

GRIEF, GAPS, GAINS AND GIVING

ONE PARENT'S JOURNEY FOLLOWING AN USHER SYNDROME DIAGNOSIS

EMILY SHEPARD



What is Usher Syndrome?



What is Usher Syndrome?

**1:6000
individuals
globally**

**1:70
carrier of
Usher**

No cure

**5-9%
children
with hearing
loss**

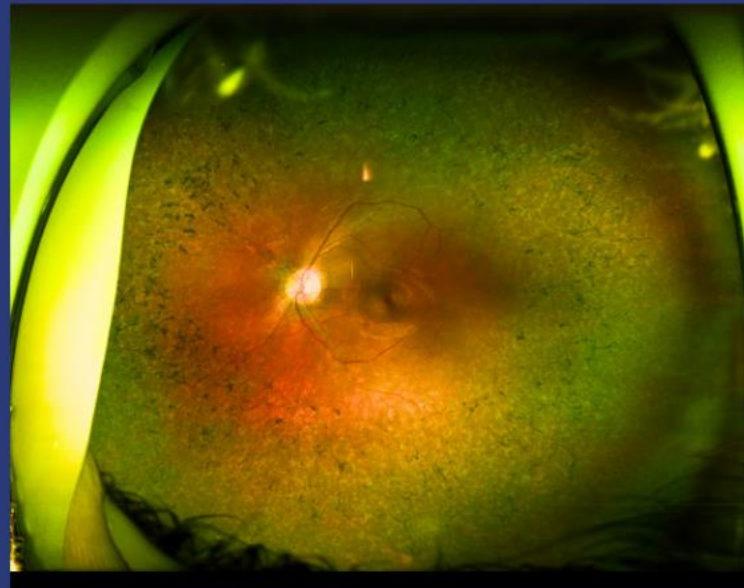
**Autosomal
recessive
inheritance**

**Most common
cause of
genetic
deafblindness**

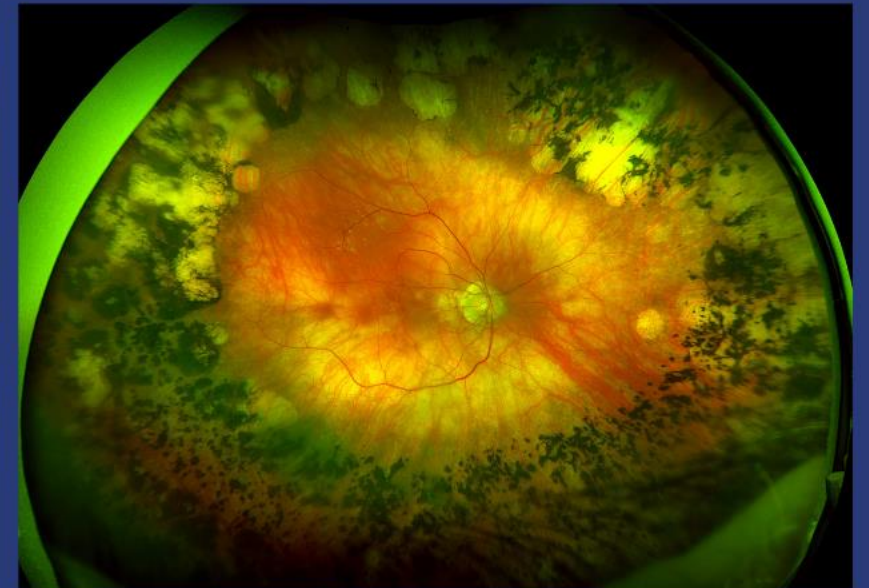
What is Retinitis Pigmentosa?



Healthy retina



Early retinitis pigmentosa



Advanced retinitis pigmentosa

UsherKids Australia

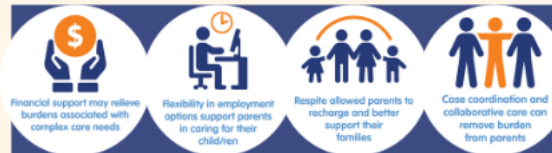
Our purpose is to see children and families living with Usher syndrome love their lives.



Our mission is to empower the Usher community through support, connection and knowledge.

WHAT ARE THE SUPPORT NEEDS OF PARENTS OF YOUNG CHILDREN WITH USHER SYNDROME?

There are substantial gaps in the services and supports provided to parents of children with Usher syndrome. UsherKids Australia led a research study exploring the support needs of parents of children with Usher syndrome. The following four central themes were unveiled.



Financial support may relieve burdens associated with complex care needs. Flexibility in employment options support parents in caring for their child/ren. Respite allowed parents to recharge and better support their families. Case coordination and collaborative care can remove burden from parents.

PRACTICAL NEEDS

Managing Usher syndrome requires coordination among various healthcare professionals, scheduling numerous appointments, and implementing treatment plans. Parents often need to navigate complex healthcare systems and seek funding for therapies. Case coordination and collaborative care can reduce burden from parents.



Parents required information to locate professionals with adequate knowledge and training. Accurate and timely information regarding diagnosis may relieve emotional distress. Parents needed additional information on vestibular dysfunction. Need for various sources of information.

INFORMATIONAL NEEDS

Parents reported dissatisfaction with the level of knowledge among medical professionals regarding Usher syndrome. They felt they are required to educate healthcare providers about the condition and appropriate management strategies.



SOCIAL NEEDS

Parents of children with Usher syndrome face isolation due to the rarity of the condition. They struggle to connect with others facing similar challenges and often feel alone in managing their child's complex needs. They described substantial benefits to connecting with other parents of children with Usher syndrome and identified support groups to be useful in facilitating such connections.

EMOTIONAL NEEDS

Parents experience significant emotional burden upon receiving the diagnosis of Usher syndrome in their child. They face grief, uncertainty about the future, and pressure to engage in early intervention strategies to maximise their child's development potential.



Confronting emotions regarding child's progressive vision loss.

Support required to cope with grief and loss.

Exploring the support needs of Australian parents of young children with Usher syndrome: qualitative thematic analysis



QR link to paper



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the royal victorian eye and ear hospital



CENTRE FOR Eye Research Australia

Awareness of Usher Syndrome and the Need for Multidisciplinary Care: A Cross-Occupational Survey of Allied Health Clinicians

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Background: Usher syndrome is the most common cause of deaf-blindness, affecting up to 1 in 6000 people. Multidisciplinary care is required to maximize outcomes for individuals and families. This study assessed awareness of Usher Syndrome amongst allied health clinicians who provide care related to the primarily affected senses of hearing and vision, ie, optometry, orthoptics and audiology.

Methods: A prospective cross-sectional online survey of clinicians working in Australian university-affiliated clinics (7 optometry, 1 orthoptics and 4 audiology) was completed between September 2021 and January 2022. Questions were asked about the cause, common symptoms, and awareness of health professions who manage Usher syndrome.

Results: The 27 audiologists, 40 optometrists, and 7 orthoptists who completed the survey included 53 females (71.6%), had an average age of 37 years (range 24–70), and had an average duration of clinical experience of 13 years (range 1–45 years). The majority of respondents correctly identified Usher syndrome as a genetic condition (86%), identified at least two of the affected senses (97%), and identified the progressive nature of the vision and hearing losses (>90%). Awareness of vestibular dysfunction and its characteristics was low, as was knowledge of the key treatment roles that speech pathologists, genetic counsellors and geneticists play in the management of Usher Syndrome. The majority of respondents also did not identify important aspects of care within their own discipline.

Conclusion: This study has shown that there is a need for targeted education to be delivered to hearing and vision care allied health clinicians to raise awareness of the vestibular impacts and aspects of vision loss experienced by people with Usher syndrome. This education needs to target the broad range of clinicians who have a key role in providing multidisciplinary care (including speech pathologists, geneticists, and genetic counsellors) and to identify the key aspects of good-quality multidisciplinary care.

Keywords: deaf-blindness, optometry, audiology, orthoptics, genetics, physiotherapy

Usher syndrome is an autosomal recessive genetic condition.¹ It is characterized by abnormal development of hair cells resulting in hearing impairment (Figure 1A), loss of retinal photoreceptor cells resulting in progressive vision loss (retinitis pigmentosa; Figure 1B), and possible vestibular dysfunction which is also due to hair cell abnormalities. Usher syndrome is thought to affect approximately 1 in 6000 individuals globally² and is the leading cause of deaf-blindness worldwide.³

Awareness of Usher Syndrome and the Need for Multidisciplinary Care: A Cross-Occupational Survey of Allied Health Clinicians



QR link
to paper

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PRESENTS

Usher Syndrome Youth Camp

2025



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OPPORTUNITIES**

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ANNUAL REPORT

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