ERG, VEP and Oculo-motility in Wilson’s disease: a study on 34 patients

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Wilson's disease is a recessive autosomal genetic disorder (mutation of ATP 7B gene on chromosome 13) resulting from a copper metabolism anomaly. It produces hepatic, neuro-psychiatric and ophthalmologic manifestations such as Kayser-Fleisher ring and sunflower cataract.

The origin of these is an excessive accumulation of copper in the liver, cornea, kidneys and (mainly) the basal ganglia, as well as in other cerebral structures such as the cerebellum and white matter.

Reports on sub-clinical involvement of pathways other than the extra-pyramidal system are rare and, despite the implications for the eye and cerebral structures, there are only a few studies on the ERG, VEP or oculo-motility (OM) in this pathology.

Materials and Methods
34 patients affected by Wilson's disease, aged from 14 to 55 years were studied. An ophthalmological clinical evaluation, and eye movement recording utilising the EOG (OM) were performed for each patient. ERGs and VEPs (ISCEV procedures) were performed on 11 patients, CTs were performed on 6 patients and cerebral MRIs on 28.

Oculo-motility recording in Wilson's disease
Patient O.R.: normal EOG - normal vertical pursuit
Patient M.B.: abnormal EOG - saccadic vertical pursuit

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EOG: ocular movement recordings
Results
- ERG normal for all 11 patients
- VEP abnormal in 5 patients and normal in 6
- OM abnormal vertical pursuit in 29 patients of 34 (85%)
  abnormal vertical opto-kinetic nystagmus (OKN)
  abnormal horizontal pursuit in 14 patients (41%).
When the MRI was abnormal (20 cases), the oculo-motility results were always abnormal. When the MRI was normal (8 cases), the oculo-motility was abnormal in 6 patients and normal in 2. Furthermore, despite a normal neurological examination, 7 patients had an abnormal oculo-motility recording.

Oculo-motility recordings in Wilson’s disease

![Graph showing oculo-motility recordings in Wilson's disease](image)

Normal ERG in Wilson’s disease - patient S.R.

![Graph showing normal ERG in Wilson's disease](image)

VEPs in Wilson's disease

![Graph showing VEPs in Wilson's disease](image)
MR imaging: typical features in Wilson's disease - patient G.D.

Cerebral MRI in 28 patients

Discussion and Conclusion

These findings confirmed that VEPs can be altered in Wilson's disease, and demonstrated the usefulness of OM recordings in its diagnosis, even in cases of normal neurological examination and normal cerebral MRI. The OM recordings and VEPs could demonstrate subclinical functional neuro-visual disorders and therefore aid early diagnosis of Wilson's disease.

Wilson's disease is the first neurological genetically discovered disease with an effective treatment. However, delayed diagnosis causes fatal complications. An early diagnosis of Wilson's disease is essential for a better prognosis, avoiding irreversible lesions in the brain, brainstem and liver. OMs and VEPs contribute to this early diagnosis.

In Wilson's disease, discontinuing the treatment (diet without copper, D-Penicillamine, TETA, Zinc therapy) will be fatal.

References