



# The Wilhelm Foundation

## Organisational Strategy and Impact Objectives

September 2024

<https://wilhelmfoundation.org>





**The Wilhelm Foundation** is a Swedish-based international organisation leading the charge in fostering collaborations among researchers, People Living With an Undiagnosed Disease (PLWUDs) and healthcare providers to find answers for individuals and families affected by undiagnosed diseases.

Wilhelm's founders, Helene and Mikk, lost three children to an undiagnosed disease which sparked an unwavering commitment to supporting anyone, anywhere living with these conditions, and in creating a supportive ecosystem that leaves no one behind. Their vision centres on creating impact where groundbreaking research, empowerment of PLWUDs, diagnosis, and the promise of medical advancements converge.



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## Foreword by Helene

As I reflect on the 25-year journey of the Wilhelm Foundation, I am filled with immense pride and gratitude. This milestone marks not just a celebration of our achievements but a testament to the unwavering determination, commitment and passion that has driven us from the very beginning. Our Foundation was born out of a deeply personal tragedy; losing three of our four children, Wilhelm, Emma, and Hugo, to an undiagnosed disease. This profound loss ignited a relentless pursuit to ensure that no family would have to endure the same pain and uncertainty.

Over the past quarter-century, the Wilhelm Foundation has grown from a small, dedicated team into an internationally recognised leader in the fight against undiagnosed diseases. We have fostered collaborations among researchers, healthcare providers, and communities of PLWUDs across the globe, striving to bring answers to those who need them most. Our work has been guided by the principles of innovation, collaboration, and compassion, and these values will continue to light our path forward.

As we stand at this pivotal moment in our history, we are not merely looking back at our legacy, but more importantly, we are looking ahead. Our new strategy, outlined in this document, is a bold vision for the future – one that builds on our past successes and addresses the evolving challenges of the undiagnosed community. It is a plan rooted in sustainability and growth, designed to expand our impact and reach even further.



Our mission remains clear: to end the diagnostic odyssey for PLWUDs everywhere, with a special focus on low- and middle-income countries. We will achieve this by driving international collaboration, fostering innovation, and empowering PLWUDs and their families. Our vision of a world without undiagnosed diseases is not a distant dream but a tangible goal, and we are committed to making it a reality.

Looking forward, we will continue to champion initiatives like the Global Undiagnosed Day and the Undiagnosed Hackathon, which have already begun to revolutionise the way we approach these medical mysteries. As co-founder and permanent Board member of the Undiagnosed Diseases Network International (UDNI), we will continue to strengthen our partnerships and seek new ones, always with the aim of breaking down silos and fostering a global network of support and knowledge.

Our future is bright, and our resolve is stronger than ever. We are poised to make even greater strides in the coming years, driven by the same spirit that has sustained us for the past 25. Together, we will turn our vision of reducing the number of adults and children who remain undiagnosed after comprehensive investigations from **60% to 0%** into a reality. This journey is far from over, and I am confident that with your support, we will continue to make a profound difference in the lives of those affected by undiagnosed diseases.

Thank you for being a part of this remarkable journey.

**Here's to the next 25 years and beyond.**

With heartfelt gratitude,



**Helene Cederroth**

**President and Founder, The Wilhelm Foundation**



## New Mission, Vision and Values

This year, the Wilhelm Foundation undertook the task of articulating a new mission and vision for the organisation, a decision driven by the marking of our 25th anniversary.

As we revisited our history, celebrated important milestones, and reflected on our legacy, we were also able to have a better understanding of our impact, and a clearer view of future possibilities. Our renewed focus deeply aligns with our core values and the evolving needs of our undiagnosed community, setting an inspiring course for the years to come.

### MISSION

**At the Wilhelm Foundation, our mission is to end the diagnostic odyssey for PLWUDs everywhere, focusing especially on low- and middle-income countries (LMICs).**

### VISION

**Our vision at the Wilhelm Foundation is a world without undiagnosed diseases. A future where families everywhere find timely answers, support, and effective treatments.**

### Our Core Values



## The Current Undiagnosed Environment

“THE WILHELM FOUNDATION is a shining example of what an advocacy group can achieve. Dedicated to the investigation and care of individuals with rare and undiagnosed diseases, the Wilhelm Foundation will expand its efforts and influence to inspire physicians and scientists to pursue medical diagnoses and catalyse genetic discoveries. This is, and will continue to be, of enormous benefit to the world’s undiagnosed community.”

**WILLIAM GAHL**

CO-FOUNDER OF UNDIAGNOSED DISEASES NETWORK INTERNATIONAL  
MEMBER, NATIONAL HUMAN GENOME RESEARCH INSTITUTE

Around 350 million people worldwide live with an undiagnosed disease<sup>1</sup>. This presents an enormous public health challenge and imposes a substantial burden on PLWUDs and their families, and healthcare professionals worldwide. Despite advances in medicine, an alarming 60% of PLWUDs remain without answers even after exhaustive investigations, including whole-genome testing.

This gruelling diagnostic odyssey can drag on for decades, leading to severe and devastating health issues, along with crushing emotional, financial, and social burdens. PLWUDs often face stigma, isolation, and uncertainty, coupled with delays in accessing appropriate therapies – a situation made worse by a lack of awareness and limited access to advanced genomic diagnostic technologies, especially in LMICs.



Timely and accurate diagnoses are lifesaving. They can significantly improve patient care by providing a clearer understanding of potential symptoms or complications and guiding early decisions on management options, including targeted therapies, cancer risk surveillance, genetic counselling, and prenatal diagnosis.

The rapid advancement of precision therapies for previously untreatable rare diseases highlights the critical necessity for accurate diagnoses. Furthermore, receiving a diagnosis can offer immense relief to PLWUDs and their caregivers, shedding light on the condition and enabling participation in patient organisations. It also opens doors for further diagnoses, potentially transforming lives and alleviating the overwhelming burdens these individuals face.

## Our Strategic Goals and Ambitions

“THE WILHELM FOUNDATION truly champions global efforts in the field of rare and undiagnosed diseases. I feel privileged and honoured to be part of this big family and contribute to ending the diagnostic odyssey for patients and their families. Collaboration across borders – this is our vision to solve the unsolved, to diagnose the undiagnosed!”

**TINATIN TKEMALADZE**

PROFESSOR AT TBILISI STATE MEDICAL UNIVERSITY, GEORGIA



**From 60% towards 0%** – we must act on and reduce the 60% of PLWUDs who remain undiagnosed even after comprehensive investigations including whole-genome testing.

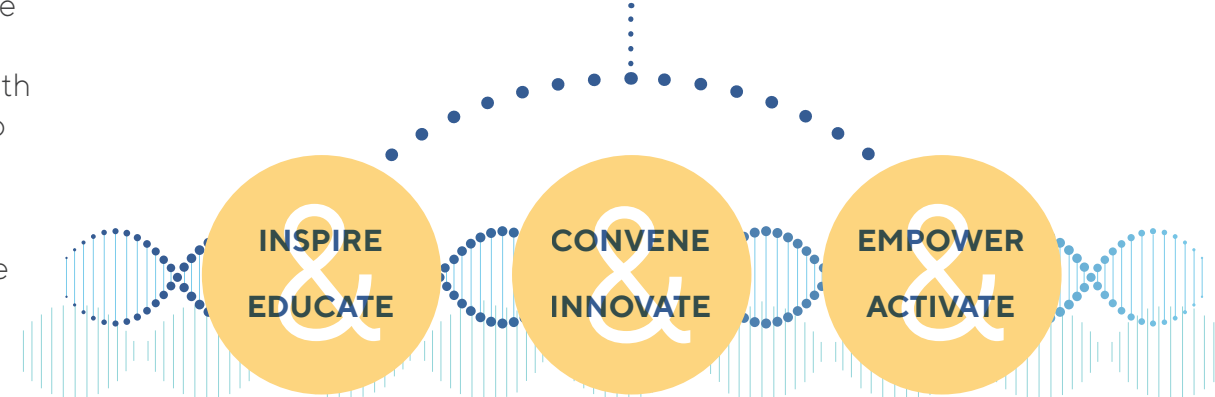
Additionally, there is still much important work to be done to ensure that the remaining 40% are able to get a diagnosis with the available solutions that we have today<sup>2</sup>. That includes improving awareness around, accessibility to and reduction of the costs of genetic testing. Further, we need health systems with good infrastructure, including recognised standards and scaled expertise, to support the processing, delivery and analysis of these tests.

In order to create meaningful impact, it is therefore vital that the Wilhelm Foundation focuses resources and efforts on specific activities that unite the community, provides education to key stakeholders and enhances much needed connectivity across health systems.

The central, transformative promise of the 2030 Agenda for Sustainable Development – to leave no one behind – fuels the work of the Wilhelm Foundation in addressing urgent needs for undiagnosed diseases in LMICs.

Using this ‘north star’ we have developed three, interconnected, strategic goals that will now shape all of the work carried out by the Wilhelm Foundation into the future.

### LEAVE NO ONE BEHIND



<sup>2</sup> Stranneheim, H., Lagerstedt-Robinson, K., Magnusson, M. et al. Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. *Genome Med* 13, 40 (2021). <https://doi.org/10.1186/s13073-021-00855-5>



## Our Strategic Goals and Ambitions:

### INSPIRE & EDUCATE

This strategic imperative will ensure that the work of the Wilhelm Foundation unites the community, bringing it together to raise awareness of the diagnostic odyssey that is experienced by children, adults and their families. This imperative will also guide us in addressing the most urgent unmet needs across undiagnosed diseases; educating the experts and providing healthcare professionals with more knowledge on how to tackle such needs will be central to our strategy.

#### EXAMPLES OF HOW WE DO THIS:

##### Global Undiagnosed Day

Global Undiagnosed Day is marked on the 29th of April to celebrate PLWUDs and their families, and to remember the ones we have lost.

The Undiagnosed Day campaign serves as an international rallying point for advocacy, giving a voice to this often-overlooked community. Every year the Wilhelm Foundation coordinates this global effort, bringing together healthcare professionals, researchers, policymakers, patient advocacy groups, and the public. The result is the creation of a global movement dedicated to unravelling the mysteries of undiagnosed diseases and improving the lives of those affected.

##### The Undiagnosed Photo Project

A global photography project collaboration with award winning photographer, Rick Guidotti, from Positive Exposure, to show the beauty and diversity of those affected by undiagnosed disease, with the aim of effecting change in societal attitudes towards individuals living with genetic, physical, behavioural or intellectual differences.



## Our Strategic Goals and Ambitions:

### CONVENE & INNOVATE

This strategic imperative will ensure that the work of the Wilhelm Foundation proves to be pivotal in fostering dynamic international partnerships between key international stakeholders. We must break down siloed thinking when it comes to creating meaningful impact in undiagnosed diseases. By bringing together healthcare professionals, policymakers, non-governmental organisations, PLWUDs and civil society together we can drive innovative thinking and foster an environment for collaboration.

#### EXAMPLES OF HOW WE DO THIS:

##### **Undiagnosed Diseases Network International (UDNI)**

The Wilhelm Foundation co-founded UDNI in 2014 with the aim of engaging a growing number of research and Undiagnosed Diseases Programs all over the world to collaborate, share and debate data, while fostering the participation of PLWUDs. Since 2014, the UDNI has organised 10+ international conferences to present achievements and continue discussing solutions for the many challenges in the field. The Wilhelm Foundation remains active in the UDNI network and its workstreams, ensuring - whenever possible - collaboration and integration with its own projects.

##### **Undiagnosed Hackathon**

The Undiagnosed Hackathon (UH) is an annual initiative launched by the Wilhelm Foundation, that brings together PLWUDs, medical professionals, researchers, geneticists, data scientists, industry representatives and AI experts to collaboratively analyse undiagnosed cases and attempt to identify diagnoses or insights that could lead to diagnoses but also innovative ways to tackling the issue at hand.



## Our Strategic Goals and Ambitions:

### CONVENE & INNOVATE: Undiagnosed Hackathon - *continued*

Participants from all over the world form multidisciplinary teams and spend an intensive 48 hours diving into patient data including medical records, newly generated genomic data and reports, applying analytical methods and innovative approaches with the goal of cracking previously unsolved medical mysteries.

The event is founded on the idea that innovation thrives at the intersection of different disciplines, with people from different backgrounds that transcend organisations and country boundaries. Through a cross-pollination of ideas, the event sparks creative solutions which may not emerge within traditional silos.

The first UH was hosted by the Wilhelm Foundation in collaboration with the Karolinska Institute, Phenotips, and the Undiagnosed Diseases Network International. It took place in 2023 in Sweden, where 4 out of 11 families received diagnoses.

The second UH was organised by the Wilhelm Foundation in collaboration with Radboud University Medical Center and the Undiagnosed Diseases Network International. It took place in June, 2024 in the Netherlands, and 10 out of 26 families received diagnoses within 48 hours.

The UH highlights what is possible to achieve when silos are broken, new data is generated and shared globally, and when there are meaningful partnerships with PLWUDs. Also, it shows us the transformative potential of leveraging advanced technologies such as long-read RNA and DNA sequencing and Optical Genome Mapping, which enable deeper genetic analysis, overcoming limitations of traditional short-read sequencing and helping identify structural variations often missed by other methods.

In summary, this event is proving critical to reaching our strategic goals with the community because it is helping to find new solutions for the 60% of PLWUDs who remain undiagnosed even after comprehensive investigations including whole-genome testing. It is these new solutions and new ways of thinking that will create the change we need.



## Our Strategic Goals and Ambitions:

### EMPOWER & ACTIVATE

This strategic imperative will ensure the work of the Wilhelm Foundation provides a catalyst for action. Words and promises are not enough, we must act to empower all stakeholders in the undiagnosed diseases community so that we enable and sustain connectivity, promote best practice in action, share positive experiences and stay close to the community we serve.

#### EXAMPLES OF HOW WE DO THIS:

##### Champions Initiative

Scaling the diagnosis of PLWUDs in LMICs presents a unique set of challenges, including limited resources, infrastructure, stigma and awareness. With that in mind, the Wilhelm Foundation has been promoting a strategic and collaborative approach to enhance diagnostic capabilities and patient outcomes in the Global South and for Diversity, Equity and Inclusion (DE&I).

Examples include:

- **Pakistan:** covered costs of training of a genetic counsellor for 1 year.
- **Congo:** facilitated experience exchange programme in Harvard, USA, of two Congolese healthcare professionals. Supported the creation of a Rare Disease Center in Kinshasa with provision of \$12,500 of support.
- **Undiagnosed Hackathon and UDNI Congresses:** provision of funds to ensure attendance of a number of participants from LMICs.

##### Community engagement

At the Wilhelm Foundation, we believe in the mandate of understanding, listening to, and actively engaging with the individuals and families at the heart of our mission. As such the principle of community engagement runs as a red thread throughout all of our initiatives including: Undiagnosed Hackathon, Undiagnosed Photo Project, Facebook, Zoom calls, a Swedish Parents Support Group incl. Family Camps, and keeps us close to the community we serve.



## 25 Years of Milestones: Our Impact Timeline

“*THE WILHELM FOUNDATION is the essential and vital nucleus around which the various components of undiagnosed diseases aggregate with mutual trust, enthusiasm and generosity: experts, young scientists, physicians, patients, families, caregivers and patient associations.*”

**DOMENICA TARUSCIO**

FORMER DIRECTOR OF THE NATIONAL CENTRE FOR RARE DISEASES  
ISTITUTO SUPERIORE DI SANITÀ, ITALY

EVER SINCE The Wilhelm Foundation was founded 25 years ago, we have maintained a focus on creating impact, fostering collaboration and driving innovation. Working with our community, adults and children living with undiagnosed diseases, and their families we are proud to look back at **25 years of impact...**



**1999** - Wilhelm (*left*) passes away, the Wilhelm Foundation is founded

**2001** - Our first key fundraising events, including a concert in Össeby-Gärns church

**2003** - First *Silver-Lining* trips internationally for families impacted by undiagnosed diseases and we start to raise awareness of the Wilhelm Foundation beyond Sweden

**2005** - Wilhelm Foundation receives endorsement from Professor Emeritus Bengt Hagberg and other Swedish professors

**2007** - Further endorsements from the scientific community with help and support from specialists

**2009** - Wilhelm Foundation launches social media channels

**2011** - We launch the English language website. More *Silver Linings* given to families impacted by undiagnosed diseases, including a trip to Disneyland

**2013** - Wilhelm Foundation celebrates the first Undiagnosed Children's Day in April. First meeting with William A. Gahl, Director Undiagnosed Diseases Program (UDP) NHGRI, National Institutes of Health

**2000** - Emma (*centre*) passes away, the Wilhelm Foundation obtains and achieves 90-account equivalent to 501(c)(3) and the fund is controlled by the Swedish Fundraising Control

**2002** - Hugo (*right*) passes away. Driving awareness and telling the story of the Wilhelm Foundation through the largest circulating Swedish national newspapers. Further fundraising initiatives

**2004** - Scaling up of Wilhelm Foundation's awareness and fundraising initiatives including our first *postcards* campaign

**2006** - Establishing Ambassadors and Business Friends

**2008** - Further drive for fundraising and awareness through the media and a further *postcards* campaign

**2010** - Significant publicity opportunity as Wilhelm Foundation appears on Malou von Sivers Show (the equivalent of Oprah Winfrey in Sweden) - more and more families start contacting us for help and guidance

**2012** - We start to scale our work internationally and become a member of EURORDIS



## IMPACT MILESTONES

**2014** - First International Conference on Rare and Undiagnosed Diseases - Rome. Wilhelm Foundation invited to the Vatican in Rome to meet the Pope's medical advisor. Presentation at satellite meeting at the European Society of Human Genetics (ESHG)

**2016** - Assess expansion of UDP and attending 4th International Conference on Rare and Undiagnosed Diseases in Tokyo, Japan

**2018** - Wilhelm Foundation receives EURORDIS Black Pearl Award celebrating outstanding achievements for PLWUDs

**2020** - Awarded Swedish Care Hero. We co-author three medical publications

**2022** - We launch our first *Champions Initiative* with LMICs

**2024** - Our events reach a whole new level.

- **Second international Undiagnosed Hackathon** - Nijmegen, The Netherlands
- **Undiagnosed Photo Project Exhibition** - New York City, USA
- **Undiagnosed Day** - April 29th, we hold an in-person (hybrid) meeting with Harvard and included parallel track international talks and presentations.

**2015** - Undiagnosed Diseases Network International (UDNI) founded. International connectivity continues to enhance at 2nd International Conference on Rare and Undiagnosed Diseases in Hungary and Wilhelm Foundation attends a two-day workshop in Perth, Australia with the aim of starting The Undiagnosed Diseases Program (UDP)

**2017** - Rare Diseases International meeting with the World Health Organization in Geneva. First Undiagnosed Photo Project Exhibition with families impacted by undiagnosed diseases.

**2019** - Attend United Nations headquarters meeting in New York with NGO Committee for Rare Diseases. First partnership with photographer Rick Guidotti, Positive Exposure, to launch a global photography project, *The Undiagnosed International*, in India

**2021** - Achieve Chan Zuckerberg Initiative *Rare as One* partner status

**2023** - Proudly launch our first international *Undiagnosed Hackathon* with attendance from 95 participants from 28 countries and six continents



## Partnerships

“IT IS A HUGE CHALLENGE to serve the most vulnerable families with rare diseases on the most underserved continent, but it is also my greatest privilege! We partner with the Wilhelm Foundation because they embody the values we strive towards on a daily basis: equity, collaboration, and genuine care in action.

With the help of Helene, Mikk and the Wilhelm Foundation, so many of the world's most neglected children have a chance to not only find a diagnosis, but access to a community around the Wilhelm Foundation which aims to improve every aspect of their lives. We are eagerly anticipating the decadal plan of the Foundation, and hope to play an impactful part in the activities to benefit the undiagnosed rare disease community in Africa, and worldwide.”

**PROFESSOR SHAHIDA MOOSA**

ASSOCIATE PROFESSOR OF MEDICAL GENETICS (SU)  
AND HEAD OF MEDICAL GENETICS (TYGERBERG HOSPITAL), SOUTH AFRICA



## Key Partners

The Wilhelm Foundation strategically partners with important actors in the field of health/science to accelerate its mission and goals.

Collaboration is one of our core values and is critical in tackling undiagnosed diseases through harnessing collective expertise, resources, and networks of various organisations, driving innovative solutions and comprehensive support strategies. Through working together we can amplify our impact, improve

access to diagnostic technologies, and provide holistic care and advocacy for those affected by undiagnosed diseases.

The Wilhelm Foundation partners with a growing number of groups around the world, including the National Institutes of Health (NIH), Karolinska Institutet, Illumina, the Chan Zuckerberg Initiative, and European Reference Networks (ERNs), to amplify our impact, break down silos and pave the way for innovative solutions that bring diagnosis to our families across the world.



**DELL MEDICAL SCHOOL:**  
Center For Rare Disease,  
University Of Texas At Austin



**INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM (IRDIRC)**



**CHAN ZUCKERBERG INITIATIVE**



**ERN ITHACA:**  
European Reference Network For Rare Malformation Syndromes, Intellectual And Other Neurodevelopmental Disorders



**ERN JARDIN PROJECT:**  
The Joint Action On Integration Of Erns Into National Healthcare Systems



**EURORDIS:**  
Mental Health & Wellbeing Working Group



**KAROLINSKA INSTITUTE:**  
Swedish Diagnostic Research Project



**NATIONAL INSTITUTES OF HEALTH**



**RARE CARE AUSTRALIA**



**RARE DISEASE DAY**



**RARE DISEASES INTERNATIONAL:**  
Mapping Rare Initiative



## Impact Ahead - the next three years

“*IMAGINE A WORLD without the uncertainty and isolation of being undiagnosed. Imagine what that would do for 100s of millions who are undiagnosed on our planet. Imagine connecting brilliant hearts and minds across our planet to solve the world’s most challenging medical mysteries. Imagine what we need to do to close the remaining diagnosis gap. Well, this is what the Wilhelm Foundation has done through driving this agenda and the action forward.*

*10 years ago, the Wilhelm Foundation started their remarkable journey of networking partners across the globe and supporting them to address the global undiagnosed disease emergency. A truly global network of the world’s leading doctors, scientists and institutions; and most importantly connected and empowered people living with an undiagnosed disease. Just imagine what the Wilhelm Foundation will do over the next 10 years. Just imagine how you can help... and then act together with the remarkable Wilhelm Foundation! ”*

**GARETH BAYNAM**

MEDICAL DIRECTOR AT RARE CARE CENTER

PROGRAM DIRECTOR AT UNDIAGNOSED DISEASE PROGRAM, WESTERN AUSTRALIA



## Impact Ahead - the next three years

Through the publication of this three-year strategy, we now have a clear framework for how we continue to make impact, foster collaboration and drive innovation on behalf of the undiagnosed diseases community.

This has also shaped a series of priorities for action over the coming three years as we take a longer-term view of the challenges that lie ahead.

### The Wilhelm Foundation will:

**WORK TIRELESSLY** to “close the gap” from **60% towards 0%** of PLWUDs who remain undiagnosed.

- o In the short term this means catalysing and engaging in new research and Undiagnosed Diseases Programs and continuing to promote the active participation of PLWUD.
- o Further, we will drive innovative thinking and foster an environment for collaboration in undiagnosed diseases through initiatives such as our Undiagnosed Hackathon, which we hope to expand as we welcome more participants each year and develop new tools, outputs and global data sharing.
- o Finally, we will leverage the UDNI conferences as well as other global and national meetings to explore new ways of tackling the current barriers.

**SCALE DIAGNOSIS** in LMICs to enhance diagnostic capabilities and patient outcomes through continuing efforts such as the Champions Initiative and experience exchange programmes.

- o In the short term this means welcoming more Champions to facilitate discussion, knowledge exchange and exploration of opportunities to boost the diagnostic ecosystem.

- o We will also continue to promote and find ways to support the attendance of experts from LMICs at international conferences and meetings, as well as ensure some of our own efforts take place in the Global South to address the current visa and passport inequities which pose as a significant barrier to global health education, practice and participation.

**COLLABORATE** with the undiagnosed diseases communities and key partners to ensure the diagnostic odyssey is prioritised and acted upon in rare diseases.

- o In the short term we aim to increase the number of organisations we partner with to deliver our strategic ambitions.
- o We will also support and promote successes of our collaborators via our social media channels and conferences.

**ENCOURAGE AND ENABLE** governments and policymakers from more parts of the world to take action on reducing the burden of undiagnosed diseases on children and adults.

- o In the short term, this means working with international partners such as Rare Diseases International, EURORDIS, IRDIRC and other umbrella initiatives to ensure that the priorities of PLWUD feature prominently in international, regional and national commitments.

**INSPIRE AND EMPOWER** PLWUDs and their families to raise their voice in a growing chorus around the world.

- o In the short term this means involving more countries and partners in yearly awareness raising events such as the Global Undiagnosed Day and the Undiagnosed Photo Project.
- o We will also continue our national efforts, always putting PLWUDs at the centre of all that we do.

## Being Accountable - Financial Statement

### **Accountability is vitally important to the Wilhelm Foundation.**

The Foundation ensures that when we invest in undertaking a programme of work, we also invest in evaluating the success of these programmes in achieving the desired outcomes.

Since its inception in 1999 Helene and Mikk Cederroth, with the help of volunteers, have worked tirelessly with an unwavering commitment to using resources and the funds raised effectively and efficiently.

The Wilhelm Foundation has raised and secured restricted funds to run its programmatic activities and events with its partners, leaving a small operational surplus in order to build up a level of sustainable reserves where needed.

We will continue to carefully manage our finances and carry out fundraising efforts aimed at supporting our three strategic pillars. We hope through the publication of this strategic framework we can generate and secure income to help drive our future ambitions for impact.





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