684,527 on chromosome 12.

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