22Q11.2 DELETION SYNDROME

A 3-in-1 Medical Reference

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

TO INTERNET REFERENCES

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22Q11.2 DELETION SYNDROME

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

JAMES N. PARKER, M.D.
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FORWARD

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 22q11.2 deletion syndrome 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 22q11.2 deletion syndrome, 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 22q11.2 deletion syndrome, 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 22q11.2 deletion syndrome. 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 22q11.2 deletion syndrome. 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 22q11.2 deletion syndrome, 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.

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 22q11.2 deletion syndrome.

The Editors

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

CHAPTER 1. STUDIES ON 22Q11.2 DELETION SYNDROME

Overview

In this chapter, we will show you how to locate peer-reviewed references and studies on 22q11.2 deletion syndrome. For those interested in basic information about 22q11.2 deletion syndrome, 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 22q11.2 deletion syndrome 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 22q11.2 deletion syndrome is provided. [2]

The Genetics Home Reference has recently published the following summary for 22q11.2 deletion syndrome:

What Is 22q11.2 Deletion Syndrome?[3]

22q11.2 deletion syndrome is a disorder caused by the deletion of a small piece of chromosome 22. The deletion occurs near the middle of the chromosome at a location designated q11.2.

The features of this syndrome vary widely, even among affected members of the same family, and involve many parts of the body. Characteristic signs and symptoms include heart defects that are often present from birth, an opening in the roof of the mouth (a cleft palate) or other defects in the palate, recurrent infections caused by problems with the

[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=22q112deletionsyndrome**.

immune system, and mild differences in facial features. Affected individuals may also have kidney abnormalities, low levels of calcium in the blood (which can result in seizures), a decrease in blood platelets (thrombocytopenia), significant feeding difficulties, and autoimmune disorders such as rheumatoid arthritis and Graves' disease. Skeletal differences are possible, including abnormalities of the spinal bones (vertebrae), extra fingers or toes (polydactyly), and premature fusion of certain bones of the skull (craniosynostosis).

Many children with 22q11.2 deletion syndrome have developmental delays and learning disabilities. Later in life, they are at an increased risk of developing mental illnesses such as schizophrenia, depression, anxiety, and bipolar disorder. Additionally, affected children are probably more likely than children without 22q11.2 deletion syndrome to have developmental disorders (such as autism) that affect communication and social interaction.

Because the signs and symptoms of 22q11.2 deletion syndrome are so varied, different groupings of features were once described as separate conditions. Doctors named these conditions DiGeorge syndrome, velocardiofacial syndrome (also called Shprintzen syndrome), and conotruncal anomaly face syndrome. In addition, some children with the 22q11.2 deletion were diagnosed with the autosomal dominant form of Opitz G/BBB syndrome and Cayler cardiofacial syndrome. Once the genetic basis for these disorders was identified, doctors determined that they were all part of a single syndrome with many possible signs and symptoms. To avoid confusion, this condition is usually called 22q11.2 deletion syndrome, a description based on its underlying genetic cause.

How Common Is 22q11.2 Deletion Syndrome?

22q11.2 deletion syndrome affects an estimated 1 in 4,000 newborns. This condition actually may be more common, however, because some people with the deletion have few signs and symptoms and therefore may not have been diagnosed.

What Are the Genetic Changes Related to 22q11.2 Deletion Syndrome?

22q11.2 deletion syndrome is a chromosomal condition related to **chromosome 22** (**http://ghr.nlm.nih.gov/chromosome=22**).

The **COMT** (**http://ghr.nlm.nih.gov/gene=comt**) and **TBX1** (**http://ghr.nlm.nih.gov/gene=tbx1**) genes are associated with 22q11.2 deletion syndrome.

Most people with 22q11.2 deletion syndrome are missing about 3 million base pairs (the building blocks of DNA) on one copy of chromosome 22 in each cell. This region contains about 30 genes, many of which have not been well characterized. A small percentage of affected individuals have shorter deletions in the same region. This condition is often described as a contiguous gene deletion syndrome because a deletion in chromosome 22 leads to the loss of many genes that are close together.

Researchers are working to identify all of the genes that contribute to the features of 22q11.2 deletion syndrome. They have determined that the loss of a particular gene on chromosome 22, TBX1, is probably responsible for many of the syndrome's characteristic signs (such as heart defects, a cleft palate, distinctive facial features, and low calcium levels). The loss of another gene, COMT, in the same region of chromosome 22 may help explain the increased

risk of behavioral problems and mental illness. Additional genes in the deleted region likely contribute to the varied features of 22q11.2 deletion syndrome.

Where Can I Find Additional Information about 22q11.2 Deletion Syndrome?

You may find the following resources about 22q11.2 deletion syndrome helpful. These materials are written for the general public.

NIH Publications - National Institutes of Health

- National Center for Biotechnology Information: Genes and Disease: **http://www.ncbi.nlm.nih.gov/books/bv.fcgi?call=bv.View.ShowSection&rid=gnd.section.150**
- National Institute on Deafness and Other Communication Disorders: **http://www.nidcd.nih.gov/health/voice/velocario.asp**

MedlinePlus - Health Information

- Health Topic: Cleft Lip and Palate: **http://www.nlm.nih.gov/medlineplus/cleftlipandpalate.html**
- Health Topic: Congenital Heart Disease: **http://www.nlm.nih.gov/medlineplus/congenitalheartdisease.html**
- Health Topic: Immune System and Disorders: **http://www.nlm.nih.gov/medlineplus/immunesystemanddisorders.html**

Educational Resources - Information Pages

- American Heart Association: **http://www.americanheart.org/presenter.jhtml?identifier=3018193**
- California Department of Developmental Services: **http://www.ddhealthinfo.org/ggrc/doc2.asp?ParentID=5167**
- Center for Craniofacial Development and Disorders, Johns Hopkins Medicine: **http://www.hopkinsmedicine.org/craniofacial/Education/DefinedArticle.cfm?ArticleID=100&Source=Family&LayArticle=Yes**
- Children's Hospital and Regional Medical Center, Seattle, Washington: **http://craniofacial.seattlechildrens.org/conditions/velocar.asp**
- Children's Hospital of Philadelphia: **http://www.chop.edu/consumer/jsp/division/generic.jsp?id=74634**
- Cincinnati Children's Hospital Medical Center: **http://www.cincinnatichildrens.org/health/heart-encyclopedia/disease/syndrome/vcfs.htm**

- Emory University School of Medicine:
  http://www.genetics.emory.edu/pdf/Emory_Human_Genetics_Congenital_Heart_Defects_22q.PDF
- Madisons Foundation:
  http://www.madisonsfoundation.org/content/3/1/display.asp?did=64
- Orphanet:
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=567
- Swedish Information Center for Rare Diseases:
  http://www.sos.se/smkh/2003-110-6/2003-110-6.htm
- UC Davis Children's Hospital:
  http://www.ucdmc.ucdavis.edu/children/services/cleft/health/anomalies/velocardiofacial.html

Patient Support - for Patients and Families

- 22q and You Newsletter (Children's Hospital of Philadelphia):
  http://www.cbil.upenn.edu/VCFS/22qandyou/
- Chromosome Deletion Outreach:
  http://www.chromodisorder.org
- National Organization for Rare Disorders:
  http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=Velocardiofacial+Syndrome
- Resource list from the University of Kansas Medical Center:
  http://www.kumc.edu/gec/support/velo.html
- Velo-Cardio-Facial Syndrome Educational Foundation, Inc.:
  http://www.vcfsef.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=22q11deletion
- Gene Tests - DNA tests ordered by healthcare professionals:
  http://www.genetests.org/query?testid=2550
- Genetic Tools - Teaching cases:
  http://www.genetests.org/servlet/access?fcn=y&filename=/tools/cases/22q11del-17/
- ClinicalTrials.gov - Linking patients to medical research:
  http://clinicaltrials.gov/search/condition=%2222q11.2+deletion+syndrome%22?recruiting=false
- PubMed - Recent literature:
  http://ghr.nlm.nih.gov/condition=22q112deletionsyndrome/show/PubMed;jsessionid=F161A88C20724941BF724D2EDE53F5D6

- OMIM - Genetic disorder catalog: **http://ghr.nlm.nih.gov/condition=22q112deletionsyndrome/show/OMIM;jsessionid=F161A88C20724941BF724D2EDE53F5D6**

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through an affected relative: cast a wide FISHing net! Genet Med. 2001 Jan-Feb;3(1):23-9. PubMed citation

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- Sullivan KE. The clinical, immunological, and molecular spectrum of chromosome 22q11.2 deletion syndrome and DiGeorge syndrome. Curr Opin Allergy Clin Immunol. 2004 Dec;4(6):505-12. Review. PubMed citation
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- Yamagishi H, Srivastava D. Unraveling the genetic and developmental mysteries of 22q11 deletion syndrome. Trends Mol Med. 2003 Sep;9(9):383-9. Review. PubMed citation

Summaries of the chromosome and genes related to 22q11.2 deletion syndrome are provided below:

What Is Chromosome 22?[4]

Chromosome 22 is one of the 23 pairs of chromosomes in humans. People normally have two copies of this chromosome. Chromosome 22 is the second smallest human chromosome, spanning about 50 million base pairs (the building blocks of DNA) and representing between 1.5 percent and 2 percent of the total DNA in cells.

In 1999, researchers working on the Human Genome Project announced they had determined the sequence of base pairs that make up this chromosome. Chromosome 22 was the first human chromosome to be fully sequenced.

Identifying genes on each chromosome is an active area of genetic research. Because researchers use different approaches to predict the number of genes on each chromosome, the estimated number of genes varies. Chromosome 22 likely contains between 500 and 800 genes.

Genes on chromosome 22 are among the estimated 20,000 to 25,000 total genes in the human genome.

There are many genetic conditions related to genes on chromosome 22.

[4] Adapted from the Genetics Home Reference of the National Library of Medicine: **http://ghr.nlm.nih.gov/chromosome=22;jsessionid=F161A88C20724941BF724D2EDE53F5D6**.

What Chromosomal Conditions Are Related to Chromosome 22?

The following conditions are caused by changes in the structure or number of copies of chromosome 22.

22q11.2 Deletion Syndrome

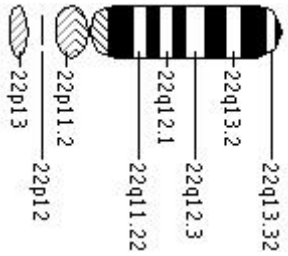
Most people with 22q11.2 deletion syndrome are missing about 3 million base pairs on one copy of chromosome 22 in each cell. The deletion occurs near the middle of the chromosome at a location designated as q11.2. This region contains about 30 genes, but many of these genes have not been well characterized. A small percentage of affected individuals have shorter deletions in the same region.

Other Chromosomal Conditions

Other changes in the number or structure of chromosome 22 can have a variety of effects. Mental retardation, delayed development, delayed or absent speech, distinctive facial features, and behavioral problems are common features. Frequent changes to chromosome 22 include an extra piece of the chromosome in each cell (partial trisomy), a missing segment of the chromosome in each cell (partial monosomy), and a circular structure called a ring chromosome 22. A ring chromosome occurs when both ends of a broken chromosome are reunited. Rearrangements (translocations) of genetic material between chromosomes can also lead to extra or missing material from chromosome 22. The most common of these translocations involves chromosomes 11 and 22.

Is There a Standard Way to Diagram CHROMOSOME 22?

Geneticists use diagrams called ideograms as a standard representation for chromosomes. Ideograms show a chromosome's relative size and its banding pattern. A banding pattern is the characteristic pattern of dark and light bands that appears when a chromosome is stained with a chemical solution and then viewed under a microscope. These bands are used to describe the location of genes on each chromosome.



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What Is the Official Name of the COMT Gene?[5]

The official name of this gene is "catechol-O-methyltransferase."

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

What Is the Normal Function of the COMT Gene?

The COMT gene provides instructions for making an enzyme called catechol-O-methyltransferase. The gene makes two versions of this enzyme. The longer form, called membrane-bound catechol-O-methyltransferase (MB-COMT), is chiefly produced by nerve cells in the brain. A shorter form, called soluble catechol-O-methyltransferase (S-COMT), functions primarily in other tissues, including the liver, kidneys, and blood.

In the brain, catechol-O-methyltransferase helps break down certain chemical messengers called neurotransmitters. These chemicals conduct signals from one nerve cell to another. Catechol-O-methyltransferase is particularly important in an area at the front of the brain (the prefrontal cortex) that organizes and coordinates information from other parts of the brain. This region is involved with personality, planning, abstract thinking, emotion, and working (short-term) memory. To function efficiently, the prefrontal cortex requires signalling by neurotransmitters such as dopamine and norepinephrine. Catechol-O-methyltransferase helps maintain appropriate levels of these neurotransmitters in this part of the brain.

What Conditions Are Related to the COMT Gene?

22q11.2 Deletion Syndrome - Associated with the COMT Gene

Most cases of 22q11.2 deletion syndrome are caused by the deletion of a small piece of chromosome 22. This region of the chromosome contains about 30 genes, including the COMT gene. As a result of the deletion, people with this disorder have only one copy of these genes in each cell instead of the usual two copies. A loss of one copy of the COMT gene leads to a reduction in the amount of catechol-O-methyltransferase. Researchers believe that reduced activity of this enzyme in the prefrontal cortex may help explain the

[5] Adapted from the Genetics Home Reference of the National Library of Medicine: **http://ghr.nlm.nih.gov/gene=comt**.