9 yrs old Shakul with Wardeenburg Syndrome, a rare condition affecting his eyes and hearing

Seeing the Unseen: A Global Call to Action on Rare Eye Diseases

This is a *case for action* on rare eye diseases designed to inform and mobilise health advocates and policy makers to prioritise rare eye diseases in the health systems. The case for action includes 2 sections- *Policy* section which includes policy opportunity, overview on why eye health matters, five key policy recommendations; and *Case* section which includes two lived experiences of person living with rare eye diseases and two country examples on national provision on rare eye diseases.

We would like to thank Rare Diseases International for their valuable review and expert input into the development of the advocacy brief.

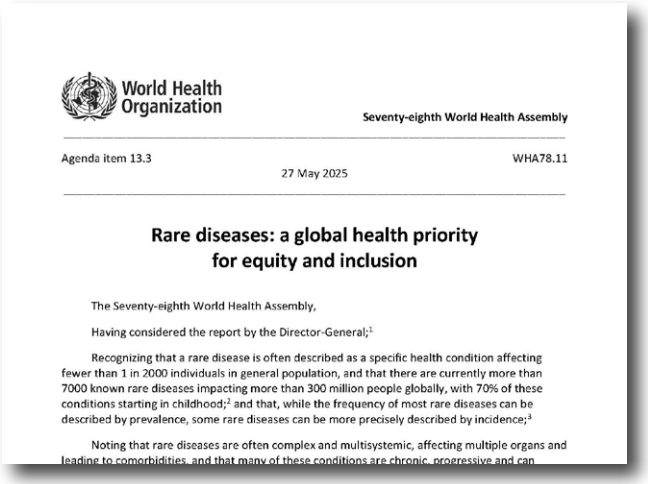
Section 1:

Policy Priorities for Rare Eye Diseases

Introduction

A rare disease is described as a specific health condition affecting 1 in 2000 individuals or fewer in general population. There are currently over 7000 known rare diseases impacting more than 300 million globally.[1] Most of the conditions start in childhood and are chronic, progressive, and can consequently result in disabilities and premature death. People living with rare diseases, in addition to the impact of the condition, face discriminatory behaviours and various barriers. This poses significant challenges to family, education, employment and well-being.

Recognising this, the WHO member states adopted a historic resolution on 'Rare Diseases: as a Global Health Priority for Equity and Inclusion' at the 78th World Health Assembly (WHA78.11) in May 2025. The resolution calls for urgent national and global action including strengthening health systems focused on removing barriers, ensuring timely diagnosis and treatment, and providing access to a wide range of affordable, high-quality services for people living with rare diseases. WHO is mandated to deliver a 10-year Global Action Plan on rare diseases by 2028.



World Health Organization

Seventy-eighth World Health Assembly

Agenda item 13.3 — WHA78.11

27 May 2025

Rare diseases: a global health priority for equity and inclusion

The Seventy-eighth World Health Assembly,

Having considered the report by the Director-General;[1]

Recognizing that a rare disease is often described as a specific health condition affecting fewer than 1 in 2000 individuals in general population, and that there are currently more than 7000 known rare diseases impacting more than 300 million people globally, with 70% of these conditions starting in childhood;[2] and that, while the frequency of most rare diseases can be described by prevalence, some rare diseases can be more precisely described by incidence;[3]

Noting that rare diseases are often complex and multisystemic, affecting multiple organs and leading to comorbidities, and that many of these conditions are chronic, progressive and can

Why Eye Health Matters in the Rare Diseases Agenda

- Over 900 rare eye diseases have been identified, ranging from retinitis pigmentosa, Graves' disease, Thyroid Eye Disease, Leber congenital amaurosis to Stargardt disease and inherited optic neuropathies.[2]
- Many of these conditions lead to progressive vision loss or blindness, often beginning in childhood or early adulthood.
- Rare eye diseases are often overlooked in global health agendas and remain underdiagnosed and underfunded. Rare eye diseases are frequently misdiagnosed or undiagnosed due to lack of awareness, limited diagnostic capacity, and weak referral systems especially in low- and middle-income countries.
- Early diagnosis, specialised treatment pathways, low vision rehabilitation, assistive technologies, specific policies to address patients' unmet needs, and inclusive education and employment policies bring significant support to improving quality of life.

This is a pivotal moment to ensure that the specific needs of people living with rare eye diseases are fully integrated into national health strategies and that the global action plan includes explicit commitments to address rare eye diseases.

Policy Recommendations

We urge eye health advocates, policy makers to consider following 5 key policy recommendations:

1. Integrate eye health into national rare diseases strategies

Eye health must be formally recognized as a critical component of national rare disease strategies and policies. Vision loss from rare eye diseases often leads to compounded disability, heightened social exclusion, and increased economic vulnerability. It also places significant pressure on social systems and creates reliance on carers, who in turn may face hardship, including loss of income, personal health impacts, and reduced financial stability.

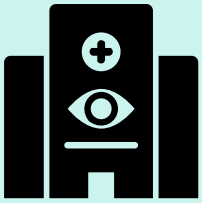
National plans should:

- Explicitly address the prevention, diagnosis, treatment, management, and rehabilitation of rare eye conditions within broader health and social policies.
- Ensure that people living with rare eye diseases are not sidelined in the pursuit of Universal Health Coverage and sustainable development.

2. Raise awareness of rare eye diseases among health professionals, policymakers, and the public

Awareness of rare eye diseases remains low across healthcare systems, leading to misdiagnoses, delayed interventions, and inadequate support. Targeted awareness campaigns are needed to raise awareness and educate:

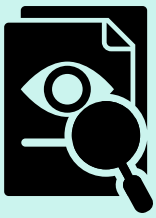
- Healthcare professionals on recognizing symptoms, making timely and appropriate referrals, and understanding the psychosocial impacts of vision loss.
- Policymakers on the impact and burden of rare eye diseases both societally as well as economically, and the importance of investing in inclusive and appropriate health services.
- The public to reduce stigma, promote early health-seeking behaviour, and foster societal inclusion for people living with vision impairment.



3. Strengthen Primary Health Care for early detection and referral

Primary health care systems are the frontline of health services and must be equipped to detect and manage rare eye diseases early. This requires:

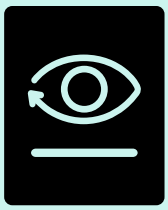
- Training healthcare workers to recognize early signs of rare eye diseases and provide timely referrals to specialists.
- Integrating vision screening for children and adults into routine PHC services.
- Incorporating genetic counselling at the PHC level to inform patients and families about inherited eye conditions and available diagnostic pathways.



4. Invest in research, data collection, and innovation

There is an urgent need to bridge knowledge gaps through:

- Establishing patient registries and strengthening and connecting Centers of Excellence to track prevalence, outcomes, and lived experiences of people with rare eye diseases, while also ensuring that patients in areas with limited healthcare resources can quickly access expert advice.
- Supporting genetic research and clinical trials to develop better diagnostic tools, treatments, and potential cures.
- Prioritize research efforts in low-resource and underrepresented populations, where the burden is often higher, yet data and services are lacking.



5. Ensure equitable access to diagnostics, treatment, and assistive technologies

Equity in eye health means ensuring that everyone regardless of geography or income level has access to the services they need. Governments should:

- Include low vision services, assistive devices, and rehabilitation in national health insurance schemes and benefits packages.
- Facilitate access to affordable diagnostic services, including genetic testing, to enable earlier and more accurate diagnoses.
- Promote availability of assistive technologies, such as magnifiers, screen readers, and mobility aids, that enhance independence and quality of life.

Section 2:

Cases

Lived Experiences: The Human Face of Rare Eye Diseases

Audre's Journey – Lithuania (Stargardt Disease)

At just 10 years old, Audre's world blurred: words faded on pages, and faces became unrecognizable. Forced to leave mainstream education for a school for the blind, she lived for nearly 30 years without a diagnosis. Misdiagnoses, inconclusive explanations, and a lack of genetic testing defined her journey until she was finally diagnosed with Stargardt disease at age 38.

"The constant loss of vision without knowing the cause filled me with panic... We need more education about these conditions not just for patients, but for families and society. Every child or adult deserves an early diagnosis, proper care, and the assurance they will not be left to face it alone."

Her resilience now fuels her advocacy through Lithuania's Retina organization and Blind Association, but her story reflects the uncertainty and invisibility faced by countless others.

"The constant loss of vision without knowing the cause filled me with panic...

Monika's Journey – Austria (Thyroid Eye Disease)

In 2011, Monika was diagnosed with Thyroid Eye disease, an autoimmune thyroid condition that can severely affect the eyes. Initially told little about potential risks, she was unprepared when her eyes began to protrude, double vision set in, and doctors warned she might go blind without immediate surgery. Between 2013 and 2017, she underwent 10 major surgeries on her eyes and face.

"It was terrible to hear the word blind... The doctor said: this is the first and last possible surgery."

Monika lost her job and financial security, while her family endured years of stress and uncertainty. Even in Austria, access to affordable, specialized care was limited. Determined to help others, she founded the Thyroid Eye Disease Patient Association in 2017, now supporting more than 40 people.

"If I had received the right information at the beginning, I would have been less afraid. Now I tell others: take it seriously, avoid stress, and don't delay care."

Her advocacy highlights systemic gaps even in strong health systems and why early information, access, and inclusion matter globally.

Kindly approved by Audre Grybauskaite and Monika Schindler-Schnitzl

Country Examples: Paving the Way

India's commitment to people living with a rare disease

India's National Policy for Rare Diseases 2021 represents a significant step toward addressing the health and social needs of over 70 million people estimated to be living with a rare diseases in the country. The policy categorizes rare diseases based on treatment availability and clinical evidence, covering a wide spectrum of conditions, several of which involve eye and vision-related complications. Complementing this is a hospital-based National Rare Disease Registry, developed by the Indian Council of Medical Research (ICMR) to strengthen data and planning.

The policy promotes awareness about rare disease, all levels of prevention, capacity building of health professionals, and optimal screening and early diagnosis. There are 11 centres of excellences for rare disease across the country, providing a platform for multidisciplinary care, including ophthalmological evaluation, genetic counselling and long term follow up. The policy provisions financial support for treatment and supports crowd funding under the *Rastriya Arogya Nidhi* scheme.

India's policy reflects a strong commitment to equity, access, and system-wide coordination, setting up a model for improving outcomes for people living with rare diseases.

South Korea: Integrating Rare Eye Diseases into National Frameworks

South Korea offers a compelling example of how rare diseases, including those affecting the eye, can be effectively integrated into national health systems.The country has established a strong institutional framework led by the Ministry of Health and Welfare and the Korea Disease Control and Prevention Agency (KDCA), supported by the National Health Insurance Service (NHIS) and a network of designated tertiary hospitals. This system coordinates policy, financing, service delivery, and research for over 1,000 recognized rare and incurable conditions.

The National Rare Disease Registry managed by KDCA currently includes 14 rare eye conditions, such as retinitis pigmentosa, Leber congenital amaurosis, and cone-rod dystrophies. The registry supports surveillance, guides clinical responses, and informs national planning.

While MOHW's policies may not explicitly reference rare eye diseases, their inclusion in the KDCA registry ensures visibility and response. This integrated model with mechanisms for diagnosis, care, and financial support offers a practical framework for elevating rare eye diseases within broader health and rare disease strategies. It reinforces the case for ensuring that rare eye conditions are not overlooked in national and global health agendas.

Looking ahead

Both country examples show that national leadership is possible, but integration of eye health remains limited. The lived experiences echo a global truth: people living with rare eye diseases cannot wait. Policymakers, health leaders, and advocates must act now to ensure that rare eye diseases are seen, recognized, and addressed within universal health coverage and rare diseases policies.

The Global Summit for Eye Health 2026 will be a critical opportunity to spotlight rare eye diseases within the broader momentum initiated by the 78 WHA resolution on Rare Diseases. The summit convening governments, CSOs, private sector, research and academia, people with lived experience, and so on creates a high-level platform to hold governments accountable, reinforce rare eye diseases inclusion in the Global Action Plan, and catalyse sustained national-level advocacy.

GLOBAL SUMMIT FOR EYE HEALTH

IAPB November 2025

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1. Cover image - Photo submitted by Marjin Fidder to the IAPB WSD Photo competition
2. Page 7 - Photo submitted by Julie-Anne Davies to the IAPB WSD Photo competition

Endnotes

1 The Lancet Global Health. The landscape for rare diseases in 2024. Editorial. 10.1016/S2214-109X(24)00056-1.

2 The need for widely available genomic testing in rare eye diseases: an ERN-EYE position statement https://pubmed.ncbi.nlm.nih.gov/33743793/

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