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Viewpoint article - Beyond Clinical Assessments: Advocating for Early Support for Children with Usher Syndrome in Australia

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Abstract

Usher syndrome is a genetic condition characterised by congenital hearing loss and progressive vision loss in childhood to early adolescence, resulting in prospective deafblindness; vestibular dysfunction may also occur. Advances in genomic testing now enable diagnosis well before vision loss is identified, highlighting families who could benefit from early psychosocial, educational, and practical support. However, eligibility for funded early-intervention, disability, and educational services in Australia remains largely tied to clinically measurable impairment. This misalignment creates a paradox: the earlier Usher syndrome is diagnosed, the longer families may wait for support, leaving children not yet deaf or blind ‘enough’ to qualify, despite a confirmed lifelong, multisystem condition.

We argue that the prospective nature of deafblindness in Usher syndrome warrants early, specialised, and transdisciplinary intervention at the point of genetic diagnosis. We outline the risks associated with delayed support, highlight the benefits of early intervention, and propose a proactive model of care integrating deafblind specialists, education systems, genetic services, allied health, and family-centred supports. To ensure equitable and timely care, eligibility frameworks must evolve to reflect contemporary genomic capabilities and the future-focused needs of children with Usher syndrome and their families.

Keywords

Usher syndrome, deafblindness, early intervention, eligibility frameworks, transdisciplinary care

Introduction

Usher syndrome is a rare genetic condition characterised by dual diagnoses of bilateral congenital moderate-to-profound sensorineural hearing loss and progressive vision loss in childhood-to-early adolescence from retinitis pigmentosa (Castiglione & Möller, 2022). Deafblindness associated with Usher syndrome can be considered prospective, with early-onset deafness and later-onset vision loss (Meekins-Doherty et al., 2023). Usher syndrome accounts for approximately half of hereditary deafblindness cases (Castiglione & Möller, 2022), with an estimated prevalence of 4-17 per 100,000 (Kimberling et al., 2010). In some cases, Usher syndrome is also associated with vestibular dysfunction, resulting in difficulties with balance and gross motor development (Cushing & Papsin, 2018). The combined impacts of dual sensory loss are multiplicative, or exponential in nature (Anthony, 2016).

Early and specialised support is therefore critical. Yet, while advances in genomic technologies now allow for early diagnosis before visual symptoms appear, access to educational and disability support varies and usually depends on clinical evidence of impairment. This misalignment leaves families waiting until vision or hearing loss progresses to meet eligibility thresholds. In this article, we argue that for Usher syndrome, prospective deafblindness itself should be sufficient justification for early, specialised, transdisciplinary intervention.

Early diagnosis through genetic testing

Genetic testing now enables early diagnosis of Usher syndrome in infancy, sometimes well before visual symptoms are identified (Brodie et al., 2021). This holds several benefits for clinical management, directing further evaluation, refining genetic counselling, and improving patient outcomes (Brodie et al., 2021). The availability of genetic results may mean the person avoids expensive imaging, alongside decreased radiation and sedation exposure (Alford et al., 2014). As with other genetic syndromes that affect multiple systems, identifying Usher syndrome through genetic testing facilitates the investigation and early management of associated comorbidities (Alford et al., 2014; Urbančič et al., 2020).

Early diagnosis of Usher syndrome brings promise and challenge. Early diagnosis allows families time to prepare for the future by engaging practical and psychosocial support (Johansen et al., 2024). For growing families, this support may take the form of providing reproductive information about the likelihood of Usher syndrome occurring again, and supports them in making informed choices that feel right for their family, including options that may lower this likelihood if they wish (Kuliev et al., 2020). For others, this testing can support participation in leading research in the presymptomatic stage of the condition such as clinical trials (Toms et al., 2020).

However, it appears to be a trend in the current context that systems governing educational and disability support have found it challenging to keep pace with the growing number of individuals now diagnosed prior to the onset of combined hearing and vision impairment. Across many jurisdictions, eligibility for funded services is still tied to clinical evidence of impairment such as audiograms, visual acuity, or visual field measurement thresholds with organisational and bureaucratic barriers that inhibit flexibility, choice, and control for families and supports (Backhouse, 2017; Johansen et al., 2024). Barriers to timely access exist and there are instances where children have been excluded from critical funded support services (Backhouse, 2017; Johansen et al., 2024). Despite a confirmed genetic diagnosis, a lack of sufficiently measurable impairment means they are deemed not yet blind or deaf ‘enough’ to warrant these funded services.

Reliance on measurable impairment creates a troubling paradox: the more successful we are at diagnosing Usher syndrome early, the longer families may be forced to wait for services. This defeats one of the primary purposes for providing a presymptomatic diagnosis. To reconcile this discrepancy, a genetic diagnosis of Usher syndrome should, in itself, justify access to early, specialised, and transdisciplinary deafblind support and intervention. This approach reflects the prospective and progressive nature of deafblindness in Usher syndrome. Families should be able to choose whether, when, and to what extent they are ready and wish to access this support.

Current policy and practice

Internationally, access to early intervention and formal supports for children with deafblindness is highly variable, and clear, contemporary guidelines are limited. Studies have highlighted significant unmet needs, psychological distress and lack of structured support for families (Bodsworth et al.,

2011). Existing guidelines in some regions are outdated. For example, UK guidance for local authorities on supporting deafblind people is now more than a decade old (Department of Health and Social Care, 2014). Even in countries with well-established systems, such as Sweden, families report that supports can be difficult to obtain in practice (Ehn et al., 2019).

In Australia, current approaches to support from educational and disability systems are determined by eligibility criteria based on the presence of current impairment, often based on the outcome of clinical assessments of individual systems (NDIS, 2024; New South Wales Department of Education, 2024). However, this approach ignores the functional or lived experience of the person. For example, despite a diagnosis of Usher syndrome, children may only qualify for vision services once visual acuity falls below a threshold, or when a functional vision assessment demonstrates clear difficulty accessing the curriculum. This approach highlights the disconnect between policy frameworks which are reactive and deficit-based versus advances in genomic medicine which are proactive and predictive. Such a reactive approach can restrict access to critical early intervention, and strategies that can influence quality of life of the person and their family, from the time of diagnosis rather than point of measurable impairment.

The Impact on Families and Children

Without robust eligibility criteria that accurately reflect both current and future needs of people with Usher syndrome, the likelihood of psychosocial impacts escalates. Without true recognition of the individual's needs, they and their family will feel like they are falling through the cracks (Johansen et al., 2024). The benefits of early intervention are well documented and, with genetic testing facilitating an early diagnosis, such intervention can and should occur as early as the family and child are ready and willing. When early intervention is delayed, the child is likely to experience poorer outcomes in domains such as education, developing independence and overall wellbeing (Arcous et al., 2020; Ehn et al., 2018; Ellis & Hodges, 2013), and important safety issues may be unaddressed (Cushing & Papsin, 2018). This delay in early intervention should not be allowed to happen.

Usher syndrome presents a compelling argument for an effective team approach (Ayton et al., 2023; Maxwell et al., 2025). All professionals must understand the family experience, and the impact the family's wellness has on what professionals, and the family, are collectively striving to achieve. Families need access to coordinated, transdisciplinary support which includes genetic counsellors, educators, audiologists, O&M specialists, orthoptists, optometrists, ophthalmologists, psychologists, speech pathologists, occupational therapists and physiotherapists (Meekins-Doherty et al., 2023). This support must be financially equitable and accessible to all; without financial access, people will be left unsupported. With early support, children with Usher syndrome can fully participate in education and social settings, which positively impacts their long-term outcomes, including employability and quality of life (Cmar, 2015; Ehn et al., 2016, 2018).

Further, the diagnosis of Usher syndrome, as with any significant congenital impairment, can have lasting effects on an individual's sense of identity, making access to early intervention particularly critical during adolescence and early adulthood (Couzens et al., 2015; Evans & Baillie, 2022). This is in line with the World Federation of the Deafblind's recommendation for referral to support services as early as possible (World Federation for the Deafblind, 2022). This allows the child to grow knowing that Usher syndrome and its sensory impacts are part of them and their future. This gradual approach or "little bits of information often" is advised when talking to children about Usher syndrome. This aligns with similar approaches of genetic counselling and provision of healthcare information to minors (Metcalfe et al., 2011; Stuttgen et al., 2021). This approach avoids a sudden and potentially confronting disclosure and instead facilitates a gradual, developmentally appropriate integration of the diagnosis and implications for the child and family.

Rethinking Eligibility Criteria Approaches

The current approach to funded support for Usher syndrome which applies a, "wait until there is measurable loss," framework is flawed. We should be asking "why wait"? The benefits of early intervention are known including cost-effectiveness, as early intervention mitigates downstream educational and social costs. We should be looking to other conditions where services and support are successfully offered at the time of diagnosis. For example, early intervention is routinely available to autistic children, and in the case of paediatric oncology psychosocial services are provided at the point of diagnosis (Ehrhardt et al., 2023; Landier et

al., 2004; Trembath et al., 2022). In the case of Usher syndrome, we encourage a rethink of current eligibility criteria and propose the adoption of a prospective diagnostic approach that incorporates genetic and syndromic knowledge. From this we are able to conclude that a functional, anticipatory model of eligibility, which also recognises future trajectory and prospect of deafblindness, rather than relying solely on currently measurable deficits will directly benefit those impacted by Usher syndrome and their families. We can consider this akin to many other genetic conditions where pre-symptomatic diagnosis is performed, in part, for the explicit purpose of informing those affected of risk management strategies to curb the impact of the disease on both the individual and the population (e.g., cardiac disease, cancer etc).

Another critical recommendation we make is to reconsider the way deafness and vision impairment are assessed separately for people with Usher syndrome, this approach is commonly adopted by the National Disability Insurance Scheme (NDIS). In Australia, the term 'imputing disability' has been introduced as part of the National Consistent Collection of Data (NCCD) such that school teams can make a judgement that a student has an undiagnosed disability that has a functional impact on their learning based on their observations (Imputing Disability for the NCCD, 2021). However, children with Usher syndrome are unlikely to be picked up due to their later onset of symptoms related to retinitis pigmentosa, with marked vision deterioration. This does a significant disservice to people and reflects a poor understanding of the complex impact of dual sensory loss. It is clear such systems do not recognise that combined hearing and vision loss is experienced as greater than the sum of its parts. Unquestionably this approach must change.

Calling for a Transdisciplinary, Proactive Model of Support

Alongside early diagnosis, a transdisciplinary and proactive model of support is recommended. To be effective, this model must be grounded in collaboration between policymakers, clinicians, educators, and advocacy groups. The model must foster:

- Early access to deafblind specialists.
- Education system readiness, which includes specialist education staff with experience in deafness, blindness, and communication.
- Family-centred services, psychosocial support, peer support networks.
- Integration of genetics into rehabilitation pathways, opening access to allied health team members.

To be truly effective, all members of the transdisciplinary team must have awareness of the prospective deafblindness experienced in Usher syndrome and take all opportunities to collaborate (Ayton et al., 2023).

Conclusion

Usher syndrome is a condition that is lifelong and has a significant impact on the person, their family and the life they want. With the right support, the impact of Usher syndrome can be lessened. However, this relies on policy makers within government and organisations providing early intervention, education and disability services to recognise the need for early, specialised support and understand its benefits. It also requires them to be brave to make decisions that may seem to benefit only a small group of children and families, but which ultimately have far-reaching, long-lasting positive impacts well beyond the individual child. They must take the bold step of aligning legislation and funding with contemporary science, to ensure children and families receive timely, appropriate, and equitable support. Now is the time for recognising the current primary reliance, in high income countries, such as Australia, on clinical outcomes is outdated and harmful in the case of Usher syndrome. With genetic testing providing an early diagnosis, we call for a rethink of existing eligibility frameworks. It is also time to recognise the prospective nature of the deafblindness that is associated with Usher syndrome. This call to action not only honours the lived experience of families but also ensures children with Usher syndrome are given the best possible chance to thrive.

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