May 2023

Policy change for Wilson's disease

Action statement

Wilson's disease is a rare genetic condition that affects the body's ability to metabolise copper. It has a range of potentially debilitating symptoms that can have a devastating impact on the lives of the people affected and those of their families. Wilson's disease is progressive and, if left untreated, usually fatal.[1] However, early diagnosis and appropriate treatment can give a person living with Wilson's disease a similar life expectancy to that of the general population.[2]

The European Liver Patients' Association (ELPA) and the Wilson's Disease Policy Network jointly organised a meeting on 28 February 2023 to raise awareness of the challenges that people living with Wilson's disease face.

During the meeting, patient advocates, healthcare professionals and policymakers identified policy priorities that must be addressed to support people living with Wilson's disease:

1. European institutions should urgently support better coordination of data collection on Wilson's disease.
2. Policymakers must address inequalities in access to best-practice care for people with Wilson's disease across Europe.
3. Health system leaders must raise awareness of Wilson's disease among key groups of healthcare professionals to increase the speed of diagnosis.
4. Policymakers, health system leaders, professional societies and patient associations should work together to address treatment non-adherence, especially among high-risk groups.
5. Policymakers, health system leaders and industry should ensure that cost is not a barrier to people with Wilson's disease accessing the most appropriate treatments.

1. European institutions should urgently support better coordination of data collection on Wilson's disease

The lack of a robust evidence base on Wilson's disease means that clinical decision-making is directed by guidelines that are largely based on expert opinion rather than rigorous randomised clinical trials.[3] Given how rare Wilson's disease is, data pooling and cross-national data-sharing are crucial to ensure the availability of a large data set that can be used to inform robust decisions on treatment and care. Coordinating clinical trial data with real-world evidence (e.g. from prospective registries that follow people with Wilson's disease over extended periods) would create a strong basis on which to develop high-quality guidelines. It would also support healthcare professionals in working closely with each patient to make evidence-based clinical decisions that best meet their needs.

European institutions must do more to support the development, long-term funding and harmonisation of population-based registries for rare diseases, such as Wilson's disease, which are available to everyone involved in rare diseases. They could do this by encouraging the creation of a uniform methodology, which uses Orphanet nomenclature, for all national registries and by funding its implementation. Alternatively, they could increase the use of disease codes to ensure consistency, produce high-quality epidemiological data and mitigate language barriers. The differences in legal frameworks and languages across countries continue to pose challenges, but it is imperative that the coordination of Wilson's disease data collection is rapidly intensified.

2. Policymakers must address inequalities in access to best-practice care for people with Wilson's disease across Europe

Best-practice care requires a multidisciplinary team that provides a range of clinical and social services based on the latest available evidence.[4] Many people can access this care through specific centres of expertise, such as those affiliated with European Reference Networks (ERNs). However, these centres are not evenly distributed throughout Europe, leading to potential inequalities in the level of comprehensive, high-quality care to which a person with Wilson's disease will have access.[5]

Policymakers in countries where there are gaps in care should consider supporting the affiliation of additional centres with the ERNs. This would give the centres real-time access to the ERNs' knowledge-sharing benefits and, ultimately, bolster their ability to provide the same level of high-quality multidisciplinary care across the continent.

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In addition, the ERNs should consider offering learning and training opportunities to non-affiliated centres to support the expansion of multidisciplinary care. Strengthened coordination between the different ERNs relevant to Wilson's disease, such as those responsible for rare liver disease (ERN RARE-LIVER), rare neurological conditions (ERN-RND) and rare metabolic disorders (MetabERN), could improve the delivery of multidisciplinary care for people with Wilson's disease. The European Rare Disease Research Coordination and Support Action consortium could be used to facilitate this level of coordination.

3. Health system leaders must raise awareness of Wilson's disease among key groups of healthcare professionals to increase the speed of diagnosis

Increasing the awareness of Wilson's disease among key groups of healthcare professionals – such as those working in primary care, paediatrics, gastroenterology/hepatology, neurology and psychiatry – is key to tackling diagnostic delays. Delays in diagnosis and the misdiagnosis of Wilson's disease can have a devastating effect.[6] The earlier a person receives the correct diagnosis, the sooner they can start treatment. Prompt treatment can prevent the disease from progressing to the point where the symptoms become severe and potentially debilitating.

For this reason, it should be a priority to improve diagnosis in children, which may be achieved by screening children with liver diseases. As Wilson's disease is genetic, family screening is also an important tool for diagnosis in both children and adults.[1] The potential future use of newborn screening for Wilson's disease could provide an enormous opportunity for early diagnosis, but more research is needed before this can be developed.

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4. Policymakers, health system leaders, professional societies and patient associations should work together to address treatment non-adherence, especially among high-risk groups

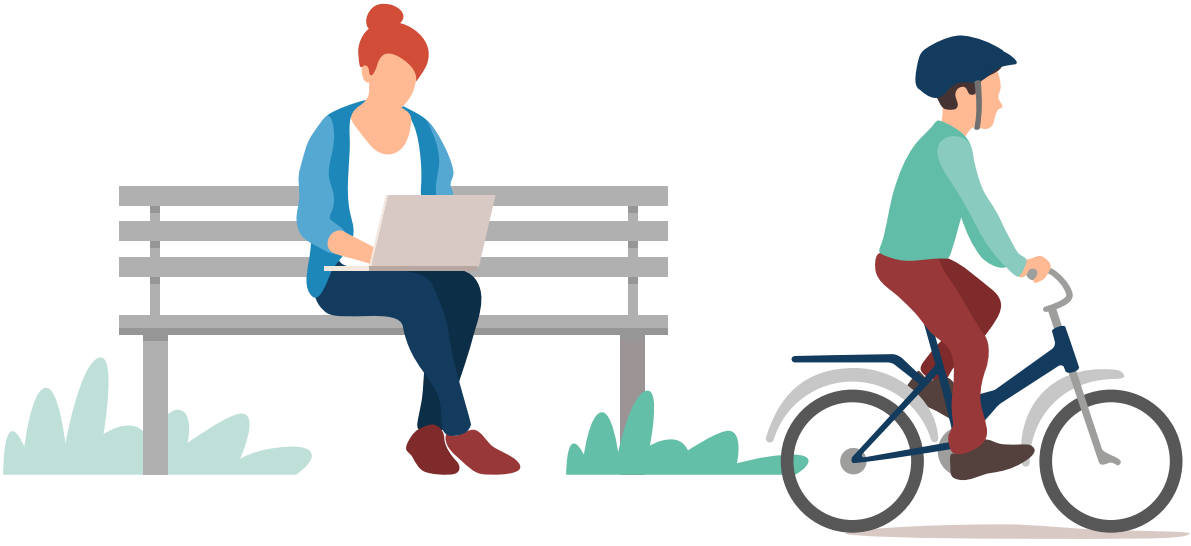
Despite the risk of disease progression and even death, 20–45% of people with Wilson's disease do not follow their treatment regimen for the recommended period.[7] The reasons vary among individuals and by treatment type, but they include treatment side effects, inconvenient dosing, cost and forgetting to take the medication.[1 8-10] As some groups, such as young people, are particularly at risk of non-adherence, education about the dangers to their health may need to be targeted.[2] System changes are also needed, both to enhance the communication between people with Wilson's disease and healthcare professionals and to support more regular and effective monitoring. Improved monitoring can reduce the risk of treatment non-adherence.

5. Policymakers, health system leaders and industry should ensure that cost is not a barrier to people with Wilson's disease accessing the most appropriate treatments

There have been reports of shortages of some medications for Wilson's disease in Europe. In light of this, it is crucial that policymakers, health system leaders and industry work together to secure restriction-free access to the most appropriate treatments available for each person with Wilson's disease, within the constraints of national budgets.

In addition, it is vital that people with Wilson's disease have free access to support services such as speech therapy, physiotherapy and psychological therapy, if needed. Currently, these services are not always covered by national reimbursement. This leads to people paying out of pocket for private sessions, which is not financially possible for everyone. A lack of access to these services can lead to the significant worsening of symptoms and a lower quality of life.

The Wilson's Disease Policy Network and ELPA call on all stakeholders across Europe, including national governments, the European Parliament and the European Commission, to recognise the unmet needs of people living with Wilson's disease and take the priority actions outlined in this action statement to address them.



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