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(303) 866-6681 or (303) 866-6605

Fact Sheet

Hunter Syndrome

The primary content of this information was retrieved from: http://www.hunterpatients.com/

What is Hunter syndrome? Hunter syndrome, also known as mucopolysaccharidosis II (MPS II), is a genetic condition that primarily affects males. It is one of several related lysosomal storage diseases. The condition interferes with the body's ability to break down and recycle specific mucopolysaccarides (a group of compounds composed of protein and complex sugars), also known as glycosaminoglycans or GAG.

In Hunter syndrome, GAG builds up in cells throughout the body due to a deficiency or absence of the enzyme iduronate-2-sulfatase (I2S). This buildup interferes with the way certain cells and organs in the body function and leads to a number of serious symptoms.

What are the Symptoms of Hunter syndrome? The symptoms of Hunter syndrome (MPS II) are generally not apparent at birth, but usually begin to become noticeable after the first year of life.
Common early symptoms of Hunter syndrome may include:

√ umbilical or inguinal hernia
√ ear infections
√ runny nose
√ colds

Since these symptoms are common among many infants, they are not likely to lead a doctor to make an immediate diagnosis of Hunter syndrome. As the build-up of GAG continues throughout the cells of the body, signs of Hunter syndrome become more visible and include:

- ✓ a distinctive coarseness in facial features including a prominent forehead, a nose with a flattened bridge, and an enlarged tongue
- ✓ a large head
- ✓ an enlarged abdomen
- ✓ pebbly, ivory-colored skin lesions on upper arms, legs, and upper back
- ✓ frequent ear infections (conductive hearing loss)
- ✓ poor peripheral vision / vision loss due to build up of spinal fluid
- ✓ progressive sensorineural (inner ear) hearing loss
- ✓ respiratory infections / limited lung capacity
- ✓ cardiac (heart) problems with heart valves
- ✓ epilepsy (seizure disorder)
- ✓ breathing problems while sleeping / disorganized sleep patterns
- ✓ joints of fingers, arms, and legs held in partial flexion carpel tunnel syndrome / motor fatigue

- ✓ short stature, dysplasia, and joint stiffness
- ✓ developmental delays

The rate and progression of the symptoms may be different for each person with Hunter syndrome and there is a broad range in the type and severity of the symptoms.

What is the Biochemistry of Hunter Syndrome? The biochemistry of Hunter syndrome is related to a problem in a part of the body's connective tissue known as the extracellular matrix. This matrix is made up of a variety of sugars and proteins and helps to form the architectural framework of the body. The matrix surrounds the cells of the body in an organized meshwork and functions as the glue that holds the cells of the body together. One of the parts of the extracellular matrix is a complex molecule called a proteoglycan. Like many components of the body, proteoglycans need to be broken down and replaced. When the body breaks down proteoglycans, one of the resulting products is mucopolysaccharides, otherwise known as GAG. There are several types of GAG, each found in certain characteristic places in the body:

GAG	LOCATION IN BODY
Hyaluronic acid	Various connective tissues, skin, cartilage, synovial fluid
Chondroitin sulfate	Cartilage, cornea, bone, skin, arteries
Dermatan sulfate	Skin, blood vessels, heart, heart valves
Heparan sulfate	Lung, arteries, cell surfaces
Heparin	Lung, liver, certain immune system cells
Keratan sulfate	Cartilage, cornea, intervertebral disks

In Hunter syndrome, the problem concerns the breakdown of two GAG: (a) dermatan sulfate and (b) heparan sulfate. The first step in the breakdown of dermatan sulfate and heparan sulfate requires the lysosomal enzyme I2S. This enzyme is either partially or completely inactive in individuals with Hunter syndrome. As a result, GAG builds up in cells throughout the body, particularly in tissues that contain large amounts of dermatan sulfate and heparan sulfate. As this buildup progresses, it interferes with how certain cells and organs in the body function and results in several serious health concerns. The rate of GAG buildup is not the same for all people with Hunter syndrome, resulting in a wide spectrum of medical problems

What are the Genetics of Hunter syndrome? Hunter syndrome (MPS II) affects an estimated close to 1 in 155,000 live male births. Since Hunter syndrome is an inherited disorder (X-linked recessive) that primarily affects males, it is passed down from one generation to the next generation. Nearly every cell in the human body has 46 chromosomes, with 23 derived from each parent. The I2S gene is located on the X chromosome. Females have two X chromosomes, one inherited from each parent, whereas males have one X chromosome that they inherit from their mother and one Y chromosome inherited from their father.

If a male has an abnormal copy of the I2S gene, he will develop Hunter syndrome. A male can obtain an abnormal copy of the I2S gene in one of two ways. One way occurs when the mother is a carrier (she has one abnormal and one normal I2S gene) and she passes along the abnormal gene to her son. Another way occurs during egg and sperm formation, a mutation can develop in the I2S gene on the son's X chromosome. In this second case, the mother is not a carrier and the risk of a spontaneous mutation occurring again in a future sibling is low but not zero. Females can carry one abnormal copy of the I2S gene and are usually not affected. Hunter syndrome has been reported to occur in females.

How is Hunter syndrome Diagnosed? Many of the early signs and symptoms of Hunter syndrome are commonly seen in infants and toddlers, but the path to diagnosis often takes time. The diagnosis often

includes the following gathering information:

- ✓ Symptoms that usually appear between 2 to 4 years of age
- ✓ Visible signs and symptoms of Hunter syndrome (MPS II) are usually the first clues
- ✓ Laboratory testing results that provide additional evidence that an MPS disorder is present that may be used to refer parents to a medical geneticist for further testing
- ✓ A definitive diagnosis of Hunter syndrome is made by measuring the iduronate-2 sufatase (12S) enzyme activity through a blood test.

What are Needed Treatment and Educational Supports for Hunter Syndrome? The child will likely need to be in the ongoing care of medical practitioners to address respiratory, pain management, eye care, and orthopedic needs. Audiology evaluations should be completed on a regular basis to ensure that the child receives any needed hearing aids and/or listening devices. The educational team may require the services of a teacher trained in the area of blindness/ visual impairment and/or a teacher trained in deafness, if the student is eligible for special education services. The child may require specialized instruction, aids, and/or equipment and appropriate related services to assure access to the general education curriculum. In the event of a combined vision and hearing loss, the child will benefit from educational techniques specific to deafblindness. In the event of a combined vision and hearing loss, the child will benefit from educational techniques specific to deafblindness and should be referred to the Colorado Services for Children and Youth with Combined Vision and Hearing Loss Project.

Resources:

Brochure on Hunter syndrome can be found at:
http://www.hunterpatients.com/pdf/hunter_syndrome_brochure.pdf

Hunter Patients: http://www.hunterpatients.com/

For more information about the Colorado Services for Children and Youth with Combined Vision and Hearing Loss Project contact:

Tanni Anthony
Phone: 303-866-6681
Email: anthony_t@cde.state.co.us

Gina Quintana
Phone: 303-866-6605
Email: quintana_g@cde.state.co.us

Colorado Department of Education
Exceptional Student Leadership Unit
1560 Broadway, Suite 1175
Denver, CO 80202

Fax: 303-866-6767

Web Page Address: http://www.cde.state.co.us/cdesped/SD-DB.asp

Fact Sheets from the Colorado Services to Children and Youth with Combined Vision and Hearing Loss Project are to be used by both families and professionals serving individuals with vision and hearing loss. The information applies to children, birth through 21 years of age. The purpose of the Fact Sheet is to give general information on a specific topic. The contents of this Fact Sheet were developed under a grant from the United States Department of Education (US DOE) #H326C080044. However, these contents do not necessarily represent the policy of the US DOE and you should not assume endorsement by the Federal Government. More specific information for an individual student can be provided through personalized technical assistance available from the project. For more information call (303) 866-6681 or (303) 866-6605. Updated: 1/12