



# Wilson Disease Watch

January-June 2011. vol 1

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# Corpus callosum abnormalities in Wilson's disease

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*I. J Neural Neurosurg Psychiatry (2010).*

## Summary

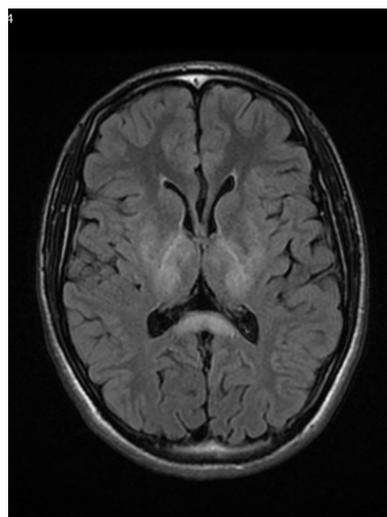
**Introduction:** Neurological presentations of Wilson's disease (WD) are associated with brain lesions classically localized in globus pallidus, putamen, thalamus, mesencephalon, pons, and dentate nucleus. Less attention have been paid to cortico-subcortical white matter abnormalities.

**Objective:** Evaluation of the frequency of corpus callosum (CC) lesions in patients with neurological symptoms related to WD and description of their clinical consequences.

**Method:** We included all patients with neurological expression of WD, followed in the French national centre for WD who had a brain MRI between March 2006 and December 2008. Localization of brain lesions were analysed and the frequency of lesions in CC evaluated. All patients were assessed using UWDRS (Unified Wilson's Disease Rating Scale). For patients with abnormalities located in CC, a clinical dysconnexion syndrome was investigated.

**Results:** Among 81 patients (45 men, 36 females, mean age: 34.8 years, from 12 to 74 years) with neurological expression, 42% had white matter lesions. 23.4 % of patients presented CC lesions, limited to the posterior part or splenium. The severity of disability estimated by UWDRS was correlated with the presence of CC lesions on MRI.

**Conclusion:** Abnormalities in CC are not unusual (23.4%). Together with lesions of basal ganglia, CC signal changes should suggest a WD diagnosis.



## Comments

This study reports hypersignal FLAIR in the splenium of corpus callosum in 23.4% of Wilson's disease patients presenting neurological symptoms. This localisation does not, apparently, induce any specific symptoms but is correlated with the handicap as evaluated by UWDRS.

## Take home message

Together with lesions in basal ganglia, corpus callosum lesions strongly suggest the diagnosis of Wilson's disease.

## Interest for patients

Topography or some lesions observed on Brain MRI would strongly suggest the diagnosis of Wilson's disease associated with neurological symptoms

# Zinc monotherapy is not as effective as chelating agents in treatment of Wilson disease

Weiss KH, Gotthardt DN, Klemm D, Merle U, Ferenci-Foerster D, Schaefer M, Ferenci P, Stremmel W. Gastroenterology, 2011.

## Summary

### BACKGROUND & AIMS:

Wilson disease is a genetic disorder that affects copper storage, leading to liver failure and neurologic deterioration. Patients are treated with copper chelators and zinc salts, but it is not clear what approach is optimal because there have been few studies of large cohorts. We assessed long-term outcomes of different treatments.

### METHODS:

Patients in tertiary care centers were retrospectively analyzed (n = 288; median follow-up time, 17.1 years) for adherence to therapy, survival, treatment failure, and adverse events from different treatment regimens (chelators, zinc, or a combination). Hepatic treatment failure was defined as an increase in activity of liver enzymes (aspartate aminotransferase, alanine aminotransferase, and  $\gamma$ -glutamyltransferase) >2-fold the upper limit of normal or >100% of baseline with an increase in urinary copper excretion.

### RESULTS:

The median age at onset of Wilson disease was 17.5 years. Hepatic and neuropsychiatric symptoms occurred in 196 (68.1%) and 99 (34.4%) patients, respectively. Hepatic treatment failure occurred more often from zinc therapy (14/88 treatments) than from chelator therapy (4/313 treatments;  $P < .001$ ). Actuarial survival, without transplantation, showed an advantage for chelating agents ( $P < .001$  vs zinc). Changes in treatment resulted mostly from adverse events, but the frequency did not differ between groups. Patients who did not respond to zinc therapy showed hepatic improvement after reintroduction of a chelating agent.

## Comments

Treatments with chelating agents or zinc salt are effective in most patients with Wilson's disease; this retrospective study suggests that chelating agents are better at preventing hepatic deterioration.

### Interest for patients

It is important to identify patients who do not respond to zinc therapy and have increased activities of liver enzymes, indicating that a chelating agent should be added to the therapeutic regimen.

# EuroWilson DG Sanco

## Operating Grant - Network meeting

January 28th, 2011 - Luxembourg



### ***In 2011, the general objectives of the EuroWilson network are:***

- To improve information by regularly updating the website [www.eurowilson.org](http://www.eurowilson.org).
- To increase knowledge of the disease
- To improve access to diagnosis, treatment and high quality of care by developing European guidelines



EuroWilson has received 167k€ funding from the EU Commission DG Sanco. This funding will be principally used to collect follow-up data in the registry, for network meetings, patient group meetings and updating the website.

### **Regarding the database the key messages were to:**

- collect follow-up of the important cohort of patients in the database.
- include siblings screened for WD
- finalise the on-going publications



# Rare Disease Day

February 28th 2011 - Paris



*Rare Disease Day*

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*Rare Disease Day is an annual, awareness-raising event co-ordinated by [EURORDIS](#) at the international level and National Alliances of Patient Organisations at the national level*

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Rare Disease Day was first launched on 29th February 2008. On this day patient groups, from different countries, collaborated on a large-scale awareness raising campaign.

They organised events including walks and press conferences.

Each year Rare Disease Day has enjoyed an ever growing number of participating countries and patient organisations.

In Lariboisière Hospital, in Paris, during Rare Disease Day 2011 on 28th February, the EuroWilson Network was represented by Dr Jean-Marc Trocello, Network director and by Serge Renaud, President of the French patient association.



# EuroWilson

## Patient Representatives Meeting

June 10th, 2011 - Paris

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**THE AIM OF THE MEETING WAS PROVIDE THE FORUM FOR SHARING EXPERIENCE BETWEEN THE DIFFERENT NATIONAL ASSOCIATIONS OF WILSON'S DISEASE IN EUROPE**

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EuroWilson invited representatives from several patient associations in Europe to join together for a meeting in Paris on 10th June 2011.

Each organization gave a brief account of its key activities and history. There was general agreement that EuroWilson can best support the patient group by:

- 1/ Translating and disseminating documents
- 2/ Improving the visibility of the associations in each country through its website [www.eurowilson.org](http://www.eurowilson.org)
- 3/ Developing a survey to patients to better understand needs and expectations
- 4/ Organising meetings

For more information on the Wilson's disease patient organizations, please go to their websites:

### **FRANCE**

[www.abpmaladiewilson.fr](http://www.abpmaladiewilson.fr)

### **GERMANY**

[www.morbus-wilson.de](http://www.morbus-wilson.de)

### **ITALY**

[www.malattiadwilson.it](http://www.malattiadwilson.it)

### **SPAIN**

[www.enfermedaddewilson.org](http://www.enfermedaddewilson.org)

### **SWITZERLAND**

The Swiss WD organisation for German speaking patients

### **UK**

[www.wilsonsdisease.org](http://www.wilsonsdisease.org)

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**PRESENTATION OF A SURVEY FOR THE WD PATIENTS TO BETTER UNDERSTAND THEIR EXPECTATIONS OF THE NETWORK AND HEALTHCARE IN EUROPE**

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### **ACTION POINTS**

- To send back comments about patient survey to have a final version to be translated as soon as possible
- To translate the dietary advice leaflet for WD patients into several languages
- To translate and disseminate WDSG-UK pamphlet



ASOCIACIÓN  
ESPAÑOLA DE  
FAMILIARES Y  
ENFERMOS DE  
WILSON



# Agenda 2011

## January

January, 28th, 2011 : European meeting, EuroWilson, Luxembourg

## February

February, 28th, 2011 : Rare disease day

## May

European Guidelines with EASL

## June

June, 10th, 2011: European patient representatives meeting, Paris

## July

July, 5th, 2011 : Executive board meeting, Paris

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# Actualities of European Database

- European charter will be proposed to the executive board in July
- The network recruited a Database Manager in June the 1<sup>st</sup>, 2011, Emeline Ruano
- A new owner of the database programm (TechniData) is now able to find solutions for the CPS Cards.  
Each card will be either renewed or released.
- The set up of the “follow up” in the database will begin soon