Metabolic Diseases

Abstract:

A metabolic disease is a disease that disrupts normal metabolism. Metabolism is the process of converting food into energy on a cellular level and there are thousands of enzymes needed to carry out the process of metabolism. Metabolism involves the processing or transport of proteins (amino acids), carbohydrates (sugars and starches), or lipids (fatty acids), and when this process is disrupted it can lead to disease. As a result, some people may produce too much or too little of an enzyme to remain healthy. Metabolism creates the energy that allows the body to grow, reproduce, repair damage, and respond to the environment. Metabolic diseases weaken these actions in many ways, and this leads to broken links within the normal process of metabolizing foods for energy. There are more than 1,300 known metabolic diseases.

Learning Objectives:

1. Explain what a metabolic disease is.
2. List some of the metabolic diseases.
3. Describe the difference between metabolic disease and metabolic syndrome.
4. Identify symptoms associated with metabolic diseases.

Introduction

Metabolism is all chemical reactions involved in maintaining the living state of the cells within the body. There are two categories of metabolism within the human body. First, is catabolism, which is producing energy by breaking down larger molecules into smaller ones, such as breaking down a carbohydrate into glucose. Secondly, anabolism is the synthesis of all compounds needed to build new cells, maintain body tissues, and store energy. The interruption of these processes can cause havoc on the body and lead to metabolic disease.

Metabolic Diseases

Food is broken down in a series of steps by cellular enzymes into products with a different biochemical structures. These products then become the substrate or the next step for the next enzyme in a metabolic pathway. If an enzyme is missing or does not work properly, the pathway is broken and this inability to break products down properly leads to metabolic diseases. Low activity of a certain enzyme could result in the accumulation of that enzyme and at high levels can be toxic to the body. When this happens enzyme pathways that are normally less active may become activated forming atypical, potentially toxic byproducts. Each cell in the body contains thousands of metabolic pathways, if a process is interrupted or changed the cascading effect can be detrimental to many processes that follow. The consequences of these metabolic interruptions may be severe; intellectual disability, seizures, decreased muscle tone, organ failure, blindness, and deafness may occur, depending on which enzyme is dysfunctional.

Causes of Metabolic Disease

Metabolism is a complex process that involves many biochemicals, tissues, and organs. This means there are many opportunities for something to go wrong and cause a metabolic disorder. Some examples of causes are:

1. *Genetics*: Genes can influence metabolic processes in a variety of ways. Clients with Gaucher's disease have a genetic mutation that limits the

production of glucocerebrosidase, leading to a buildup of fat around the body.

2. *Organ dysfunction:* Organs involved in metabolism can fail to function properly. Clients with diabetes can occur when the pancreas is unable to produce sufficient insulin to regulate blood glucose levels.
3. *Mitochondrial dysfunction:* Mitochondria are small parts of cells that primarily produce energy. Mutations of the mitochondria can affect how well mitochondria function and how much energy they can produce.

Metabolic disorders do not always have a clear cause. Type 1 diabetes results from the immune system mistakenly attacking cells in the pancreas. It is still unclear what caused this problem with the immune system in the first place.

Metabolic Syndrome vs Metabolic Disease

Metabolic syndrome is a group of conditions that together raise the risk of coronary heart disease, diabetes, stroke, and other serious health problems. Metabolic syndrome is also called insulin resistance syndrome. You may have metabolic syndrome if you have three or more of the following conditions.

1. A large waistline: Extra fat in your stomach area is a bigger risk factor for heart disease than extra fat in other parts of your body.
2. High blood pressure: If blood pressure rises and stays high for a long period of time, it can damage the cardiovascular system including the heart and blood vessels. High blood pressure can also cause plaque to accumulate in your vessels. Plaque can cause heart and blood vessel diseases which can lead to a heart attack or stroke.
3. High blood sugar levels: Large sugar molecules can damage your blood vessels and raise the risk of blood clots. Blood clots can lead to heart and blood vessel diseases.

4. High blood triglycerides: Triglycerides are a type of fat found in the blood. High levels of triglycerides normally raise the levels of LDL cholesterol (bad cholesterol) which increases the risk of heart disease.
5. Low HDL cholesterol (good cholesterol) allows LDL cholesterol to settle in the blood vessels and form plaque. Having adequate HDL blood cholesterol levels is important because they remove LDL cholesterol from the blood vessels and protect the vessels from plaque buildup.

Metabolic syndrome is common in the United States. About 1 in 3 adults have metabolic syndrome. Metabolic syndrome is preventable by keeping weight within a healthy range, getting adequate exercise, having a healthy diet consisting of fruits and vegetables, and not smoking tobacco.

A metabolic disease occurs when chemical reactions, that are different from the normal reactions in your body, disrupt the normal process of metabolism. This could mean there are too many of some substances or enzymes and not enough of others, throwing off the body's normal balance. This affects the breakdown of amino acids, carbohydrates, or lipids. Another group, mitochondrial diseases, affects the parts of the cells that produce energy for the body. Metabolic disease occurs when some organs, such as your liver or pancreas, become diseased or do not function normally.

Types of Metabolic Diseases

There are more than 1,300 known metabolic diseases as stated earlier but we will focus on a few. Metabolic diseases can be congenital, acquired, or both. Below is a list of just some of the metabolic diseases.

Congenital:

1. Fabry disease
2. Phenylketonuria (PKU)
3. Tay-Sachs disease
4. Pompe disease
5. Niemann-Pick disease
6. Morquio syndrome

7. Hunter syndrome
8. Lesh-Nyhan syndrome
9. Hurler syndrome
10. Hartnup
11. Gaucher disease

Congenital and Acquired:

1. Diabetes
2. Obesity
3. Dyslipidemia
4. Hyperthyroidism
5. Hypoparathyroidism
6. Hypothyroidism
7. Cushing's syndrome
8. Hemochromatosis
9. Hyperparathyroidism

Fabry Disease (congenital)

Fabry disease is a lack of or non-functioning enzymes needed to metabolize lipids, fat-like substances. It narrows your blood vessels, which leads to issues with the skin, kidneys, heart, brain, and nervous system.

Type I and Type II Diabetes (congenital/acquired)

There are two main types of diabetes, Type I and Type II. Type II diabetes is more common than Type I diabetes. In both I and II diabetes, the body has a deficiency or a lack of a hormone called insulin. Insulin helps move glucose/blood sugar into the cells where it can be utilized for energy for the body. The blood sugar level of a client with Type I or type II diabetes becomes elevated either due to a lack of insulin production or the body is resistant to the insulin that is being produced by the pancreas. When blood glucose is not controlled, serious short-term and long-term complications are likely to occur.

Type 1 diabetes is an autoimmune disorder and is also referred to as insulin-dependent diabetes. It is an autoimmune disorder because the

pancreas is damaged by the body's own antibodies and leads to little, or no production of insulin needed to carry on the normal process of turning blood glucose into energy for the body. Juvenile-onset diabetes is another name for type 1 diabetes because the onset is usually in childhood. It is believed that genetics might play a role in type 1 diabetes as well as issues with the cells in the pancreas called Beta Islet Cells that produce insulin.

Tay-Sachs Disease (congenital)

Tay-Sachs is a rare genetic disorder passed in the HEXA gene from both parents. Tay-Sachs disease is typically found in people with certain ancestries, such as Eastern European Jews. There is an absence of an enzyme that helps break down fatty substances. When these fatty substances build up to toxic levels in the brain and spinal cord, they affect the function of the nerve cells. Tay-Sachs disease is usually fatal by around 3 to 5 years of age. Rarer types of Tay-Sachs disease start later in childhood or early adulthood.

Hurler Syndrome (congenital)

Hurler syndrome causes an abnormal accumulation of complex sugars (sugar molecules that are strung together in long, complex chains) in their cells, which affects many body systems. Hurler syndrome is divided into two subtypes, "severe" and "attenuated" (less severe). Hurler syndrome is just one of around 50 diseases classified as lysosomal storage disorders (LSD). These disorders have genetic variations that disrupt the normal activity of lysosomes (enzymes) in the cell that degrade and recycle molecules such as fats and sugars.

Niemann-Pick Disease (congenital)

Niemann-Pick diseases cause infants to develop enlarged liver, difficulty feeding, and extensive nerve destruction. Profound brain damage occurs by six months of age and infants rarely live beyond 18 months.

Morquio Syndrome (congenital)

Morquio syndrome is a rare genetic condition caused by a gene mutation that affects a child's bones and spine, organs, and physical abilities. Children with this condition are missing or do not produce enough of the enzymes that break down sugar chains naturally produced in the body. Both parents must have the recessive gene to pass it on to their children.

The life expectancy depends on the severity of symptoms. Severe cases may contribute to death in late childhood or early adolescence. If it is a milder form, clients may live into adulthood. Spinal cord compression and airway obstruction are major causes of death for clients with Morquio syndrome.

Gaucher's Disease (congenital)

Gaucher's disease is considerably more common in the descendants of Jewish people from Eastern Europe, although individuals from any ethnic group may be affected. The Jewish population has the highest prevalence with an incidence of approximately 1 in 450 persons and affects around 6,000 people in the United States. Gaucher's disease is a genetic mutation that limits the production of an enzyme that results in an accumulation of lipids in the liver, spleen, and bone marrow. Gaucher disease can leave a client with pain, fatigue, jaundice, bone damage, anemia, and even death.

There are several types of the disease. Type 1 Gaucher disease is the most common form of this condition. Type 1 doesn't affect the central nervous system (brain and spinal cord). Type 1 can range from mild to severe and symptoms occur anywhere from childhood to adulthood.

Types 2 and 3 Gaucher disease affect the central nervous system and are known as neuronopathic forms of the disorder. Type 2 Gaucher disease is usually aggressive and causes life-threatening medical problems beginning in infancy. Type 3 Gaucher disease is slower in the progress of the disease than Type 2 but will also affect the nervous system.

There are two subclasses of Gaucher disease type 2. The perinatal lethal form has life-threatening complications that start before or in early infancy. Most infants do not live beyond a few days following birth. The second is the cardiovascular type 2, causing hardening of the valves. This form also affects

the eyes, and bones, and causes the spleen to enlarge (splenomegaly). With this form of the disease, a client could live into their sixties.

Pompe Disease (congenital)

Pompe disease is a rare genetic disorder in which the body has the inability to break down complex sugars. As glycogen steadily builds up in the tissues, it causes tissue and organ damage, and eventually leads to progressive muscle deterioration. When the muscles for breathing become affected assistance with a respiratory infection may be required. Life expectancy has a large range depending on when symptoms begin. Symptoms beginning in infancy could give a range of 1-8 years and those with late-onset could live to be beyond 30 years of age.

Hemochromatosis (congenital/acquired)

Hemochromatosis is a condition that affects how the body absorbs iron. This can result from a mutation in the HFE gene or excessive iron from blood transfusions or a diet high in iron-rich foods. Hemochromatosis which is hereditary causes your body to absorb too much iron from the food you eat. Excess iron is stored in your organs, especially your liver, heart, and pancreas. High levels of iron can lead to life-threatening conditions, such as liver disease, heart problems, and diabetes.

The genes that cause hemochromatosis are inherited, but only a few people who have the genes ever develop serious problems. Hemochromatosis usually presents in midlife.

Phenylketonuria (PKU) (congenital)

PKU develops because the body does not have the ability, or has a reduced ability, to produce phenylalanine hydroxylase. This enzyme is important for processing amino acids. Amino acids are essential in building proteins, for bodily growth and development. The body's normal process is to break down excess amino acids and remove them but people with PKU are unable to do so. These excess amino acids can build up in the body and cause damage to the brain. Early treatment is necessary to prevent severe complications from PKU.

About one in 15,000 babies are born with PKU in the United States. Although it is principally a childhood disorder, in rare cases, the first signs of PKU may develop in late adulthood resembling common neurological diseases.

Cushing Disease (congenital/acquired)

Cushing disease (also called Cushing's disease) occurs when the body produces too much cortisol. Cortisol is a hormone that is produced by the adrenal glands and plays an important role in the stress response. Maintaining an adequate balance of cortisol is essential for optimal health. The most common cause of Cushing disease is the overuse of steroid drugs, such as prednisone, but it can also occur from the overproduction of cortisol by the adrenal glands.

Hyperparathyroidism (acquired)

Hyperparathyroidism occurs when the parathyroid glands in the neck (near the thyroid gland) produce too much parathyroid hormone. Parathyroid hormone is produced by these 4 small glands which help the body manage the calcium levels in the blood and bones. If too much of the parathyroid hormone is made, it will pull calcium from the bones into the bloodstream. If calcium levels in the blood become too high a condition called hypercalcemia will occur. When there is too much calcium in the blood the bones weaken because of the shift from the bones to the bloodstream. Kidney stones could form due to excess circulating calcium, and calcium will interfere with heart rhythm and how the brain functions.

Mitochondrial Disorders

Mitochondrial disorders are a group of conditions that prevent the mitochondria (powerhouse of the cells) from producing adequate energy for the cells which interferes with their normal function. This is a genetic mutation that passes through families. These disorders can affect any part of the body because mitochondria are prevalent throughout the entire body. Mitochondria play a crucial role in metabolism and if not functioning appropriately can lead to many metabolic issues.

Mitochondria are specialized and perform specific functions within certain tissues. The mitochondria produce energy within the heart called ATP. This occurs when levels of calcium are increased and the communication between cells amplifies.

Mitochondrial processes are influenced by diet and nutrition and as a result effects of mitochondrial-associated diseases. Diet provides energy through carbohydrates and fats. Metabolic processes result in byproducts, and the liver, and the kidneys are no exception. They produce ketone bodies and ketone bodies are a rich source of energy for the body. Ketone bodies can be utilized by the body as an alternative source of energy when needed.

The ketogenic diet is a diet that is high in fats and low in carbohydrates. The diet is designed to produce ketone bi-products through low carbohydrate consumption and fats as the primary source of energy. The ketogenic diet has been used when carbohydrate utilization has become compromised. In this type of therapy, ketone bodies can serve as an energy-providing source. The ketogenic diet was used in an epilepsy study and had been successful in preventing epileptic seizures, a symptom of mitochondrial disorder. By developing a diet that increases metabolic function in cases of defects in mitochondrial function, a single specific diet plan may have the ability to treat a wide range of mitochondrial disorders.

Symptoms of Metabolic Disease

Fabry Disease

Symptoms vary depending on the type. Some symptoms are mild and might not appear until later in life. Males tend to have more severe symptoms than females. Fabry disease symptoms include:

1. numbness, tingling, burning, or pain in the hands or feet
2. extreme pain during physical activity
3. heat or cold intolerance
4. abnormal opacity of the eye (cornea), which does not change someone's vision

5. dizziness
6. flu-like symptoms, including fatigue, fever, and body aches
7. gastrointestinal problems, such as diarrhea, constipation, and abdominal pain
8. hearing loss or ringing in ears (tinnitus)
9. high levels of protein in the urine (proteinuria)
10. raised red or purplish skin lesions (angiokeratoma) on your chest, back, and in the genital area
11. sweating less (hypohidrosis) or not at all (anhidrosis)
12. swelling (edema) in the legs, ankles, or feet

Diabetes

Symptoms of diabetes can vary depending on the type as well as the severity of blood sugar control.

1. urinate (pee) a lot, often at night
2. thirsty
3. lose weight without trying
4. increased appetite
5. blurry vision
6. numbness or tingling hands or feet
7. fatigue
8. very dry skin
9. sores that heal slowly or not at all
10. more infections than usual

Type 1 Diabetes:

Clients who have type 1 diabetes could also have symptoms such as nausea, vomiting, or stomach pains. These symptoms can develop in just a few weeks or months. Symptoms can be severe. Type 1 diabetes normally starts when you're a child, but can develop at any age of life.

Type 2 Diabetes:

Type 2 diabetes symptoms can take years to develop and sometimes symptoms do not appear. Type 2 diabetes usually starts when you're an adult,

though more and more children and teens are developing it due to inactivity and dietary choices.

Tay-Sachs Disease

Signs and symptoms start to show up at about 3 to 6 months of age. As the disease progresses, development slows and muscles begin to weaken. Over time, this leads to seizures, vision and hearing loss, paralysis, and other major issues. Children with this form of Tay-Sachs disease typically live only a few years.

Hurler Syndrome

Symptoms fall on a spectrum. Some children have only a few, mild symptoms while others can experience significant complications of the disease.

Symptoms may include:

1. clouding of the front part of the eye (corneal clouding)
2. frequent upper respiratory infections
3. enlarged tonsils and/or adenoids
4. distinct facial features (coarse facial features)
5. hernias

Other symptoms children might experience over time include:

1. thickening and dysfunction of the heart valves
2. abnormal curvature of the spine (scoliosis, kyphosis)
3. enlargement of the liver and spleen (hepatosplenomegaly)
4. accumulation of fluid around the brain (hydrocephalus)
5. compression of the spinal cord
6. progressive neurological decline

Niemann-Pick

Depending on the type of Niemann-Pick disease someone has, signs and symptoms may include:

1. clumsiness and difficulty walking
2. excessive muscle contractions (dystonia) or eye movements
3. sleep disturbances
4. difficulty swallowing and eating
5. recurrent pneumonia

The three main types of Niemann-Pick disease are types A, B, and C. The signs and symptoms you experience depend on the type and severity of your condition. Some infants with type A will show signs and symptoms within the first few months of life. Those with type B may not show signs for years and have a better chance of surviving to adulthood. People with type C may not experience any symptoms until adulthood.

Morquio Syndrome

Signs and symptoms of Morquio syndrome typically appear between ages 1 and 3. Children with Morquio syndrome may develop:

1. scoliosis or kyphosis
2. knock knees
3. heart and vision problems
4. enlarged liver
5. short height

Gaucher's Disease

There are different types of Gaucher disease, and signs and symptoms of the disease vary widely, even within the same type. Type 1 is by far the most common.

Siblings, even identical twins, with the disease can have different levels of severity. Some people who have Gaucher disease have only mild or no symptoms.

Most clients who have Gaucher disease have varying degrees of the following problems:

1. abdominal complaints
2. skeletal abnormalities

3. blood disorders

Gaucher disease in its rare form can affect the brain, which can cause abnormal eye movements, muscle rigidity, swallowing difficulties, and seizures. One rare subtype of Gaucher disease begins in infancy and typically results in death by 2 years of age.

Pompe Disease

A baby between a few months old and age 1 has early-onset, or infantile, Pompe disease. This could look like:

1. trouble eating and not gaining weight
2. poor head and neck control
3. rolling over and sitting up later than expected
4. breathing problems and lung infections
5. enlarged and thickening heart or heart defects
6. enlarged liver
7. enlarged tongue

In older clients, symptoms may start as late as 60 years of age. This is known as late-onset Pompe disease. This type tends to move slowly, and it doesn't usually involve the heart. Signs and symptoms could include:

1. feeling weak in the legs, trunk, and arms
2. shortness of breath, a hard time exercising, and lung infections
3. trouble breathing while you sleep
4. curve in your spine
5. enlarged liver
6. enlarged tongue that makes it hard to chew and swallow
7. stiff joints

Hemochromatosis

Some clients with hereditary hemochromatosis never have symptoms. Early signs and symptoms often overlap with those of other common conditions.

Signs and symptoms may include:

1. joint pain
2. abdominal pain
3. fatigue
4. weakness
5. diabetes
6. loss of sex drive
7. impotence
8. heart failure
9. liver failure
10. bronze or gray skin color
11. memory fog

Phenylketonuria (PKU)

Children with untreated PKU appear normal at birth. But by age 3 to 6 months, they begin to lose interest in their surroundings. By age 1 year, children are developmentally delayed, and their skin has less pigmentation than someone without the condition. If clients with PKU do not restrict the phenylalanine in their diet, they develop severe intellectual and developmental disabilities.

Other symptoms include:

1. behavioral or social problems
2. seizures, shaking, or jerking movements in the arms and legs
3. stunted or slow growth
4. skin rashes, like eczema
5. small head size, called microcephaly
6. musty odor in urine, breath, or skin that is a result of the extra phenylalanine in the body
7. fair skin and blue eyes, due to the body's failure to transform phenylalanine into melanin

Cushing Disease

Common signs and symptoms for a client with Cushing disease may include:

1. weight gain and fatty tissue deposits, particularly around the midsection and upper back, in the face (moon face), and between the shoulders (buffalo hump)
2. pink or purple stretch marks (striae) on the skin of the abdomen, thighs, breasts, and arms
3. thinning, fragile skin that bruises easily
4. slow healing cuts, insect bites, and infections
5. acne

Signs and symptoms for female clients with Cushing disease may include:

1. thicker or more visible body and facial hair (hirsutism)
2. irregular or absent menstrual periods

Signs and symptoms for male clients with Cushing disease may include:

1. decreased sex drive
2. decreased fertility
3. erectile dysfunction

Other possible signs and symptoms of Cushing disease may include:

1. severe fatigue
2. muscle weakness
3. depression, anxiety, and irritability
4. loss of emotional control
5. cognitive difficulties
6. new or worsened high blood pressure
7. headache
8. infections
9. skin darkening
10. bone loss, leading to fractures over time
11. in children, impaired growth

Hyperparathyroidism

Symptoms may be so mild and nonspecific that they don't seem related to parathyroid function, or they may be severe. The range of signs and symptoms include:

1. weak bones that break easily (osteoporosis)
2. kidney stones
3. excessive urination
4. stomach (abdominal) pain
5. tiring easily or weakness
6. depression or forgetfulness
7. bone and joint pain
8. frequent complaints of illness with no clear cause
9. nausea, vomiting, or loss of appetite

Mitochondrial Diseases

Symptoms of mitochondrial diseases can include:

1. poor growth
2. muscle weakness, muscle pain, low muscle tone, exercise intolerance
3. vision and/or hearing problems
4. learning disabilities, delays in development
5. autism spectrum disorder
6. heart, liver, or kidney diseases
7. gastrointestinal disorders, swallowing difficulties, diarrhea or constipation, unexplained vomiting, cramping, and reflux
8. diabetes
9. increased risk of infection
10. neurological problems, seizures, migraines, and strokes
11. movement disorders
12. thyroid problems
13. respiratory (breathing) problems
14. lactic acidosis (a buildup of lactate)
15. dementia

Metabolism is a complex process that involves many chemical reactions that have results in many different parts of the body. With so many complex processes occurring there are many opportunities for something to go wrong and cause a metabolic disorder.

Treatments of Metabolic Diseases

Treatments for metabolic diseases will vary depending on the diagnosis.

Fabry Disease

There isn't a cure for Fabry disease. Medications for pain and stomach problems can ease symptoms. Two treatments are utilized to prevent the buildup of fatty substances. The goal is not to cure the disease but to prevent heart problems, kidney disease, and other life-threatening complications.

1. Enzyme replacement therapy: The enzyme will help with the build-up of fatty substances within the body.
2. Oral chaperone therapy: Chaperones are small molecules that repair faulty alpha-GAL enzymes.

Researchers are actively developing several new therapies using genetic engineering and stem cell technologies.

Diabetes

Management of type 2 diabetes includes:

1. healthy eating
2. regular exercise
3. weight loss
4. diabetes medication or insulin therapy if needed
5. blood sugar monitoring

Healthy Eating

There is no specific diabetes diet but making adjustments can be helpful in keeping the blood sugar within or close to within normal limits.

1. regular schedule for meals and healthy snacks
2. smaller portion sizes
3. more high-fiber foods, such as fruits, non-starchy vegetables, and whole grains
4. fewer refined grains, starchy vegetables, and sweets
5. modest servings of low-fat dairy, low-fat meats, and fish
6. healthy cooking oils, such as olive oil or canola oil
7. fewer calories

Physical Activity

Exercise is important for losing weight or maintaining a healthy weight. It also helps with regulating blood sugar levels.

Aerobic exercise: It is important to choose an aerobic exercise that you enjoy for consistency. 30 minutes or more of moderate aerobic exercise on most days of the week, or at least 150 minutes a week is the recommended amount for adults. Children should have 60 minutes of moderate to vigorous aerobic exercise daily.

Resistance exercise: Resistance exercise would include weightlifting, yoga, and calisthenics. These exercises will increase strength, balance, and help with motion throughout the day.

Limit inactivity: Interrupting extended times of inactivity, such as sitting at the computer. Incorporating movement or stretching regularly throughout the day will help stabilize blood sugar levels.

Weight loss: By reducing even 5% of body weight there may be improvements in blood sugar levels, cholesterol, triglycerides, and blood pressure. And, the more weight reduction the better chances of disease control.

Monitor blood sugar: Your healthcare provider will help determine when and how often blood sugar should be checked. If administering insulin, blood sugars may have to be taken multiple times during the day.

Diabetes medications

Drug treatments for type 2 diabetes include the following:

1. Metformin works by lowering glucose production in the liver and improving the body's sensitivity to insulin, so the body uses insulin more effectively.
2. Some people experience B-12 deficiency and may need to take supplements.
3. Sulfonylureas help your body secrete more insulin.
4. Glinides stimulate the pancreas to secrete more insulin.
5. Thiazolidinediones make the body's tissues more sensitive to insulin.
6. DPP-4 inhibitors help reduce blood sugar levels but tend to have a very modest effect.
7. GLP-1 receptor agonists are injectable medications that slow digestion and help lower blood sugar levels.
8. SGLT2 inhibitors affect the blood-filtering functions in your kidneys by inhibiting the return of glucose to the bloodstream.

Insulin Therapy

Different types of insulin vary on how quickly they begin to work and how long they have an effect.

Weight-loss Surgery

Weight-loss surgery changes the shape and function of your digestive system. Weight-loss surgery could be an option for adults living with type 2 diabetes who have a body mass index (BMI) of 35 or higher.

Tay-Sachs Disease

There is no cure for Tay-Sachs disease, and the goal of treatment is support and comfort. Symptoms of slowed development usually appear around six months of age. Symptoms progress until they lead to death, often around age four.

Supportive treatments include:

1. Medication: To reduce symptoms and prevent complications.
2. Respiratory care: Accumulated mucus in the lungs is common and chest physiotherapy (CPT), exercise, and other techniques can help remove

mucus from the lungs. Medications to reduce saliva production may also be prescribed.

3. Nutrition and hydration: Trouble swallowing or the development of respiratory problems may occur so a feeding tube may be inserted through the nose and into the stomach.
4. Physical therapy: Helps keep joints flexible and maintains as much ability to move as possible.
5. Occupational therapy: Activities and supportive devices to help with daily functioning.
6. Speech and language therapy: Swallowing issues.

Hurler Syndrome

Most therapies for Hurler syndrome are directed toward the treatment of complications and are not specific to an underlying abnormality.

1. Enzyme replacement therapy
2. Hematopoietic stem cell transplant (HSCT)
3. Additional management of Hurler syndrome is supportive and includes surgical interventions

Niemann-Pick Disease

No cure currently exists and there is no treatment for type A or B. Physical therapy is an important part of treatment for type C to help maintain mobility as long as possible.

Morquio Syndrome

For type A, the IV medicine called Vimizim, which replaces the missing enzyme, may be tried. Enzyme replacement therapy is not available for type B.

Gaucher's Disease

There is no cure for Gaucher's disease. But treatment can help you control your symptoms. Your treatment will depend on what type of Gaucher disease you have. Treatment may include:

1. Enzyme replacement therapy (types 1 and 3)

2. Medication
3. Regular physical exams and bone density screening to check your disease
4. Bone marrow transplant
5. Surgery to remove all or part of your spleen
6. Joint replacement surgery
7. Blood transfusions
8. Enzyme replacement therapy

Pompe Disease

In 2006, the FDA approved the first treatment known as enzyme replacement therapy (ERT), which is still the only treatment approved for Pompe disease.

Hemochromatosis

Phlebotomy can be used to reduce iron levels and chelation for those who cannot undergo blood removal through phlebotomy. When chelating the medication binds excess iron, allowing your body to expel iron through your urine or stool. Chelation is not commonly used in hereditary hemochromatosis.

Phenylketonuria (PKU)

The main treatments for PKU include:

1. A lifetime diet with a very limited intake of foods with phenylalanine
2. Taking a PKU formula, a special nutritional supplement containing essential protein
3. Medications, for certain clients with PKU

Because the amount of phenylalanine that a client with PKU can safely eat is so low, it's important to avoid all high-protein foods, such as:

1. milk
2. eggs
3. cheese
4. nuts

5. soy products (soybeans, tofu, tempeh, and milk)
6. beans and peas
7. poultry, beef, pork, and any other meat
8. fish
9. potatoes and grains

PKU medications

The drug Kuvan may be used in combination with a PKU diet. Enzyme therapy, Palynziq, for adults with PKU when current therapy does not adequately reduce the level of phenylalanine.

Cushing Disease

Treatments for Cushing syndrome are designed to lower the high level of cortisol in your body. The best treatment depends on the cause of the syndrome. The long-term use of corticosteroid medications can cause Cushing syndrome so reducing the dosage of the drug over a period of time, while still managing the condition for which you take it is the ideal treatment for this situation.

Surgery

If the cause of Cushing syndrome is a tumor, removal of the tumor may be recommended. Pituitary tumors are typically removed by a neurosurgeon, who may perform the procedure through the nose. For tumors that are located in the adrenal glands, lungs, or pancreas, they can be removed using minimally invasive surgical techniques, with smaller incisions.

Radiation therapy

Radiation can be used for clients that are not candidates for surgical removal of a tumor or if the tumor was not completely removed during surgery. Radiation can be given in small doses over a six-week period, or one large one-time dose can be administered to prevent reoccurring radiation exposure to surrounding tissues.

Medications

Medications are another option that can be used to control cortisol production when surgery and radiation have not been successful. Medications may also be pre-surgical protocol if the client is very ill and symptoms need to be decreased or stabilized prior to surgery.

Hyperparathyroidism

Your health care provider may recommend no treatment and regular monitoring if:

1. calcium levels are only slightly elevated
2. kidneys are working well, and no kidney stones detected
3. bone density is within the standard range or only slightly below the range
4. no other symptoms that may improve with treatment

Regularly scheduled tests to monitor blood-calcium levels and bone density will need to be completed to assess and detect any changes that may need treatment.

Surgery

Surgery is the most common treatment for primary hyperparathyroidism and in most cases cures the condition. Only those glands that have a tumor or are enlarged and not functioning properly will be removed. The parathyroid glands are two pairs of small, oval-shaped glands within the neck.

Complications from surgery aren't common but include:

1. damage to nerves controlling the vocal cords
2. long-term low calcium levels requiring the use of calcium and vitamin D supplements due to removal or damage to all parathyroid glands

Medications

Medications to treat hyperparathyroidism can be utilized when surgery isn't an option or hasn't been successful. Medication may mimic circulating calcium in the blood, tricking the parathyroid glands to release less parathyroid hormone. Other medications may help keep the balance of calcium and phosphorus minerals so that the parathyroid glands don't have to work hard.

Lifestyle and home remedies

Monitor how much calcium and vitamin D you get in your diet. The daily recommended amount of calcium for adults ages 19 to 50 and men ages 51 to 70 is 1,000 milligrams (mg) of calcium a day. That calcium recommendation increases to 1,200 mg a day for women aged 51 and older and men aged 71 and older. The daily recommended amount of vitamin D is 600 international units (IUs) of vitamin D a day for people ages 1 to 70 and 800 IUs a day for adults aged 71 and older.

To prevent the formation of kidney stones, drink enough water to produce nearly clear urine. This will facilitate the removal of excess calcium in the urinary tract and not allow it to sit static within the kidneys and bladder.

Regular exercise, including strength training, helps maintain strong bones. Smoking on the other hand may increase bone loss and increase the risk for other serious health problems. Smoking cessation is an important treatment option for hyperparathyroidism. Finally, avoid calcium-raising medications such as lithium or diuretics like thiazide-type diuretics and amiloride.

Mitochondrial diseases

There is no cure for mitochondrial diseases. Treatments can vary from client to client depending on the type of mitochondrial disease and how severe it is. Treatments for mitochondrial disease may include:

1. vitamins and supplements
2. exercise
3. energy conservation

4. speech therapy, physical therapy, respiratory therapy, and occupational therapy
5. avoid exposure to heat/cold
6. adequate sleep
7. avoid stress when possible
8. eat on a regular basis
9. avoid alcohol, smoking, and monosodium glutamate (MSG)

Summary

Some metabolic diseases are managed with lifestyle changes, such as healthy eating and avoiding too much sugar, and others require medication. Some inherited metabolic disorders are treated by long-term nutritional supplements, while metabolic disorders that arise as a result of another disease or condition often resolve with the treatment of the underlying condition. No matter the cause, appropriate treatment or support is essential for either health improvement or possibly comfort for the client. Not all metabolic diseases are treatable, and many are incurable but having knowledge about these diseases will help you as the caregiver support and assist the client and their family on a more meaningful level.