DOWNLOAD PDF THE COMPLETE DIRECTORY FOR PEOPLE WITH RARE DISORDERS 2002/03

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Complete Directory for People with Rare Disorders /03 by National Organization for Rare Disorders, Laura Mars, Grey House Publishing (Creator) starting at. Complete Directory for People with Rare Disorders /03 has 0 available edition to buy at Alibris.

Genetic Syndromes What causes genetic disorders? Each cell in the human body contains 23 pairs of chromosomes. You inherit one set of the pair from your father and one set from your mother. Each chromosome is made up of many genes , about 2, in each chromosome, for a total of 50, genes in each cell. These genes make all the proteins in the body, which promote development and growth, and carry out all body functions. When one or more of these genes or chromosomes are missing or mutated , or if extra chromosomes are present, the proteins may not get made, may be made incorrectly, or too many may be made. Any of these situations can cause abnormal development and growth and can result in a genetic syndrome. Sometimes these abnormal genes or chromosomes are passed down from a parent, and sometimes they occur spontaneously without reason. For help in understanding the terms used in genetics, see the talking glossary of genetic terms from the National Human Genome Research Institute. Here, you can listen to detailed explanations of these complicated terms. What are genetic syndromes? A syndrome is a disease or disorder that has more than one identifying feature or symptom. Each particular genetic syndrome will have many typical features, depending on which aspects of development are affected by the abnormal genes or chromosomes. A child might be born with obvious body deformities, abnormal organ function for example: However, many of the genetic syndromes start to take effect only once the baby has been born and is starting to feed and grow. These babies may look and act entirely normal at birth, but then develop problems later on in life. This is a big and complex topic, and we are learning more and more about genetic syndromes every day. How can I find out more to help me understand this stuff? Here are some links to sites that explain more about genes and genetic syndromes: What Are Genetic Disorders?

Chapter 2 : HON - List of Rare Diseases

C Cataract A condition in which the lens of the eye, which is normally clear, becomes cloudy or opaque. Cataracts generally form slowly and without pain. They can affect one or both eyes. Over time, a cataract may interfere with vision, causing images to appear blurred or fuzzy and colors to seem faded. Most cataracts are related to aging. In fact, cataracts affect more than 50 percent of all adults by age 80 and are the primary cause of vision loss in people 55 and older. People with early cataract may benefit from new eyeglasses, bright lighting, anti-glare sunglasses, or magnifying lenses. If, despite such devices, cataract interferes with daily activities, surgery is the only effective treatment. Cataract surgery, which is common, involves removal of the cloudy lens and replacement with an artificial lens. People with Charles Bonnet syndrome may see a wide range of images, from simple patterns to people, animals, and buildings. The visual disturbances associated with this syndrome are not signs of mental illness, and people realize that the images they are seeing are not real. There is no cure for Charles Bonnet syndrome. However, the symptoms often stop on their own. People who have Charles Bonnet syndrome should consult with an eye care specialist because treatment for vision disorders may help. It affects males more than females. It is an autosomal dominant disorder caused by mutations in the CRB1 gene. Choroidal Neovascularization Choroidal neovascularization refers to new and abnormal blood vessels that grow, multiply, and develop into a cluster beneath the macula. The macula is the part of the retina that provides the clearest central vision. Choroideremia Rare disorder that causes progressive loss of the choroid, an important layer under the retina that is responsible for some of its blood supply. Choroideremia is an inherited disorder that generally affects males only. It commonly begins as night blindness in childhood and gradually advances to increasing vision loss. Most people with this disorder are able to retain good vision until age 40 or There is no treatment for choroideremia, but people who have the disorder may find it helpful to use optical, electronic, or computer-based devices for low vision. For example, someone with only a tiny defect in the iris may have normal vision. However, a person with large defects in the retina and optic nerve may have limited vision. Children whose vision is impaired by coloboma may benefit from using reading materials that have large black print and well-spaced letters and words. They may also find it helpful to read one line at a time with the aid of a cutout reading window. Color blindness is not really a form of blindness, but rather a deficiency in color perception. It usually affects both eyes and is much more common in males than in females. There is no treatment or cure for this problem, but a color-blind person can learn to adapt in various ways. For example, a color-blind driver can remember that the light positioned at the top of a traffic light is the red one. It is beneficial to diagnose color blindness in children at an early age so that steps can be taken to avoid learning problems related to color perception. People with cone-rod dystrophy typically experience decreased sharpness of vision followed by a loss of peripheral vision and color perception. The most common form of cone-rod dystrophy is retinitis pigmentosa. There is no treatment or cure for this disease, which is also referred to as cone-rod degeneration, progressive cone-rod dystrophy, and retinal cone dystrophy. Some congenital eye conditions, such as retinitis pigmentosa , are passed on through genes. Others, such as vision loss due to German measles, result from a disease or deficiency during pregnancy. Sometimes, as in the case of coloboma , the cause of a congenital eye defect is not known. Congenital eye defects can impair vision or even cause blindness. Some conditions are immediately apparent in an infant, while others may not become known until later in life. Conjunctivitis Conjunctivitis is inflammation of the conjunctiva, which is the thin translucent tissue that lines the inner surface of the eyelid and the outer surface of the sclera, which is the white part of the eye. There are many different causes of conjunctivitis. Some types of conjunctivitis are infectious, while others are not. These can generally be differentiated from one another based on history and an examination by an eye doctor.

Conjunctivitis Corneal disease Disease or disorder that affects the cornea, the clear, curved surface that covers the front of the eye. The effects of corneal disease vary. Some corneal conditions cause few, if any, vision problems. For example, infections of the cornea can often be treated with antibiotics. However, if the cornea becomes cloudy, light cannot penetrate the eye to reach the retina, and severe visual impairment, or even blindness, may result. Corneal dystrophies are usually inherited conditions in which one or more parts of the cornea lose their clarity due to a buildup of cloudy material. Keratoconus is the most common corneal dystrophy in the United States. When corneal disease causes the cornea to become permanently clouded or scarred, doctors may be able to restore vision with a corneal transplantâ€”surgical replacement of the old cornea with a new one. Although the eye is normal, the brain cannot properly process the information it receives. The degree of vision loss may be mild or severe and can vary greatly, even from day to day. Also known as cerebral visual impairment, cortical visual impairment CVI may be temporary or permanent. People with cortical visual impairment have difficulty using what their eye sees. For example, they may have trouble recognizing faces, interpreting drawings, perceiving depth, or distinguishing between background and foreground. Children with cortical visual impairment are often able to see better when told in advance what to look for. Cortical visual impairment is also known as neurological visual impairment NVI.

Chapter 3 : Dysphagia | Swallowing Disorder | MedlinePlus

The complete directory for people with rare disorders: a comprehensive guide to over 1, rare disorders from the National Organization for Rare Disorders, Inc.

Where can I get more information? What is Creutzfeldt-Jakob disease? Creutzfeldt-Jakob disease CJD is a rare, degenerative, fatal brain disorder. It affects about one person in every one million per year worldwide; in the United States there are about cases per year. CJD usually appears in later life and runs a rapid course. Typical onset of symptoms occurs at about age 60, and about 70 percent of individuals die within one year. In the early stages of the disease, people may have failing memory, behavioral changes, lack of coordination, and visual disturbances. As the illness progresses, mental deterioration becomes pronounced and involuntary movements, blindness, weakness of extremities, and coma may occur. There are three major categories of CJD. In sporadic CJD, the disease appears even though the person has no known risk factors for the disease. This is by far the most common type of CJD and accounts for at least 85 percent of cases. In hereditary CJD, the person may have a family history of the disease and test positive for a genetic mutation associated with CJD. In acquired CJD, the disease is transmitted by exposure to brain or nervous system tissue, usually through certain medical procedures. CJD belongs to a family of human and animal diseases known as the transmissible spongiform encephalopathies TSEs or prion diseases. Spongiform refers to the characteristic appearance of infected brains, which become filled with holes until they resemble sponges when examined under a microscope. Kuru was identified in people of an isolated tribe who practiced ritual cannibalisms in Papua, New Guinea and has now almost disappeared. Kuru is considered an acquired prion disease. To date, about cases of vCJD, mostly in the United Kingdom, have been reported related to consuming beef but none in which the disease was acquired in the U. Other TSEs are found in specific kinds of animals. These include BSE, mink encephalopathy, feline encephalopathy, and scrapie, which affects sheep and goats. Chronic wasting disease CWD affects elk and deer and is increasingly prevalent in certain areas in the United States. To date no transmission of CWD to humans has been reported. Although sporadic TSE includes five distinct subtypes of sporadic CJD and sporadic fatal insomnia sFI , overall they are characterized by rapidly progressive dementia. Initially, individuals experience problems with muscle coordination, personality changes including impaired memory, judgment, and thinking , and impaired vision. People with the disease, especially with FFI, also may experience insomnia, depression, or unusual sensations. They often develop involuntary muscle jerks called myoclonus, and they may go blind. They eventually lose the ability to move and speak, and enter a coma. Pneumonia and other infections often occur in these individuals and can lead to death. Variant CJD begins primarily with psychiatric symptoms, affects younger individuals than other types of CJD, and has a longer than usual duration from onset of symptoms to death. However, CJD causes unique changes in brain tissue which can be seen at autopsy. Current scientific consensus maintains that abnormal forms of normal cellular proteins called prions cause CJD in people and TSE in animals. The normal, harmless prion is usually designated PrPC C stands for cellular and the abnormal, infectious form which causes the disease is PrPSc Sc stands for prototypical prion diseaseâ€”scrapie. Proteins are long chains of amino acids that have to fold together into a unique shape or conformation to gain function in the cells. Research findings indicate that the infectious prion originates from a normal protein whose conformation has changed to one that causes the disease. The normal prion protein is found throughout the body but is most abundant in the nervous system. Its overall role is not fully understood. It is believed that the harmless to infectious protein conformational change is common to the all major forms of human prion disease, including CJD. In the acquired form of the disease, the PrPSc comes from the outside the body, for example, through contaminated meat as is seen in vCJD. It then clings to and changes the conformation of the normal prion protein of the host and progressively spreads in domino-like fashion toward the brain where it causes lesions. As the mutated PrPC replicates itself, it spontaneously changes shape into the infectious form. Prions themselves do not

contain genetic information and do not require genes to reproduce themselves. Several different mutations in the prion gene have been identified. The particular mutation found in each family affects how frequently the disease appears and what symptoms are most noticeable. However, not all people with mutations in the prion protein gene develop CJD. In the sporadic form, the infectious prions are believed to be made by an error of the cell machinery that makes proteins and controls their quality. These errors are more likely to occur with aging, which explains the general advanced age at onset of CJD and other prion diseases. Once they are formed, abnormal prion proteins aggregate, or clump together. Investigators think these protein aggregates lead to the nerve cell loss and other brain damage seen in CJD. However, they do not know exactly how this damage occurs. CJD cannot be transmitted through the air or through touching or most other forms of casual contact. Spouses and other household members of people with sporadic CJD have no higher risk of contracting the disease than the general population. However, exposure to brain tissue and spinal cord fluid from infected persons should be avoided to prevent transmission of the disease through these materials. In some cases, CJD has spread to other people from grafts of dura mater a tissue that covers the brain , transplanted corneas, implantation of inadequately sterilized electrodes in the brain, and injections of contaminated pituitary growth hormone derived from human pituitary glands taken from cadavers. Doctors call these cases that are linked to medical procedures iatrogenic cases. Since , all human growth hormone used in the United States has been synthesized by recombinant DNA procedures, which eliminates the risk of transmitting CJD by this route. Many people are concerned that it may be possible to transmit CJD through blood and related blood products such as plasma. Some animal studies suggest that contaminated blood and related products may transmit the disease, although this has never been shown in humans. Recent studies suggest that while there may be prions in the blood of individuals with vCJD, this is not the case in individuals with sporadic CJD. Scientists do not know how many abnormal prions a person must receive before he or she develops CJD, so they do not know whether these fluids are potentially infectious or not. They do know that, even though millions of people receive blood transfusions each year, there are no reported cases of someone contracting sporadic CJD from a transfusion. Even among people with hemophilia a rare bleeding disorder in which the blood does not clot normally , who sometimes receive blood plasma concentrated from thousands of donors, there are no reported cases of CJD. While there is no evidence that blood from people with sporadic CJD is infectious, studies have found that infectious prions from BSE and vCJD accumulate in the lymph nodes which produce white blood cells , the spleen, and the tonsils. At present, four cases of vCJD infection have been identified following transfusion of red blood cells from asymptomatic donors who subsequently died from vCJD. Recently, one case of likely transmission of vCJD infection by concentrates of blood-clotting protein has been reported in an elderly individual with hemophilia in the United Kingdom. The possibility that blood from people with vCJD may be infectious has led to a policy preventing individuals in the United States from donating blood if they have resided for more than three months in a country or countries where BSE is common. Both brain biopsy and autopsy pose a small, but definite, risk that the surgeon or others who handle the brain tissue may become accidentally infected by self-inoculation. Special surgical and disinfection procedures can markedly reduce this risk. How is CJD diagnosed? Several tests can help diagnose CJD. RT-QuIC is based on an ultrasensitive detection of the pathogenic prion protein in the cerebrospinal fluid of individuals affected by CJD and other forms of human prion diseases. This new advanced test demonstrates a very high sensitivity and specificity of the disease. RT-QuIC differs from traditional surrogate markers of prion disease â€” and tau proteinsâ€”in that it detects directly a disease-defining pathogenic prion protein as opposed to a surrogate marker of rapid neurodegeneration. Detection of these traditional surrogate marker proteins is accurate in approximately three-fourths of cases. Magnetic resonance imaging MRI has recently been found to be accurate in about 90 percent of cases. The only way to confirm a diagnosis of CJD is by brain biopsy or autopsy. This procedure may be dangerous for the individual, and the operation does not always obtain tissue from the affected part of the brain. Because a correct diagnosis of CJD does not help the individual, a brain biopsy is discouraged unless it is needed to rule out a treatable disorder. In an autopsy, the

whole brain is examined after death. Currently, there is no treatment that can cure or control CJD, although studies of a variety of drugs are now in progress. Current treatment for CJD is aimed at easing symptoms and making the person as comfortable as possible. Opiate drugs can help relieve pain if it occurs, and the drugs clonazepam and sodium valproate may help relieve myoclonus. During later stages of the disease, intravenous fluids and artificial feeding also may be used. To reduce the already very low risk of CJD transmission from one person to another, people should never donate blood, tissues, or organs if they have suspected or confirmed CJD, or if they are at increased risk because of a family history of the disease, a dura mater graft, or other factor. Normal sterilization procedures such as cooking, washing, and boiling do not destroy prions. Although there is no evidence that caregivers, healthcare workers, and those who prepare bodies for funerals and cremation have increased risk of prion diseases when compared to general population, they should take the following precautions when they are working with a person with CJD: Cover cuts and abrasions with waterproof dressings. Use disposable bedclothes and other cloth for contact with the person. If disposable materials are not available, regular cloth should be soaked in undiluted chlorine bleach for an hour or more, and then washed in a normal fashion after each use. Use face protection if there is a risk of splashing contaminated material such as blood or cerebrospinal fluid. Soak instruments that have come in contact with the person in undiluted chlorine bleach for an hour or more, then use an autoclave pressure cooker to sterilize them in distilled water for at least one hour at - degrees Celsius. The mission of the National Institute of Neurological Disorders and Stroke NINDS is to seek fundamental knowledge of the brain and nervous system and to use that knowledge to reduce the burden of neurological disease. Researchers are examining and characterizing the prions associated with CJD and other human and animal prion diseases and trying to discover factors that influence prion infectivity and transmission, and how the disorder damages the brain. For example, researchers are investigating the cellular mechanisms involved in abnormal prion formation and accumulation, as well as their replication by select cellular subsets in the brain. Other projects are examining how abnormal prions cross the protective blood-brain barrier and spread throughout the central nervous system, and tests that measure the biological activity of prions. Findings may identify new therapeutic targets to treat prion diseases. Scientists are conducting biochemical analyses of brain tissue, blood, spinal fluid, urine, and serum in the hope of determining the nature of the transmissible agent or agents causing CJD. To help with this research, they are seeking biopsy and autopsy tissue, blood, and cerebrospinal fluid from individuals with CJD and related diseases. The following investigators have expressed an interest in receiving such material:

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Chapter 4 : Agenesis of the corpus callosum - Wikipedia

Similar to other Grey House publications (e.g., The Complete Directory for People with Chronic Illness, 3d ed.), this resource offers a unique compilation of data for lay readers, healthcare professionals, and librarians on over rare disorders.

What is a Rare Disease? Key figures A disease or disorder is defined as rare in Europe when it affects fewer than 1 in A disease or disorder is defined as rare in the USA when it affects fewer than , Americans at any given time. One rare disease may affect only a handful of patients in the EU European Union , and another may touch as many as , Characteristics of rare diseases Over rare diseases are characterised by a broad diversity of disorders and symptoms that vary not only from disease to disease but also from patient to patient suffering from the same disease. Relatively common symptoms can hide underlying rare diseases leading to misdiagnosis and delaying treatment. Quintessentially disabling, the patients quality of life is affected by the lack or loss of autonomy due to the chronic, progressive, degenerative, and frequently life-threatening aspects of the disease. The fact that there are often no existing effective cures adds to the high level of pain and suffering endured by patients and their families. Common problems faced The lack of scientific knowledge and quality information on the disease often results in a delay in diagnosis. Also the need for appropriate quality health care engenders inequalities and difficulties in access to treatment and care. This often results in heavy social and financial burdens on patients. As mentioned, due to the broad diversity of disorders and relatively common symptoms which can hide underlying rare diseases, initial misdiagnosis is common. In addition, symptoms differ not only from disease to disease, but also from patient to patient suffering from the same disease. Due to the rarity and diversity of rare diseases, research needs to be international to ensure that experts, researchers and clinicians are connected, that clinical trials are multinational and that patients can benefit from the pooling of resources across borders. How can things change? Although rare disease patients and their families face many challenges, enormous progress is being made every day. The ongoing implementation of a better comprehensive approach to rare diseases has led to the development of appropriate public health policies. Important gains continue to be made with the increase of international cooperation in the field of clinical and scientific research as well as the sharing of scientific knowledge about all rare diseases, not only the most "recurrent" ones. Both of these advances have led to the development of new diagnostic and therapeutic procedures. Rare Disease Day is a great example of how progress continues to be made, with events being held worldwide each year. However, the road ahead is long with much progress to be made.

Chapter 5 : Rare Disease Day Â® - Article

Full text Full text is available as a scanned copy of the original print version. Get a printable copy (PDF file) of the complete article (K), or click on a page image below to browse page by page.

Chapter 6 : Diseases & Conditions A-Z Index - A

The Complete Directory For People With Rare Disorders 99 Coping with chronic, rare, and invisible diseases and, coping with chronic, rare, and.

Chapter 7 : Creutzfeldt-Jakob Disease Fact Sheet | National Institute of Neurological Disorders and Stroke

â€¢ The Complete Directory for People with Rare Disorders: A Comprehensive Guide to Over 1, Rare Disorders from the National Organization for Rare Disorders Lakeville, CT: Grey House Pub.,

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Chapter 8 : Autoimmune Disease List â€¢ AARDA

The Complete Directory For People With Rare Disorders 03 A Comprehensive Guide To Over 1 Rare Disorders From The National Complete Guide Market Breadth.

Chapter 9 : EURORDIS - The Voice of Rare Disease Patients in Europe

If you are seeking information about a rare disease that is not in this database, we would suggest contacting the Genetic and Rare Diseases Information Center (GARD) at the National Institutes of Health. NIH has the most complete database of rare diseases in the U.S.