Condition Details

Learn more about the conditions on the ScreenPlus panel.

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Acid Sphingomyelinase Deficiency (ASMD), also called Niemann-Pick disease types A and B

What is it?

ASMD is a rare disease that causes the buildup of a fatty substance in parts of the body. People with ASMD-B have enlarged livers and spleens, as well as lung and liver disease that can get worse over time. The most severe form, ASMD-A, also affects the brain and usually results in death during infancy. There is also a form of ASMD that is intermediate in severity.

Gene and enzyme function

Mutations in the *SMPD1* gene cause ASMD. The *SMPD1* gene normally instructs the body to make acid sphingomyelinase, an enzyme that helps break down a fatty substance called sphingomyelin. In people with ASMD-B, there is a buildup of sphingomyelin in liver, spleen, lungs, and bone marrow; in infants with ASMD-A and in children with the intermediate form of ASMD, sphingomyelin also builds up in the brain.

More information about symptoms

People with ASMD-B (also known as Niemann Pick B) usually have large livers and spleens that are often first noted during childhood. Over time, some individuals with ASMD-B have breathing difficulties, liver disease, bleeding problems, lipid abnormalities, and weak bones. Many people with ASMD-B live into adulthood. Infants with ASMD-A (also known as Niemann Pick A) are usually diagnosed during the first year of life because they have large livers and spleens, don't gain weight well, and fall behind in their development. Most babies with ASMD-A die between 2-3 years of age. Individuals with the intermediate form have brain involvement in addition to ASMD-B symptoms, but the brain disease progresses at a slower pace than in ASMD-A.

How common is ASMD?

All forms of ASMD are extremely rare disorders, estimated to affect about 1:250,000 individuals.

How is it inherited?

All forms of ASMD are inherited in an **autosomal recessive inheritance** pattern.

Is there treatment?

This information is current as of November 2020. Decisions about if, when, and how to manage this condition should always be made in conjunction with your health care provider.

Treatment

There is currently no FDA-approved therapy for ASMD.

Clinical Trials

Click **HERE** to see **clinical trials** studying an investigational **enzyme replacement therapy** for ASMD-B.

Additional Information and Support

- Genetics Home Reference & Medline Plus (ASMD)
- National Organization for Rare Diseases (ASMD)
- National Niemann Pick Disease Foundation

Source

- Genetics Home Reference (GHR) and MedlinePlus

Ceroid Lipofuscinosis Type 2 (CLN2)

What is it?

CLN2 is a brain disease that affects young children. Children with CLN2 have seizures, slow development, problems with coordination, and vision loss. Without treatment, people with CLN2 usually do not survive past their teenage years.

Gene and enzyme function

Mutations in the *TPP1* gene cause CLN2. The *TPP1* gene normally instructs the body to make tripeptidyl peptidase 1, an enzyme that breaks down peptides (protein fragments). In people with CLN2, these peptides are not broken down, so they build up in the brain.

More information about symptoms

CLN2, also known as a form of Batten disease, typically presents in children between 2-4 years of age. The initial signs in children with CLN2 are usually seizures and difficulties with coordination. People with CLN2 also develop muscle twitches, vision loss, problems with motor skills and speech development, intellectual disability, and behavioral problems. CLN2 also causes loss of previously acquired skills. Without treatment, children with CLN2 usually do not live past their teens. Some people with CLN2 do not develop symptoms until later in childhood and may survive into adulthood.

How common is CLN2?

CLN2 is an extremely rare disorder. Its frequency in the population is not known.

How is it inherited?

CLN2 is inherited in an **autosomal recessive inheritance** pattern.

Is there treatment?

This information is current as of November 2020. Decisions about if, when, and how to manage this condition should always be made in conjunction with your health care provider.

Treatment

There is an FDA-approved enzyme replacement therapy for people with CLN2.

Click **HERE** for more information about CLN2 treatment.

Clinical trials

Click **HERE** to see **clinical trials** evaluating investigational **enzyme replacement therapy** and **gene therapy** for CLN2.

Additional Information and Support

- Genetics Home Reference & Medline Plus (CLN2)
- Batten Disease Support and Research Association

Source

- Genetics Home Reference (GHR) and MedlinePlus

Cerebrotendinous Xanthomatosis (CTX)

What is it?

People with CTX have abnormal storage of fats in different parts of the body resulting in neurological symptoms that usually start in early adulthood. Other CTX symptoms such as chronic diarrhea and corneal clouding usually begin during childhood.

Gene and enzyme function

Mutations in the *CYP27A1* gene cause CTX. The *CYP27A1* gene normally instructs the body to make sterol 27-hydroxylase, an enzyme that helps break down cholesterol. In people with CTX, cholesterol and another fatty substance, cholestanol, accumulate as fatty nodules called xanthomas. The xanthomas are most commonly found in the brain and tendons, which connect muscle to bone.

More information about symptoms

People with CTX may develop chronic diarrhea and cataracts during childhood. Nerve problems typically develop in early adulthood and include seizures, movement disorders, speech impairment, loss of sensation in arms and legs, decline in intellectual function, hallucinations, and depression. Xanthomas (abnormal yellowish fatty deposits in the skin) typically develop during adolescence or early adulthood and are most commonly found in the tendons of the hands, elbows, knees, neck, and heels. They may interfere with flexibility at these locations. People with CTX also have increased risk for heart disease.

How common is CTX?

CTX is an extremely rare disorder, estimated to affect 1:1,000,000 individuals.

How is CTX inherited?

CTX is inherited in an **autosomal recessive inheritance** pattern.

Is there treatment?

This information is current as of November 2020. Decisions about if, when, and how to manage this condition should always be made in conjunction with your health care provider.

Treatment

There is an FDA-approved treatment for people with CTX

Click **HERE** for more information about CTX treatment

Clinical trials

Click **HERE** to see **clinical trials** evaluating an investigational medication for CTX.

Additional Information and Support

- Genetics Home Reference and Medline Plus (CTX)
- National Organization for Rare Diseases (CTX)
- United Leukodystrophy Foundation

Source

- Genetics Home Reference (GHR) and MedlinePlus

Fabry Disease

What is it?

Fabry disease is a rare disorder that causes a buildup of a fatty substance in the parts of the body. Buildup of this substance in the heart, kidneys, and blood vessels may cause life-threatening complications including heart attacks, kidney disease, and stroke.

Gene and enzyme function

Mutations in the *GLA* gene cause Fabry disease. The *GLA* gene normally instructs the body to make alpha-galactosidase A, an enzyme that breaks down a fatty substance known as globotriaosylceramide. In people with Fabry disease, there is buildup of globotriaosylceramide in the heart, kidneys, nerves, and small blood vessels.

More information about symptoms

The classic form of Fabry disease causes kidney disease, heart disease, and strokes, which typically appear in early adulthood. Many boys with Fabry disease develop pain in the hands and feet during childhood, a characteristic skin rash, decreased ability to sweat, cloudiness in the eye's cornea, gastrointestinal problems, and hearing loss. Less severe forms of Fabry disease may appear during adulthood and affect mainly the heart or kidneys.

How common is Fabry disease?

Fabry disease is a rare disorder, estimated to affect 1:40,000 – 60,000 males. The prevalence in females is unknown. It is more common and usually more severe in males.

How is Fabry disease inherited?

Fabry disease is inherited in an **X-linked inheritance** pattern. Fabry disease in females can range from asymptomatic to as severe as classically-affected males.

Is there treatment?

This information is current as of November 2020. Decisions about if, when, and how to manage this condition should always be made in conjunction with your health care provider.

Treatment

There is FDA-approved **enzyme replacement therapy** and an FDA-approved **pharmacologic chaperone therapy** for people with Fabry disease who have specific GLA mutations. Your health care provider might also consider additional therapies to help control symptoms.

Click **HERE** for more information about Fabry disease treatment.

- Fabrazyme
- Migalastat

Clinical trials

Click **HERE** to see **clinical trials** studying investigational **enzyme replacement therapy**, **substrate reduction therapies**, and **gene therapies** for Fabry disease.

Additional Information and Support

- Genetics Home Reference and Medline Plus (Fabry Disease)
- National Organization for Rare Diseases (Fabry Disease)
- Fabry International Network
- Fabry Support and Information Group
- National Fabry Disease Foundation

Source

- Genetics Home Reference (GHR) and MedlinePlus

Gaucher Disease

What is it?

Gaucher disease is a rare disease that results in the accumulation of a fatty substance in parts of the body. People with the most common type of Gaucher disease, type 1, may have enlarged livers and spleens, bone disease, and bleeding issues. Type 2 Gaucher disease also involves progressive neurologic disease and is usually fatal in early childhood. Children with type 3 Gaucher disease have disease that is intermediate in severity between types 1 and 2.

Gene and enzyme function

Mutations in the *GBA* gene cause Gaucher disease. The *GBA* gene normally instructs the body to make beta-glucocerebrosidase, an enzyme that breaks down a fatty substance known as glucocerebroside. In people with type 1 Gaucher disease, there is buildup of glucocerebroside in the liver, spleen, and bone marrow. People with type 2 and 3 Gaucher also have glucocerebroside buildup in the brain.

More information about symptoms

People with type 1 Gaucher disease can show symptoms anytime from childhood to adulthood. Symptoms can range from mild to severe and include enlarged liver and spleen, anemia, easy bruising, and bone problems including fractures and pain. People with type 2 or 3 Gaucher disease have additional disease in the brain. Symptoms include abnormal eye movements, seizures, and muscle problems. Type 2 is more severe and presents in infancy, while type 3 presents in childhood and tends to progress more slowly. Unfortunately, most babies with type 2 Gaucher disease die between 1-3 years of age. The very severe, perinatal lethal form causes life-threatening complications before birth, and infants may survive for only a few days. People with the rare cardiovascular form primarily have disease in the heart but may also have eye abnormalities, bone problems, and an enlarged spleen. People with this form may survive into adulthood.

How common is Gaucher disease?

Gaucher disease is a rare disorder, estimated to affect 1:50,000 – 100,000 individuals. Type 1 Gaucher disease is more common in individuals of Ashkenazi Jewish origin.

How is Gaucher disease inherited?

Gaucher disease is inherited in an **autosomal recessive inheritance** pattern.

Is there treatment?

This information is current as of November 2020. Decisions about if, when, and how to manage this condition should always be made in conjunction with your health care provider.

Treatment

There are FDA-approved **enzyme replacement therapies** and **substrate reduction therapies** for people with type 1 Gaucher disease. Unfortunately, there are no approved treatments for the brain manifestations of types 2 and 3 Gaucher disease.

Click **HERE** for more information Gaucher disease treatment

- Cerezyme® (imiglucerase)
- VPRIV® (velaglucerase alpha)
- Elelyso® (taliglucerase alpha)
- Cerdelga® (eliglustat)
- Zavesca® (miglustat)

Clinical trials

Click HERE to see **clinical trials** studying investigational treatments for Gaucher disease. Therapies being studied include **gene therapy** and **substrate reduction therapy**.

Additional Information and Support

- Genetics Home Reference & Medline Plus (Gaucher Disease)
- National Organization for Rare Diseases (Gaucher Disease)
- National Gaucher Foundation

Source

- Genetics Home Reference (GHR) and MedlinePlus

GM1 Gangliosidosis

What is it?

GM1 gangliosidosis is a rare disorder that causes progressive developmental disabilities, skeletal abnormalities, enlarged liver and spleen, vision loss, bone disease, and distinctive facial features. Children with the most severe form of GM1 gangliosidosis usually do not survive past early childhood. There are also less severe forms of GM1 gangliosidosis that progress more slowly.

Gene and enzyme function

GM1 gangliosidosis results from mutations in the *GLB1* gene, which provides instructions for making beta-galactosidase. Beta-galactosidase is an enzyme that normally breaks down a fatty substance, GM1 ganglioside. When beta-galactosidase is deficient, GM1 ganglioside builds up in the cells in the brain, liver, spleen, and other organs. This toxic buildup causes disease symptoms.

More information about symptoms

The most common form of GM1 gangliosidosis is Type 1, which is usually diagnosed in the first year of life when babies are noted to have developmental delays which progressively worsen. Over time, infants with type 1 GM1 gangliosidosis develop enlarged livers and spleens, seizures, and skeletal disease. Some also develop "coarse" facial features and heart disease, and most do not survive past early childhood. Type 2 GM1 gangliosidosis may present during late infancy or early childhood, with a slower progression. The least severe form is Type 3 GM1 gangliosidosis, which may present during the teenage years.

How common is GM1 gangliosidosis?

GMI gangliosidosis is a very rare disorder, and is estimated to occur in 1:100,000 to 1:200,000 newborns.

How is GM1 gangliosidosis inherited?

GMI gangliosidosis is inherited in an **autosomal recessive** manner.

Is there treatment?

This information is current as of November 2020. Decisions about if, when, and how to manage this condition should always be made in conjunction with your health care provider.

Treatment

There is currently no FDA approved treatment for GM1 gangliosidosis

Clinical Trials

Click **HERE** to see **clinical trials** studying investigational **gene therapy** for GM1 gangliosidosis.

Additional Information and Support

- Genetics Home Reference & Medline Plus (GM1 Gangliosidosis)
- Cure GM1
- Hunter's Hope

Source

- Genetics Home Reference (GHR) and MedlinePlus

Lysosomal Acid Lipase Deficiency (LAL-D)

What is it?

People with LAL-D are missing an enzyme that helps break down fats. LAL-D causes disease in the liver, which can be life-threatening in infants. A more common, less severe form of LAL-D can begin later in life.

Gene and enzyme function

Mutations in the *LIPA* gene cause LAL-D. The *LIPA* gene normally instructs the body to make lysosomal acid lipase, an enzyme that breaks down fats. In people with LAL-D, there is buildup of fats in the liver.

More information about symptoms

Babies with the severe form of LAL-D (also known as Wolman disease) have enlarged livers and spleens, poor weight gain, jaundice, and gastrointestinal problems, including fat in the stool. Babies develop liver disease, multi-organ failure, and severe malnutrition and generally do not survive past one year. Babies with the severe form may also have calcium deposits in the adrenal glands, anemia, and developmental delay. In the later-onset form of LAL-D (also known as cholesteryl ester storage disease) symptoms can appear anytime from childhood to adulthood. People with later-onset LAL-D generally have enlarged livers and may also have enlarged spleens, liver disease, gastrointestinal problems, and increased risk for heart attack and stroke.

How common is LAL-D?

LAL-D is a very rare disorder, estimated to affect 1:40,000 – 300,000 individuals. The later-onset form is more common.

How is LAL-D inherited?

LAL-D is inherited in an **autosomal recessive inheritance** pattern.

Is there treatment?

This information is current as of November 2020. Decisions about if, when, and how to manage this condition should always be made in conjunction with your health care provider.

Treatment

There is an FDA-approved **enzyme replacement therapy** for people with LAL-D.

Click **HERE** for more information about LAL-D treatment

Clinical trials

Click **HERE** to see if there are any **clinical trials** studying investigational treatments for LAL-D

Additional Information and Support

- Genetics Home Reference & Medline Plus (LAL-D)
- National Organization for Rare Disorders (LAL-D)
- American Liver Foundation
- LAL-D Aware

Source

- Genetics Home Reference (GHR) and MedlinePlus

Metachromatic Leukodystrophy (MLD)

What is it?

Metachromatic leukodystrophy due to arylsulfatase A deficiency is a rare, progressive neurologic disease that causes intellectual disability, loss of motor skills, and other abnormalities. The most common form of MLD is usually fatal in childhood. Milder forms can present anytime from childhood to adulthood.

Gene and enzyme function

Most people with MLD due to arylsulfatase A deficiency have mutations in the *ARSA* gene. The *ARSA* gene normally instructs the body to make arylsulfatase A, an enzyme that breaks down fats known as sulfatides. In people with MLD, there is a toxic buildup of sulfatides in the brain that causes the clinical signs and symptoms. A small number of people with MLD have mutations in the *PSAP* gene, not the *ARSA* gene. ScreenPlus will not detect MLD caused by mutations in *PSAP*.

More information about symptoms

People with MLD experience progressive decline in nerve function, resulting in progressive loss of intellectual function and motor skills. Additional features may include loss of sensation in the arms and legs, incontinence, seizures, paralysis, loss of speech, blindness, and hearing loss. Babies with the most common form of MLD, the late infantile form, generally do not survive past childhood. The juvenile and adult onset forms of MLD can present with difficulty talking, seizures, as well as behavior and personality changes.

How common is MLD?

MLD is a rare disorder, estimated to affect 1:40,000 – 160,000 individuals.

How is MLD inherited?

MLD is inherited in an **autosomal recessive inheritance** pattern.

Is there treatment?

This information is current as of November 2020. Decisions about if, when, and how to manage this condition should always be made in conjunction with your health care provider.

Treatment

Some people with MLD are currently treated with **bone marrow transplantation**.

Clinical trials

Click **HERE** to see **clinical trials** studying an investigational **enzyme replacement therapy** and an investigational **gene therapy** for MLD.

Additional information and support

- Genetics Home Reference & Medline Plus (MLD)
- National Organization for Rare Disorders (MLD)
- Cure MLD
- MLD Foundation
- Hunter's Hope
- United Leukodystrophy Foundation

Source

- Genetics Home Reference (GHR) and MedlinePlus

Mucopolysaccharidosis Type II (MPS II), also called Hunter Syndrome

What is it?

MPS II is a rare disorder in which complex sugar molecules build up in many parts of the body. Individuals with MPS II usually have developmental disabilities, a distinctive facial appearance, and significant health issues. Without treatment, children with the severe form of MPS II do not survive.

Gene and enzyme function

Mutations in the *IDS* gene cause MPS II. The *IDS* gene normally instructs the body to make iduronate 2-sulfatase, an enzyme that breaks down large sugar molecules known as glycosaminoglycans (GAGs). In people with MPS II, GAGs accumulate in the liver, spleen, heart, skeleton, skin, lungs, and brain, resulting in disease symptoms.

More information about symptoms

People with the more severe form of MPS II are usually diagnosed during infancy. They have distinctive "coarse" facial features, a narrowed airway, enlarged livers and spleens, umbilical or inguinal hernias, short stature, a particular skin rash, and bone disease. They may also have reduced vision, hearing loss, lung disease, heart disease, developmental disability, and behavioral disturbances. People with the less severe form of MPS II may have a slower disease progression.

How common is MPS II?

MPS II is a very rare disorder, estimated to affect 1:100,000 – 170,000 males.

How is MPS II inherited?

MPS II is inherited in an **X-linked recessive inheritance** pattern.

Is there treatment?

This information is current as of November 2020. Decisions about if, when, and how to manage this condition should always be made in conjunction with your health care provider.

Treatment

There is an FDA-approved enzyme replacement therapy that improves some of the symptoms of MPS II, but does not treat the neurologic features.

Click **HERE** for more information about MPS II treatment

Clinical trials

Click **HERE** to see **clinical trials** studying an investigational **enzyme replacement therapy** and an investigational **gene therapy** for MPSII.

Additional information and support

- Genetics Home Reference & Medline Plus (MPS-II)
- National Organization for Rare Disorders (MPS-II)
- National MPS Society

Source

- Genetics Home Reference (GHR) and MedlinePlus

Mucopolysaccharidosis Type IIIB (MPS IIIB), also called Sanfilippo Syndrome Type IIIB

What is it?

MPS IIIB is a rare disorder in which complex sugar molecules build up in the brain, causing significant developmental disabilities and behavioral issues which tend to worsen over time.

Gene and enzyme function

Mutations in the *NAGLU* gene cause MPS IIIB. The *NAGLU* gene normally instructs the body to make alpha-N-acetylglucosaminidase, an enzyme that breaks down large sugar molecules known as glycosaminoglycans (GAGs). In people with MPS IIIB, there is buildup of GAGs in the brain and spinal cord.

More information about symptoms

Children with MPS IIIB are usually diagnosed during the preschool years. Common features include delayed speech, behavioral problems, sleep problems, and intellectual disability. Autistic features and seizures may also develop, and children with MPS IIIB may progressively lose some motor skills as they get older. Some children with MPS IIIB have mildly coarse facial features, enlarged liver, vision and hearing loss, and umbilical or inguinal hernias.

How common is it?

MPS IIIB is a rare disorder, estimated to affect fewer than 1:70,000 individuals.

How is MPS IIIb inherited?

MPS IIIB is inherited in an **autosomal recessive inheritance** pattern.

Is there treatment?

This information is current as of November 2020. Decisions about if, when, and how to manage this condition should always be made in conjunction with your health care provider.

Treatment

There is currently no FDA approved treatment for MPS IIIB.

Clinical trials

Click **HERE** to see **clinical trials** studying an investigational **enzyme replacement therapy** and an investigational **gene therapy** for MPS IIIB.

Additional information and support

- Genetics Home Reference & Medline Plus (MPS-IIIB)
- National Organization for Rare Disorders (MPS-IIIB)
- National MPS Society
- Cure Sanfilippo Foundation

Source

- Genetics Home Reference (GHR) and MedlinePlus

Mucopolysaccharidosis Type IVA (MPS IVA), also called Morquio Syndrome Type IVA

What is it?

MPS IVA is a rare disorder in which complex sugar molecules build up mainly in the skeleton, causing progressive bone disease, short stature, and other skeletal abnormalities.

Gene and enzyme function

Mutations in the *GALNS* gene cause MPS IVA. The *GALNS* gene normally instructs the body to make N-acetylgalactosamine 6-sulfatase, an enzyme that breaks down large sugar molecules known as glycosaminoglycans (GAGs). In people with MPS IVA, there is buildup of GAGs mainly in the bones.

More information about symptoms

MPS IVA is usually diagnosed during infancy because of bone disease, which typically causes malformations of the spine, joints, and short stature. Some children with MPS IVA also have heart disease, clouding in the eyes' corneas, mildly enlarged liver, narrowed airway, umbilical or inguinal hernias, and hearing loss. MPS IVA does not usually affect intelligence. Those with severe symptoms may only survive until late childhood or adolescence, while those with milder symptoms may survive into adulthood.

How common is MPS IVA?

MPS IV is an extremely rare disorder, estimated to affect 1:200,000 – 300,000 individuals.

How is MPS IVA inherited?

MPS IVA is inherited in an **autosomal recessive inheritance** pattern.

Is there treatment?

This information is current as of November 2020. Decisions about if, when, and how to manage this condition should always be made in conjunction with your health care provider.

Treatment

There is an FDA-approved **enzyme replacement therapy** for MPS IVA.

Click **HERE** for more information about treatment for MPS IVA

Clinical trials

Click **HERE** to see if there are any new **clinical trials** for MPS IVA

Additional information and support

- Genetics Home Reference & Medline Plus (MPS-IVA)
- National Organization for Rare Disorders (MPS-IVA)
- National MPS Society

Source

- Genetics Home Reference (GHR) and MedlinePlus

Mucopolysaccharidosis Type VI (MPS VI), also called Maroteaux-Lamy Syndrome

What is it?

MPS VI is a rare disorder in which complex sugar molecules build up in many parts of the body. Individuals with MPS VI usually have a distinctive appearance and disease involvement in many parts of the body, including potentially life-threatening changes in the heart and airway. People with MPS VI usually have normal intelligence. Without treatment, children with the most severe form of MPSVI do not survive into adulthood.

Gene and enzyme function

Mutations in the *ARSB* gene cause MPS VI. The *ARSB* gene normally instructs the body to make arylsulfatase B, an enzyme that breaks down large sugar molecules known as glycosaminoglycans (GAGs). In people with MPS VI, there is buildup of GAGs in different tissues in the body.

More information about symptoms

Children with the more severe form of MPS VI are usually diagnosed during early childhood. They usually have bone disease, short stature, joint deformities, “coarse” facial features, and enlarged livers and spleens. They may also have a narrowed airway, umbilical or inguinal hernias, reduced vision, hearing loss, and heart disease. MPS VI does not affect intelligence. Those with severe symptoms may only survive until late childhood or adolescence, while those with milder symptoms may survive into adulthood.

How common is MPS VI?

MPS VI is an extremely rare disorder, estimated to affect 1:250,000 – 600,000 individuals.

How is MPS VI inherited?

MPS VI is inherited in an **autosomal recessive inheritance** pattern.

Is there treatment?

This information is current as of November 2020. Decisions about if, when, and how to manage this condition should always be made in conjunction with your health care provider.

Treatment

There is an FDA-approved enzyme replacement therapy for MPS VI.

Click **HERE** for more information about treatment for MPS VI.

Clinical trials

Click **HERE** to see if there are any investigational **clinical trials** for MPSVI.

Additional Information and Support

- Genetics Home Reference & Medline Plus (MPS-VI)
- National Organization for Rare Disorders (MPS-VI)
- National MPS Society

Source

- Genetics Home Reference (GHR) and MedlinePlus

Mucopolysaccharidosis Type VII (MPS VII), also called Sly Syndrome

What is it?

MPS VII is a rare disorder in which complex sugar molecules build up in many parts of the body. Individuals with MPS VII usually have a distinctive appearance and disease involvement in many parts of the body, including potentially life-threatening changes in the heart and airway. Many individuals with MPS VII have some intellectual disability.

Gene and enzyme function

Mutations in the *GUSB* gene cause MPS VII. The *GUSB* gene normally instructs the body to make beta-glucuronidase, an enzyme that breaks down large sugar molecules known as glycosaminoglycans (GAGs). In people with MPS VII, there is buildup of GAGs is different tissues in the body.

More information about symptoms

Babies with the most severe form of MPS VII have excessive fluids in the body, and usually die soon after birth. Other children with MPS VII develop symptoms during early childhood, which may include coarse facial features, enlarged livers and spleens, heart disease, umbilical or inguinal hernias, short stature, and bone disease. They may also have a narrowed airway, reduced vision, and hearing loss. Many individuals with MPS VII also have developmental disabilities, although some have relatively normal intelligence.

How common is MPS VII?

MPS VII is one of the rarest types of MPS. It is estimated to affect 1:250,000 individuals.

How is MPS VII inherited?

MPS VII is inherited in an **autosomal recessive inheritance** pattern.

Is there treatment?

This information is current as of November 2020. Decisions about if, when, and how to manage this condition should always be made in conjunction with your health care provider.

Treatment

There is an FDA-approved **enzyme replacement therapy** for MPS VII.

Click **HERE** for more information about treatment for MPS VII

Clinical trials

Click **HERE** to see if there are any investigational **clinical trials** for MPSVII

Additional Information and Support

- Genetic Home Reference & Medline Plus (MPS-VII)
- National Organization for Rare Disorders (MPS-VII)
- National MPS Society

Source

- Genetics Home Reference (GHR) and MedlinePlus

Niemann-Pick Disease Type C (NPC)

What is it?

NPC is a rare disorder that causes a buildup of cholesterol and other fatty substances in parts of the body. Most people with NPC have neurologic disease that may include progressive developmental delays, unsteady gait, seizures, swallowing difficulties, and psychiatric symptoms. NPC may also cause liver and lung disease.

Gene and transporter function

Mutations in the *NPC1* gene or *NPC2* gene cause NPC (type C1 and C2, respectively). The *NPC1* and *NPC2* genes normally instruct the body to make NPC intracellular cholesterol transporter 1 and NPC intracellular cholesterol transporter 2, proteins that help move cholesterol and other fats within cells. In people with NPC, there is abnormal buildup of cholesterol and fats in the liver, spleen, and brain, resulting in the clinical signs and symptoms.

More information about symptoms

While NPC may vary from person to person, common features of NPC include vertical gaze palsy, enlarged liver and spleen. Over time, individuals with NPC can develop problems with coordination, speech and swallowing, intellectual disability, dementia, seizures, liver disease, and lung disease. Signs of NPC can present from infancy through adulthood, and survival depends on severity and onset of symptoms.

How common is NPC?

NPC is a very rare disorder, estimated to affect 1:150,000 individuals.

How is NPC inherited?

NPC is inherited in an **autosomal recessive inheritance** pattern.

Is there treatment?

This information is current as of November 2020. Decisions about if, when, and how to manage this condition should always be made in conjunction with your health care provider.

Treatment

There is currently no FDA approved treatment for NPC

Clinical trials

Click **HERE** for information about **clinical trials** for NPC, which are studying a variety of treatment approaches.

Additional Information and Support

- Genetics Home Reference & Medline Plus (NPC)
- National Organization for Rare Disorders (NPC)
- Firefly Fund
- National Niemann Pick Disease Foundation
- Ara Parseghian Medical Research Fund

Source

- Genetics Home Reference (GHR) and MedlinePlus

Term Definitions

Autosomal recessive: Most genes in the human body have two copies, one inherited from each parent. **Autosomal recessive inheritance** means that *both* copies of a certain gene have abnormalities ("mutations") that affect how the gene functions, which may result in disease symptoms.
(link to https://ghr.nlm.nih.gov/primer/inheritance/inheritancepatterns)

X-linked disorders are caused by abnormalities ("mutations") in genes on the X chromosome. Most males have one X chromosome and one Y chromosome (XY). Because they have only one X chromosome, a mutation on in a disease gene on the X-chromosome will cause disease symptoms. Most females, on the other hand, have two X chromosomes (XX), so a mutation in a disease gene on one of the X chromosomes will be balanced by the healthy copy of the gene on the other X chromosome. In general, for X-linked disorders, males are more likely to show disease symptoms than biologic females.

Clinical trials are research studies that test a specific intervention. A clinical trial that is studying a new investigational drug must demonstrate that it is safe and effective before it can be approved by the United States Food and Drug Administration (FDA).

Enzyme replacement therapy involves replacing the specific enzyme that is not functioning properly in a particular disease. Most enzyme replacement therapies are given intravenously, although some are delivered directly into the central nervous system.

Substrate reduction therapy is a way to reduce the toxic buildup of a substance by blocking its production. Most substrate reduction therapies are oral drugs.

Gene therapy is a way to replace an abnormal disease gene with a healthy copy that functions properly. Some newer gene therapies work by inactivating a mutated gene as a way to reduce the disease manifestations. Because gene therapy is a relatively new field of medicine, many gene therapies are still under clinical investigation.

Pharmacologic chaperone therapy is a way to treat certain disorders using drugs that support or stabilize the structure of an otherwise abnormal protein, enabling it to work better.

Bone marrow transplantation is a medical procedure to replace a person's bone marrow in an attempt to cure certain rare, life-threatening diseases.