

# ANNEX 3

Information obtained on Internet (not an exhaustive research- general introduction to medical information available via the Internet)

## **RARE DISEASES INFORMATION**

### **1. Vascular Dementias**

[http://www.mental-health-matters.com/disorders/dis\\_details.php?disID=30](http://www.mental-health-matters.com/disorders/dis_details.php?disID=30)

Vascular (formerly arteriosclerotic) dementia, which includes multi-infarct dementia, is distinguished from dementia in Alzheimer's disease by its history of onset, clinical features, and subsequent course. Typically, there is a history of transient ischaemic attacks with brief impairment of consciousness, fleeting pareses, or visual loss. The dementia may also follow a succession of acute cerebrovascular accidents or, less commonly, a single major stroke. Some impairment of memory and thinking then becomes apparent. Onset, which is usually in later life, can be abrupt, following one particular ischaemic episode, or there may be more gradual emergence. The dementia is usually the result of infarction of the brain due to vascular diseases, including hypertensive cerebrovascular disease. The infarcts are usually small but cumulative in their effect.

The diagnosis presupposes the presence of a dementia as described above. Impairment of cognitive function is commonly uneven, so that there may be memory loss, intellectual impairment, and focal neurological signs. Insight and judgement may be relatively well preserved. An abrupt onset or a stepwise deterioration, as well as the presence of focal neurological signs and symptoms, increases the probability of the diagnosis; in some cases, confirmation can be provided only by computerized axial tomography or, ultimately, neuropathological examination.

Associated features are: hypertension, carotid bruit, emotional lability with transient depressive mood, weeping or explosive laughter, and transient episodes of clouded consciousness or delirium, often provoked by further infarction. Personality is believed to be relatively well preserved, but personality changes may be evident in a proportion of cases with apathy, disinhibition, or accentuation of previous traits such as egocentricity, paranoid attitudes, or irritability.

**Vascular Dementia (see AE info already available)**

## **Subcortical Vascular Dementia**

There may be a history of hypertension and foci of ischaemic destruction in the deep white matter of the cerebral hemispheres, which can be suspected on clinical grounds and demonstrated on computerized axial tomography scans. The cerebral cortex is usually preserved and this contrasts with the clinical picture, which may closely resemble that of dementia in Alzheimer's disease. (Where diffuse demyelination of white matter can be demonstrated, the term "Binswanger's encephalopathy" may be used.)

Other websites of interest:

<http://www.uku.fi/neuro/ab0234.htm>

Very detailed article/study of vascular dementia which includes the subcortical type

[http://www.thomasland.com/\\_nonsearch/TSR07305.pdf](http://www.thomasland.com/_nonsearch/TSR07305.pdf)

[http://www.mental-health-matters.com/disorders/dis\\_details.php?disID=30](http://www.mental-health-matters.com/disorders/dis_details.php?disID=30)

<http://www.cpa.ca/factsheets/cognitive.htm>

Website with details of a drug trial for the treatment of subcortical vascular dementia

<http://www.bscc.co.uk/newsdigest/news0502.html>

## **Multi-infarct dementia**

[http://www.ninds.nih.gov/health\\_and\\_medical/disorders/multi-infarctdementia\\_doc.htm](http://www.ninds.nih.gov/health_and_medical/disorders/multi-infarctdementia_doc.htm)

### **What is Multi-Infarct Dementia?**

Multi-infarct dementia (MID), a common cause of dementia in the elderly, occurs when blood clots block small blood vessels in the brain and destroy brain tissue. Probable risk factors are high blood pressure and advanced age. CADASIL (cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy) is an inherited form of MID. This disease can cause stroke, dementia, migraine-like headaches, and psychiatric disturbances. Symptoms of MID, which often develop in a stepwise manner, include confusion, problems with recent memory, wandering or getting lost in familiar places, loss of bladder or bowel control (incontinence), emotional problems such as laughing or crying inappropriately, difficulty following instructions, and problems handling money. Usually the damage is so slight that the change is noticeable only as a series of small steps. However over time, as more small vessels are blocked, there is a gradual mental decline. MID, which typically begins between the ages of 60 and 75, affects men more often than women.

### **Is there any treatment?**

Currently there is no treatment for MID that can reverse the damage that has already occurred. Treatment focuses on prevention of additional brain damage by controlling high blood pressure.

### **What is the prognosis?**

Prognosis for patients with MID is generally poor. Individuals with the disease may improve for short periods of time, then decline again. Early treatment and management of blood pressure may prevent further progression of the disorder.

## **What research is being done?**

The NINDS supports and conducts a wide range of research on dementing disorders such as MID and on cerebrovascular disease. The goals of this research are to improve the diagnosis of these disorders and to find ways to treat and prevent them.

## **Other websites of interest:**

<http://www.mentalhealth.com/dis/p20-or02.html>  
<http://www.nmha.org/infoctr/factsheets/102.cfm>  
<http://www.nlm.nih.gov/medlineplus/ency/article/000746.htm>

This is a fairly interesting site with an online journal written by someone with multi-infarct dementia...not sure how useful it is though...

<http://www.geocities.com/resa526/journal2001.html>  
<http://www.alzheimers.org/pubs/mid.htm>

This address is another part of the "slide show" website- brain scans etc:

[http://brighamrad.harvard.edu/education/online/BrainSPECT/Main\\_Slide\\_Show/Main\\_SS\\_9.html](http://brighamrad.harvard.edu/education/online/BrainSPECT/Main_Slide_Show/Main_SS_9.html)  
[http://www.psychnet-uk.com/dsm\\_iv/multi\\_infarct\\_dementia.htm](http://www.psychnet-uk.com/dsm_iv/multi_infarct_dementia.htm)  
[http://www.allsands.com/Health/Diseases/multiinfarctde\\_xmr\\_gn.htm](http://www.allsands.com/Health/Diseases/multiinfarctde_xmr_gn.htm)  
<http://www.acnp.org/G4/GN401000146/CH143.html>

Interesting website on clinical trials:

<http://www.clinicaltrials.gov/ct/gui/info/resources;jsessionid=32B4F0314303B2840850F5A888B24615>

Easy to use website with info on carers, general caring for the elderly and several dementia-related diseases:

<http://www.helpguide.org/dementias/multiinfarct.asp>

Informative website with chapters on all the rare diseases:

[http://www.dasninternational.org/regular/reg\\_information.html](http://www.dasninternational.org/regular/reg_information.html)

Address for Los Angeles Alzheimer's association:

<http://www.alzla.org>

Address for the Texan Department of Health, Alzheimer's Program:

<http://www.tdh.state.tx.us/osp/alz.htm>

List of contact numbers for Alzheimer associations around the USA:

[http://www.tdh.state.tx.us/osp/a\\_info.htm](http://www.tdh.state.tx.us/osp/a_info.htm)

Alzheimer's association San Diego (USA):

[http://www.sanhttp://www.health-nexus.com/multi-infarct\\_dementia.html](http://www.sanhttp://www.health-nexus.com/multi-infarct_dementia.html)  
[http://www.alz.org/about\\_disease/overview.html](http://www.alz.org/about_disease/overview.html)

### **Vascular Dementia of Acute Onset**

[http://www.mental-health-matters.com/disorders/dis\\_details.php?disID=30](http://www.mental-health-matters.com/disorders/dis_details.php?disID=30)

Usually develops rapidly after a succession of strokes from cerebrovascular thrombosis, embolism, or haemorrhage. In rare cases, a single large infarction may be the cause.

### **Mixed Cortical and Subcortical Vascular Dementia**

[http://www.mental-health-matters.com/disorders/dis\\_details.php?disID=30](http://www.mental-health-matters.com/disorders/dis_details.php?disID=30)

Mixed cortical and subcortical components of the vascular dementia may be suspected from the clinical features, the results of investigations (including autopsy), or both.

### **Binswanger Disease**

[http://www.ninds.nih.gov/health\\_and\\_medical/disorders/binswang\\_doc.htm](http://www.ninds.nih.gov/health_and_medical/disorders/binswang_doc.htm)

#### **What is Binswanger's Disease?**

Binswanger's disease is a rare form of dementia characterized by cerebrovascular lesions in the deep white-matter of the brain, loss of memory and cognition, and mood changes. Patients usually show signs of abnormal blood pressure, stroke, blood abnormalities, disease of the large blood vessels in the neck, and disease of the heart valves. Other prominent features of the disease include urinary incontinence, difficulty walking, clumsiness, slowness of conduct, lack of facial expression, and speech difficulties. These symptoms, which tend to begin after the age of 60, are not always present in all patients and may sometimes appear only as a passing phase. Seizures may also be present.

#### **Is there any treatment?**

There is no specific course of treatment for Binswanger's disease. Treatment is symptomatic, often involving the use of medications to control high blood pressure, depression, heart arrhythmias and low blood pressure.

#### **What is the prognosis?**

Binswanger's disease is a slowly progressive condition for which there is no cure. The disorder is often marked by strokes and partial recovery. Patients with this disorder usually die within 5 years after its onset.

#### **What research is being done?**

The NINDS conducts and supports a wide range of research on dementing disorders, including dementias of old age such as Binswanger's disease. The goals of this research are to improve the diagnosis of dementias and to find ways to treat and prevent them. The National Institute on Aging and the National Institute of Mental Health also support research related to the dementias.

[http://www.rarediseases.org/search/rdbdetail\\_abstract.html?disname=Binswanger%27s%20Disease](http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=Binswanger%27s%20Disease)

### **Synonyms of Binswanger's Disease**

- Binswanger's Encephalopathy
- Multi-infarct Dementia, Binswanger's Type
- SAE
- Subcortical Arteriosclerotic Encephalopathy
- Vascular Dementia, Binswanger's Type
- Disorder Subdivisions

## **General Discussion**

Binswanger's Disease is a progressive neurological disorder characterized by degeneration of the white matter of the brain. Affected individuals usually experience a gradual loss of motor, cognitive, and behavioral abilities during a ten year period. In some cases, symptoms and physical findings associated with Binswanger's Disease may stabilize or improve for a brief time; however, in most cases, progression of the disorder usually returns. Affected individuals experience progressive memory loss and deterioration of intellectual abilities (dementia), strokes, paralysis of side of the body (hemiparesis), electrical disturbance in the brain (seizures), and/or an abnormally slow, unsteady walk (abnormal gait). Affected individuals often become depressed, uncaring (apathetic), inactive, unable to make decisions (abulic), hardly speak, and show poor judgment. In addition, affected individuals may exhibit difficulty forming words (dysarthria), swallowing difficulties (dysphagia), and inability to control the release of urine (incontinence). In some cases, affected individuals may demonstrate abnormalities that are similar to those seen in Parkinson Syndrome, such as tremors; short, shuffling steps; loss of trunk mobility; and/or loss of coordination (synergy) between upper limb and trunk movements when walking. Individuals with Binswanger's Disease may be at greater risk to develop narrowing and hardening of blood vessels (arteriosclerosis) and high blood pressure (hypertension) than the general population. The exact cause of Binswanger's Disease is not known.

## **Other websites of interest:**

<http://www.clevelandclinic.org/health/health-info/docs/1200/1256.asp?index=6016>

<http://healthlink.mcw.edu/article/921389325.html>

<http://serendip.brynmawr.edu/bb/neuro/neuro01/web2/Ledoux.html>

Address for the National Organisation of Rare Diseases:

[http://www.rarediseases.org/search/rdbdetail\\_abstract.html?disname=Binswanger's%20Disease](http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=Binswanger's%20Disease)

<http://hvelink.saint-lukes.org/library/healthguide/IllnessConditions/topic.asp?hwid=nord614>

<http://archneur.ama-assn.org/issues/v56n9/fpdf/ned8355.pdf>

Alzheimer's association of Toronto (Binswanger's disease plus other rare diseases):

<http://www.asmt.org/RD2.htm>

## **Dementia with Lewy Bodies (see also AE info)**

<http://www.emedicine.com/neuro/topic91.htm>

### **Synonyms and related keywords:**

Lewy body variant of Alzheimer disease, diffuse Lewy body disease, senile dementia of the Lewy body type

DLB is a progressive degenerative dementia. Clinical features that help distinguish DLB from AD include the following:

Fluctuations in cognitive function with varying levels of alertness and attention

Visual hallucinations

Parkinsonian motor features

Although extrapyramidal features may occur late in the course of AD, they appear relatively early in DLB.

Whereas patients with AD virtually always have anterograde memory loss as a prominent symptom and sign early in the course of the illness, anterograde memory loss may be less prominent in DLB.

Executive function deficits (ie, problems with attention and visuospatial functioning) may be more prominent in DLB than in AD.

Other symptoms that may alert clinicians to the diagnosis of DLB (versus AD) include the following:

Nonvisual hallucinations

Delusions

Unexplained syncope

Rapid eye movement sleep disorder

Neuroleptic sensitivity

### **Physical:**

Patients usually have impaired cognition consistent with dementia.

An important observation during mental status testing is that the patient has periods of being alert, coherent, and oriented that alternate with periods of being confused and unresponsive to questions (although awake). This fluctuation is a relatively specific feature of DLB.

Retrieval from memory may be relatively worse than memory storage.

Patients may do relatively well with confrontation naming tests and poorly on tests of visuospatial skills (eg, drawing a clock, copying figures).

Patients may have some parkinsonian signs but usually not enough to meet the criteria for diagnosis of PD.

Mild gait impairment is relatively frequent and should not be ascribed to "old age" or osteoarthritis.

Resting tremor occurs less frequently than in PD.

Myoclonus may occur before severe dementia.

### **Medical Care:**

Selegine, Vitamin E, levodopa, risperidone, olanzapine and clozapine are the standard medications used to treat this disease.

### **Consultations:**

Spouses, family members, and caregivers of patients with DLB frequently realize that the DLB patient behaves differently than typical patients with AD. Primary caregivers (or neurologists not specializing in dementia) frequently are unable to adequately explain these differences. In such situations, referral to a dementia specialist can be helpful.

**Diet:**

No dietary restrictions are indicated except for patients with severe disease who have swallowing impairment.

**Activity:**

Physical therapy and exercise classes can be useful to maintain mobility.  
Advise families of potential problems faced by patients with DLB who drive.

**Prognosis:**

DLB is a disorder of inexorable progression.

Rate of progression varies, and some investigators think that progression is faster than in AD.

Patients eventually die from complications of immobility, poor nutrition, and swallowing difficulties.

Other websites of interest:

<http://www.zarcrom.com/users/alzheimers/odem/lewy-d.html>

<http://www.nottingham.ac.uk/pathology/lewy/lewyhome.html>

<http://www.alznorcal.org/research/resrchlewy.html>

<http://www.lewybodydisease.org/>

**CADASIL**

<http://www.thedoctorsdoctor.com/diseases/cadasil.htm>

CADASIL stands for *cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy*. It is a disease of young adults and presents with migraines with or without an aura, mood disturbances, focal neurologic deficits, strokes, and dementia. Most patients will show symptoms by age 60 years. There are recurrent subcortical ischemic events causing permanent deficits in as many as 2/3 of patients. Unfortunately, there are no laboratory screening tests except for a mild elevation of cerebrospinal fluid protein in 25% of cases. Recently, electron microscopic studies performed upon a skin biopsy may yield the diagnostic changes (see the outline below).

**Synonyms:**

- Hereditary multi-infarct dementia
- Chronic familial vascular encephalopathy
- Familial disorder with subcortical ischemic strokes
- Agnogenic medial arteriopathy
- Familial Binswanger's disease

**Age Range-meridian:**

30-40 years

Death usually occurs 12-23 years after onset of symptoms

Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) is a hereditary cerebrovascular disease leading to cognitive decline and dementia.

CADASIL usually begins with migraine in about one third of the patients. More severe manifestations, transient ischemic attacks or recurrent strokes, appear between 30 and 50 years of age .

**Other websites of interest:**

<http://www.thedoctorsdoctor.com/diseases/cadasil.htm>

<http://www.genetics.ucla.edu/schanenlab/cadasil.html>

<http://brainpath.medsch.ucla.edu/abstracts/vol12/1203/1203a9.htm>

Journal study of CADASIL patients

<http://www.ahsnet.org/journal/1999/02/Feb99abstrts.pdf>

**Corticobasal Degeneration**

[http://www.ninds.nih.gov/health\\_and\\_medical/disorders/cortico\\_doc.htm](http://www.ninds.nih.gov/health_and_medical/disorders/cortico_doc.htm)

**What is Corticobasal Degeneration?**

Corticobasal degeneration is a progressive neurological disorder characterized by nerve cell loss and atrophy (shrinkage) of multiple areas of the brain including the cerebral cortex and the basal ganglia. Corticobasal degeneration progresses gradually. Initial symptoms, which typically begin at or around age 60, may first appear on one side of the body (unilateral), but eventually affect both sides as the disease progresses. Symptoms include signs of parkinsonism such as poor coordination, akinesia (an absence of movements), rigidity (a resistance to imposed movement), and disequilibrium (impaired balance); and limb dystonia (abnormal muscle postures). Other symptoms such as cognitive and visual-spatial impairments, apraxia (loss of the ability to make familiar, purposeful movements), hesitant and halting speech, myoclonus, and dysphagia (difficulty swallowing) may also occur. The patient is unable to walk. Symptoms vary among patients.

**Is there any treatment?**

There is no treatment available to slow the course of corticobasal degeneration, and the symptoms of the disease are generally resistant to therapy. Antiparkinsonian drugs do not produce any significant or sustained improvement. Clonazepam may help the myoclonus. Occupational, physical, and speech therapy may help in managing disability.

**What is the prognosis?**

The course of corticobasal degeneration is one of inexorable progression until death, usually 6 to 8 years after diagnosis. Death is generally caused by pneumonia or other complications of severe debility, such as sepsis or pulmonary embolism.

**What research is being done?**

The NINDS supports and conducts research studies on degenerative disorders such as corticobasal degeneration. The goals of these studies are to increase scientific understanding of these disorders and to find ways to prevent, treat, and cure them.

**Other websites of interest:**

<http://www.emedicine.com/NEURO/topic77.htm>

<http://www.cmdg.org/Movement/Parkinsons/Plus/CBGD/cbgd.htm>

<http://www.njneuro.org/movedis/cbgd.htm>

<http://www.neurologyindia.com/vol48-4/le3.pdf>



website with journals on corticobasal degeneration

<http://jama.ama-assn.org/issues/v284n14/fpdf/jbk1011.pdf>

Med Help website. Need to get user name etc to log in, but contains journals, clinical trials etc

[http://www.medscape.com/viewarticle/405229\\_print](http://www.medscape.com/viewarticle/405229_print)

clinical trials website

<http://clinicaltrials.gov/ct/gui/show/NCT00017940?order=17>

### **Normal Pressure Hydrocephalus**

<http://www.allaboutnph.com/>

For most patients the cause of NPH cannot be determined. In some cases, history of previous brain injury or surgery can result in hydrocephalus. Examples are brain hemorrhage, aneurysm, trauma, tumors or cysts, infections or subdural hematomas. In other cases, the imbalance in the production or absorption of CSF causes the hydrocephalus.

Diagnosis of NPH is often difficult due to the symptoms being similar to other disorders. In many cases the NPH is thought to be mild dementia, Alzheimer's, Parkinson's or simply old age factors. Many cases go completely unrecognized and are never treated.

Usually, NPH causes the ventricles to enlarge due to increased CSF within the skull. If a person exhibits symptoms of hydrocephalus a physician may perform several tests to determine if shunting is an option. The most common diagnostic tools are neuro-imaging devices such as CT or MRI and a careful clinical assessment. Once the diagnosis of NPH is suspected there is no single perfect test to determine if a patient will respond to the shunt.

Characterized by three primary symptoms, NPH patients usually exhibit gait disturbance (difficulty walking), dementia, and urinary incontinence. However, not all symptoms are always apparent.

Because these three symptoms are often associated with the aging process in general, and a majority of the NPH population is older than 60 years, people often assume that they must live with the problems or adapt to the changes occurring within their bodies. Symptoms can be present for months or even years before a person sees a physician. The symptoms of NPH seem to progress with time. The rate of progress is variable, and it is often a critical loss of function, or disability, that brings patients to their doctors. It seems that the longer the symptoms have been present, the less likely it is that treatment will be successful. As a general rule, the earlier the diagnosis, the better the chance for successful treatment, but some people experiencing symptoms for years can improve with treatment.

Gait disturbances range in severity, from mild imbalance to the inability to stand or walk at all. For many patients, the gait is wide-based, short, slow and shuffling. People may have trouble picking up their feet, making stairs and curbs difficult and frequently resulting in falls. Gait disturbance is often the most pronounced symptom and the first to become apparent.

Mild dementia can be described as a loss of interest in daily activities, forgetfulness, difficulty dealing with routine tasks and short-term memory loss. People do not usually lose language skills, but they may deny that there are any problems. Not everyone will have an obvious mental impairment.

Impairment in bladder control is usually characterized by urinary frequency and urgency in mild cases, whereas a complete loss of bladder control (urinary incontinence) can occur in more severe cases. Urinary frequency is the need to urinate more often than usual, sometimes as often as every one to two

hours. Urinary urgency is a strong, immediate sensation of the need to urinate. This urge is sometimes so strong that it cannot be held back, resulting in incontinence. In very rare cases, fecal incontinence may occur. Some patients never display signs of bladder problems.

### **Diagnostic procedures**

Diagnostic procedures for normal pressure hydrocephalus *may* include one or more of these tests: ultrasound, computerized tomography (CT), magnetic resonance imaging (MRI), lumbar puncture or tap, continuous lumbar CSF drainage, intracranial pressure (ICP) monitoring, measurement of cerebrospinal fluid outflow resistance or isotopic cisternography, and neuropsychological testing.

### **Progressive Supranuclear Palsy**

[http://www.ninds.nih.gov/health\\_and\\_medical/disorders/psp.htm](http://www.ninds.nih.gov/health_and_medical/disorders/psp.htm)

### **What is Progressive Supranuclear Palsy?**

Progressive supranuclear palsy (PSP) is a rare brain disorder that causes serious and permanent problems with control of gait and balance. The most obvious sign of the disease is an inability to aim the eyes properly, which occurs because of lesions in the area of the brain that coordinates eye movements. Some patients describe this effect as a blurring. PSP patients often show alterations of mood and behavior, including depression and apathy as well as progressive mild dementia. It must be emphasized that the pattern of signs and symptoms can be quite different from person to person. The symptoms of PSP are caused by a gradual deterioration of brain cells in a few tiny but important places at the base of the brain, in the region called the brainstem. PSP is often misdiagnosed because some of its symptoms are very much like those of Parkinson's disease, Alzheimer's disease, and more rare neurodegenerative disorders, such as Creutzfeldt-Jakob disease. The key to establishing the diagnosis of PSP is the identification of early gait instability and difficulty moving the eyes, the hallmark of the disease, as well as ruling out other similar disorders, some of which are treatable. Although PSP gets progressively worse, no one dies from PSP itself.

### **Is there any treatment?**

There is currently no effective treatment for PSP, although scientists are searching for better ways to manage the disease. In some patients the slowness, stiffness, and balance problems of PSP may respond to antiparkinsonian agents such as levodopa, or levodopa combined with anticholinergic agents, but the effect is usually temporary. The speech, vision, and swallowing difficulties usually do not respond to any drug treatment.. Another group of drugs that has been of some modest success in PSP are antidepressant medications. The most commonly used of these drugs are Prozac, Elavil, and Tofranil. The anti-PSP benefit of these drugs seems not to be related to their ability to relieve depression. Non-drug treatment for PSP can take many forms. Patients frequently use weighted walking aids because of their tendency to fall backward. Bifocals or special glasses called prisms are sometimes prescribed for PSP patients to remedy the difficulty of looking down. Formal physical therapy is of no proven benefit in PSP, but certain exercises can be done to keep the joints limber. A surgical procedure, a gastrostomy, may be necessary when there are swallowing disturbances. This

surgery involves the placement of a tube through the skin of the abdomen into the stomach (intestine) for feeding purposes.

### **What is the prognosis?**

PSP gets progressively worse but is not itself directly life-threatening. It does, however, predispose patients to serious complications such as pneumonia secondary to difficulty in swallowing (dysphagia). The most common complications are choking and pneumonia, head injury, and fractures caused by falls. The most common cause of death is pneumonia. With good attention to medical and nutritional needs, however, most PSP patients live well into their 70s and beyond.

### **What research is being done?**

Therapeutic trials with free radical scavengers (agents that can get rid of potentially harmful free radicals) are being planned for the future. Research is ongoing on Parkinson's and Alzheimer's diseases. Better understanding of those common, related disorders will go a long way toward solving the problem of PSP, just as studying PSP may help shed light on Parkinson's and Alzheimer's diseases.

### **Other websites of interest:**

society of PSP (USA)

<http://www.psp.org/>

BBC news on SPS plus links to relevant stories

[http://news.bbc.co.uk/2/hi/health/medical\\_notes/461557.stm](http://news.bbc.co.uk/2/hi/health/medical_notes/461557.stm)

Website of PSP association, Europe

<http://www.pspeur.org/>

### **Multiple System Atrophy**

[http://www.ninds.nih.gov/health\\_and\\_medical/disorders/shydrgr doc.htm](http://www.ninds.nih.gov/health_and_medical/disorders/shydrgr doc.htm)

### **Synonym(s):**

Multiple System Atrophy with Postural Hypotension

Reviewed 07-01-2001

### **What is Shy-Drager Syndrome?**

Multiple system atrophy (MSA) with postural hypotension, also called Shy-Drager syndrome, is a progressive disorder of the central and sympathetic nervous systems. The disorder is characterized by postural (or orthostatic) hypotension—an excessive drop in blood pressure when the patient stands up, which causes dizziness or momentary blackouts. MSA has been classified clinically into three types, olivopontocerebellar atrophy (OPCA), which primarily affects balance, coordination, and speech; a parkinsonian form (striatonigral degeneration), which can resemble Parkinson's disease because of slow movement and stiff muscles; and a mixed cerebellar and parkinsonian form. In all three forms of MSA, the patient can have orthostatic hypotension. Orthostatic hypotension and symptoms of autonomic failure such as constipation, impotence in men, and urinary incontinence usually predominate early in the course of the disease. Constipation may be unrelenting and hard to manage. Shy-Drager syndrome may be difficult to diagnose in the early stages. For the majority of patients, blood pressure is low when the patients stand up and high when the patients lie down. Other symptoms

that may develop include impaired speech, difficulties with breathing and swallowing, and inability to sweat.

### **Is there any treatment?**

Orthostatic hypotension in Shy-Drager syndrome is treatable, but there is not known effective treatment for the progression central nervous system degeneration. The general treatment course is aimed at controlling symptoms. Antiparkinsonian medication, such as L-dopa, may be helpful. To relieve low blood pressure while standing, dietary increases of salt and fluid may be beneficial. Medications to elevate blood pressure, such as salt-retaining steroids, are often necessary, but they can cause side effects and should be carefully monitored by a physician. Alpha-adrenergic medications, non-steroidal anti-inflammatory drugs, and sympathomimetic amines are sometimes used. Sleeping in a head-up position at night reduces morning orthostatic hypotension. An artificial feeding tube or breathing tube may be surgically inserted for management of swallowing and breathing difficulties.

### **What is the prognosis?**

Shy-Drager syndrome usually ends in the patient's death by 7 to 10 years after diagnosis. Breathing problems such as aspiration, stridor (high-pitched breathing sounds due to airway obstruction), or cardiopulmonary arrest are common causes of death.

### **What research is being done?**

The NINDS carries out and funds research about disorders of the autonomic nervous system, including Shy-Drager syndrome. This research is aimed at discovering ways to diagnose and treat disorders of the autonomic nervous system and ultimately to cure or prevent them.

### **Other websites of interest:**

<http://www.ndrf.org/MSA.htm>

<http://neuro-www.mgh.harvard.edu/forum/ShyDragerMenu.html>

## **2. Dementia in Other Diseases Classified Elsewhere**

### **Pick's Disease**

<http://www.nnpdf.org/>

Niemann-Pick Disease ("Niemann-Pick") is actually a term for a group of diseases which affect metabolism and which are caused by specific genetic mutations. The three most commonly recognized forms of the disease are Types A, B and C.

### **Type A**

Niemann-Pick begins in the first few months of life. Symptoms may include:

- feeding difficulties
- a large abdomen within 3 to 6 months
- progressive loss of early motor skills
- cherry red spot in the eye
- (generally) a very rapid decline leading to death by two to three years of age.

### **Type B**

is biochemically similar to Type A but the symptoms are more variable. Abdominal enlargement may be detected in early childhood but there is almost no neurological involvement, such as loss of motor skills. Some patients may develop repeated respiratory infections.

### **Type C**

Niemann-Pick usually affects children of school age, but the disease may strike at any time from early infancy to adulthood. Symptoms may include:

jaundice at (or shortly after) birth

an enlarged spleen and/or liver

difficulty with upward and downward eye movements (Vertical Supranuclear Gaze Palsy).

VSPG is highly suggestive of Type C.

unsteadiness of gait, clumsiness, problems in walking ("ataxia")

difficulty in posturing of limbs ("dystonia")

slurred, irregular speech ("dysarthria")

learning difficulties and progressive intellectual decline ("dementia")

sudden loss of muscle tone which may lead to falls ("cataplexy")

tremors accompanying movement and, in some cases, seizures.

A child showing signs before one year of age may not live to school age. Children showing symptoms after entering school may live into their mid to late teens, with few surviving into their twenties.

### **For Types A and B**

Niemann-Pick the ASM gene has been isolated and extensively studied - it resides on chromosome 11 in man. Many of the molecular abnormalities in this gene which cause Types A and B NPD have been identified and DNA testing and prenatal diagnosis has begun.

Research into therapies for Types A and B NPD has progressed rapidly since the early 1990's. Mount Sinai School of Medicine is conducting research on bone marrow transplantation, enzyme replacement therapy, and gene therapy. All of these therapies have had some success against **Type B** Niemann-Pick in a laboratory environment. Unfortunately, none of the potential therapies has been effective against Type A.

*Bone marrow transplantation* has proven effective in mouse models for many aspects of Type B when the transplant occurs early in life. It is unclear if transplantation later in life is also effective.

*Enzyme replacement* could not be evaluated until the discovery of the ASM gene allowed researchers to produce large quantities of the enzyme for evaluation. Test results on mice indicate that enzyme replacement has the potential to be an effective treatment for Type B NPD.

**Type C** Niemann-Pick, no specific treatment is available. A healthy low-cholesterol diet is recommended. However research into low-cholesterol diets and cholesterol-lowering drugs do not indicate that these halt the progress of the disease or change cholesterol metabolism at the cellular level.

**Other websites of interest:**

National Pick's Disease Foundation, UK:

<http://www.nnpdf.org/npdg-uk/index.html>

Website about "Jacob", a child who has Niemann-Pick Disease:

<http://www.jacob-quinn.com/>

<http://www.parseghian.org/apmrfweb/>

<http://health.yahoo.com/health/dc/001207/0.html>

**Creutzfeldt-jacob Disease**

[http://www.mdtravelhealth.com/infectious/mad\\_cow\\_disease.html](http://www.mdtravelhealth.com/infectious/mad_cow_disease.html)

**New variant Creutzfeldt-Jacob disease and bovine spongiform encephalopathy****("mad cow disease")**

New variant Creutzfeldt-Jacob disease (CJD) is a degenerative neurologic disease acquired by eating beef from cows with a related illness known as bovine spongiform encephalopathy ("mad cow disease"). The disorder was initially described in the United Kingdom. As of December 2001, there were more than 100 definite or probable cases of new variant CJD reported from the United Kingdom, five from France, and one from the Republic of Ireland. "Mad cow disease" has also been identified in Austria, Belgium, Czech Republic, Denmark, Finland, Germany, Greece, Italy, Liechtenstein, Luxembourg, the Netherlands, Portugal, Slovak Republic, Slovenia, Spain, Switzerland, and Japan, but human cases have not been reported from these countries to date.

New variant CJD appears to be caused by abnormal proteins called prions, which do not resemble bacteria, viruses, or other conventional causes of human and animal disease. Transmission appears to have been amplified through the use of ruminant body tissues as cattle feed. This practice was subsequently banned throughout the European Union.

The incubation period of new variant CJD appears to be several years or longer. Initial symptoms may be psychiatric, similar to depression or (less often) psychosis. Patients may complain of unusual sensory symptoms, such as "stickiness" of the skin. Later symptoms include unsteady gait, involuntary movements, dementia, and mutism, progressing inevitably to death within 1-2 years. There is no treatment.

New variant CJD can only be acquired by eating beef or beef products; i.e if you do not consume beef or beef products while visiting a country which has reported "mad cow disease", you cannot become infected. There is no evidence of any risk from pork, lamb, milk or milk products. Transmission by blood transfusion or blood products has not been reported. The Centers for Disease Control does not advise against eating European beef, but suggests that travelers who wish to reduce their risk may either abstain from beef completely while in Europe or eat only solid pieces of muscle meat, such as steak, rather than products like sausage or chopped meat that might be contaminated. Cooking, drying, or freezing does not inactivate the agent that causes new variant CJD or prevent its transmission.

**Other websites of interest:**

BBC web site address with all related news story links

<http://www.bbc.co.uk/cgi-bin/search/results.pl?q=creutzfeldt-jacob+disease&uri=%2Fhome%2Ftoday%2Findex.live.shtml&x=13&y=14>

Web site with stats of CJD in the UK, annual reports...

<http://www.cjd.ed.ac.uk/>  
<http://cjdfoundation.org/>

Web site that concentrates on bovine form, but also has large amounts of general news articles and also a list of leading researchers, scientists etc of CJD

<http://www.mad-cow.org/>  
[http://www.mdtravelhealth.com/infectious/mad\\_cow\\_disease.html](http://www.mdtravelhealth.com/infectious/mad_cow_disease.html)  
[http://www.medical-library.org/journals2a/creutzfeldt\\_jacob2.htm](http://www.medical-library.org/journals2a/creutzfeldt_jacob2.htm)

Address with links to web sites covering care of CJD patients

<http://www.austinareaaorn.org/creutzfeldt.htm>  
<http://www.avera.org/adam/ency/article/000788.htm>

"Slide show" web site...brain tissues, MRI scans etc

<http://www.biomedicine.dote.hu/cselenyi/phd3/sld015.htm>

Web site concerning policies, legislation on bovine form/protection of humans...

<http://olpa.od.nih.gov/legislation/107/session1/planimaldisease.asp>

Pseudo dictionary of abbreviations used in general CJD terminology

<http://www.defra.gov.uk/animalh/bse/glossary.html>  
<http://omni.ac.uk/browse/mesh/detail/C0022336L0022336.html>  
[http://www.thehealthyforum.com/bodymapssec/brain\\_cjd.html](http://www.thehealthyforum.com/bodymapssec/brain_cjd.html)  
<http://clinlabs.path.queensu.ca/ic/cjd.htm>

**Gerstmann-Sträussler-Scheinker Syndrome**

No websites found on this disease, however, not to be confused with Gerstmann Syndrome (check).

**Dementia in Huntington's Disease**

<http://www.hdny.org/>

**What is Huntington's Disease?**

Huntington's disease (HD) is a hereditary brain disorder that affects approximately 30,000 people in the United States. The symptoms of HD typically progress slowly and may include a movement disorder, cognitive impairment, and psychiatric symptoms.

**What are the symptoms of HD?**

Three domains are commonly affected: cognitive (thinking), motor and behavior. The symptoms and progression of the disease vary from person to person; even members of the same family may have different symptoms. Some individuals can have mild involuntary movements (chorea) and have more of the emotional/behavioral symptoms or vice versa. The symptoms of HD usually appear during the late 30's to mid-40's, but sometimes children or the elderly can have symptoms.

**Behavioural problems:**

- irritability
- depression
- anxiety
- aggressive outbursts
- mood swings
- social withdrawal

**Motor problems:**

- fidgety
- incoordination
- involuntary movements
- (chorea, dystonia)

**difficulties with:**

- speech
- swallowing
- balance, walking

**Cognitive dysfunction:**

- Short term memory problems
- Problems organizing, coping, concentrating

**How is HD inherited?**

HD is a genetic disorder that is passed down from one generation to the next. Each child of a parent with HD has a 50% chance of inheriting the gene that causes HD. People who inherit the gene will eventually develop Huntington's disease. Those individuals who do not inherit the gene will not develop the disease, nor will their children.

**Other websites of interest:**

<http://www.yourgenesyourhealth.org/ygyh/mason/ygyh.html?syndrome=hd>

Huntington's Disease Society of America:

<http://www.hdsa.org/>

<http://www.hda.org.uk/>

<http://www.interlog.com/~rlaycock/2nd.html>

Website on caring for people with Huntington's Disease:

<http://www.kumc.edu/hospital/huntingtons/>

<http://www.hdlighthouse.org/>

Website with a list of useful contact numbers, mainly ones in the USA:

<http://www.lib.uchicago.edu/~rd13/hd/electron.html>

Canada's Huntington's Society's website:

<http://www.hsc-ca.org/>

The Scottish Huntington's website:

<http://www.hdscotland.org/>

German Huntington's website:

[http://www.gwdg.de/~usancke/DHH/engl\\_ind.html](http://www.gwdg.de/~usancke/DHH/engl_ind.html)

General Caregiver Website:

<http://www.caregiver.org/>



## **Dementia in Human Immunodeficiency Virus Disease**

<http://www.emedicine.com/NEURO/topic447.htm>

### **Synonyms and related keywords:**

AIDS dementia complex, ADC, HIV-1-associated cognitive/motor complex, AIDS encephalopathy, HIV encephalopathy, subacute HIV encephalitis, HIV-associated dementia complex, AIDS-related dementia, HIV dementia, acquired immunodeficiency syndrome, AIDS

### **History:**

ADC affects cognitive, behavioral, and motor function.

Patients often present with the insidious onset of reduced work productivity, poor concentration, mental slowness, decreased libido, and forgetfulness.

Apathy and withdrawal from hobbies or social activities are common and must be differentiated from depression.

Rare features include sleep disturbances, psychosis (with mania), and seizures.

Motor problems include imbalance, clumsiness, and weakness.

Early symptoms are subtle and may be overlooked.

Later, these symptoms evolve into a global dementia with memory loss and language impairment. This can lead to a vegetative state.

### **Physical:**

The neuropsychological examination reveals features more suggestive of subcortical dementia, as seen in Parkinson disease, than of cortical dementia of the Alzheimer type.

Early on, neuropsychological examination reveals psychomotor slowing, memory loss, and word-finding difficulties. Subcortical involvement with impaired retrieval and manipulation of acquired knowledge is prominent.

Later, severe psychomotor retardation and language impairment become obvious, leading to akinetic mutism.

### **Neurological examination**

Early on, the neurological examination may be normal or reveal subtle impairment of rapid limb and eye movements.

Later, frontal lobe release signs, tremor, hyperreflexia, clonus, spasticity, weakness, and poor coordination develop. These signs may reflect a concomitant vacuolar myelopathy or neuropathy.

The terminal stage of ADC, after progression over several months, includes severe psychomotor retardation and dementia, apraxia, paraparesis, and akinetic mutism. Death ensues within a few months of reaching this stage.

Seizures are rare and warrant exclusion of other conditions.

Price and Brew in 1988 outlined a clinical staging of ADC, summarized by the following:

Stage 0 (normal) - Normal mental and motor function

Stage 0.5 (equivocal/subclinical): Symptoms may be absent, minimal, or equivocal, with no impairment of work or performance of activities of daily living (ADL). Mild signs (snout response, slowed ocular or extremity movements) may be present. Gait and strength are normal.

Stage 1 (mild): Patient able to perform all but the more demanding aspects of work or ADL but has unequivocal evidence of functional, intellectual, or motor impairment. Signs or symptoms may include diminished performance on neuropsychological testing. Patient can walk without assistance.

Stage 2 (moderate): Patient able to perform basic activities of self-care but cannot work or maintain the more demanding aspects of daily life. Patient is ambulatory but may require a single prop.

Stage 3 (severe): Patient has major intellectual incapacity (cannot follow news or personal events, cannot sustain complex conversation, considerable slowing of all outputs). Motor disability precludes walking unassisted (ie, without walker or personal support); walking usually slowed and accompanied by clumsiness of arms.

Stage 4 (end stage): Patient is in a nearly vegetative state. Intellectual and social comprehension and output are at a rudimentary level. Patient is nearly or absolutely mute. Patient is paraparetic or paraplegic with urinary and fecal incontinence.

Although the presence or absence of focal neurological signs is a useful diagnostic criterion, it may be misleading, since these patients often have more than one neurological condition.

#### **Medical Care:**

No specific treatment is available for cognitive decline in AIDS.

Metabolic causes of cognitive decline, such as vitamin deficiencies, thyroid dysfunction, and liver and renal dysfunction, should be corrected in consultation with internal medicine specialists.

Antiretroviral agents, in particular HAART, protect against ADC, can induce remission of ADC, and have reduced the prevalence of ADC. Optimal doses in the presence of ADC remain unclear, however. If ADC develops during treatment with antiretroviral agents, additional or alternative agents should be tried.

Sometimes depression and behavioral disturbances such as hallucinations or delusions require pharmacotherapy. However, caution is required when patients with ADC are treated with psychoactive drugs, such as antidepressants, neuroleptics, and anxiolytics, because of enhanced susceptibility to sedative properties and possible paradoxical reactions. Such symptoms should be treated cooperatively by specialists in internal medicine, neurology, and psychiatry.

#### **Diet:**

In general, patients with AIDS should be encouraged to maintain a balanced diet. Often this requires input from a nutritionist.

#### **Activity:**

Patients should be encouraged to remain as active as their underlying disease permits.

**Other websites of interest:**

<http://www.aegis.com/topics/oi/oi-adc.html>  
<http://www.sfaf.org/treatment/beta/b31/b31adc.html>  
<http://www.neuro.wustl.edu/narc/adx.html>  
<http://www.med.harvard.edu/AANLIB/cases/case14/mr1-tc1/010.html>

This one has info on lots of the other diseases as well:

<http://omni.ac.uk/browse/mesh/detail/C0001849L0001849.html>  
<http://www.projinf.org/fs/dementia.html>  
[http://www.thebody.com/treat/neuro\\_dementia.html](http://www.thebody.com/treat/neuro_dementia.html)

This address is a « slide show » of brain scans etc of patients with ADC:

[http://brighamrad.harvard.edu/education/online/BrainSPECT/Main\\_Slide\\_Show/Main\\_SS\\_19.html](http://brighamrad.harvard.edu/education/online/BrainSPECT/Main_Slide_Show/Main_SS_19.html)  
<http://www.aidsinfonet.org/504-dementia.html>  
[http://pni.med.jhu.edu/Past\\_Research/aids\\_dementia.htm](http://pni.med.jhu.edu/Past_Research/aids_dementia.htm)  
[http://www.niv.ac.za/virusa/aidsa/v7\\_3.htm](http://www.niv.ac.za/virusa/aidsa/v7_3.htm)  
<http://www.retroconference.org/2002/Abstract/13381.htm>  
<http://www.fpnotebook.com/HIV47.htm>  
<http://www.hivcybermall.org/doctorsedu/AIDS%20Dementia%20Complex.htm>

This site has articles on the results of drug trials of a whole array of drugs:

<http://www.hivpositive.com/f-Oi/OppInfections/4-Neuro/4-Neu-Demen.html>

**Dementia in Parkinson's Disease**

<http://www.pdf.org/aboutdisease/overview/index.html>

**Parkinson's disease: An Overview**

Parkinson's disease (PD) is a disorder of the central nervous system that affects between one and one-and-a-half million Americans. Because it is not contagious and does not have to be reported by physicians, the incidence of the disease is often underestimated.

PD may appear at any age, but it is uncommon in people younger than 30, and the risk of developing it increases with age. It occurs in all parts of the world, and men are affected slightly more often than women.

**Primary Symptoms**

Following is a list of the primary symptoms of Parkinson's disease. It is important to note that not all patients experience the full range of symptoms; in fact, most do not.

**Rigidity** is an increased tone or stiffness in the muscles. Unless it is temporarily eased by anti-Parkinson's medications, rigidity is always present. However, it increases during movement. It is often responsible for a mask-like expression of the face. In some patients, rigidity leads to sensations of pain, especially in the arms and shoulders.

**Tremor** is the symptom the public most often identifies with PD, but in fact, up to 25% of patients experience very slight tremor or none at all. When it is present, the tremor may be worse on one side of the body. Besides affecting the limbs, it sometimes involves the head, neck, face, and jaw.

**Bradykinesia** means slowness of movement. This symptom is characterized by a delay in initiating movements, caused by the brain's slowness in transmitting the necessary instructions to the appropriate parts of the body. When the instructions have been received, the body responds slowly in carrying them out.

### **The role of the patient**

Treating Parkinson's disease is not exclusively the doctor's job; there is much a patient can do to stay as well as possible for as long as possible

### **Exercise:**

For people with Parkinson's, regular exercise and/or physical therapy are essential for maintaining and improving mobility, flexibility, balance and a range of motion, and for warding off many of the secondary symptoms mentioned above. Exercise is as important as medication for the management of PD.

### **Support groups:**

These groups play an important role in the emotional well-being of patients and families. They provide a caring environment for asking questions about Parkinson's, for laughing and crying and sharing stories and getting advice from other sufferers, and for forging friendships with people who understand each other's problems.

### **Staying active:**

PD seems to advance more slowly in people who remain involved in their pre-Parkinson's activities, or who find new activities to amuse them and engage their interest. In a word, getting joy out of life has proved to be good for the health

### **Commonly prescribed medications**

Levodopa, symmetrel, anticholinergics, selegine, dopamine agonists, COMT inhibitors.

### **Side effects from medications**

Like the symptoms of PD itself, the side effects caused by Parkinson's medications vary from patient to patient. They may include dry mouth, nausea, dizziness, confusion, hallucinations, drowsiness, insomnia, and other unwelcome symptoms. Some patients experience no side effects from a drug, while others have to discontinue its use because of them.

### **Surgical interventions**

#### **Pallidotomy:**

This procedure has a long history in the treatment of Parkinson's disease, but it fell out of favor with the advent of levodopa. In recent years it has gained new popularity, mainly because magnetic imaging now allows it to be performed with far greater precision. Pallidotomy is indicated for patients who have developed dyskinetic movements in reaction to their medications. It targets the source of these unwanted movements, the *globus pallidus*, and uses an electrode to destroy the trouble-causing cells. As with any surgical procedure, there are risks involved. The most serious is the possibility of stroke; other risks include partial loss of vision, speech and swallowing difficulties, and confusion.

#### **Brain tissue transplants:**

Although they have produced encouraging results, transplantation surgeries are still in the experimental stage. The experiments began with fetal tissue, but now scientists are also working with genetically engineered cells and a variety of animal cells that can be made to produce dopamine.

**Deep brain stimulation:**

Like pallidotomy, this technique also seeks to stop uncontrollable movements. It is based on the technology of cardiac pacemakers. Electrodes are implanted in the thalamus or globus pallidus and connected to a pacemaker-like device, which the patient can switch on or off as symptoms dictate.

**Other websites of interest:**

Caregiver info:

<http://www.parkinsonscare.com/>

Website for the American Parkinson's Society:

<http://www.apdaparkinson.org/>

Address for World Parkinson's Association:

<http://www.wpda.org/>

<http://www.va.gov/padrecc/>

<http://www.whatsnewinpd.com/>

<http://www.parkinsonsinfo.com/>

Paper on Parkinson's patients living at home:

<http://www.cnsonline.org/www/archive/parkins/park-02.txt>

Website on young sufferers of Parkinson's:

<http://www.young-parkinsons.org.uk/>

<http://www.endotoxin.qmxhome.de/>

<http://www.parkinson.org/>

<http://www.parkinsonsnet.com/>

Parkinson's Institute of Ireland:

<http://www.officeobjects.com/PARKINSONS/>

Society for Neuroscience website...research papers, updates, news etc:

<http://apu.sfn.org/briefings/parkinsons.html>

More info on clinical trials:

<http://www.arkansasclinicaltrials.com/>

**Cerebral Lipidoisis****Synonyms**

- Infantile Gaucher Disease
- Acute Cerebral Gaucher Disease
- Acid Beta-glucosidase Deficiency
- Cerebroside Lipidosis
- Cerebrosidosis
- Familial Splenic Anemia
- Gaucher-Schlagenhauer
- Glucocerebrosidase deficiency
- Glucocerebrosidosis
- Glucosyl Ceramide Lipidosis
- Histiocytosis, lipid, kersin type
- Norrbottnian Gaucher Disease

<http://www.gaucher.org.uk/living.htm>

### **What is the basis of Gaucher disease?**

The human body contains specialized cells called macrophages that remove worn-out cells by degrading them to simple molecules for recycling. This process is analogous to eating and digesting food. The macrophages "eat" worn-out cells and degrade them inside cell compartments called lysosomes that serve as the "digestive tracts" of cells. The enzyme glucocerebrosidase is located within the lysosomes and is responsible for breaking down glucocerebroside into glucose and a fat called ceramide.

People with Gaucher disease lack the normal form of the glucocerebrosidase enzyme and are unable to break down glucocerebroside. Instead, the glucocerebroside remains stored within the lysosomes, preventing the macrophages from functioning normally. Enlarged macrophages containing undigested glucocerebroside are called Gaucher cells. These cells are the hallmark of this disease. Gaucher disease is the most common of 10 so-called "storage" disorders. The most widely known of these disorders is Tay-Sachs disease.

Generalized fatigue

Lack of energy and stamina

Abdomen

Enlarged spleen

Enlarged liver

Pain

Compression of the lungs

Skeletal system

Growth retardation in children

Pain and degeneration of joints and bone-covering tissue

Loss of bone density leading to

widening of bones along the knee joint

curvature of the bones

spontaneous fractures

Acute bone infarctions - "bone crises"

Bone necrosis (death of tissue)

Lungs

Decreased ability to provide oxygen to the blood

Kidneys

Disruption of normal function

Skin

Yellow-brown pigmentation

Non-raised, round, purplish-red spots, especially around the eyes

Blood

Increased bleeding tendency such as nosebleeds and bruising

Subnormal levels of

blood platelets

red blood cells

white blood cells

Elevated levels of

acid phosphatase

plasma proteins

Digestive

Loss of appetite

Intestinal complaints

Most people with Gaucher disease do not develop **all** of the possible symptoms.

In addition, the severity of the disease varies **enormously**.

## **Is there treatment for Gaucher disease?**

### **What is enzyme replacement therapy?**

Because people with Gaucher disease are deficient in glucocerebrosidase enzymatic activity, the most direct and logical therapeutic approach to this inherited disease is to supplement or to replace the missing enzyme. Dr. Roscoe Brady pioneered the development of this therapy at the National Institute of Neurological Disorders and Stroke. Initial research on the natural glucocerebrosidase enzyme showed that it was not particularly effective when administered by infusion to people with Gaucher disease. The majority of the enzyme did not reach the "Gaucher cells" in the body. Dr. Brady developed a form of the glucocerebrosidase enzyme that was modified to increase targeting and uptake in the macrophages, the cells where the enzyme is needed. Modified glucocerebrosidase enzyme (Ceredase) was evaluated in clinical trials which showed that repeated infusions of the enzyme reduced the signs and symptoms of the disease, and reversed the disease progression. This development was a very exciting one and represented the first true therapeutic breakthrough. Ceredase received FDA approval in 1992.

The production of the modified glucocerebrosidase enzyme using a recombinant cell line has been achieved, clinical testing has shown it to be effective and Cerezyme received FDA approval in November 1996. Since then Ceredase has been phased out and replaced by the recombinant product Cerezyme for 95% of patients.

The administration of macrophage-targeted glucocerebrosidase is required at regular intervals throughout an individual's lifetime. As such, the enzyme is an effective therapy, rather than a cure. Currently enzyme replacement therapy is being used to treat 2,000 Gaucher sufferers worldwide.

### **Managing symptoms**

In the past, patient care and therapy for Gaucher disease was directed at managing (ie, relieving) the symptoms resulting from the accumulation of Gaucher cells in the various organs. Careful monitoring of the liver and/or spleen size and blood counts by a patient's physician helps to determine the appropriate therapy.

Depending on symptoms, therapy includes the following measures, either alone or in combination: bed rest, non-aspirin analgesics (aspirin inhibits the blood from clotting and is not advisable for people with Gaucher disease) and anti-inflammatories for acute and chronic pain, biofeedback techniques for pain management, hyperbaric oxygen therapy for the treatment of bone crisis, splenectomy (either complete or partial) for severe anemia, low platelet counts and mechanical obstruction. Sometimes bleeding from a minor wound may require medical intervention to avoid a major blood loss. Oxygen therapy may be necessary for people who have reduced blood supply to the lungs. Low blood count resulting from overactivity of a Gaucher cell-containing spleen is sometimes treated by blood and/or platelet transfusions, or by iron therapy to alleviate the anemia. In the case of a severe and persistent lowering of the blood count, physicians may decide to remove all or part of the spleen. Neither of these approaches is totally satisfactory. Iron therapy increases the risk of hemochromatosis (an iron surplus disease), while spleen removal increases the susceptibility to bacterial diseases and may lead to increased liver and bony symptoms. For these reasons, spleen removal is usually delayed as long as possible and partial spleen removal (which may be more difficult to perform) may be recommended over total removal.

Orthopedic evaluation and intervention may be required for many of the bone complications associated with Gaucher disease. These procedures include orthopedic surgical techniques to relieve pressure from damaged bony areas and/or the insertion of prosthetic devices (such as hip replacements) in joints that have been destroyed by the disease process.

Bone marrow transplantation has been tried as a treatment for severely ill people with Gaucher disease. Because it is such a high-risk procedure, and it requires carefully matched donors, bone marrow transplantation is not performed often. The procedure is generally reserved as a treatment option for terminally ill patients.

### **What is the state of other research on Gaucher disease?**

Currently, research is underway in three main areas: investigation of the bisphosphonate group of drugs (eg Fosamax or alendronate, Didronel or etidronate, pamidronate) for osteoporosis and bone disease; attempts to do gene therapy which if successful could provide a cure; and a trial of OGT 918 a drug which acts as an inhibitor of one of the key enzymes responsible for the formation of glycosphingolipids such as glucocerebroside.

Scientists have already identified many of the particular genetic differences in the glucocerebrosidase gene among people with Gaucher disease. Scientists may be able to correlate the particular defects with the course that Gaucher disease follows in an individual with those genes. Based on this research, physicians may be able to predict disease progression with more improved accuracy in prognosis. Efforts are underway to develop gene therapy. This approach involves introducing normal genes for glucocerebrosidase into cells of the affected person. These cells would then produce sufficient normal amounts of active glucocerebrosidase. Approaches to gene therapy are experimental at present.

### **Other websites of interest:**

<http://yalenewhavenhealth.org/library/healthguide/IllnessConditions/topic.asp?wid=nord12>

<http://www.icomm.ca/geneinfo/def-c.htm>

What seems to be an informative web site on Alzheimer's and other related diseases, but in Spanish

<http://www.homestead.com/montedeoya/clubalzheimer.html>

<http://www.medev.ch/pufa/pufa9704.htm>

<http://www.mazornet.com/genetics/gauchers.asp>

<http://www.ukselfhelp.info/c.htm>

### **Hepatolenticular Degeneration**

<http://www.geocities.com/nutriflip/Diseases/WilsonsDisease.html>

Wilson's Disease (Hepatolenticular Degeneration) is a genetic disorder that is fatal unless detected and treated before serious illness develops from copper poisoning. Wilson's Disease affects one in thirty thousand people world wide. The genetic defect causes excessive copper accumulation. Small amounts of copper are essential as vitamins. Copper is present in most foods, and most people get much more than they need. Healthy people excrete copper they don't need, but Wilson's Disease patients cannot.



The gene for Wilson's disease (ATP7B) was mapped to chromosome 13. The sequence of the gene was found to be similar to sections of the gene defective in Menkes disease, another disease caused by defects in copper transport.

The liver of a person who has Wilson's disease does not release copper into bile as it should. Bile is a liquid produced by the liver that helps with digestion. As the intestines absorb copper from food, the copper builds up in the liver and injures liver tissue. Eventually, the damage causes the liver to release the copper directly into the bloodstream, which carries the copper throughout the body. The copper buildup leads to damage in the kidneys, brain, and eyes. If not treated, Wilson's disease can cause severe brain damage, liver failure, and death.

Symptoms usually appear between the ages of 6 and 20 years, but can begin as late as age 40. The most characteristic sign is the Kayser-Fleischer ring--a rusty brown ring around the cornea of the eye that can be seen only through an eye exam. Other signs depend on whether the damage occurs in the liver, blood, central nervous system, urinary system, or musculoskeletal system. Many signs would be detected only by a doctor, like swelling of the liver and spleen; fluid buildup in the lining of the abdomen; anemia; low platelet and white blood cell count in the blood; high levels of amino acids, protein, uric acid, and carbohydrates in urine; and softening of the bones. Some symptoms are more obvious, like jaundice, which appears as yellowing of the eyes and skin; vomiting blood; speech and language problems; tremors in the arms and hands; and rigid muscles.

The disease is treated with lifelong use of D-penicillamine or trientine hydrochloride, drugs that help remove copper from tissue. Patients will also need to take vitamin B6 and follow a low-copper diet, which means avoiding mushrooms, nuts, chocolate, dried fruit, liver, and shellfish. Taking extra zinc may be helpful in blocking the intestines' absorption of copper.

Zinc and Vitamin C supplementation increases the excretion of copper. With the use of oral binders of copper eg penicillamine, Vitamin B6, and multi mineral must be taken to reduce side effects of this drug. Iron and zinc are also bound by this binder.

The newest FDA-approved drug is zinc acetate (Galzin). Zinc acts by blocking the absorption of copper in the intestinal tract. This action both depletes accumulated copper and prevents it reaccumulation. Zinc's effectiveness has been shown by 15 years of considerable experience overseas. A major advantage of zinc therapy is its lack of side effects.

The nutrients mentioned above reflect the major nutritional supplements that may help the condition. Please do remember however that nutritional supplementation is an adjunct to medical treatment and in no way replaces medical treatment.

**Other interesting websites:**

<http://www.hon.ch/HONselect/RareDiseases/C06.552.413.html>

[http://dmoz.org/Health/Conditions and Diseases/Nutrition and Metabolism Disorders/Vitamins and Minerals/Wilson's Disease/](http://dmoz.org/Health/Conditions_and_Diseases/Nutrition_and_Metabolism_Disorders/Vitamins_and_Minerals/Wilson's_Disease/)

Web site with a list of links for hepatolenticular degeneration

<http://www.bdid.com/wilson.htm>

Web site with slides of affected areas in Wilson's disease

<http://www.brisbio.ac.uk/ROADS/subject-listing/hepatolenticulardegeneration.html>

<http://www.avera.org/adam/ency/article/000785.htm>

## **Hypercalcaemia**

<http://www.nlm.nih.gov/medlineplus/ency/article/000365.htm#contentDescription>

### **Definition**

An excessive amount of calcium in the blood.

### **Causes and risks**

Calcium is an important element in the body. It is part of the mineral component of bone, and it exists as a charged particle called an ion in the blood and inside cells. Calcium is important to several body functions including bone formation, muscle contraction and the release of hormones.

Parathyroid hormone (PTH) and vitamin D regulate calcium balance in the body. PTH is produced by the parathyroid glands; four small glands located in the neck behind the thyroid gland. Vitamin D is obtained from exposure of skin to sunlight and from dietary sources such as fortified dairy products, egg yolks, fish, and fortified cereals.

Too much PTH is an important cause of hypercalcemia.

[Primary hyperparathyroidism](#) is the most common cause of hypercalcemia over all and is the most common cause of excess PTH. Familial hypocalciuric hypercalcemia (FHH) is a condition of benign hypercalcemia caused by high PTH. The drug lithium may increase PTH release and cause hypercalcemia.

Blood calcium may be high despite low levels of PTH. Some malignant tumors (for example, lung cancers, breast cancer) produce PTH-related peptide (PTHrp) that increases blood calcium. Excess vitamin D (hypervitaminosis D) from diet or granulomatous diseases may cause hypercalcemia. [Kidney failure](#), adrenal gland failure, [hyperthyroidism](#), prolonged immobilization, a class of diuretics called thiazides, and ingestion of massive amounts of calcium (milk-alkali syndrome) may also cause hypercalcemia.

The reported prevalence of hypercalcemia ranges from 0.1 to 1% of the population. The widespread ability to measure blood calcium since the 1960s has improved detection of hypercalcemia, and today most patients with hypercalcemia have no symptoms. Women over the age of 50 are most likely to be hypercalcemic, usually due to primary hyperparathyroidism.

**Prevention** Most causes of hypercalcemia cannot be prevented. Women over the age of 50 should see their health care provider regularly and have their blood calcium screened periodically.

Hypercalcemia from calcium and vitamin D supplements can be avoided by contacting your health care provider for advice if you are taking supplements without a prescription.

## **Symptoms**

Skeletal:

[Bone pain](#)

Loss of height

Bowing of the shoulders

Spinal column curvature ([kyphosis](#))

Pathological fractures

Kidney and [kidney stones](#):

[Flank pain](#)

[Frequent urination](#)

Frequent thirst

Abdominal:

Pain

[Nausea and/or vomiting](#)

Poor appetite (anorexia)

[Constipation](#)

Psychological:

Irritability

[Memory loss](#)

Apathy

[Depression](#)

[Dementia](#)

Coma

Muscular:

Weakness

Muscle twitches ([fasciculations](#))

[Muscle atrophy](#)

## **Signs and tests**

High serum total and/or ionized calcium

High serum PTH level (hyperparathyroidism)

High urine calcium (hyperparathyroidism, other causes)

Low urine calcium (FHH)

High vitamin D level (hypervitaminosis D, granulomatous diseases)

High serum PTHrp (certain cancers)

## **Treatment**

Treatment is directed at the underlying cause of hypercalcemia whenever possible. When hypercalcemia is mild and caused by primary hyperparathyroidism, patients may be followed closely by their physician over time. Severe hypercalcemia causing symptoms and requiring hospitalization is treated aggressively with the following:

Intravenous fluids

Bisphosphonates (drugs that stop bone resorption such as pamidronate or etidronate)

Calcitonin

Glucocorticoids (steroids, for hypervitaminosis D)

Hemodialysis (for hypercalcemia that is unresponsive to treatment and life-threatening)

## **Prognosis**

Prognosis depends on the underlying cause of hypercalcemia. Patients with mild hyperparathyroidism or hypercalcemia with a treatable cause (for example, primary hyperparathyroidism, dietary hypervitaminosis D) are unlikely to suffer complications from hypercalcemia. Patients with hypercalcemia in the setting conditions such as cancer or granulomatous disease may have a poor prognosis

due to the underlying disease itself rather than the hypercalcemia. The complications of prolonged hypercalcemia are uncommon today.

**Other interesting websites:**

Address contains links to clinical trials etc

<http://rarediseases.info.nih.gov/>

**Hypoparathyroidism**

Hypothyroidism (also the same as Hypocalcaemia...check)

(Equivalent to Hypoparathyroidism...check)

<http://www.hypoparathyroidism.org/definition.php>

**Important:** It is possible that the main title of the title of the article (Hypoparathyroidism) is not the name you expected. Please check the Synonym listing to find the alternative names and disorder subdivisions covered by this article.

**Synonyms:**

Tetany

Parathyroid

Information on the following can be found in the Related Disorders of this report:

Hypo-calcemia

DiGeorge Syndrome

Osteomalacia

Pseudo-Hypoparathyroidism

**General Discussion:**

Remember: The information contained in the Rare Diseases Database is provided for educational purposes only. It should not be used for diagnostic or treatment purposes. If you wish to obtain more information about this disorder, please contact your personal physician.

Hypoparathyroidism is a disorder that causes lower than normal levels of calcium in the blood due to insufficient levels of parathyroid hormone. This condition can be inherited, associated with other disorders, or it may result from neck surgery.

**Symptoms:**

Hypoparathyroidism is characterized by weakness , muscle cramps, abnormal sensations such as tingling, burning and numbness (paresthesias) of the hands excessive nervousness, loss of memory, headaches and uncontrollable cramping muscle movements of the wrists and feet. Other symptoms may be spasms of the facial muscles (Chvostek Sign), the contraction of muscles produced by mild compression of nerves (Trousseau's Sign), malformations of the teeth, including enamel and roots of the teeth; and malformed finger nails. In some hypoparathyroid conditions, there may also be pernicious anemia, dry and coarse skin, patchy hair loss (alopecia), thin, scant eyebrows, patches of skin that have lost pigment (vitiligo) and mental depression.

**Causes:**

The exact cause of Hypoparathyroidism is unknown. It can occur as a separate disorder, in association with other endocrine gland disorders that affect the thyroid, ovaries or adrenal glands, or it may be due to the removal of or damage to the parathyroid glands. It may also be inherited and transmitted through autosomal recessive genes.

Human traits, including the classic genetic diseases, are the product of the interaction of two genes, one received from the father and one from the mother. In recessive disorders, the condition does not appear unless a person inherits the same defective gene for the same trait for each parent. If a patient receives one normal gene and one gene for the disease, the person will be a carrier for the disease, but usually will show no symptoms. The risk of transmitting the disease to the children of a couple, both of whom are carriers for a recessive disorder, is twenty-five percent. Fifty percent of their children will be carriers, but healthy as described above. Twenty-five percent of their children will receive both normal genes, one from each parent and will be genetically normal.

**Affected Population:**

Hypoparathyroidism affects males and females in equal numbers. It is seen more often in children under 16 and in adults over 40.

**Related Disorders:**

Symptoms of the following disorders can be similar to those of Hypoparathyroidism. Comparisons may be useful for a differential diagnosis.

**DiGeorge Syndrome**

is a complex group of congenital malformations among which is a susceptibility of recurrent infections due to a decreased immune system and the occurrence of seizures during infancy due to low levels of calcium in the blood. The disorder results from faulty development of two of the pharyngeal pouches during early development of the fetus. The parathyroid gland which regulates the concentration of calcium in the blood, and the thymus gland which transforms certain lymphocytes into T-Cells, (responsible for cellular and long term immune reactions), are absent or abnormal in DiGeorge Syndrome.

**Hypo-calcemia**

is characterized by abnormally low levels of calcium and high levels of phosphorous in the blood. It is characterized by spasms of the facial muscles, abdominal and muscle cramps, spasms of the foot and wrist (carpopedal), and strange sensations such as tingling and burning or numbness (paresthesias) of the lips, tongue, fingers and feet. In some severe cases, there may be spasms of the larynx and generalized convulsions.

**Osteomalacia**

is a disease that causes softening of the bones due to insufficient levels of calcium. This results in the bones becoming brittle, and easily broken. It is characterized by pains in the limbs, spine and pelvis, and general weakness. It is seen mostly in adult women.

**Pseudohypoparathyroidism**

is a hereditary disorder characterized by an inadequate response to the parathyroid hormone, although this hormone is present in normal amounts. This inadequate response affects bone growth in patients with this disorder, headaches, weakness, easy fatigue, lack of energy, and blurred vision light may also occur.

**Therapies (Standard):**

Hypoparathyroidism is treated with calcium, ergocalciferol and dihydrotachysterol (forms of vitamin D). Genetic counseling may be of benefit for patients and their families if they have the inherited form of this disorder. Other treatment is symptomatic and supportive.

**Therapies (Investigational):**

At the present time, studies are being conducted on the effectiveness of vitamin-D3 as a treatment of Hypoparathyroidism. More research must be conducted to determine long term safety and effectiveness of this method. This disease entry is based upon medical information available through December 1989. Since NORD's resources are limited it is not possible to keep every entry in the Rare Diseases Data Base completely current and accurate.

**Other interesting websites:**

<http://www.unithroid.com/>

<http://www.hypoparathyroidism.org/definition.php>

### **3. Intoxications**

**Korsakoff's Syndrome**

<http://www.scope2000.co.uk/>

Heavy drinkers often eat poorly, and the metabolism of alcohol depletes the body's stores of 'B' vitamins. This can lead to a severe deficiency of vitamin B1 (thiamine), causing Wernicke-Korsakoff (Korsakov) Syndrome. In the Wernicke's phase of this syndrome, patients become drowsy and unresponsive, and their walking and eye movements become uncoordinated. Wernicke's is a medical emergency, requiring prompt administration of intravenous thiamine. If not treated in time, patients develop Korsakoff's, exhibiting marked impairment of short term memory. Patients with Korsakoff's may not remember an event that occurred ten minutes earlier; to mask their confusion and make sense of their lives, they sometimes fabricate events ('confabulation'). Patients with Korsakoff's rarely recover, and frequently require long term care.

Korsakoff's syndrome - Psychological impairment is partial rather than general; that is, a limited number of specific functions are effected, such as memory, thinking, perception, or mood. Affective syndromes occur, depressive disorders being more common than mania. Schizophrenia-like syndrome can arise in association with brain disease. Personality disorder is another highly important complication.

In some conditions, but not all, focal lesions in the brain are demonstrable.

## **Wernicke's**

<http://www.healthcentral.com/mhc/top/000771.cfm#Alternative%20names:>

### **Alternative names:**

Korsakoff psychosis; alcoholic encephalopathy; encephalopathy, alcoholic; Wernicke's disease

### **Definition:**

A brain disorder involving loss of specific brain functions, due to [thiamine deficiency](#).

### **Causes, incidence, and risk factors:**

Wernicke-Korsakoff syndrome usually affects people between 40 and 80 years old. The onset is gradual.

The syndrome is actually two disorders that may occur independently or together. Wernicke's disease involves damage to multiple nerves in both the [central nervous system](#) (brain and spinal cord) and the [peripheral](#) nervous system (the rest of the body). It may also include symptoms caused by [alcohol withdrawal](#). The cause is generally attributed to [malnutrition](#), especially lack of vitamin B-1 ([thiamine](#)), which commonly accompanies habitual [alcohol use](#) or [alcoholism](#).

### **Prevention:**

Minimal or moderate [alcohol use](#) and adequate nutrition reduce the risk of developing Wernicke-Korsakoff syndrome.

### **Symptoms:**

vision changes

[double vision](#)

[eye movements, uncontrollable](#) or twitching of the eyes

[eyelid drooping](#)

[loss of muscle coordination](#)

unsteady, uncoordinated walking

[weakness](#)

[movement, dysfunctional](#)

[hand tremor](#)

[muscle contractions](#)

[muscle atrophy](#)

[facial paralysis](#)

sensation changes

[decreased sensation](#) in the feet or hands, [numbness](#)

[abnormal sensations, tingling](#)

thin, malnourished appearance

[loss of hair](#)

[dry skin](#)

[swallowing difficulty](#)

[speech impairment](#)

[hoarseness or changing voice](#)

[mood changes](#), emotional changes, and [behavior changes](#)

[loss of memory](#), can be profound

confabulation

decreased intellect/cognitive skills

[decreased problem solving](#)

loss of ability to think abstractly

autonomic disturbances:

orthostatic [dizziness](#)

[constipation](#)

inability to tolerate cold environment

Note: Symptoms that indicate [alcohol withdrawal](#) may also be present or may develop.

### **Treatment:**

The goals of treatment are to control symptoms as much as possible and to prevent progression of the disorder. Hospitalization is required for initial control of symptoms. If the person is [lethargic](#), [unconscious](#), or [comatose](#), monitoring and care appropriate to the condition may be required. The airway should be monitored and protected as appropriate.

[Thiamine](#) (vitamin B-1) may improve symptoms of [confusion](#) or [delirium](#), difficulties with vision and eye movement, and [muscle incoordination](#). B-1 may be given by injection into a vein or a muscle, or by mouth. Thiamine does not generally improve [loss of memory](#) and intellect associated with Korsakoff psychosis.

Total abstinence from alcohol is required to prevent progressive loss of brain function and damage to [peripheral](#) nerves. A well-balanced, nourishing diet is recommended.

### **Expectations (prognosis):**

Without treatment, Wernicke-Korsakoff syndrome progresses steadily to death. With treatment, symptoms such as [incoordination](#) and [vision difficulties](#) may be controlled, and progression of the disorder may be slowed or stopped. Some of the symptoms, particularly the [loss of memory](#) and intellect/cognitive skills, may be permanent. There may be a need for custodial care if the loss of intellect/cognitive skills is severe. Other disorders related to the abuse of alcohol may also be present.

### **Other Websites of interest:**

<http://www.bme.jhu.edu/labs/chb/disorders/wernicke.html>

<http://www.whonamedit.com/synd.cfm/1352.html>

<http://www.bme.jhu.edu/labs/chb/disorders/wernicke.html>

<http://www.whonamedit.com/synd.cfm/1352.html>

<http://www.healthcentral.com/mhc/top/000771.cfm>

Web site with pictures of brain tissue in various conditions (MS, wernick's...)

<http://medlib.med.utah.edu/WebPath/CNSHTML/CNS085.html>

<http://www.medfriendly.com/wernickesdisease.html>

<http://www.austin360.com/shared/health/adam/ency/article/000771.html>

<http://psychiatry.mc.duke.edu/ijpm/26-3/Casanova.html>

<http://www.pennhealth.com/ency/article/000771.htm>

<http://www.zarcrom.com/users/alzheimers/odem/al6.html>

<http://www.nursingceu.com/NCEU/courses/wksyndrome/>

<http://www.rwjhamilton.org/Atoz/encyclopedia/article/000771.asp->

"Barnes and Noble" website/online store has a limited number of books on the above diseases.

<http://www.starkhealth.com/articles/psycho.htm>

[http://www.mrtabletdemon.co.uk/disease\\_index\\_W.htm](http://www.mrtabletdemon.co.uk/disease_index_W.htm)



Web address of Journals of toxicology in Wiley Science:  
<http://www3.interscience.wiley.com/cgi-bin/abstract/78503518/START>

### **Alcohol Related Dementia**

<http://www.netdoctor.co.uk/diseases/facts/dementia.htm>

Brain damage can be caused by drinking too much [alcohol](#). It is important that people with this type of dementia give up drinking alcohol completely to stop the disease progressing.

### **Other websites of interest:**

Web address of articles on Wiley Interscience concerning alcohol related dementia

<http://www3.interscience.wiley.com/cgi-bin/search>  
<http://www.zarcrom.com/users/alzheimers/odem/al-d.html>  
<http://www.alz.co.uk/alzheimers/alcohol.html>  
<http://www.alzheimers.asn.au/DementiaInformation/di03.html>  
<http://www.agingincanada.ca/Seniors%20Alcohol/1e3-1.htm>  
<http://www.living-your-life.com/health/alcohol-dementia.php>

BBC news articles on alcohol related topics: cost to health services, alcohol related dementia, alcoholism in the elderly...

<http://news.bbc.co.uk/1/hi/health/743878.stm>

Web site with methods to improve memory after alcohol related brain damage

[http://www.betterhealth.vic.gov.au/bhcv2/BHCLang.nsf/\(LevelFour\)/962585A35C9A70194A256B640083BA52?OpenDocument](http://www.betterhealth.vic.gov.au/bhcv2/BHCLang.nsf/(LevelFour)/962585A35C9A70194A256B640083BA52?OpenDocument)  
<http://www.psyweb.com/Mdisord/subsd.html>  
<http://www.netdoctor.co.uk/diseases/facts/dementia.htm>

Web site with a selection of "rare diseases", treatments etc...

<http://www.dementia.ie/AboutDementia/WhatIsDementia.html>

## **4. Dementia in Other Specified Diseases**

### **Multiple Sclerosis**

<http://www.nationalmssociety.org/about%20ms.asp>

### **What Is Multiple Sclerosis?**

Multiple sclerosis is a chronic, often disabling, disease of the central nervous system. Symptoms may be mild, such as numbness in the limbs, or severe—paralysis or loss of vision.

Most people with MS are diagnosed between the ages of 20 and 40, but the unpredictable physical and emotional effects can be lifelong. The progress, severity, and specific symptoms of MS in any one person cannot yet be predicted, but advances in research and treatment give hope to those affected by the disease.

The National Multiple Sclerosis Society is dedicated to ending the devastating effects of multiple sclerosis.

Symptoms of MS are **unpredictable and vary** from person to person and from time to time in the same person. For example: One person may experience abnormal fatigue, while another might have severe vision problems. A person with MS could have loss of balance and muscle coordination making walking difficult; another person with MS could have slurred speech, tremors, stiffness, and bladder problems. Even severe symptoms may disappear completely and the person will regain lost functions.

### **Symptoms:**

Bladder dysfunction, bowel dysfunction, cognitive problems, dizziness and vertigo, emotional problems, gait problems, headache, hearing loss, itching, numbness, pain, seizures, sexual dysfunction, spasticity, speech and swallowing disorders, tremours, visual problems.

There is no laboratory test, symptom, or physical finding which, when present or positive, always means a person has MS. The diagnosis of MS must be made by a careful process that demonstrates findings that are consistent with MS, and also rules out other causes.

### **Prognosis**

People with MS can expect one of four courses of disease. MS tends to take one of four clinical courses, each of which might be mild, moderate, or severe.

[Occupational Therapy](#)

[Physical Therapy](#)

[Speech Therapy](#)

[Rehabilitation](#)

[Exercise](#)

[Diet](#)

[Vitamin Therapy](#)

And various medications

### **Other interesting websites:**

<http://www.healthtalk.com/msen/>

Multiple Sclerosis societies of the UK

<http://www.mssociety.org.uk/index.html>

Web site with info. on caring and treating MS

<http://www.mscares.org/>

Web site with articles, personal accounts etc to help understand MS

<http://www.understandingms.com/>

Address of MS society America

<http://www.msaa.com/>

Address of MS society Australia

<http://www.msaustralia.org.au/>

<http://www.noah-health.org/english/illness/neuro/muscler.html>

Web site with links to various MS support groups

<http://www.infosci.org/>

General web site on MS with personal accounts, treatment, famous people with MS, MS news...

<http://www.mult-sclerosis.org/>

Address of "Barnes and Noble" MS section

<http://r1.us.rmi.yahoo.com/rmi/http://search.barnesandnoble.com/booksearch/results.asp?rmivars%3ftarget=top?WRD=multiple+sclerosis&userid=531GLUMVNJ&sourceid=00002123466036150646&bdate=08%2D27%2D2002+10%3A20%3A12>

Address of MS society Ireland

<http://www.ms-society.ie/>

Web site with definitions, helpful descriptions etc of MS related words

<http://www.msonly.com/glossary.html>

MS support web site

<http://pages.cthome.net/Marrone/>

Address with various treatments of MS

<http://www.msadvances.com/>

MS chat room

<http://www.msadvances.com/>

<http://mscare.com/>

MS net guide (resources, trials, links, treatments...)

<http://www.msnetguide.com/>

## **Neurosyphilis**

<http://www.emedicine.com/neuro/topic684.htm>

**Synonyms and related keywords:** neurosyphilis, acute syphilitic meningitis, meningovascular syphilis, tabes dorsalis, general paresis, optic atrophy

Symptoms of neurosyphilis, principally of central origin, include the following, by frequency:

Personality change - 33%

Ataxia - 28%

Stroke - 23%

Ophthalmic symptoms (eg, blurred vision, reduced color perception, impaired acuity, visual dimming, photophobia) - 17%

Urinary symptoms (eg, bladder incontinence) - 17%

Lightning pains - 10%

Headache - 10%

Dizziness - 10%

Hearing loss - 10%

Seizures - 7%

Signs of neurosyphilis, by decreasing frequency, include the following:

Hyporeflexia - 50%

Sensory impairment (eg, decreased proprioception, loss of vibratory sense) - 48%

Pupillary changes (anisocoria, Argyll Robertson pupils) - 43%

Cranial neuropathy - 36%

Dementia, mania, or paranoia - 35%

Romberg sign - 24%

Charcot joint - 13%

Hypotonia - 10%

Optic atrophy 7%

Neurosyphilis is divided into 2 general categories: (1) early involvement of the CNS limited to the meninges and (2) parenchymal involvement. The 6 delineated groups are as follows:

Asymptomatic

Acute syphilitic meningitis

Meningovascular syphilis

Tabes dorsalis (parenchymal)

General paresis (of the insane [GPI])

Optic atrophy

These syndromes overlap, rendering combined forms. Early neurosyphilis affects mesodermal structures (ie, mainly meninges and vessels), whereas late neurosyphilis affects brain and spinal cord parenchyma

Adequate treatment of neurosyphilis is based largely on achieving treponemicidal levels of penicillin (PCN) in the CSF. *T pallidum* is highly susceptible to PCN, which is the drug of choice for all stages of syphilis. Serum levels of PCN should be maintained for many days, since treponemes divide slowly in early syphilis (in experimental settings, they divide in 30-33 h) and PCN acts only on dividing cells. PCN acts by interfering with the synthesis of cell walls and is active only against organisms that, like *T pallidum*, synthesize their cell walls in growth and division. The intensity of therapy should be based on the presence or absence of CNS involvement and HIV infection.

PCN has some ameliorative effect in every stage of neurosyphilis. Meningovascular disease responds most dramatically. Intravenous (IV) PCN requires hospital admission, which involves loss of time from work, high cost, and risks of the hospital environment. For inpatient treatment, accurate diagnosis of neurosyphilis is imperative.

#### **Other interesting websites:**

<http://neuroland.com/id/neurosyph.htm>

Psychiatrist's web site

<http://www.priory.com/psych/neurosyphilis.htm>

<http://www.hivpositive.com/f-Oi/OppInfections/4-Bacterial/4-Syphilis.html>

<http://www.medterms.com/script/main/art.asp?articlekey=4558>

<http://www.emedicine.com/neuro/topic684.htm>

Address with reports/articles on two neurosyphilis patients

<http://ams.cu.edu.tr/January2000Vol9No1/hacer.html>

<http://www.healthcentral.com/mhc/top/000703.cfm>

[http://www.upcmd.com/dot/examples/01080/criteria\\_syphilis.html](http://www.upcmd.com/dot/examples/01080/criteria_syphilis.html)

Address for "Barnes and Noble" section of books on neurosyphilis

[http://r1.us.rmi.yahoo.com/rmi/http://search.barnesandnoble.com/booksearch/results.asp?rmivars%3ftarget= top?sourceid=00339421585264447149&bfdte=08-27-2002+10:52:43&WRD=neurosyphilis](http://r1.us.rmi.yahoo.com/rmi/http://search.barnesandnoble.com/booksearch/results.asp?rmivars%3ftarget=top?sourceid=00339421585264447149&bfdte=08-27-2002+10:52:43&WRD=neurosyphilis)

<http://www.enlmedical.com/article/000703.htm>

<http://www.xrefer.com/entry/127612>

Case report of neurosyphilis

[http://www.acta-clinica.com/Volume\\_39\\_2000/8/8.htm](http://www.acta-clinica.com/Volume_39_2000/8/8.htm)

<http://www.rwjhamilton.org/Atoz/encyclopedia/article/000703.asp>

## **Polyarteritis**

<http://www.healthcentral.com/mhc/top/001438.cfm#Alternative%20names:>

### **Alternative names:**

periarteritis nodosa

### **Definition:**

A serious blood vessel disease in which small- and medium-sized arteries become swollen and damaged.

### **Causes, incidence, and risk factors:**

Polyarteritis nodosa is a vascular disease of unknown cause. It typically strikes adults, not children. The end result of arterial damage is subsequent damage to the tissue the arteries supply. In this disease, secondary symptoms are a result of damage to the organs being affected, often the skin, heart, kidneys and nervous system.

Generalized symptoms include [fever](#), [fatigue](#), [weakness](#), [loss of appetite](#), [weight loss](#). [Muscle aches](#) ([myalgia](#)) and [joint aches](#) ([arthralgia](#)) are common. The skin may show [rashes](#), [swelling](#), [ulcers](#), and lumps (nodular lesions). Nerve involvement may cause sensory changes with [numbness](#), pain, burning and weakness. [Central nervous system](#) involvement may cause strokes or [seizures](#). Kidney involvement can produce varying degrees of [renal failure](#). Involvement of the arteries of the heart may cause a [heart attack](#) ([acute myocardial infarction](#)), [heart failure](#) and inflammation of the sack around the heart ([pericarditis](#)).

### **Prevention:**

There is no known prevention for this disease.

### **Symptoms:**

[fatigue](#)  
[weakness](#)  
[fever](#)  
[abdominal pain](#)  
[decreased appetite](#) ([anorexia](#))  
[weight loss, unintentional](#)  
[muscle aches](#) ([myalgia](#))  
[joint aches](#) ([arthralgia](#))

### **Signs and tests:**

There are no specific laboratory tests for the diagnosis of polyarteritis nodosa. The diagnosis is generally based upon clinical findings and a few laboratory studies which help to confirm the diagnosis.

[CBC](#) (may demonstrate an elevated [white blood count](#))

[ESR](#) (often elevated)

tissue [biopsy](#) (demonstrates inflammation in small arteries - arteritis)

immunoglobulins (may be increased)

### **Treatment:**

[prednisone](#)  
[cyclophosphamide](#)

**Expectations (prognosis):**

Treatment is mandatory for long-term survival. The five-year survival rate in treated patients may approach 50%.

**Complications:**

[stroke](#)

[renal failure](#)

[heart attack](#)

[intestinal necrosis](#) and perforation

**Other interesting websites:**

<http://www.angelfire.com/pa3/autoimmunedisease/aifeindex.html>

<http://www.merck.com/pubs/mmanual/section5/chapter50/50m.htm>

case study of patient with PAN

<http://dermatology.cdlib.org/DOJvol7num1/NYUcases/polyarteritis/kim.html>

[http://www.medhelp.org/HealthTopics/Polyarteritis\\_Nodosa.html](http://www.medhelp.org/HealthTopics/Polyarteritis_Nodosa.html)

[http://www.biostructural.com/polyarteritis\\_nodosa.shtm](http://www.biostructural.com/polyarteritis_nodosa.shtm)

**Systematic Lupus Erythematosus**

<http://cerebel.com/lupus/overview.htm>

**Presenting Signs and Symptoms**

80% of patients with SLE will present with involvement of the skin or joints. A common presenting complaint is a photosensitive rash often with alopecia. Alternatively, patients may present with arthralgia or frank arthritis. However, patients may present with fever accompanied by single organ involvement, such as inflammatory serositis, glomerulonephritis, neuropsychiatric disturbance or hematological disorder (i.e. autoimmune hemolytic anemia or thrombocytopenia). Rarely, patients present with severe, generalized acute lupus crisis with multiorgan involvement.

The course of SLE and common complications of the illness are best understood by reviewing the individual major areas of potential disease involvement.

<http://www.geocities.com/sarahmcraig/preventing.html>

Your doctor has put together a treatment plan that is designed specifically for you and your lupus. This probably includes physical and emotional rest, aggressive treatment of infections, good nutrition, and avoidance of direct sunlight and other sources of ultraviolet light. Your doctor may have also prescribed medications to control disease symptoms and other health problems that you might have. One of the most important ways you can help yourself is to understand your treatment plan and the things you need to do to keep your disease under control.

**Other interesting websites:**

<http://cerebel.com/lupus/overview.htm>

web site on "Prism", a trial/experiment/study of SLE

<http://www.prism-lupus.org/>

"layman's" website...check accuracy, but useful if correct as user friendly

<http://www.geocities.com/sarahmcraig/>

## **Trypanosomiasis**

[http://www.cdc.gov/ncidod/dpd/parasites/trypanosomiasis/factsheet\\_trypanosomiasis.htm](http://www.cdc.gov/ncidod/dpd/parasites/trypanosomiasis/factsheet_trypanosomiasis.htm)

There are two types of Trypanosomiasis (sleeping sickness)- African and American (the developed world). The African one originates from the bite of the tsetse fly, the "modern" one is thought to be caught from dogs, cats, fleas... Medication for Trypanosomiasis is available, but there is no preventive action as yet against the disease which can be effectively taken. Death will occur if the condition is not treated.

Other interesting websites:

<http://www.who.int/health-topics/afrtryps.htm>

-website with links to both African and American types of Trypanosomiasis

<http://www.healthubs.com/trypanosomiasis/>

<http://www.biosci.ohio-state.edu/~parasite/trypanosoma.html>

## **Vitamin B12 Deficiency**

<http://google.yahoo.com/bin/query?p=vitamin+B12+deficiency&hc=0&hs=0>

## **Dementia and Down's Syndrome**

[http://www.dsscotland.org.uk/information\\_frameset.htm](http://www.dsscotland.org.uk/information_frameset.htm)

Relatives and carers frequently play an important part in the process of identifying illness and seeking diagnosis. They are often the ones, with daily contact, who first become aware of the changes in someone's personality and behaviour.

When considering a diagnosis that might be dementia, a GP may refer on to a Consultant in Learning Disability or a Psychogeriatrician. All possible causes for changes in behaviour must be considered. For example, hypothyroidism, depression, even a chest infection can cause symptoms similar to dementia, but they are treatable. Clearly, full medical assessment is important.

Diagnosis of dementia comes about through a process of eliminating other possible causes of changes in mood and behaviour whilst also clarifying more specific symptoms, such as memory loss.

When dementia is diagnosed, it can come as a blow to family and friends. It can be a great shock: a disappointment that brings fear for the future. As yet, there is no cure for Alzheimer's disease. However, much can be done to ease the impact of the illness, and to maintain the best possible lifestyle for those affected. Individuals with DS and dementia may or may not themselves be aware of the changes that are taking place. Difficulties arising may or may not cause them frustration. In any case it's important to work with them: listening, respecting their views, and maintaining their dignity.

The most common cause of dementia is Alzheimer's disease. Unfortunately, it is more common in people with DS than in the general population, and can occur earlier. For all people, the risk of having Alzheimer's disease increases significantly as the individual gets older.

Alzheimer's disease results in disorder of brain function. There are characteristic physical changes in the brain; there is a loss (atrophy) of brain tissue, with significant numbers of brain cells dying.

The great majority of individuals with DS over forty will have these changes in their brains, but by no means all of them will show any signs of dementia. The majority of people with DS will never have Alzheimer's disease.

For the minority who do, typical symptoms of early dementia are problems with memory such as forgetting recent events, mislaying objects, switching on switches and forgetting about them, and so on. There can also be changes in behaviour and personality, such as withdrawal and irritability. Some individuals developing Alzheimer's disease start to have fits or seizures.

As the illness progresses, conversational skills deteriorate. The individual may become less sociable, more apathetic and withdrawn. Difficulties will arise over tasks that were previously well managed, such as with dressing and toileting. What causes Alzheimer's disease is not yet known, and as yet there is no cure. However, much can be done by carers, family and professionals to alleviate the symptoms and minimise distress.

**Other interesting websites:**

<http://www.nas.com/downsyn/>

National Down Syndrome association USA

<http://www.ndss.org/>

<http://www.ds-health.com/>

Down Syndrome journal with research papers etc:

<http://www.denison.edu/dsq/>

Website of Chicago's Down Syndrome association:

<http://www.nads.org/>

Website with newsletters, helpful caring hints, description of events etc of "Upside!" foundation:

<http://www.telebyte.com/upside/upside.html>

General website with contact numbers, useful addresses etc, mainly in the USA:

<http://www.downsyndrome.com/>

Canadian Down Syndrome Association website:

<http://www.cdss.ca/>

Scottish Down Syndrome website:

<http://www.sdsa.org.uk/>

Down Syndrome Research Foundation website...discussions, forum, research papers etc:

<http://www.dsrf.org/>



## **RARE DISEASES - websites**

Korsakoff's Syndrome

<http://google.yahoo.com/bin/query?p=korsakoff%27s+syndrome&hc=0&hs=1>

<http://www.scope2000.co.uk/>

Wernicke's disease

<http://google.yahoo.com/bin/query?p=wernicke%27s+disease&hc=0&hs=0>

<http://www.bme.jhu.edu/labs/chb/disorders/wernicke.html>

<http://www.whonamedit.com/synd.cfm/1352.html>

<http://www.healthcentral.com/mhc/top/000771.cfm>

Web site with pictures of brain tissue in various conditions (MS, Wernick's...)

<http://medlib.med.utah.edu/WebPath/CNSHTML/CNS085.html>

<http://www.medfriendly.com/wernickesdisease.html>

<http://www.austin360.com/shared/health/adam/ency/article/000771.html>

<http://psychiatry.mc.duke.edu/ijpm/26-3/Casanova.html>

<http://www.pennhealth.com/ency/article/000771.htm>

<http://www.zarcrom.com/users/alzheimers/odem/al6.html>

<http://www.nursingceu.com/NCEU/courses/wksyndrome/>

<http://www.rwjhamilton.org/Atoz/encyclopedia/article/000771.asp>

"Barnes and Noble" website/online store has a limited number of books on the above diseases.

<http://www.starkhealth.com/articles/psycho.htm>

[http://www.mrtabletdemon.co.uk/disease\\_index\\_W.htm](http://www.mrtabletdemon.co.uk/disease_index_W.htm)

Web address of Journals of toxicology in Wiley Science:

<http://www3.interscience.wiley.com/cgi-bin/abstract/78503518/START>

Alcohol related dementia

<http://google.yahoo.com/bin/query?p=alcohol+related+dementia&hc=0&hs=0>

Web address of articles on Wiley Interscience concerning alcohol related dementia

<http://www3.interscience.wiley.com/cgi-bin/search>

<http://www.zarcrom.com/users/alzheimers/odem/al-d.html>

<http://www.alz.co.uk/alzheimers/alcohol.html>

<http://www.alzheimers.asn.au/DementiaInformation/di03.html>

<http://www.agingincanada.ca/Seniors%20Alcohol/1e3-1.htm>

<http://www.living-your-life.com/health/alcohol-dementia.php>

BBC news articles on alcohol related topics: cost to health services, alcohol related dementia, alcoholism in the elderly...

<http://news.bbc.co.uk/1/hi/health/743878.stm>

Web site with methods to improve memory after alcohol related brain damage

[http://www.betterhealth.vic.gov.au/bhcv2/BHCLang.nsf/\(LevelFour\)/962585A35C9A70194A256B640083BA52?OpenDocument](http://www.betterhealth.vic.gov.au/bhcv2/BHCLang.nsf/(LevelFour)/962585A35C9A70194A256B640083BA52?OpenDocument)

<http://www.psyweb.com/Mdisord/subsd.html>

<http://www.netdoctor.co.uk/diseases/facts/dementia.htm>

Web site with a selection of "rare diseases", treatments etc...

<http://www.dementia.ie/AboutDementia/WhatIsDementia.html>

Multiple Sclerosis

<http://google.yahoo.com/bin/query?p=multiple+scerosis&hc=0&hs=0>

<http://www.nmss.org/>

<http://www.healthtalk.com/msen/>

Multiple Sclerosis societies of the UK

<http://www.mssociety.org.uk/index.html>

Web site with info on caring and treating MS

<http://www.mscares.org/>

Web site with articles, personal accounts etc to help understand MS

<http://www.understandingms.com/>

Address of MS society America

<http://www.msaa.com/>

Address of MS society Australia

<http://www.msaustralia.org.au/>

<http://www.noah-health.org/english/illness/neuro/muscler.html>

Web site with links to various MS support groups

<http://www.infosci.org/>

General web site on MS with personal accounts, treatment, famous people with MS, MS news...

<http://www.mult-sclerosis.org/>

Address of "Barnes and Noble" MS section

[http://r1.us.rmi.yahoo.com/rmi/http://search.barnesandnoble.com/booksearch/results.asp?rmivars%3ftarget=\\_top?WRD=multiple+sclerosis&userid=531GLUMVNJ&sourceid=00002123466036150646&bfdte=08%2D27%2D2002+10%3A20%3A12](http://r1.us.rmi.yahoo.com/rmi/http://search.barnesandnoble.com/booksearch/results.asp?rmivars%3ftarget=_top?WRD=multiple+sclerosis&userid=531GLUMVNJ&sourceid=00002123466036150646&bfdte=08%2D27%2D2002+10%3A20%3A12)

Address of MS society Ireland

<http://www.ms-society.ie/>

Web site with definitions, helpful descriptions etc of MS related words

<http://www.msonly.com/glossary.html>

MS support web site

<http://pages.cthome.net/Marrone/>

Address with various treatments of MS

<http://www.msadvances.com/>

MS chat room

<http://www.msadvances.com/>

<http://mscare.com/>

MS net guide (resources, trials, links, treatments...)

<http://www.msnetguide.com/>

Neurosyphilis

<http://google.yahoo.com/bin/query?p=neurosyphilis&hc=0&hs=0>

<http://neuroland.com/id/neurosyph.htm>

Psychiatrist's web site

<http://www.priory.com/psych/neurosyphilis.htm>

<http://www.hivpositive.com/f-Oi/OppInfections/4-Bacterial/4-Syphilis.html>

<http://www.medterms.com/script/main/art.asp?articlekey=4558>

<http://www.emedicine.com/neuro/topic684.htm>

Address with reports/articles on two neurosyphilis patients

<http://ams.cu.edu.tr/January2000Vol9No1/hacer.html>

<http://www.healthcentral.com/mhc/top/000703.cfm>

[http://www.upcmd.com/dot/examples/01080/criteria\\_syphilis.html](http://www.upcmd.com/dot/examples/01080/criteria_syphilis.html)

Address for "Barnes and Noble" section of books on neurosyphilis

<http://r1.us.rmi.yahoo.com/rmi/http://search.barnesandnoble.com/booksearch/results.asp?rmivars%3ftarget=top?sourceid=00339421585264447149&bfd=08-27-2002+10:52:43&WRD=neurosyphilis>

<http://www.enlmedical.com/article/000703.htm>

<http://www.xrefer.com/entry/127612>

Case report of neurosyphilis

[http://www.acta-clinica.com/Volume\\_39\\_2000/8/8.htm](http://www.acta-clinica.com/Volume_39_2000/8/8.htm)

<http://www.rwjhamilton.org/Atoz/encyclopedia/article/000703.asp>

Creutzfeldt-jacob disease

<http://google.yahoo.com/bin/query?p=creutzfeldt-jacob+disease&hc=0&hs=0>

BBC web site address with all related news story links

<http://www.bbc.co.uk/cgi-bin/search/results.pl?q=creutzfeldt-jacob+disease&uri=%2Fhome%2Ftoday%2Findex.live.shtml&x=13&y=14>

Web site with stats of CJD in the UK, annual reports...

<http://www.cjd.ed.ac.uk/>

<http://cjd.foundation.org/>

Web site that concentrates on bovine form, but also has large amounts of general news articles and also a list of leading researchers, scientists etc of CJD

<http://www.mad-cow.org/>

[http://www.mdtravelhealth.com/infectious/mad\\_cow\\_disease.html](http://www.mdtravelhealth.com/infectious/mad_cow_disease.html)

[http://www.medical-library.org/journals2a/creutzfeldt\\_jacob2.htm](http://www.medical-library.org/journals2a/creutzfeldt_jacob2.htm)

Address with links to web sites covering care of CJD patients

<http://www.austinareaaorn.org/creutzfeldt.htm>

<http://www.avera.org/adam/ency/article/000788.htm>

"Slide show" web site...brain tissues, MRI scans etc

<http://www.biomedicine.dote.hu/cselenyi/phd3/sld015.htm>

Web site concerning policies, legislation on bovine form/protection of humans...

<http://olpa.od.nih.gov/legislation/107/session1/planimaldisease.asp>

Pseudo dictionary of abbreviations used in general CJD terminology

<http://www.defra.gov.uk/animalh/bse/glossary.html>

<http://omni.ac.uk/browse/mesh/detail/C0022336L0022336.html>

[http://www.thehealthyforum.com/bodymapssec/brain\\_cjd.html](http://www.thehealthyforum.com/bodymapssec/brain_cjd.html)

<http://clinlabs.path.queensu.ca/ic/cjd.htm>

Cerebral Lipidosis

<http://google.yahoo.com/bin/query?p=cerebral+lipidosis&hc=0&hs=0>

<http://yalenewhavenhealth.org/library/healthguide/IllnessConditions/topic.asp?hwid=nord12>

<http://www.icomm.ca/geneinfo/def-c.htm>

What seems to be an informative web site on Alzheimer's and other related diseases, but in Spanish

<http://www.homestead.com/montedeoya/clubalzheimer.html>

<http://www.medev.ch/pufa/pufa9704.htm>

<http://www.mazornet.com/genetics/gauchers.asp>

<http://www.ukselfhelp.info/c.htm>

Hepatolenticular degeneration (Wilson's Disease)

<http://google.yahoo.com/bin/query?p=hepatolenticular+degeneration&hc=0&hs=0>

<http://www.hon.ch/HONselect/RareDiseases/C06.552.413.html>

<http://www.geocities.com/nutriflip/Diseases/WilsonsDisease.html>

[http://dmoz.org/Health/Conditions\\_and\\_Diseases/Nutrition\\_and\\_Metabolism\\_Disorders/Vitamins\\_and\\_Minerals/Wilson's\\_Disease/](http://dmoz.org/Health/Conditions_and_Diseases/Nutrition_and_Metabolism_Disorders/Vitamins_and_Minerals/Wilson's_Disease/)

Web site with a list of links for hepatolenticular degeneration

<http://www.bdid.com/wilson.htm>

Web site with slides of affected areas in Wilson's disease

<http://www.brisbio.ac.uk/ROADS/subject-listing/hepatolenticulardegeneration.html>

<http://www.avera.org/adam/ency/article/000785.htm>

Hypercalcaemia

<http://www.nlm.nih.gov/medlineplus/ency/article/000365.htm>

Address contains links to clinical trials etc

<http://rarediseases.info.nih.gov/>

Hypothyroidism

<http://www.unithroid.com/>

not sure if this is right for this condition...spelling??

<http://www.hypoparathyroidism.org./default.php>

<http://www.hypoparathyroidism.org/definition.php>

Polyarteritis Nodosa

<http://google.yahoo.com/bin/query?p=polyarteritis+nodosa&hc=0&hs=0>

<http://www.angelfire.com/pa3/autoimmunedisease/aifeindex.html>

<http://www.merck.com/pubs/mmanual/section5/chapter50/50m.htm>

case study of patient with PAN

<http://dermatology.cdlib.org/DOJvol7num1/NYUcases/polyarteritis/kim.html>

[http://www.medhelp.org/HealthTopics/Polyarteritis\\_Nodosa.html](http://www.medhelp.org/HealthTopics/Polyarteritis_Nodosa.html)

<http://www.healthcentral.com/mhc/top/001438.cfm>

[http://www.biostructural.com/polyarteritis\\_nodosa.shtm](http://www.biostructural.com/polyarteritis_nodosa.shtm)

Systematic Lupus Erythematosus

<http://search.yahoo.com/search?p=systemic+lupus+erythematosus>

<http://cerebel.com/lupus/overview.htm>

web site on "Prism", a trial/experiment/study of SLE

<http://www.prism-lupus.org/>

"layman's" website...check accuracy, but useful if correct as user friendly

<http://www.geocities.com/sarahmcraig/>

Trypanosomiasis

<http://search.yahoo.com/search?p=trypanosomiasis>

<http://www.who.int/health-topics/afrtryps.htm>

<http://www.cdc.gov/ncidod/dpd/parasites/trypanosomiasis/default.htm>

website with links to both African and American types of Trypanosomiasis

<http://www.healthubs.com/trypanosomiasis/>

<http://www.biosci.ohio-state.edu/~parasite/trypanosoma.html>

Vitamin B12 Deficiency

<http://google.yahoo.com/bin/query?p=vitamin+B12+deficiency&hc=0&hs=0>

Corticobasal Degeneration

<http://google.yahoo.com/bin/query?p=cortical+basal+degeneration&hc=0&hs=0>

<http://www.emedicine.com/NEURO/topic77.htm>

[http://www.ninds.nih.gov/health\\_and\\_medical/disorders/cortico\\_doc.htm](http://www.ninds.nih.gov/health_and_medical/disorders/cortico_doc.htm)

<http://www.cmdg.org/Movement/Parkinsons/Plus/CBGD/cbgd.htm>

<http://www.njneuro.org/movedis/cbgd.htm>

<http://www.neurologyindia.com/vol48-4/le3.pdf>

website with journals on corticobasal degeneration

<http://jama.ama-assn.org/issues/v284n14/fpdf/jbk1011.pdf>

-Med Help website. Need to get user name etc to log in, but contains journals, clinical trials etc

[http://www.medscape.com/viewarticle/405229\\_print](http://www.medscape.com/viewarticle/405229_print)

-clinical trials website

<http://clinicaltrials.gov/ct/gui/show/NCT00017940?order=17>

Normal Pressure Hydrocephalus

<http://search.yahoo.com/search?p=normal+pressure+hydrocephalus>

<http://www.allaboutnph.com/>

Progressive Supranuclear Palsy

<http://search.yahoo.com/search?p=progressive+supranuclear+palsy>

[http://www.ninds.nih.gov/health\\_and\\_medical/disorders/psp.htm](http://www.ninds.nih.gov/health_and_medical/disorders/psp.htm)

society of PSP (USA)

<http://www.psp.org/>

BBC news on SPS plus links to relevant stories

[http://news.bbc.co.uk/2/hi/health/medical\\_notes/461557.stm](http://news.bbc.co.uk/2/hi/health/medical_notes/461557.stm)

Website of PSP association, Europe

<http://www.pspeur.org/>

Multiple System Atrophy

<http://search.yahoo.com/search?p=Multiple+system+atrophy>

<http://www.ndrf.org/MSA.htm>

[http://www.ninds.nih.gov/health\\_and\\_medical/disorders/shydrger\\_doc.htm](http://www.ninds.nih.gov/health_and_medical/disorders/shydrger_doc.htm)

<http://neuro-www.mgh.harvard.edu/forum/ShyDragerMenu.html>

CADASIL

<http://google.yahoo.com/bin/query?p=CADASIL&hc=0&hs=0>

<http://www.thedoctorsdoctor.com/diseases/cadasil.htm>

<http://www.genetics.ucla.edu/schanenlab/cadasil.html>



<http://brainpath.medsch.ucla.edu/abstracts/vol12/1203/1203a9.htm>

journal study of CADASIL patients

<http://www.ahsnet.org/journal/1999/02/Feb99abstrts.pdf>

Subcortical Vascular Dementia

<http://google.yahoo.com/bin/query?p=subcortical+vascular+dementia&hc=0&hs=0>

<http://www.uku.fi/neuro/ab0234.htm>

very detailed article/study of vascular dementia which includes the subcortical type

[http://www.thomasland.com/\\_nonsearch/TSR07305.pdf](http://www.thomasland.com/_nonsearch/TSR07305.pdf)

[http://www.mental-health-matters.com/disorders/dis\\_details.php?disID=30](http://www.mental-health-matters.com/disorders/dis_details.php?disID=30)

<http://www.cpa.ca/factsheets/cognitive.htm>

website with details of a drug trial for the treatment of subcortical vascular dementia

<http://www.bscc.co.uk/newsdigest/news0502.html>

### **Gerstmann-Sträussler-Scheinker Syndrome**

No websites on this, several on Gerstmann Syndrome which (check source) is not the same thing.

### **Aids Dementia Complex.**

<http://google.yahoo.com/bin/query?p=aids+dementia+complex&hc=0&hs=0>

<http://www.aegis.com/topics/oi/oi-adc.html>

<http://www.sfaf.org/treatment/beta/b31/b31adc.html>

<http://www.neuro.wustl.edu/narc/adcd.html>

<http://www.med.harvard.edu/AANLIB/cases/case14/mr1-tc1/010.html>

This one has info on lots of the other diseases as well:

<http://omni.ac.uk/browse/mesh/detail/C0001849L0001849.html>

<http://www.projinf.org/fs/dementia.html>

<http://www.emedicine.com/NEURO/topic447.htm>

[http://www.thebody.com/treat/neuro\\_dementia.html](http://www.thebody.com/treat/neuro_dementia.html)

This address is a « slide show » of brain scans etc of patients with ADC:

[http://brighamrad.harvard.edu/education/online/BrainSPECT/Main\\_Slide\\_Show/Main\\_SS\\_19.html](http://brighamrad.harvard.edu/education/online/BrainSPECT/Main_Slide_Show/Main_SS_19.html)

<http://www.aidsinfonyet.org/504-dementia.html>

[http://pni.med.jhu.edu/Past\\_Research/aids\\_dementia.htm](http://pni.med.jhu.edu/Past_Research/aids_dementia.htm)

[http://www.niv.ac.za/virusa/aidsa/v7\\_3.htm](http://www.niv.ac.za/virusa/aidsa/v7_3.htm)

<http://www.retroconference.org/2002/Abstract/13381.htm>

<http://www.fpnotebook.com/HIV47.htm>

<http://www.hivcybermall.org/doctorsedu/AIDS%20Dementia%20Complex.htm>

This site has articles on the results of drug trials of a whole array of drugs:

<http://www.hivpositive.com/f-Oi/OpInfections/4-Neuro/4-Neu-Demen.html>

### **Alcohol related dementia**

<http://google.yahoo.com/bin/query?p=alcohol+related+dementia&hc=0&hs=0>

<http://www.alzheimers.asn.au/DementiaInformation/di03.html>

<http://www.zarcrom.com/users/alzheimers/odem/al-d.html>

<http://www.alz.co.uk/alzheimers/alcohol.html>

Tasmania's Alzheimer's Association's website:

<http://www.tased.edu.au/tasonline/tasalz/alcdem.htm>

<http://www.alzvic.asn.au/ad13.htm>

<http://www.stir.ac.uk/Departments/HumanSciences/AppSocSci/DS/management.htm>

<http://www.alzwissc.org/alcoholanddem.html>

<http://www.psyweb.com/Mdisord/subsd.html>

<http://alcoholresearch.lsumc.edu/effects/Marchiafava-Bignami.asp>

[http://www.medicouncilalcol.demon.co.uk/handbook/chapter\\_4.htm](http://www.medicouncilalcol.demon.co.uk/handbook/chapter_4.htm)

### **Binswanger's disease:**

<http://google.yahoo.com/bin/query?p=Binswangers+disease&hc=0&hs=0>

<http://www.clevelandclinic.org/health/health-info/docs/1200/1256.asp?index=6016>

<http://healthlink.mcw.edu/article/921389325.html>

<http://serendip.brynmawr.edu/bb/neuro/neuro01/web2/Ledoux.html>

Address for the National Organisation of Rare Diseases:

[http://www.rarediseases.org/search/rdbdetail\\_abstract.html?disname=Binswanger's%20Disease](http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=Binswanger's%20Disease)

<http://hvelink.saint-lukes.org/library/healthguide/IllnessConditions/topic.asp?hwid=nord614>

<http://archneur.ama-assn.org/issues/v56n9/fpdf/ned8355.pdf>

Alzheimer's association of Toronto (Binswanger's disease plus other rare diseases):

<http://www.asmt.org/RD2.htm>

Lewy body dementia:

<http://search.yahoo.com/search?p=lewy+body+dementia>

<http://www.zarcrom.com/users/alzheimers/odem/lewy-d.html>

<http://www.emedicine.com/neuro/topic91.htm>

<http://www.nottingham.ac.uk/pathology/lewy/lewyhome.html>

<http://www.alznorcal.org/research/resrchlewy.html>

<http://www.lewybodydisease.org/>

### **Multi-infarct dementia**

<http://google.yahoo.com/bin/query?p=multi+infarct+dementia&hc=0&hs=0>

[http://www.ninds.nih.gov/health\\_and\\_medical/disorders/multi-infarctdementia\\_doc.htm](http://www.ninds.nih.gov/health_and_medical/disorders/multi-infarctdementia_doc.htm)

<http://www.mentalhealth.com/dis/p20-or02.html>

<http://www.nmha.org/infoctr/factsheets/102.cfm>

<http://www.nlm.nih.gov/medlineplus/ency/article/000746.htm>

This is a fairly interesting site with an online journal written by someone with multi-infarct dementia...not sure how useful it is though...

<http://www.geocities.com/resa526/journal2001.html>

<http://www.alzheimers.org/pubs/mid.htm>

This address is another part of the "slide show" website- brain scans etc:

[http://brighamrad.harvard.edu/education/online/BrainSPECT/Main\\_Slide\\_Show/Main\\_SS\\_9.html](http://brighamrad.harvard.edu/education/online/BrainSPECT/Main_Slide_Show/Main_SS_9.html)

[http://www.psychnet-uk.com/dsm\\_iv/multi\\_infarct\\_dementia.htm](http://www.psychnet-uk.com/dsm_iv/multi_infarct_dementia.htm)

[http://www.allsands.com/Health/Diseases/multiinfarctde\\_xmr\\_gn.htm](http://www.allsands.com/Health/Diseases/multiinfarctde_xmr_gn.htm)

<http://www.acnp.org/G4/GN401000146/CH143.html>

Interesting website on clinical trials:

<http://www.clinicaltrials.gov/ct/gui/info/resources;jsessionid=32B4F0314303B2840850F5A888B24615>

Easy to use website with info on carers, general caring for the elderly and several dementia-related diseases:

<http://www.helpguide.org/dementias/multiinfarct.asp>

Informative website with chapters on all the rare diseases:

[http://www.dasninternational.org/regular/reg\\_information.html](http://www.dasninternational.org/regular/reg_information.html)

Address for Los Angeles Alzheimer's association:

<http://www.alzla.org>

Address for the Texan Department of Health, Alzheimer's Program:

<http://www.tdh.state.tx.us/osp/alz.htm>

List of contact numbers for Alzheimer associations around the USA:

[http://www.tdh.state.tx.us/osp/a\\_info.htm](http://www.tdh.state.tx.us/osp/a_info.htm)

Alzheimer's association San Diego (USA):

[http://www.sanhttp://www.health-nexus.com/multi-infarct\\_dementia.html](http://www.sanhttp://www.health-nexus.com/multi-infarct_dementia.html)  
[http://www.alz.org/about\\_disease/overview.html](http://www.alz.org/about_disease/overview.html)

### **Pick's Disease:**

<http://search.yahoo.com/search?p=pick%27s+disease>

National Niemann-Pick's Disease Foundation:

<http://www.nnpdf.org/>

National Pick's Disease Foundation, UK:

<http://www.nnpdf.org/npdg-uk/index.html>

Website about "Jacob", a child who has Niemann-Pick Disease:

<http://www.jacob-quinn.com/>

<http://www.parseghian.org/apmrfweb/>

<http://health.yahoo.com/health/dc/001207/0.html>

## **Down Syndrome**

<http://search.yahoo.com/search?p=down+syndrome>

<http://www.nas.com/downsyn/>

National Down Syndrome association USA

<http://www.ndss.org/>

<http://www.ds-health.com/>

Down Syndrome journal with research papers etc:

<http://www.denison.edu/dsq/>

Website of Chicago's Down Syndrome association:

<http://www.nads.org/>

Website with newsletters, helpful caring hints, description of events etc of "Upside!" foundation:

<http://www.telebyte.com/upside/upside.html>

General website with contact numbers, useful addresses etc, mainly in the USA:

<http://www.downsyndrome.com/>

Canadian Down Syndrome Association website:

<http://www.cdss.ca/>

Scottish Down Syndrome website:

<http://www.sdsa.org.uk/>

Down Syndrome Research Foundation website...discussions, forum, research papers etc:

<http://www.dsrf.org/>

## **Huntington's disease**

<http://search.yahoo.com/search?p=huntington%27s+disease>

<http://www.yourgenesyourhealth.org/ygyh/mason/ygyh.html?syndrome=hd>

Huntington's Disease Society of America:

<http://www.hdsa.org/>

<http://www.hda.org.uk/>

<http://www.interlog.com/~rlaycock/2nd.html>

Website on caring for people with Huntington's Disease:

<http://www.kumc.edu/hospital/huntingtons/>

<http://www.hdlighthouse.org/>

<http://www.hdny.org/>

Website with a list of useful contact numbers, mainly ones in the USA:

<http://www.lib.uchicago.edu/~rd13/hd/electron.html>

Canada's Huntington's Society's website:

<http://www.hsc-ca.org/>

The Scottish Huntington's website:

<http://www.hdscotland.org/>

German Huntington's website:

[http://www.gwdg.de/~usancke/DHH/engl\\_ind.html](http://www.gwdg.de/~usancke/DHH/engl_ind.html)

General Caregiver Website:

<http://www.caregiver.org/>

**Parkinson's disease:**

<http://search.yahoo.com/search?p=parkinsons+disease>

<http://www.pdf.org/index.cfm>

Caregiver info:

<http://www.parkinsonscare.com/>

Website for the American Parkinson's Society:

<http://www.apdaparkinson.org/>

Address for World Parkinson's Association:

<http://www.wpda.org/>

<http://www.va.gov/padrecc/>

<http://www.whatsnewinpd.com/>

<http://www.parkinsonsinfo.com/>

Paper on Parkinson's patients living at home:

<http://www.cnsonline.org/www/archive/parkins/park-02.txt>

Website on young sufferers of Parkinson's:

<http://www.young-parkinsons.org.uk/>

<http://www.endotoxin.gmxhome.de/>

<http://www.parkinson.org/>

<http://www.parkinsonsnet.com/>

Parkinson's Institute of Ireland:

<http://www.officeobjects.com/PARKINSONS/>

Society for Neuroscience website...research papers, updates, news etc:

<http://apu.sfn.org/briefings/parkinsons.html>

More info on clinical trials:

<http://www.arkansasclinicaltrials.com/>

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