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THE BURDEN OF WILSON DISEASE: RESULTS FROM A FRENCH PATIENT QUESTIONNAIRE

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BACKGROUND AND AIMS

- Wilson disease (WD):
  *an autosomal recessive disorder of impaired copper transport and excretion, caused by loss of function of the ATP7B copper-binding protein
  *ATP7B mutations impair the hepatic copper excretion pathway, resulting in toxic copper accumulation in the liver, brain and other tissues
  *fatal if left undiagnosed and untreated
- WD affects approximately 15,000 patients in Europe. In France, a recent observational population-based study reported 906 prevalent cases
- At the French National Reference Centre for Wilson Disease (FNRCWD), a patient questionnaire was developed to evaluate how WD influences the everyday life of patients and their families, to better understand their needs

METHODS

- a 40-questions survey
- completed either online, by mail or at the FNRCWD
- between 6 November 2017 and 6 January 2018
- covering 6 topics: diagnosis, impact, care, treatment, costs and information access

236 patients with WD
52% between 18-40y
Female: 58%

INFORMATION ACCESS

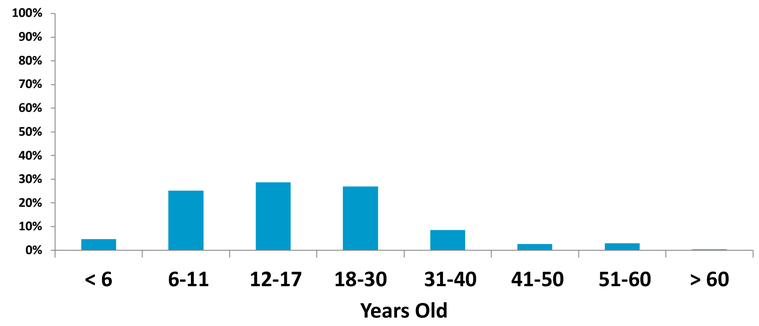
- 61% find relevant information about WD at the time of diagnosis
- 70% were satisfied with the information found
- During the last 6 months, 39% looked for medical information about WD and the majority found appropriate answers from specialist doctors, by reading the FNRCWD or WD patient association websites
- 51% find important to be in contact and exchange with other WD patients

COSTS

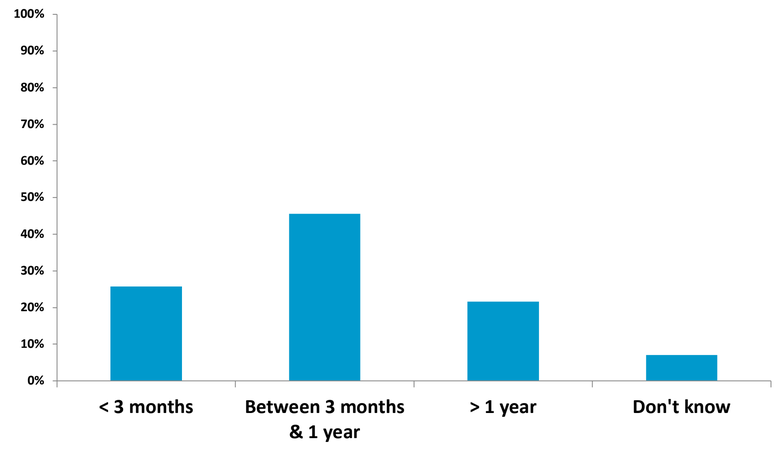
Only 4% of patients reported reimbursement issues

DIAGNOSIS

- **Age at diagnosis**



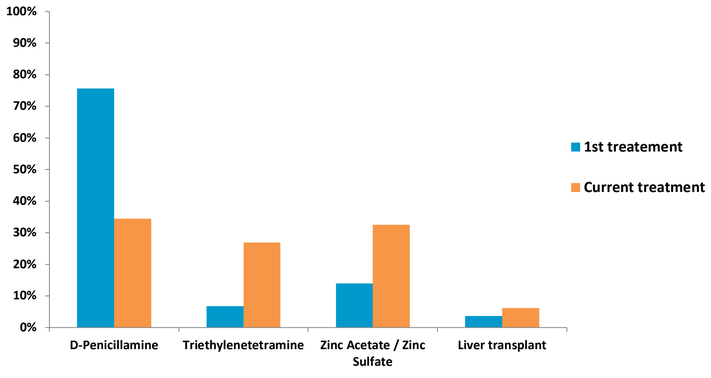
- **Most common first symptoms:** fatigue, liver problems, tremor and writing difficulties
- **Time to diagnosis after noticing first symptoms:**



- Diagnosis mostly made by a gastroenterologist (42%), neurologist (31%) or pediatrician (11%)

TREATMENT

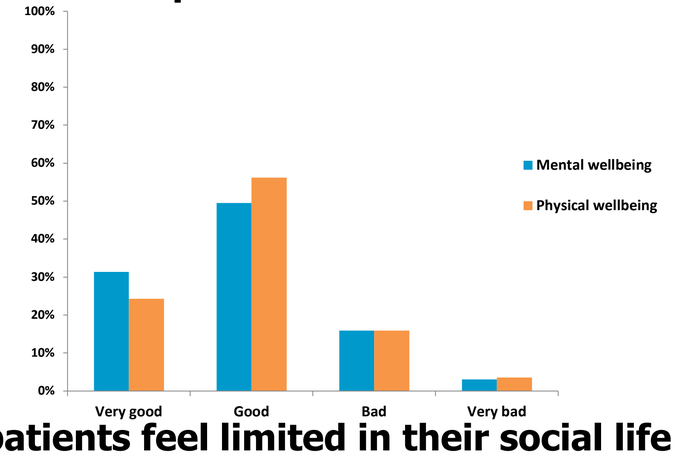
- During the first 6 months of treatment, symptoms:
  * improved (32%) or were stabilized (46%)
  * worsened in 22%
- Evolution of treatment: **60% had their treatment changed since the diagnosis, mostly due to side effects**



- At the time of questionnaire completion:
  * 52% reported difficulties taking pills 2 or 3 times a day
  * 5% reported side effects affecting daily life and leading to a cessation of treatment

IMPACT

- **Mental & Physical wellbeing at the time of questionnaire completion**



- **26% of patients feel limited in their social life**
- 6% of patients need a caregiver to assist them in their daily activities. **Caregiver is a family member in 92%**

CARE

- 2/3 of patients diagnosed during childhood find the transition from the child to adult care satisfactory
- 76% were under the care of a neurologist or gastroenterologist, at least twice a year
- 67% had at least one consultation in the FNRCWD

CONCLUSIONS

⇒ This patient questionnaire was a success as it was completed by almost 25% of French patients with WD
⇒ Diagnosis may be delayed in some patients and there is a need to monitor the efficacy and safety of treatment but good mental and physical wellbeing may be achieved in most patients
⇒ Regular follow-up by medical specialists is needed and compliance remains a fundamental issue

* *This survey has been made with the support of Wilson Therapeutics*