

Addressing Challenges and Improving Outcomes for Wolfram Syndrome Patients International Wolfram Syndrome Consortium

A. Action Items in 2023

1. Develop Clinical Guidelines for Wolfram Syndrome Management

- Adapt existing format for rare diseases and finalize guidelines.
- Publish a downloadable version on patient organizations' websites.
- Incorporate a gene panel-based screening strategy.
- Collaborate with patient organizations to disseminate guidelines to targeted specialists.

2. Compile a List of International Clinical Trial Sites for Wolfram Syndrome

- Identify motivated and experienced physicians to conduct trials.
- Ensure a diverse and representative selection of trial sites.

3. Explore Novel Biomarkers for Wolfram Syndrome

- Conduct research to identify new biomarkers related to disease progression.
- Implement blood draw protocols at multiple centers every six months to monitor biomarker levels.

4. Connect Patient Registries

- Develop a plan to connect existing patient registries for Wolfram syndrome.
- Collaborate with patient organizations to gather and share patient data.

5. Create an Innovative Clinical Trial Protocol for Novel Therapies, including Gene Therapy

- Develop a clinical trial protocol for novel therapies, including gene therapy.
- Identify potential partners for collaboration on the clinical trial.

B. Action Items in 2009

1. Patient registry
2. Longitudinal study
3. Biomarkers
4. Service clinic (multi-disciplinary)
5. Clinical guidelines
6. Animal and Cell models
7. Drug targets/Drug development
8. International Collaboration
9. Share data and reagents
10. Cure

C. Three pillars

1. Improving Patient Outcomes:

To enhance patient outcomes for those with Wolfram syndrome, it is crucial to develop comprehensive clinical guidelines that address early diagnosis, treatment options, and interventions for improving quality of life (QOL). This entails standardizing diagnostic criteria, establishing best practices for management, and exploring innovative therapies such as gene

therapy and stem cell treatments. Additionally, promoting multidisciplinary care and offering psychological support can significantly improve patients' QOL.

2. Engaging the Medical Community:

Raising awareness and fostering collaboration among the broader medical community is vital in tackling Wolfram syndrome. This can be achieved by hosting conferences, workshops, and webinars that provide up-to-date information and promote the exchange of knowledge. Encouraging research and funding opportunities, as well as facilitating collaborations between clinicians, researchers, and patient advocacy groups, can contribute to advancements in understanding and treating this rare disease.

3. Advocating for Public Policy Changes:

To benefit Wolfram syndrome patients, advocacy for public policy changes should focus on improving access to care, promoting research funding, and supporting affected families. This may include pushing for insurance coverage of specialized treatments, establishing centers of excellence, and implementing early screening programs. Working with policymakers and leveraging the power of patient advocacy groups can help in effecting these changes, ultimately benefiting those living with this rare condition.