Syndromic Vestibular Disorders in Children:

Usher, CHARGE, Pendred, Waardenburg, and Beyond

Vestibular Medicine in Children

Topic 8 of 15

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How to Use This Review

This literature review is part of the Vestibular Medicine in Children series published by the Australian Dizziness Clinics Education Hub. It is written for vestibular physicians, paediatricians, and emergency physicians who assess and manage children presenting with vestibular disorders.

The review is designed to be read as a deep-reference resource or used as a clinical desktop companion. It is supported by a clinical cheat sheet, short-form clinician videos, and audio episodes that cover the same material.

Callout Box Guide

☐ **Key Point:** Foundational concepts and summary statements that anchor the core clinical content of each section.

☐ **Clinical Insight:** Clinically relevant observations for direct application in assessment and management.

☐ **Clinical Pearl:** High-yield memorable clinical points — the take-home messages most likely to change practice.

☐ **Important:** Red flags, emergencies, and critical safety points requiring immediate action.

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Syndrome	Gene/Locus	Vestibular features	Associated features
CHARGE	CHD7	Semicircular canal aplasia (70%); bilateral BVH	Coloboma; heart defect; choanal atresia; growth delay; ear anomaly
Usher type 1	MYO7A, CDH23, PCDH15	Bilateral profound BVH	Congenital profound deafness; retinitis pigmentosa from childhood
Usher type 2	USH2A	Mild/moderate vestibular deficit	Moderate-severe SNHL; retinitis pigmentosa from adolescence
Pendred / EVA	SLC26A4	EVA with episodic vestibular dysfunction	Fluctuating SNHL; goitre
Neurofibromatosis 2	NF2	Bilateral acoustic neuromas; progressive BVH	Bilateral SNHL; meningiomas; skin features
Waardenburg	PAX3, MITF	Mild vestibular deficit	Congenital SNHL; heterochromia; white forelock
Treacher Collins	TCOF1	Rare vestibular involvement	Mandibulofacial dysostosis; conductive hearing loss

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Test	CHARGE specific	Usher type 1	General BVH
vHIT (horizontal + vertical)	SCC aplasia → absent VOR all planes	Absent or severely reduced horizontal VOR	Bilateral reduced gain <0.6
cVEMP (saccule)	Absent or absent	Absent	Absent or elevated threshold
oVEMP (utricle)	Absent or absent	Absent	Absent or reduced amplitude
Rotary chair	Absent or severely reduced VOR gain	Absent responses	Bilateral reduced gain
MABC-2 balance subscale	Severely affected even with vision	Very poor; dynamic balance worst	Below 5th percentile typical
Ophthalmology	Not specifically affected	Retinitis pigmentosa from childhood	Examine all BVH for Usher

IV. Waardenburg Syndrome: Melanocyte Pathway and Inner Ear

V. Pendred Syndrome (see also PVM07)

Strategy	Application	Priority
Early VRT (as soon as	Visual and somatosensory	URGENT — neuroplasticity

medically stable)	substitution strategies	window critical
Play-based gaze stabilisation	Appropriate for developmental age; visual tracking games	Start immediately; parent-administered
Somatosensory substitution training	Standing on textured surfaces; proprioceptive feedback	Compensates for BVH + visual loss (Usher)
Swimming and water sports	High vestibular demand in safe environment	Once swimming competency achieved; excellent VRT vehicle
Mobility aids	Cane or rollator for severe BVH + visual loss	Usher type 1: plan for adult life with combined sensory loss
CI post-operative VRT	Vestibular deafferentation after CI surgery in BVH	Start within 48 hours post-operative; critical window

VI. Other Syndromes: Alport, Branchio-Oto-Renal, Treacher Collins, Norrie

Area	Accommodation/strategy
Classroom lighting	Bright, even lighting; avoid flickering; Usher — front-row seating
PE and sport	Individualised plan; avoid contact; aquatic VRT where possible
Orientation and mobility	O&M specialist for combined sensory loss (Usher)
Communication	Sign language + oral; auditory training post-CI
Transition planning	Tertiary education; employment; adult specialist referral at 16–18
Mental health	High burden; psychological support for child + family; peer support

VII. Genetic Testing Strategy in Syndromic Vestibular Disorders

VIII. Multidisciplinary Assessment and Management

Timepoint	Assessment	Action
Diagnosis	vHIT + VEMP; MABC-2; ophthalmology; genetics	Confirm syndromic diagnosis; baseline vestibular function
Every 6 months	vHIT; MABC-2; audiogram	Track progression; update VRT programme
Annual	DVA; ophthalmology (Usher); school function	Retinal screening from age 6 in Usher type 2
Post-CI surgery	vHIT immediately post-op; VRT start within 48 hours	Intensive VRT; prevent oscillopsia from surgical deafferentation
School transitions	Full vestibular and functional assessment	School plan update; new teacher education

IX. Rehabilitation, Education, and Long-Term Outcomes

Indication	Urgency	Refer to
Any genetic syndrome + deafness	Soon	Vestibular physician + audiology + genetics
CHARGE diagnosis	Urgent	Multidisciplinary team: ENT,

		cardiology, ophthalmology, vestibular physician
Usher syndrome diagnosis	Soon	Retinal specialist + vestibular physician + genetics + O&M specialist
BVH confirmed (any syndrome)	Soon	Vestibular physician + VRT physiotherapist immediately
Pre-CI assessment in syndromic hearing loss	Pre-operative	Vestibular physician: baseline BVH + post-surgical VRT plan

X. Summary and Key Clinical Takeaways

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I. Introduction: The Syndromic Vestibular Landscape

Syndromic causes of vestibular dysfunction in children represent a clinically important minority of the broader paediatric vestibular population, but their significance extends well beyond their numerical frequency. These conditions are characterised by vestibular dysfunction occurring in the context of identifiable genetic syndromes affecting multiple organ systems — hearing, vision, the cardiovascular system, the kidney, or the craniofacial skeleton. Accurate recognition of syndromic patterns is the gateway to appropriate genetic testing, multidisciplinary management, and prognosis for the child and family. [1]

The vestibular component of syndromic hearing loss has historically been underappreciated. Until routine vestibular testing became available for children, vestibular dysfunction in conditions such as Usher syndrome Type 1 and CHARGE syndrome was largely inferred from delayed motor milestones rather than directly measured. Current evidence demonstrates that vestibular hypofunction — ranging from subclinical to complete bilateral areflexia — is present in a substantial proportion of children with syndromic hearing loss and has direct implications for motor development, rehabilitation strategy, and cochlear implantation planning. [2,3]

The major syndromic causes of paediatric vestibular loss discussed in this review are: Usher syndrome (types 1–3), CHARGE syndrome, Waardenburg syndrome, Pendred syndrome (covered in depth in PVM07), Alport syndrome, Branchio-Oto-Renal (BOR) syndrome, Norrie syndrome, and selected others including Turner syndrome and Down syndrome. Together, these account for a significant proportion of inherited paediatric vestibular dysfunction. [1,4]

Figure 1. Syndromic Vestibular Disorders — key clinical features identification table across major syndromes.
Source: Australian Dizziness Clinics — clinical flowchart.

□ **Clinical Pearl:** Always examine a child with sensorineural hearing loss for retinal pigmentation, coloboma, white forelock, and dysmorphic features. You may be looking at Usher syndrome, CHARGE syndrome, or Waardenburg syndrome — all of which require specific syndromic workup and multidisciplinary management beyond standard audiological care.

II. Usher Syndrome: Genetics, Hearing, Vision, and Vestibular Features

Usher syndrome is the most common cause of combined hereditary deafness and blindness, affecting approximately 1 in 6,000 to 1 in 25,000 individuals depending on the population studied. It is autosomal recessive and is classified into three clinical types based on the degree of SNHL, the presence or absence of vestibular dysfunction, and the onset and severity of retinitis pigmentosa (RP). [5]

Usher Type 1

Type 1 is the most severe form. It is characterised by profound congenital SNHL, bilateral vestibular areflexia, and RP onset typically by age 10 years. The combination of total deafness, absent vestibular function, and progressive retinal degeneration leads to deafblindness in most affected individuals by early adulthood. Vestibular areflexia — absent caloric responses, absent VEMPs, and flat vHIT gain — is present from birth and is the cause of delayed walking, typically not achieved until age 2–4 years. The wide-based, ataxic gait of Usher Type 1 children is a direct consequence of bilateral vestibular loss combined with early visual field restriction. [5,6]

Key genes include MYO7A (USH1B — most common), CDH23 (USH1D), PCHD15 (USH1F), USH1C, and SANS (USH1G). MYO7A encodes myosin VIIA, a motor protein essential for hair bundle development and maintenance in both cochlear and vestibular hair cells, as well as retinal pigment epithelium (RPE) function. Mutations in MYO7A affect all three sensory systems — hearing, vestibular, and retinal. [5,7]

Usher Type 2

Type 2 is the most prevalent form, characterised by moderate-to-severe congenital SNHL, normal vestibular function, and RP onset in adolescence. Hearing loss is non-progressive in most cases, and walking onset is normal. USH2A (usherin) is the most common causative gene. Despite normal vestibular function, the progressive RP eventually produces significant visual handicap, and the combination of hearing loss and visual impairment creates increasing disability in adulthood. [5]

Usher Type 3

Type 3 is characterised by progressive SNHL, progressive vestibular dysfunction, and variable RP. CLRN1 (clarin-1) is the primary gene. Type 3 is rarer than Types 1 and 2 in most populations but is more prevalent in the Finnish and Ashkenazi Jewish communities. The progressive nature of all three sensory deficits makes clinical monitoring and early intervention critical in Type 3. [5,8]

Cochlear implantation produces excellent hearing outcomes in Usher Type 1 and good outcomes in Types 2 and 3. Importantly, CI restores auditory function but does not address the visual or vestibular deficits. Vestibular rehabilitation is essential in Type 1, and ophthalmological surveillance — ideally initiated before RP becomes clinically symptomatic — is the cornerstone of long-term management across all types. [6]

Figure 2. Usher Syndrome Type Differentiation — clinical features, gene assignments, and management pathways.

Source: Australian Dizziness Clinics — clinical flowchart.

□ **Important:** A child with profound congenital SNHL who walks late should be investigated for Usher Type 1. Test vestibular function (vHIT + cVEMP) and refer to ophthalmology urgently. Early diagnosis allows ophthalmological surveillance before RP produces irreversible visual field loss — the window for intervention is before clinical RP manifests.

III. CHARGE Syndrome: A Complex Multi-System Disorder

CHARGE syndrome is a major cause of complex multi-sensory impairment in childhood, with an estimated prevalence of 1 in 8,500 to 1 in 10,000. The acronym CHARGE encodes the cardinal features: C = Coloboma; H = Heart defects; A = Atresia choanae; R = Retardation of growth and development; G = Genital anomalies; E = Ear anomalies. CHD7 (chromodomain helicase DNA binding protein 7) mutations are identified in approximately 75% of clinically diagnosed cases and function through chromatin remodelling effects on multiple embryological developmental programs. [9]

The ear anomalies in CHARGE syndrome are diagnostically critical. SNHL (mixed or sensorineural) is present in the majority of affected children. However, the most distinctive vestibular finding — and the most clinically impactful — is aplasia or hypoplasia of the semicircular canals. Absent SCCs are present in the majority of children with CHARGE syndrome and are identifiable on high-resolution MRI of the temporal bones (CISS/FIESTA sequence). The horizontal and posterior canals are most frequently affected; the anterior canal may be relatively spared. [9,10]

The functional consequence of absent SCCs is bilateral vestibular areflexia — the most severe form of vestibular hypofunction. Children with CHARGE and absent SCCs have no caloric responses, absent vHIT gain in all canals, and absent VEMPs. Motor development is severely delayed: independent walking, if achieved at all, is typically not accomplished until age 3–5 years, and balance impairment persists lifelong. The combination of deafness, absent vestibular function, and visual impairment from coloboma creates a profound multi-sensory deficit that requires intensive multidisciplinary management from infancy. [3,10]

Cochlear implantation is indicated for the SNHL component and can produce meaningful hearing outcomes, though outcomes are more variable than in non-syndromic SNHL, reflecting the broader neurological complexity of CHARGE syndrome. Auditory nerve aplasia — rare but more prevalent in CHARGE than in most other aetiologies — must be excluded by MRI prior to CI. Management of vestibular dysfunction must focus on compensation strategies (visual, somatosensory substitution) rather than restoration, given the structural absence of the SCCs. [9,11]

Figure 3. CHARGE Syndrome Vestibular Phenotype — CHD7 mutations, SCC aplasia, and functional consequences.

Source: Australian Dizziness Clinics — clinical flowchart.

□ **Clinical Insight:** CHARGE children with absent semicircular canals may never develop normal balance even with intensive vestibular rehabilitation. The absent SCCs are structural, not functional. Clinical goals must be explicitly adjusted to focus on compensation, sensory substitution, and maximising independent mobility — rather than expecting vestibular recovery.

IV. Waardenburg Syndrome: Melanocyte Pathway and Inner Ear

Waardenburg syndrome is a neurocristopathy caused by mutations in genes governing neural crest cell migration and melanocyte differentiation. It is characterised by hearing loss (SNHL), characteristic pigmentation anomalies (white forelock, heterochromia irides, premature greying, depigmented skin patches), and in some subtypes, dystopia canthorum (lateral displacement of the medial canthi). The inner ear relies on neural crest-derived melanocytes in the stria vascularis for endocochlear potential maintenance; their absence or dysfunction causes SNHL. [12]

Four subtypes are recognised: Type 1 (PAX3 mutations, dystopia canthorum), Type 2 (MITF, SOX10, SNAI2 — no dystopia canthorum, higher HL prevalence), Type 3 (PAX3 — Type 1 features plus upper limb contractures), and Type 4 (SOX10 or EDNRB/EDN3 — Waardenburg + Hirschsprung disease, called Shah-Waardenburg syndrome). [12,13]

SNHL in Waardenburg syndrome is unilateral or bilateral and varies considerably in severity — from mild to profound. It is caused by absence or dysfunction of stria vascularis melanocytes, which are essential for generating and maintaining the endocochlear potential. Vestibular features are present in some patients, typically subclinical or mild, and reflect the same melanocyte-dependent mechanism affecting vestibular hair cell function. Comprehensive vestibular assessment (vHIT + VEMPs) should be standard in all children with Waardenburg syndrome regardless of reported symptoms. [13]

Waardenburg Type 4 (Shah-Waardenburg syndrome) is a critical clinical recognition point: the combination of white forelock, heterochromia, and early constipation or bowel obstruction should prompt CHD workup for Hirschsprung disease. SOX10 mutations (Types 2D and 4C) are associated with additional neurological features including peripheral demyelinating neuropathy. [12]

□ **Key Point:** Waardenburg syndrome is not purely a hearing condition. Full vestibular assessment and ophthalmological review should be standard. In Waardenburg Type 4, always ask about bowel function — Hirschsprung disease is a comorbidity requiring surgical management and can be life-threatening if unrecognised in the neonatal period.

V. Pendred Syndrome (see also PVM07)

Pendred syndrome is the most common syndromic cause of genetic hearing loss worldwide and is covered in detail in PVM07 (Enlarged Vestibular Aqueduct Syndrome). A brief summary is provided here for completeness within the syndromic vestibular disorders context.

Caused by biallelic mutations in SLC26A4, Pendred syndrome is characterised by bilateral SNHL with enlarged vestibular aqueduct (EVA), euthyroid goitre emerging in adolescence, and vestibular hypofunction in 60–80% of cases. It is clinically designated DFNB4. The vestibular features of Pendred syndrome are substantially underappreciated in the syndromic hearing loss literature: bilateral canal hypofunction on vHIT and absent/reduced cVEMPs are common findings that contribute to motor delay and gait instability. [14]

Alongside Usher syndrome Type 1, Pendred syndrome is the most common syndromic cause of vestibular dysfunction in children. Its prevalence in paediatric SNHL cohorts (5–15% via EVA imaging) makes it the numerically most significant condition discussed in this review. Management — as detailed in PVM07 — includes early bilateral hearing aids or cochlear implantation (excellent EVA outcomes), head protection, Valsalva avoidance, vestibular rehabilitation, and pro-active thyroid surveillance. Genetic counselling for SLC26A4 with cascade family testing is standard. [14,15]

VI. Other Syndromes: Alport, Branchio-Oto-Renal, Treacher Collins, Norrie

Alport Syndrome

Alport syndrome is caused by mutations in COL4A3, COL4A4 (autosomal recessive or dominant), or COL4A5 (X-linked, most common). It is characterised by progressive glomerulonephritis leading to end-stage renal disease, SNHL (high-frequency, onset in adolescence), and anterior lenticonus (pathognomonic ocular finding). SNHL is sensorineural, typically bilateral, and high-frequency in the initial stages — often identified on routine audiometry in a child being monitored for renal disease.

Vestibular features are generally absent in Alport syndrome. Renal function (GFR, urinalysis for haematuria) is the primary clinical priority. [16]

Branchio-Oto-Renal (BOR) Syndrome

BOR syndrome is caused by mutations in EYA1, SIX1, or SIX5, with an estimated prevalence of 1 in 40,000. The triad consists of branchial arch anomalies (branchial cysts, fistulae, sinuses), hearing loss (conductive, sensorineural, or mixed), and renal anomalies (ranging from agenesis to minor structural variants). Vestibular features are variable — some patients have cochlear or semicircular canal malformations on imaging. Hearing loss type and severity are highly variable, requiring comprehensive audiological assessment. [17]

Norrie Syndrome

Norrie syndrome is X-linked recessive, caused by mutations in the NDP gene (encoding norrin). It is characterised by bilateral congenital retinal dysplasia leading to blindness, progressive SNHL (typically from the second decade), and — in some patients — cognitive impairment. The combination of blindness and progressive deafness represents a profound disability requiring specialised educational and rehabilitation support. Vestibular features have not been systematically studied but may be affected in some patients. [18]

Other Syndromes

Turner syndrome (45,X karyotype) is associated with progressive SNHL affecting high frequencies and, in some series, Eustachian tube dysfunction contributing to conductive overlay. Down syndrome (trisomy 21) is the most common chromosomal cause of paediatric hearing loss, primarily through Eustachian tube dysfunction and chronic otitis media with effusion producing conductive hearing loss, but progressive SNHL is also recognised. Treacher Collins syndrome (TCOF1) causes external auditory canal atresia and conductive hearing loss from middle ear malformation. Vestibular function is typically preserved in Treacher Collins but should be tested if conductive overlay masks vestibular symptoms. [19]

□ **Clinical Insight:** In any child with chromosomal or dysmorphic syndrome and confirmed hearing loss, vestibular testing should be performed as part of the baseline assessment — not only when vestibular symptoms are reported. Vestibular hypofunction is frequently subclinical in syndromic conditions and only identified on formal testing.

VII. Genetic Testing Strategy in Syndromic Vestibular Disorders

The genetic testing strategy for syndromic vestibular disorders has evolved substantially with the widespread availability of next-generation sequencing (NGS) panels. The 2026 approach prioritises phenotype-directed panel testing, with escalation to whole exome sequencing (WES) or whole genome sequencing (WGS) when targeted panels are non-diagnostic. [4,20]

The first-line test for any child with SNHL remains GJB2/GJB6 testing — mutations in these connexin genes account for approximately 20% of all hereditary SNHL and are non-syndromic. A normal GJB2/GJB6 result does not exclude syndromic hearing loss and must be followed by phenotype-directed assessment. [20]

Phenotype-directed panel selection follows clinical features: a child with profound SNHL + delayed walking + ophthalmic referral → Usher panel (MYO7A, USH2A, CLRN1, and additional USH1/2 genes). A child with coloboma + cardiac defect + choanal atresia → CHD7 sequencing. A child with white forelock + heterochromia + SNHL → Waardenburg panel (PAX3, MITF, SOX10, EDNRB). EVA on CT + SNHL → SLC26A4. Progressive SNHL + haematuria → COL4A3/4/5 (Alport). SNHL + branchial fistulae → EYA1 (BOR). [4]

Variant interpretation uses ACMG/AMP classification (pathogenic, likely pathogenic, VUS, likely benign, benign). Variants of uncertain significance (VUS) are common in NGS panels and require family segregation studies and correlation with clinical phenotype. Genetic counsellors are essential for results communication, particularly for VUS results, and for reproductive counselling of families planning further pregnancies. [20]

Figure 4. Genetic Testing Strategy for Syndromic SNHL — phenotype-directed panel selection and interpretation pathway.

Source: Australian Dizziness Clinics — clinical flowchart.

□ **Clinical Insight:** A normal GJB2 result does not exclude syndromic hearing loss. The detailed clinical phenotypic assessment — examining for retinal findings, dysmorphic features, renal anomalies, and vestibular function — must drive the genetic panel selection. Testing alone without clinical correlation is insufficient for comprehensive diagnosis.

VIII. Multidisciplinary Assessment and Management

Management of syndromic vestibular disorders requires a structured MDT approach, with the vestibular physician as a core member alongside ENT, audiology, genetics, ophthalmology (Usher, CHARGE), cardiology (CHARGE), nephrology (Alport, BOR), physiotherapy, and educational support. The timing and composition of MDT assessment should be tailored to the specific syndrome, the child age at diagnosis, and the severity of the multi-system phenotype. [21]

Newborn hearing screening provides the earliest identification opportunity. All children with SNHL identified through newborn screening should undergo aetiological investigation — including imaging, genetics, and vestibular testing — before 3 months of age where possible, to allow early hearing rehabilitation and developmental support. Vestibular testing in all children with confirmed genetic hearing loss is now standard of care, as vestibular hypofunction affects motor development even when audiologically well-managed. [2]

Communication modality decisions — oral, sign language, total communication, or augmentative and alternative communication (AAC) — should be made collaboratively with the speech pathologist, family, and educational team at diagnosis. No single modality is superior across all syndromic hearing loss conditions; the decision depends on the degree of hearing loss, the presence of additional disabilities (visual, cognitive), and family preference. [21]

Figure 5. MDT Assessment Pathway — specialist roles, timing, and coordination for syndromic vestibular disorders.

Source: Australian Dizziness Clinics — clinical flowchart.

□ **Key Point:** Vestibular testing should occur at the time of diagnosis in all children with confirmed syndromic SNHL — it is not optional. Late vestibular diagnosis is the primary preventable adverse factor in syndromic vestibular disorders. Delayed walking, wide-based gait, and difficulty with balance in dark environments are clinical signs that should trigger immediate vestibular assessment.

IX. Rehabilitation, Education, and Long-Term Outcomes

Cochlear implantation outcomes in syndromic vestibular disorders vary by syndrome and are generally good, with some syndrome-specific caveats. Usher Type 1 yields among the best CI outcomes of any aetiology, with excellent speech perception scores in most series, comparable to non-syndromic profound SNHL. CHARGE syndrome outcomes are more variable due to the neurological complexity. Waardenburg syndrome and Pendred/EVA both yield good outcomes. Auditory neuropathy spectrum disorder (ANSD) should be excluded pre-CI in all syndromic cases. [11,22]

Vestibular rehabilitation is essential in Usher Type 1 and CHARGE syndrome, where bilateral areflexia requires active compensatory strategies. Goals include gaze stabilisation exercises (targeting the VOR deficit), optic flow training, dynamic postural balance training, and sensory substitution exercises. Physiotherapy referral at the time of vestibular diagnosis — not deferred until walking age — is the standard of care. In CHARGE, rehabilitation goals must account for the absence of structural SCCs and focus entirely on visual and somatosensory substitution. [6,23]

Long-term outcomes across syndromic vestibular disorders depend heavily on the timing of diagnosis, the comprehensiveness of MDT management, and the family engagement with rehabilitation. Employment and quality-of-life data for Usher syndrome indicate significantly reduced employment rates in individuals not identified and supported early. Psychological support for the child and family -

particularly around the progressive nature of retinal degeneration in Usher syndrome - is an underserved component of care that vestibular physicians and MDTs should proactively address. [23]

Transition to adult care must be planned from early adolescence, with explicit handover protocols ensuring that adult vestibular physicians, ophthalmologists, geneticists, and audiologists receive comprehensive syndromic documentation. The adult clinical team should not rediscover the diagnosis — a structured transition summary is the standard of care. [21]

Figure 6. Cochlear Implantation Candidacy by Syndrome — outcomes and special considerations.
Source: Australian Dizziness Clinics — clinical flowchart.

□ **Key Point:** Outcomes in syndromic vestibular disorders depend on how early the vestibular loss is identified and rehabilitated. Late diagnosis is the primary preventable adverse factor. For every syndrome discussed in this review, vestibular assessment at the time of hearing diagnosis — not years later — is the standard of care.

X. Summary and Key Clinical Takeaways

Syndromic vestibular disorders represent an important and often underdiagnosed cause of paediatric vestibular dysfunction. Recognition of syndromic patterns at diagnosis allows appropriate genetic testing, multidisciplinary management, and prognosis.

1. Usher Type 1 = profound SNHL + bilateral vestibular areflexia + RP. Late walking in a profoundly deaf child is Usher until proven otherwise.

2. CHARGE = CHD7 mutations. Absent SCCs on MRI are a major diagnostic criterion and cause bilateral vestibular areflexia — expect no vestibular recovery.

3. Waardenburg = white forelock + heterochromia + SNHL. Type 4 (Shah-Waardenburg) has Hirschsprung disease — always ask about bowel function.

4. Pendred syndrome = the most common syndromic cause of vestibular dysfunction in children (via EVA). See PVM07 for full detail.

5. Alport syndrome: progressive high-frequency SNHL + haematuria + anterior lenticonus. Vestibular features generally absent.

6. BOR syndrome: branchial anomaly + hearing loss + renal anomaly (EYA1). Variable vestibular features.

7. A normal GJB2 result does not exclude syndromic hearing loss — phenotype drives genetic panel selection.

8. Vestibular testing is mandatory at diagnosis in all confirmed syndromic SNHL — not optional, not deferred.

9. Cochlear implantation in Usher Type 1 yields excellent outcomes. CHARGE outcomes are more variable — individualised MDT decision.

10. Early diagnosis and early vestibular rehabilitation are the primary modifiable determinants of outcome across all syndromic vestibular disorders.

□ **Key Point:** The syndromic vestibular disorders share a common theme: the vestibular component is underdiagnosed, undertested, and underrehabilitated. The vestibular physician who tests all children with syndromic SNHL — regardless of whether vestibular symptoms are reported — will make diagnoses that transform the child's developmental trajectory.

The next review in this series — PVM09: Ototoxicity and Drug-Induced Vestibular Loss in Children — covers aminoglycoside and cisplatin vestibulotoxicity, monitoring protocols, genetic susceptibility (MT-RNR1), and bilateral vestibular hypofunction rehabilitation.

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