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Chapter 1 : List of Genetic Disorders

Many human diseases have a genetic component. Some of these conditions are under investigation by researchers at or associated with the National Human Genome Research Institute (NHGRI). Below is a list of selected genetic, orphan and rare diseases.

Her specialty is transfusion medicine. Her degree at Sam Houston State University includes emphasis in journalism and photography. She writes for Examiner. Also known as Austinstar, she writes articles on art, travel and the health sciences. DNA and the genes that cause the worst diseases. Inherited genetic diseases affect millions worldwide. Family health history is the biggest predictor of genetic disease. Research is ongoing to detect and treat genetic disease. The decision to be tested for genetic disease is often difficult and includes many moral and ethical issues. The worst genetic diseases are incurable. Video of the Day Cystic Fibrosis Cystic fibrosis is the most common, fatal genetic disease, affecting about 30, people in the United States, according to the National Human Genome Research Institute. Cystic fibrosis causes the body to produce thick, sticky mucus that clogs the lungs, leads to infection and affects the pancreas. Breathing is difficult, and digestive enzymes are blocked, which inhibit the absorption of food nutrients. Tay-Sachs Tay-Sachs causes death at any early age, usually around five years old, because of a missing enzyme called Hex-A. Tay-Sachs causes progressive destruction of the nervous system and brain. There is no treatment, only management of the symptoms. Alcoholism Considered a disease, addiction to alcohol is most likely passed down through genetic makeup. If one or both parents are addicted to alcohol, then the children of those parents appear to have a higher chance of being addicted to alcohol. As of , no specific genes have been identified as the cause of alcoholism. Breast and Colon Cancer One in nine women has a chance to develop breast cancer. The BRCA1 and BRCA2 inherited genes predispose a woman to a greater chance of breast or ovarian cancer, but not all breast cancers are due to genetic reasons. A family history of colon cancer means there is a reason to believe it may be genetic. As with other cancers, genetic reasons may not be the only reasons for developing cancer. Sickle Cell Disease Having the single expression of the gene for sickle cell does not cause the disease. The gene evolved to protect people from malaria. The sickled cells get stuck in small capillaries and destroy joints and organs. Eventually, the victim will die of organ failure. Obesity Obesity is fast becoming the number one inherited disease in the United States. It is a complicated disease occurring from genetic and environmental stimuli. The children of obese parents have a much greater than average risk of being obese. No definitive research has yet discovered the specific genetic makeup involved, yet it does appear to have elements of both inherited genetic tendencies and the dietary habits of the parents. Heart Disease Many types of heart disease are passed on genetically. One in particular, called Brugada disease, is treatable as a genetic disease. If someone in the family has Brugada, a specific abnormal heart rhythm, the chances are good for other members to carry the gene and all siblings should get an electrocardiogram test. The Brugada EKG pattern is recognizable and diagnostic. A pacemaker is implanted to correct the abnormal heartbeat. Hemophilia Hemophilia, a bleeding disorder caused by the absence of genetic clotting factors, is inherited from one or both parents. Replacing the missing factors is the way to manage the various bleeding disorders. Specific tests are available to determine which clotting factors are missing. In addition, 35, people exhibit some symptoms and 75, people carry the abnormal gene. Severe organ damage can result from lack of treatment. Removing the iron rich blood is the quickest and safest method of treating this disease.

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Chapter 2 : Genetic disorder - Wikipedia

The following is a list of genetic disorders and if known, type of mutation and the chromosome involved. Although the parlance "disease-causing gene" is common, it is the occurrence of an abnormality in these genes that causes the disease.

She is also a registered nurse with expertise in a wide range of medical conditions and treatments. Causes of genetic abnormalities include environmental, congenital and gene mutation within the cell that can occur at random. Inherited genetic diseases require a specific mutated gene pattern passed down from parent to child in order for the disease to develop. Some inherited genetic diseases pass through a family from one generation to the next as carriers, but without manifestation of the disease. Video of the Day Fragile X Syndrome Fragile X syndrome, as reported by Genetics Home Reference, a service of the United States National Library of Medicine, is a genetic condition that manifests as developmental problems such as learning disabilities and mental retardation. Symptoms of fragile X include anxiety, excessive movement and acting on impulse. Fragile X affects males more severely than females, with about one-third of the males showing signs of autism. Other symptoms of fragile X include seizures and physical abnormalities such as a long and narrow face and large ears. No cure exists for fragile X. Treatment goals include therapy--physical, educational and behavioral--and medications to assist with control of aggression and behavioral abnormalities. Gaucher Disease The University of Maryland Medical Centers reports that Gaucher disease is a rare genetic disorder characterized by the absence of the glucocerebrosidase enzyme. That causes a buildup of harmful substances in the body that affect the spleen, liver, bones and marrow. Type 1 Gaucher disease affects children and adults, and it manifests as anemia, bone disease and spleen enlargement. Type 2 causes severe neurological damage in infancy and quickly results in death. Type 3 symptoms include difficulties with the brain, spleen and liver, but affected patients may reach adulthood. Treatment includes enzyme replacement therapy and, for severe cases, bone marrow transplant. Batten Disease Batten disease, a fatal genetic disorder, requires the defective gene from each parent in order for the child to develop it. Mild changes progress to seizures and loss of vision and motor skills. Death occurs by late adolescence or the early twenties. Treatment includes symptom management such as medication for seizures, along with physical and occupational therapies.

Chapter 3 : A List of Inherited Genetic Diseases | calendrierdelascience.com

Hereditary diseases are disorders or diseases that are inherited genetically. They are also known as genetic disorders or inherited diseases and may be passed on within the family if there is a mutant gene calendrierdelascience.com of Hereditary DiseasesThere are a nu.

Reviewed by our Medical Team â€" There are many theories about why genetic diseases occur. There are four different types of genetic disorders. They are [1]: Single-gene is mutated Chromosomal changes-entire areas of the chromosome can be missing or misplaced. The maternal genetic material in mitochondria can mutate as well. We know that some ethnic groups are pre-disposed for certain genetic disorders people originating in the Mediterranean areas of Europe, for example are more likely to have a form of anemia that is geneticâ€"the thalassemias. In SCA, the mutation that causes the red blood cells to change shape also helps minimize infection with the malarial parasite. The effect of this is breathing difficulties and recurrent lung infections. It also involves problems in digestion and in reproduction. The symptoms vary from relatively mild to severe. CF is most common in Caucasians, particularly in Ashkenazi Jews, but is found in all ethnic groups. Down syndrome affects about 1 out of newborn babies. It can be detected by pre-natal testing. There is a pattern of features some of which are usually immediately apparent at birthâ€"these include facial characteristics, decreased muscle tone, heart and digestive system defects and developmental delays. Children with Down Syndrome are variably affected, the affects ranging from mild to moderate to severe. Any ethnic group can be affected and it is most often associated with increased age of the mother. The delays and cognitive difficulties can range from very mild to severe and are sometimes associated with autism. In Fragile X syndrome, part of the X chromosome can break apart. The area on the X chromosome that causes the fragility can be repeated on that chromosomeâ€"the more the number of repeated areas, the greater the fragility and the more serious the syndrome. These clotting problems can result in too much bleeding and the formation of abnormal clots throughout the body, most commonly in the veins. The most common is the Factor V Leiden abnormality and can be particularly a problem in pregnancy, leading to pre-eclampsia, small-for-gestational-age babies, stillbirths and problems with the placenta. Hemophilia is a well-known clotting disorderâ€"the most common types are Hemophilia A where there is a lack of clotting factor VIII , Hemophilia B where there is a lack of clotting factor IX and Von Willebrand disease where there is a lack of the Von Willebrand clotting factor. Symptoms of all these clotting disorders include[9]: Excessive bleeding of the gums, nose, gastrointestinal system and bleeding into the joints Abnormal menstrual bleeding Skin rashes Familial combined hyperlipidemia and Familial hypercholsterolemia These are inherited disorders that result in an increase in blood lipids and cholesterol. These disorders predispose to obesity, glucose intolerance and diabetes, strokes and heart disease. Treatment includes lifestyle and dietary changes. Loss of these nerve cells causes symptoms such as behavior changes, unusual, snake-like movements chorea , uncontrolled movement, difficulty walking, loss of memory, speech and cognitive functions and difficulty in swallowing. Treatment aims to limit the course of the disease. There is also an early-onset form which begins in childhood. The most common are [11]: In Duchenne muscular dystrophy, the symptoms usually are apparent before the age of 6 and may appear even earlier. Fatigue, possible mental retardation and muscle weakness, beginning in the legs and then to the upper body. There can also be associated heart problems, respiratory problems and deformities of the chest and back. The muscle weakness makes it progressively more difficult to walk and get around. That weakness gets progressively worseâ€"by the age of 12, most kids are confined to a wheelchair. Boys are more likely to inherit this disorder. Becker muscular dystrophy In Becker muscular dystrophy, the symptoms are similar to Duchenne muscular dystrophy, but are slower to appear and slower to worsen. Those symptoms include fatigue, possible mental retardatio, and muscle weakness, beginning in the legs. The muscle weakness in the upper body is not as severe as in Duchenne. Again, boys are more likely to have the disorder and are often confined to a wheelchair by the age of Sickle Cell Anemia Sickle cell anemia SCA is an inherited disease where the red blood cells, normally shaped like discs, instead form a crescent or sickled shape. Besides pain abdominal, chest and in the bones , other symptoms include fatigue, shortness of breath, increased heart rate,

delayed growth and puberty, fever and leg ulcers. Treatments include pain medication, hydroxyurea to decrease the number of pain episode, folic acid to help support the red blood cells, blood transfusions and kidney dialysis. Sickle cell disease is more common in people of African and Mediterranean backgrounds, but it is also found in people from South and Central America, the Caribbean, and the Middle East. Genetic Screening For Cancer Patients Thalassemias The thalassemias beta-thalassemia is the most common, alpha-thalassemia is less common is a group of inherited blood disorders where hemoglobin, the oxygen-carrying molecule, is not properly synthesized by the red blood cells. This results in an anemiaâ€”where the usual symptoms are fatigue, an enlarged spleen, easily broken bones, bone pain and shortness of breath. People with thalassemia may also have poor appetite, darkened urine and jaundice a yellowish discoloration of the skin or the white parts of the eyesâ€”jaundice is a signal for liver dysfunction. Infections are common in people with thalassemia. If it is not caught early enough, high levels of phenylalanine accumulate and cause mental retardation, brain damage and seizures. Treatment consists of a phenylalanine restricted diet and the use of a cofactorâ€”thetrahydrobiopterin BH4 to reduce the amount of phenylalanine in the blood. Alphaantitrypsin deficiency leads to a decreased amount of alpha-1 antitrypsin in the lungs and in the bloodâ€”this results in lung diseases such as emphysema. The earliest symptoms are shortness of breath and wheezing. Other symptoms can include weight loss, frequent respiratory infections, fatigue, and a rapid heartbeat. There are many other genetically based diseases. There are no cures for these disorders, though there is the potential for gene therapy. Many of these disorders can be treated and those with the disease can live a fuller life.

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Chapter 4 : Hereditary diseases list | General center | calendrierdelascience.com

List of Hereditary Diseases There are numerous genetic diseases and disorders that are passed from the parent to the offspring. Some of these diseases may be congenital (present at the time of birth) or can even occur after a while.

Check new design of our homepage! An A-to-Z List of Rare Genetic Diseases and Disorders Genetic diseases are present throughout the life of an individual, some of which appear very early in life. They result in many chronic conditions that have no cure. Here are the rare genetic diseases and disorders that are seen in human beings. The rarest genetic disorder known is ribosephosphate isomerase deficiency with a single-known diagnosed case. Genetic diseases or disorders are caused due to abnormalities in the genetic makeup of an individual. Such abnormalities can be caused by a minuscule, major variation or mutation in single or multiple genes, chromosomal aberrations, and rarely due to mutations in the non-chromosomal DNA of mitochondria. Genetic diseases or disorders may or may not be inheritable. They can be recessive or dominant in nature. A rare disease in one part of the world may not be rare in another. It defines such diseases strictly according to its prevalence, specifically "any disease or condition that affects less than , persons in the United States, or about 1 in Aicardi Syndrome Aicardi syndrome is a very rare genetic disorder characterized by underdeveloped or absence of the corpus callosum, the structure separating the left and right half of the brain. It is found to affect only girls as it is believed to be caused due to a defect in the X chromosome. It affects 1 in , to , individuals in the United States. Babies born with this disorder seem normal till the age of 3 to 5 months and then start showing some key symptoms of this disorder. They include retinal abnormalities and infantile spasms resulting in seizures. Individuals affected by this disease have serious developmental issues. There is no cure for this disorder. Treatment generally involves management of seizures and supporting the affected individuals through the delay in development. Alagille Syndrome Alagille syndrome is a rare genetic disorder that affects the liver, kidney, heart, and other organs of the body. Symptoms related to this syndrome are usually noticed in the early years of life. It affects around 1 in 70, newborns. It is inherited as an autosomal dominant trait, and the severity of symptoms may vary from individual to individual. Liver malfunctioning is caused by abnormalities in the bile duct like less in number or absent, narrow, or malformed , resulting in bile accumulation in the liver and thus damaging it. Symptoms include yellowish tinge on the skin and the whites of the eyes, accumulation of cholesterol beneath the skin, itching, etc. Individuals suffering from this disorder also face many heart problems. They have distinguishable facial features -- prominent forehead, pointed chin, and deep-set eyes. There is no cure for this syndrome; however, corrective surgery aimed at the functioning of heart, liver, and kidney helps to some extent. Back to Index Alkaptonuria Alkaptonuria, also known as black urine disease, is caused due to disorder in the tyrosine metabolism of the body. It affects about 1 in , to 1 million people worldwide. It is an autosomal recessive condition characterized by accumulation of alkapton or homogentisic acid toxic tyrosine byproduct in the blood, which is excreted in the urine. Urine of such patients becomes black when exposed to air. Presence of excess alkapton may result in osteoarthritis, heart disease, kidney stones, and prostate stones in men. Some of the symptoms of alkaptonuria include darkened skin and pigmented sclera white part of eye. No specific treatment has been established; however, it has been observed that consuming foods rich in vitamin C helps. It is also advisable to avoid diets rich in tyrosine and phenylalanine. It is one of the rarest-known genetic diseases with only around cases known in medical history. Indications include childhood obesity, sensorineural hearing loss, and vision impairment. It can also lead to hyperinsulinemia, hypertriglyceridemia, early onset of diabetes 2, and deafness. This syndrome can also cause several life-threatening medical complications involving liver, heart, lungs, etc. There is no cure for this syndrome; however, medication can be provided to cure specific symptoms of the syndrome. Apert Syndrome Apert syndrome is a rare genetic disorder that is apparent at birth. It results in distortion in the shape of skull and face, and sometimes, the hands and feet are webbed. It affects 1 in 65, to 88, newborns. In people affected by this syndrome, the bones of the skull fuse prematurely, a condition called craniosynostosis, while the brain keeps developing inside the abnormal skull causing pressure on the skull and face, resulting in its distortion. It also results in poor intellectual development of the individual, hearing loss, frequent ear and sinus infections,

and short stature. In most cases, the syndrome results due to random gene mutation while in some rare instances, it is inherited as an autosomal dominant trait. There is no cure for this disorder but surgery can, to some extent, cure some of its symptoms. Back to Index Batten Disease Batten disease is a rare autosomal recessive disorder that chiefly affects the nervous system and begins in the childhood stage. It is estimated to be occurring in every 2 to 4 of every , individuals. Batten disease is characterized by a buildup of pigments called lipofuscins in the body cells. Symptoms usually appear between 5 to 10 years of age when a normal child suddenly starts having vision problems and seizures. Overtime, the symptoms worsen resulting in loss of motor ability, mental retardation, loss of sight, etc. There is no cure for this condition, and it is generally fatal by the age of Bardet-Biedl Syndrome Bardet-Biedl syndrome is a rare genetic disorder affecting multiple organs. It affects 1 in , to 1 in , newborns. The signs and symptoms may differ among individuals who are affected by syndrome, even among family members. It is characterized by vision impairment, obesity, kidney anomalies, development issues, extra finger and toes, impaired motor skills, etc. This disorder is recessive in nature. There is no cure for the disorder, and usually treatment is concentrated on specific symptoms. Camurati-Engelmann Disease Camurati-Engelmann disease is a kind of bone dysplasia. It is a rare autosomal dominant genetic disorder. Only cases of the disease have been reported worldwide so far. It is characterized by thickened bones resulting in chronic pain. The skull bones also thicken resulting in pressure on the brain, leading to various neurological problems. Individuals suffering from it also complain of increased fatigue, weakness, headache, and muscle spasms. There is no cure for this condition but it can be partially treated. Anti-inflammatory and immunosuppressive agents, like glucocorticosteroids have proved to be helpful in some cases. Alternative therapies, like massage and heat therapy in conjunction with medication are also advisable. Back to Index Carpenter Syndrome Carpenter syndrome is a rare congenital disorder characterized by malformed head, face, fingers, and toes due to premature fusion of bones. It is an autosomal recessive disorder with around documented cases known till date. It is estimated to affect 1 in , individuals. Key symptoms include oddly-shaped head, fused digits, obesity, short stature, and reduced mental ability. Treatment generally consists of a number of staged surgeries to correct the malformation of bones in the early stages of life. This condition is similar to Apert Syndrome and Pfeiffer Syndrome. It is characterized by abnormal deposits of calcium in the basal ganglia and cerebral cortex. These are areas of brain control movement. Key symptoms are clumsiness, fatigue, unsteady posture, muscle cramping, uncontrolled movement, and dementia. Individuals can be affected by this syndrome at any time of their life; however, it is more prevalent in the age group of years. Only 60 families are known to be affected by this syndrome in medical literature. There is no cure for this condition. Treatment is generally symptomatic. It affects the blood vessels, resulting in immune system problems. It is estimated to occur in 1 in 50, individuals. Common symptoms of hereditary angioedema include swelling of the arms, legs, eyes, and throat, abdominal pain, and airway blockage. Treatment is usually done by following medication, hormonal treatment, and administration of painkillers. With time, the symptoms aggravate and the affected individual needs full-time care. This condition affects 1 in 10, in United States. Life expectancy after the onset of the disease is years. Indicants include involuntary movements, loss of motor abilities, cognitive difficulties, and emotional problems. There is no cure, but medication can relieve specific symptoms associated with the disorder. It affects around 1 in 80, to , individuals. This disorder is characterized by an underdeveloped or absent cerebellar vermis and a malformed brain stem. This part of the brain controls coordination and balance. Some key symptoms of this disorder are lack of muscle control, abnormal breathing pattern, jerky eye movement, intellectual disability, and physical deformities, such as cleft lip, extra finger, and toes. The severity of the symptoms varies from individual to individual. There is no cure for the disease but treatment for symptoms, like breathing difficulty can be meted out. About 1 in 30, people are estimated to be affected with this syndrome although the number could be as high as 1 in 15, It is caused due to a defect in the action of the cilia lining the respiratory tract, which results in abnormal ciliary motion.

Chapter 5 : Diseases & Conditions A-Z Index - A

Some genetic disorders are inherited from the parents, while other genetic diseases are caused by acquired changes or mutations in a preexisting gene or group of genes. Mutations can occur either randomly or due to some environmental exposure.

Each affected person usually has one affected parent. Autosomal dominant conditions sometimes have reduced penetrance , which means although only one mutated copy is needed, not all individuals who inherit that mutation go on to develop the disease. Birth defects are also called congenital anomalies. An affected person usually has unaffected parents who each carry a single copy of the mutated gene and are referred to as carriers. Examples of this type of disorder are Albinism , Medium-chain acyl-CoA dehydrogenase deficiency , cystic fibrosis , sickle-cell disease , Tay-Sachs disease , Niemann-Pick disease , spinal muscular atrophy , and Roberts syndrome. Certain other phenotypes, such as wet versus dry earwax , are also determined in an autosomal recessive fashion. X-linked dominant X-linked dominant disorders are caused by mutations in genes on the X chromosome. Only a few disorders have this inheritance pattern, with a prime example being X-linked hypophosphatemic rickets. Males and females are both affected in these disorders, with males typically being more severely affected than females. Some X-linked dominant conditions, such as Rett syndrome , incontinentia pigmenti type 2, and Aicardi syndrome , are usually fatal in males either in utero or shortly after birth, and are therefore predominantly seen in females. Exceptions to this finding are extremely rare cases in which boys with Klinefelter syndrome 47,XXY also inherit an X-linked dominant condition and exhibit symptoms more similar to those of a female in terms of disease severity. The chance of passing on an X-linked dominant disorder differs between men and women. In addition, although these conditions do not alter fertility per se, individuals with Rett syndrome or Aicardi syndrome rarely reproduce. X-linked recessive inheritance X-linked recessive conditions are also caused by mutations in genes on the X chromosome. Males are more frequently affected than females, and the chance of passing on the disorder differs between men and women. The sons of a man with an X-linked recessive disorder will not be affected, and his daughters will carry one copy of the mutated gene. X-linked recessive conditions include the serious diseases hemophilia A , Duchenne muscular dystrophy , and Lesch-Nyhan syndrome , as well as common and less serious conditions such as male pattern baldness and red-green color blindness. X-linked recessive conditions can sometimes manifest in females due to skewed X-inactivation or monosomy X Turner syndrome. Y linkage Y-linked disorders are caused by mutations on the Y chromosome. These conditions may only be transmitted from the heterogametic sex e. More simply, this means that Y-linked disorders in humans can only be passed from men to their sons; females can never be affected because they do not possess Y-allosomes. Y-linked disorders are exceedingly rare but the most well-known examples typically cause infertility. Reproduction in such conditions is only possible through the circumvention of infertility by medical intervention. Mitochondrial disease This type of inheritance, also known as maternal inheritance, applies to genes encoded by mitochondrial DNA. Because only egg cells contribute mitochondria to the developing embryo, only mothers can pass on mitochondrial DNA conditions to their children. It is important to stress that the vast majority of mitochondrial disease particularly when symptoms develop in early life is actually caused by an underlying nuclear gene defect, and most often follows autosomal recessive inheritance. Multifactorial disorders include heart disease and diabetes. Although complex disorders often cluster in families, they do not have a clear-cut pattern of inheritance. Complex disorders are also difficult to study and treat, because the specific factors that cause most of these disorders have not yet been identified. Studies which aim to identify the cause of complex disorders can use several methodological approaches to determine genotype - phenotype associations. One method, the genotype-first approach , starts by identifying genetic variants within patients and then determining the associated clinical manifestations. This is opposed to the more traditional phenotype-first approach, and may identify causal factors that have previously been obscured by clinical heterogeneity , penetrance , and expressivity. On a pedigree, polygenic diseases do tend to "run in families", but the inheritance does not fit simple patterns as with Mendelian diseases. But this does not mean that the genes

cannot eventually be located and studied. There is also a strong environmental component to many of them e.

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Chapter 6 : Types of Hereditary Diseases | Healthfully

Hereditary diseases are health problems that are passed from parents to offspring through defective genes, according to Steady Health. Some examples of hereditary diseases include hereditary hemochromatosis, Down syndrome, spherocytosis, achondroplasia, Usher syndrome, hemophilia, sickle cell anemia.

Patient Handouts Summary Genes are the building blocks of heredity. They are passed from parent to child. They hold DNA, the instructions for making proteins. Proteins do most of the work in cells. They move molecules from one place to another, build structures, break down toxins, and do many other maintenance jobs. Sometimes there is a mutation, a change in a gene or genes. This can cause a medical condition called a genetic disorder. You can inherit a gene mutation from one or both parents. A mutation can also happen during your lifetime. There are three types of genetic disorders: Single-gene disorders, where a mutation affects one gene. Sickle cell anemia is an example. Chromosomal disorders, where chromosomes or parts of chromosomes are missing or changed. Chromosomes are the structures that hold our genes. Down syndrome is a chromosomal disorder. Complex disorders, where there are mutations in two or more genes. Often your lifestyle and environment also play a role. Colon cancer is an example. Genetic tests on blood and other tissue can identify genetic disorders. National Library of Medicine Start Here.

Chapter 7 : Table of Genetic Disorders

The list of hereditary diseases will consider both types of diseases. Some of the disorders are linked to genders. These are when the condition is mostly found in either the X or Y chromosome, so more likely or wholly likely to only affect one gender.

There are four different types of genetic disorders. They are as follows: The fortunate news is that genetic disorders are relatively uncommon, but that said, they are still a real danger and some are certainly more common than others. Here we will look at some of the most common genetic diseases, in no particular order. Cystic Fibrosis Cystic Fibrosis is one of the most widespread inherited genetic disorders. It most often affects Caucasians who are Ashkenazi Jews. It occurs only when both parents are carriers, which gives their children a 1 in 4 chance of contracting the disease. Symptoms include difficulty breathing, recurrent lung infections, digestive, and reproductive issues. This disorder is caused by the mutation of more than one gene and renders a person unable to fight off any kind of germs. It is believed to be caused by the absence of, or a lack of Adenosine Deaminase ADA , as well as notable defects in T and B cell responses. Tay-Sachs Tay-Sachs causes death at any early age, usually around five years old, because of a missing enzyme called Hex-A. Tay-Sachs causes progressive destruction of the nervous system and brain. There is no treatment for the disorder. There is only management of the symptoms. Jackson-Weiss Syndrome This syndrome is caused by a premature fusion of the skull bones, which leads to a deformity of the head and face. This disorder is a very rare one and therefore its occurrence has not been regularly noted. The syndrome, which is caused by a mutation in the FGFR2 gene, presents itself by such symptomatology as an irregularly shaped skull, abnormal placement of the eyes, a bulging forehead, and foot abnormalities. Anyone with the disorder usually lives a normal lifespan. Fragile X Fragile X, a group of genetic conditions, affects families differently. The Fragile X syndrome causes inherited mental disabilities. The disorder causes a range of developmental problems including cognitive impairment and learning disabilities. Males are more severely affected by this disorder than females. Ectrodactyly Also known as Lobster Claw Hand, Ectrodactyly is marked by one or more missing digits on either the foot or hand. Individuals with the condition usually have a cleft where their middle digits should be. People with the condition can suffer from hearing loss as well. Ectrodactyly can be treated, to a certain extent, with corrective surgery. Loss of these nerve cells causes symptoms such as behavioral changes, unusual snake-like movements chorea , uncontrolled movement, difficulty walking, loss of memory, speech and cognitive functions and difficulty in swallowing. Treatment aims to limit the course of the disease. There is also an early-onset form which begins in childhood. Neurofibromatosis Neurofibromatosis NF , a nervous system disorder, causes tumors to form on nerves. About one-half of neurofibromatosis cases are inherited and the other half result from spontaneous genetic mutation. A common finding of this disorder is multiple cafe-au-lait spots brown spots on the skin all over the body. Sickle Cell Disease Having the single expression of the gene for sickle cell does not cause the disease. The gene evolved to protect people from malaria. The sickled cells get stuck in small capillaries and destroy joints and organs. Eventually, the victim will die of organ failure. Thalassemias The thalassemias beta-thalassemia is the most common, alpha-thalassemia is less common is a group of inherited blood disorders where hemoglobin, the oxygen-carrying molecule, is not properly synthesized by the red blood cells. This results in an anemiaâ€”where the usual symptoms are fatigue, an enlarged spleen, easily broken bones, bone pain and shortness of breath. People with thalassemia may also have poor appetite, darkened urine and jaundice a yellowish discoloration of the skin or the white parts of the eyesâ€”jaundice is a signal for liver dysfunction. Infections are common in people with thalassemia.

Chapter 8 : Hereditary Diseases List

Family bonds can be very strong, so strong that several genetic disorders, or conditions, are common in children through their parents or grandparents where diseases can directly be traced back through ancestry or ethnicity. For example, sickle cell anemia is one common genetic disorder that mainly.

Inability to mount initial IgM response to the capsular polysaccharides of pyogenic bacteria. In infancy, recurrent pyogenic infections, eczema, thrombocytopenia, excessive bleeding. IgG levels remain normal. Mutation in gene coding for tyrosine kinase causes failure of Pre-B cells to differentiate into B-Cells. Recurrent pyogenic infections after 6 months when maternal antibodies wear off. Can treat with polyspecific gamma globulin preparations. Angiokeratomas skin lesions over lower trunk, fever, severe burning pain in extremities, cardiovascular and cerebrovascular involvement. Hepatosplenomegaly, erosion of femoral head, mild anemia. Normal lifespan with treatment. Death before age 1. Sphingomyelin-containing foamy histiocytes in reticuloendo-thelial system and spleen. Hepatosplenomegaly, anemia, fever, sometimes CNS deterioration. Death by age 3. Hepatosplenomegaly, micrognathia, retinal degeneration, joint stiffness, mild retardation, cardiac lesions. Gargoyle-like facies, progressive mental deterioration, stubby fingers, death by age Tay-Sachs Disease Autosomal Recessive. CNS degeneration, retardation, cherry red-spot of macula, blindness amaurosis. Death before age 4. Can result from a lack of migration of neural crest cells. Depigmentation, pink eyes, increased risk of skin cancer. Urine turns dark and black on standing, ochronosis dark pigmentation of fibrous and cartilage tissues , ochronotic arthritis, cardiac valve involvement. Disease is generally benign. Cystathionine synthase defect either deficiency, or lost affinity for pyridoxine, Vit. Mental retardation, ectopia lentis, sparse blond hair, genu valgum, failure to thrive, thromboembolic episodes, fatty changes of liver. Cysteine supplementation, give excess pyridoxine to compensate for lost pyridoxine affinity. Severe CNS defects, mental retardation, death. Person smells like maple syrup or burnt sugar. Symptoms result from accumulation of phenylalanine itself. Mental deterioration, hypopigmentation blond hair and blue eyes , mousy body odor from phenylacetic acid in urine and sweat. Can be elicited by drugs primaquine, sulfonamides, aspirin , fava beans favism. More prevalent in blacks. Defect in hexokinase, glucose-phosphate isomerase, aldolase, triose-phosphate isomerase, phosphate-glycerate kinase, or enolase. Any enzyme in glycolysis pathway. Associated with hepatic fibrosis. Elevated renin and aldosterone, hypokalemic alkalosis. Deficient resorption in proximal tubules. Defective resorption in proximal tubules. Similar to Fanconi Syndrome Type I, but without the cystinosis. Adult onset osteomalacia, amino-aciduria, polyuria, glycosuria.

Chapter 9 : Genetic Disorders: MedlinePlus

Inherited metabolic disorders are genetic conditions that result in metabolism problems. Most people with inherited metabolic disorders have a defective gene that results in an enzyme deficiency.

Triple X Syndrome is a disorder that affects only females. The disorder can produce severe or subtle problems. Triple X syndrome is caused by a female inheriting not two, but three X chromosome. Triple X syndrome is not curable, thus treatment is based on symptoms if there are any. Fragile X syndrome is a disorder that can cause a series of diverse physical traits. People with fragile X syndrome have more repeated genes, then unaffected people. The large number of extra repeated gene interferes with regulations, turning off the gene, and stopping the necessary proteins from being synthesized. These proteins are usually found in nerve cells, and help in brain development and brain communication. Treatment usually includes medicine like antidepressants, antiseizure drugs, antipsychotics, and stimulants for hyperactivity and attention problems. This disorder affects only girls and women, and may be diagnosed during infancy or early childhood. There are three form of Turner syndrome: There are no specific signs and symptoms for this disorder, and they generally change as the person develops. Here are some features that appear generally at birth: As the female develops, other symptoms include: Growth hormone, estrogen therapy for sexual development, periodic checkup with a heart specialist, fertility treatment, assign specialist for preventing high-risk pregnancies, and other treatment are provided to increase comfort. Prader-Willi Syndrome is a very rare disorder that results in a number of physical and mental problems. It causes a constant sense of hunger beginning after the first year of life. Prader-Willi syndrome is caused by an unknown gene on chromosome It occurs when certain paternal inherited from the father gene are not expressing themselves. It is a form of inherited anemia. Sickle cell anemia is an autosomal recessive trait, so only a person with both parents passing down the trait can have the disease. Signs of this disease usually show up after an infant is 4 months old. This treatment however, is extremely risky and it is very hard to find a donor. Regular visits to the doctor can help in the monitoring of blood cells, and relieve symptoms. Like other chromosomal disorders, symptoms can varied dramatically from men to men. This disorder is caused by the inheritance of an extra X chromosome in men. The problem probably resulted during an error in the formation of an egg or a sperm. This does not help with infertility however. Speech therapy and special educators can be provided, in addition to psychological therapy for emotional supports. This disease is triggered by consumption of gluten, a protein. Gluten is primarily found in bread, cookies, pasta, wheat, barley, rye, and pizza crust. The body is also not able to absorb these nutrients. This prevents adequate absorption of nutrients. This disease can be easily managed by effectively changing diet to avoid gluten. Vitamin supplements may also be required to combat malnutrition. This disease is genetic in nature, and results in unusually weak muscle fibers that are susceptible to damage. This disease can be caused by mutations to hundreds of genes involved in making proteins for protecting muscle fibers. There are specifically different form of this disease. Other less common form of this disease do exist. Corticosteroids are sometimes provided to help with muscle strength and delay the progression of symptoms. Physical therapy are also recommended to help with exercising, flexibility, and posture. Most people with this disorder die during the fetal stage, and those who survive generally have a wide range of problems which severely shorten their life span. Most have a third chromosome 18, instead of the usual two. It is rather hard for doctor to provide care for children with this disease because most have early death. Children that do live beyond their first year of life may have medical and surgical aids to reduce discomfort and long term chronic condition. Cri Du Chat, French for "cry of the cat", is a disorder that is usually not inherited. The cause of this deletion is unknown. Although there is no cure, children with this disorder can have full and happy lives. Therapy is often provided for physical and language problems. Medical professionals may provide different medications depending on other risks or problems. Autism is a disorder that can cause serious developmental problems in communication and interaction. The number of autism case is growing each year, for unknown reasons. Because of the complex nature of this disorder, it is unknown what causes autism. Genetic and environmental factors have been taken into consideration. Children with this disease have problems in social skills, language, and

behavior. Signs usually occur on one side of the body before spreading to the other side. Lifestyle changes like ongoing aerobic exercises are usually recommended. Deep brain stimulation using electrodes may also be recommended for advance development of this disorder.