Enlarged Vestibular Aqueduct:

A Vestibular Physician's Deep Review of Congenital Inner Ear Pathology, Hearing Loss, and Management

Vestibular Medicine for Vestibular Physicians

Peripheral Vestibular Pathology — Module 2.5

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How to Use This Review

This Vestibular Physician Literature Review is designed for vestibular physicians, neuro-otologists, advanced ENT trainees, and senior vestibular physiotherapists seeking a comprehensive, evidence-based synthesis of enlarged vestibular aqueduct (EVA). The document integrates pathophysiology, clinical features, diagnostic criteria, investigation protocols, differential diagnosis, and evidence-graded management in a format designed for both clinical reference and structured continuing professional development.

- Sections I–III cover epidemiology, pathophysiology, and clinical presentation.
- Section IV details diagnostic criteria including Valvassori and Cincinnati standards.
- Section V provides investigation protocols with recommended sequencing.
- Sections VI–VIII address differential diagnosis and evidence-graded management.
- Sections IX–X cover prognosis, special populations, and knowledge gaps.

Callout boxes highlight key clinical pearls, practice-changing insights, and important warnings. Tables provide rapid access to diagnostic criteria, investigation panels, and evidence summaries. Figures include original clinical illustrations, real imaging examples, and purpose-built diagnostic flowcharts. All references use Vancouver format with inline [N] citations.

Callout Box Guide

Four callout box types are used throughout this review to highlight material of particular clinical importance:

□ **Key Point:**	Foundational concepts and summary statements that anchor the core scientific or clinical content of each section.
□ **Clinical Insight:**	Clinically relevant observations for direct application in vestibular medicine practice — mechanisms that inform clinical reasoning.
□ **Clinical Pearl:**	High-yield memorable clinical points — the take-home messages most likely to influence clinical decision-making in the consulting room.
□ **Important:**	Red flags, atypical presentations, and critical safety points requiring heightened clinical attention.

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I. Introduction and Epidemiology

Enlarged vestibular aqueduct (EVA) — also termed large vestibular aqueduct syndrome (LVAS) when clinically symptomatic — is the most common radiographically identifiable inner ear bony anomaly associated with sensorineural hearing loss (SNHL) in the paediatric population [1,2,3]. The vestibular aqueduct is a bony canal within the petrous temporal bone housing the endolymphatic duct, connecting the inner ear endolymphatic space to the endolymphatic sac on the posterior petrous surface. When enlarged beyond recognised radiological thresholds, this canal produces a distinctive and clinically important audiovestibular syndrome [1,4,5].

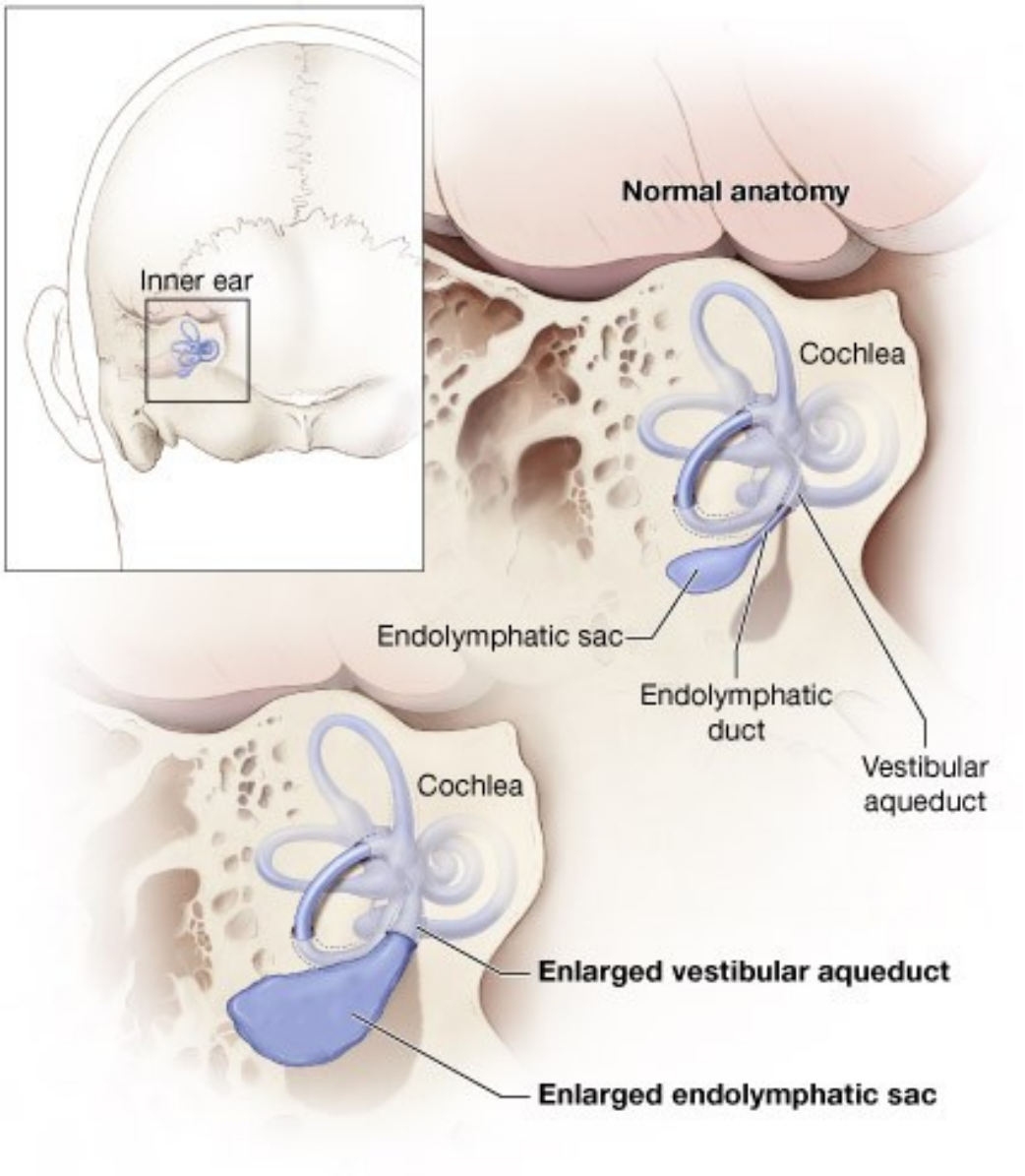

Figure 1. Normal inner ear anatomy (top) compared with enlarged vestibular aqueduct (bottom) — illustrating the widened bony canal, expanded endolymphatic sac, and their structural relationship to the cochlea and semicircular canals.

Source: Inner ear anatomy by NIH Medical Arts — National Institute on Deafness and Other Communication Disorders (NIDCD/NIH), public domain.

Epidemiology

EVA is estimated to be present in 5–15% of children with bilateral SNHL who undergo temporal bone imaging [2,5,6]. A prospective multicentre NIH cohort of 106 participants confirmed that EVA is bilateral in the majority of affected individuals, with unilateral cases constituting approximately 20–30% of presentations [7]. The condition is not exclusively paediatric — adults with EVA do present, though

hearing loss onset typically occurs in the first decade and the vestibular burden becomes apparent in the second [2,5,8].

Sex distribution data are inconsistent across cohorts. A mild female predominance (approximately 1.5:1) has been reported in some tertiary series, though other large cohorts find no significant sex difference [6,9]. Carrier frequencies for SLC26A4 mutations vary between populations — notably higher in East Asian cohorts, consistent with DFNB4 prevalence data [3,10].

□ **Key Point:** EVA is the single most common radiologically identifiable inner ear anomaly associated with childhood SNHL. It is bilateral in the majority, genetically determined in most cases, and clinically underrecognised — particularly the vestibular component.

Table 1. Epidemiology of enlarged vestibular aqueduct — key data points.

Parameter	Finding	Key Reference(s)
Prevalence in paediatric SNHL	5–15% in imaging series	Madden 2003 [2]
Laterality	Bilateral in ~70–80% of cases	NIH cohort [7]
Sex ratio	Mild female predominance (1.5:1) or equal	Arjona 2010 [6]
Age at diagnosis	Typically first decade; adults present with progressive loss	Atkinson 2015 [8]
Genetic basis (SLC26A4)	Biallelic mutations in ~50–70% non-syndromic; >95% Pendred	Griffith 2011 [3]
Associated syndromes	Pendred syndrome (SLC26A4 + thyroid); BOR syndrome (EYA1)	Kochhar 2007 [27]

II. Pathophysiology — Embryological Origins, Endolymphatic Dynamics, and SLC26A4

Embryological Origins

The vestibular aqueduct is a bony canal traversing the petrous temporal bone, housing the endolymphatic duct — the membranous channel connecting the inner ear labyrinth to the endolymphatic sac (EDS) on the posterior petrous face [11]. The normal adult aqueduct measures approximately 0.4–1.0 mm at its midpoint; values above 1.5 mm in adults (Valvassori criterion) or above 1.0 mm at the midpoint or 2.0 mm at the operculum in children (Cincinnati criteria) define enlargement [1,12].

The endolymphatic system develops from a dorsal appendage of the early otocyst during weeks 4–8 of embryogenesis [13]. Unlike cochlear and semicircular canal structures — which reach near-adult size by approximately 23 weeks of gestation — the endolymphatic duct and sac continue to remodel throughout the first years of postnatal life. Failure of this normal remodelling arrest, mediated in part by SLC26A4 expression, results in persistence of a foetal-pattern wide duct and sac [3,11,13]. The enlargement is therefore a developmental failure of resorption rather than a primary overgrowth phenomenon.

Endolymphatic Dynamics and the Third-Window Effect

The enlarged endolymphatic duct and sac create two linked pathophysiological problems: disordered endolymph homeostasis and amplified susceptibility to intracranial pressure fluctuations [14,15,16]. Normally, the endolymphatic sac regulates endolymph volume through ion transport and fluid resorption; in EVA, the sac's enlarged yet dysplastic architecture impairs this regulation, generating endolymph instability analogous — but not identical — to the hydrops of Ménière disease [16].

The enlarged bony canal additionally functions as a 'third window' — an abnormal acoustic energy pathway between the inner ear and the posterior fossa — mimicking the third-window physiology of superior semicircular canal dehiscence (SSCD) [17]. This produces the characteristic finding of an

unexplained air-bone gap on audiometry despite normal middle ear function (normal tympanogram, normal acoustic reflexes) [14,15,17]. Head trauma or rapid Valsalva manoeuvres can precipitate acute inner ear pressure shifts through this pathway, causing sudden hearing deterioration [7,16].

□ **Clinical Insight:** The air-bone gap in EVA is a third-window phenomenon, not conductive hearing loss. Normal tympanometry and acoustic reflexes in a child with an 'unexplained' air-bone gap should prompt temporal bone CT to exclude EVA and SSCD before labelling the gap conductive.

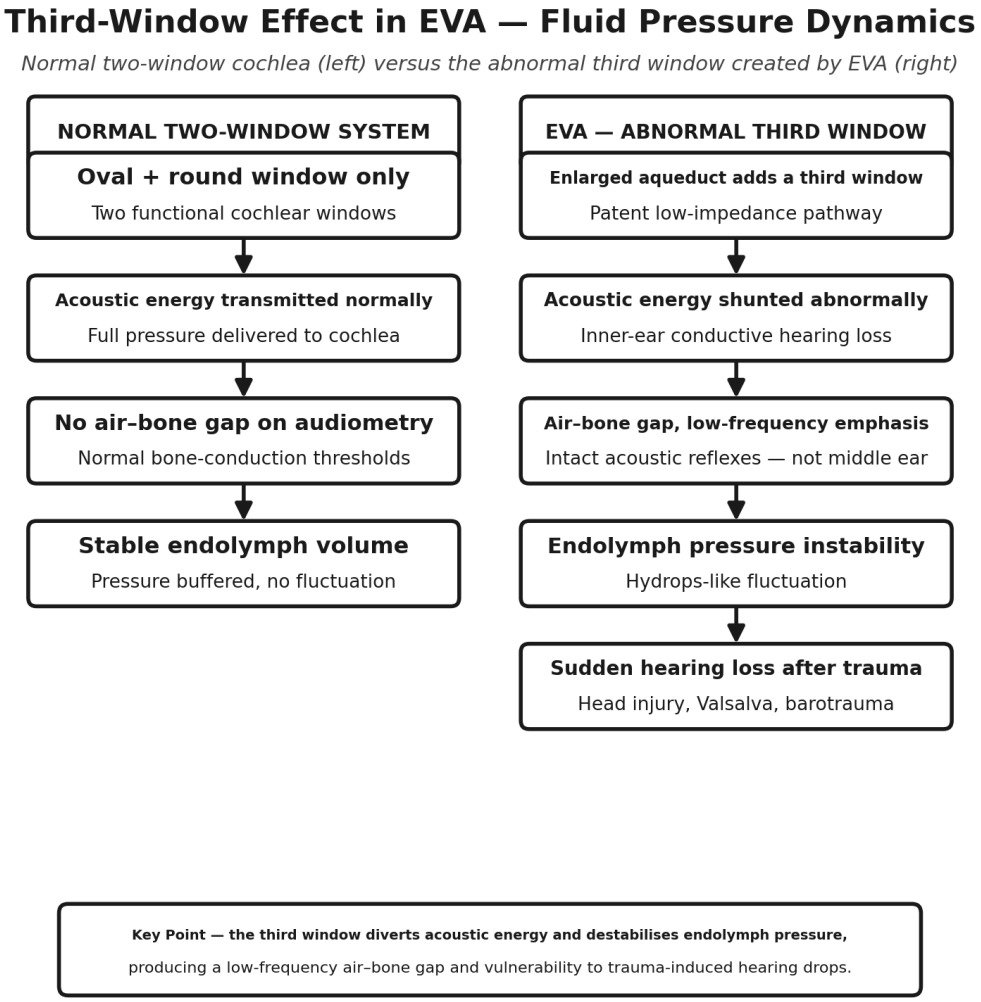

Figure 2. Third-window effect in enlarged vestibular aqueduct — normal two-window system (left) versus EVA with an abnormal acoustic pathway generating characteristic air-bone gaps and endolymphatic pressure instability (right).

Source: Adapted from Merchant et al. [14] and Griffith & Wangemann [3].

SLC26A4, Pendrin, and Genetic Associations

Genetically, EVA is strongly associated with biallelic mutations in the SLC26A4 gene on chromosome 7q22–31, encoding pendrin — a multifunctional anion exchanger ($Cl^-/I^-/HCO_3^-$) expressed in the endolymphatic sac, thyroid follicular epithelium, and renal collecting duct [3,18]. Pendrin is essential for the ionic composition and pH regulation of endolymph; loss of pendrin function produces endolymphatic acidification, disturbed ion transport, and progressive degeneration of cochlear hair cells [3,19].

SLC26A4 mutations produce two clinically distinct phenotypes separated primarily by thyroid involvement [3,10,18]:

- **Pendred Syndrome (PS):** Autosomal recessive; biallelic SLC26A4 mutations producing cochlear EVA and congenital SNHL plus thyroid goitre (and/or positive perchlorate discharge test). Mondini dysplasia frequently co-occurs. Pendred syndrome is the most common syndromic cause of congenital deafness worldwide [3,18,20].
- **DFNB4 (Non-syndromic EVA):** Autosomal recessive non-syndromic deafness type 4. SLC26A4 mutations produce EVA and hearing loss without thyroid involvement. Clinically indistinguishable from Pendred syndrome without thyroid assessment and genetic testing [10,18].

Approximately 50–70% of non-syndromic EVA patients harbour biallelic SLC26A4 mutations; the remaining cases implicate FOXI1, KCNJ10, and potential modifier loci yet to be identified [3,21,48]. A prevailing paradigm holds that EVA and hearing loss may share a common upstream cause (SLC26A4 dysfunction), rather than the enlarged aqueduct being the direct structural cause of hearing loss [3,19,21].

□ **Important:** Pendred syndrome is the most common syndromic cause of congenital deafness. Every child with EVA should have thyroid assessment (TSH, free T4) and consideration of SLC26A4 genetic testing, regardless of hearing severity. Missing Pendred has implications for thyroid monitoring, genetic counselling, and family screening.

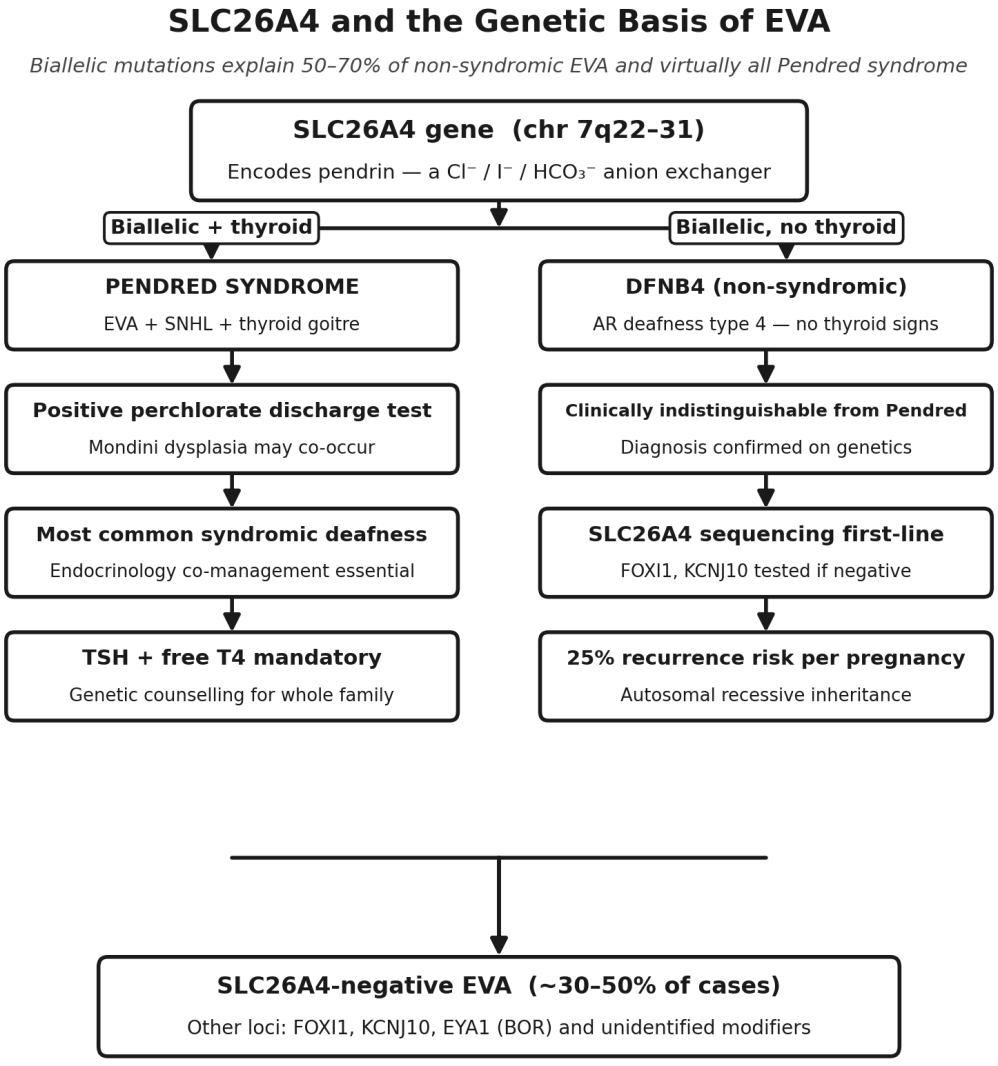

Figure 3. SLC26A4 genetic associations in EVA — Pendred syndrome versus DFNB4 non-syndromic EVA, the role of modifier genes in SLC26A4-negative cases, and the molecular basis of phenotypic variation.
Source: Adapted from Yang et al. [21] and Smith & Pyne [48].

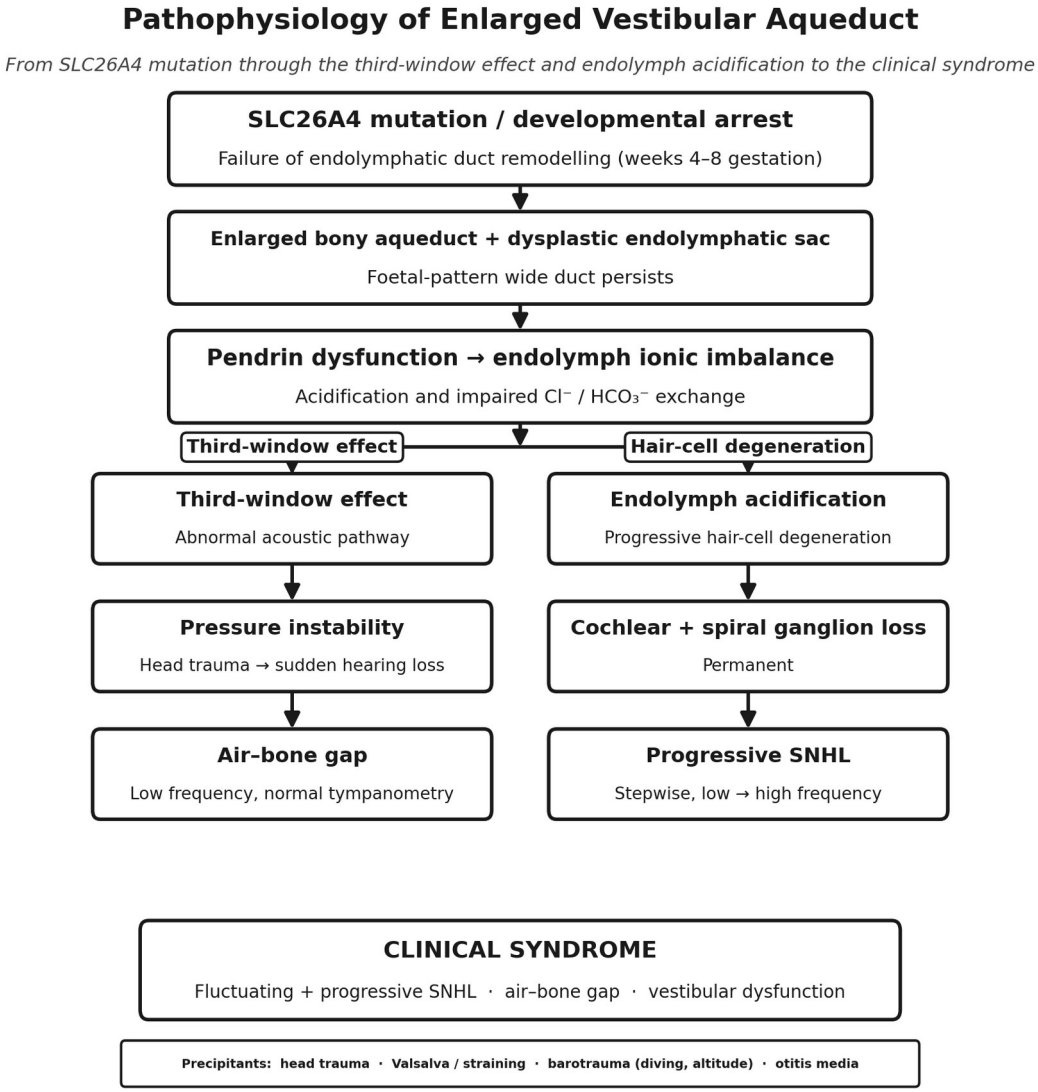

Figure 4. Pathophysiology of enlarged vestibular aqueduct — from SLC26A4 mutation through third-window effect and endolymph acidification to the clinical syndrome of progressive SNHL and vestibular dysfunction.
Source: Adapted from Griffith & Wangemann [3] and Wangemann et al. [19].

III. Clinical Features — Hearing Loss Profile, Vestibular Symptoms, and Associated Features

Hearing Loss Profile

Hearing loss is the cardinal and defining clinical feature of EVA. The loss is typically bilateral in the majority of syndromic cases (more than 90%) and in approximately 60–70% of non-syndromic cases [2,7]. The audiometric configuration is variable — some patients present with low-frequency emphasis (upsloping audiogram, reflecting third-window physiology), while others demonstrate flat or high-frequency sloping loss typical of general cochlear damage [14,15]. A hallmark feature is fluctuating and progressive hearing loss, often precipitated by minor head trauma, barotrauma, or viral illness [7,8]. This pattern distinguishes EVA from the more stable hearing loss of cochlear aplasia or auditory neuropathy.

A diagnostically important audiometric finding is an unexplained air-bone gap despite normal middle ear function — normal tympanogram and intact acoustic reflexes [14,17]. This third-window signature should not be attributed to middle ear disease without temporal bone imaging. The gap is typically most prominent at low frequencies [14,15]. Approximately 25–50% of EVA patients experience significant progressive hearing decline over 5–10 years of follow-up [7,22]. Fluctuating hearing loss episodes —

defined as acute threshold shifts with partial recovery — occur in approximately 30–40% of patients and are a hallmark of EVA natural history [47].

□ **Clinical Pearl:** Progressive SNHL + unexplained air-bone gap + normal tympanogram in a child = image the temporal bones. This triad is EVA until proven otherwise. Failure to image is the most common diagnostic error in EVA.

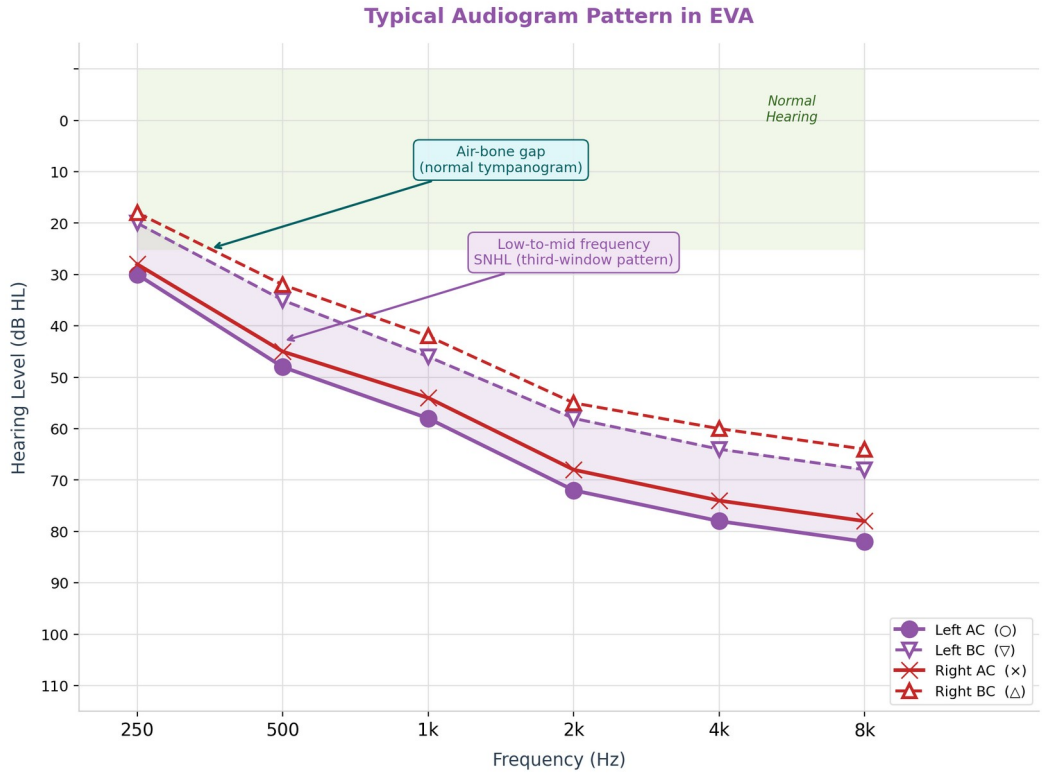

Figure 5. Typical audiogram pattern in EVA — low-to-mid frequency sensorineural hearing loss with a characteristic air-bone gap reflecting third-window physiology. Bone-conduction thresholds confirm sensorineural rather than conductive pathology.

Source: Adapted from Atkinson et al. [8] and Merchant et al. [14].

Vestibular Symptoms

Vestibular symptoms occur in a substantial but variable subset of EVA patients and are frequently underrecognised in clinical practice. The NIH prospective cohort reported vestibular symptoms in 45% and objective VFT abnormalities in 44% of 106 participants [7]. The character of vestibular symptoms in EVA differs from Ménière disease. Catastrophic episodic vertigo attacks are uncommon; instead, patients typically experience milder disequilibrium, motion sensitivity, and positional dizziness [7,24]. In children, vestibular dysfunction may manifest as motor developmental delay, delayed walking, or apparent coordination difficulties rather than overt vertigo [23,25].

Associated Features

Tinnitus is reported in approximately one-third of patients, often described as a low-pitched hum or roaring sound that may fluctuate with hearing acuity [7]. In syndromic presentations — Pendred syndrome, branchio-oto-renal (BOR) syndrome — additional features include thyroid goitre or euthyroid gland dysfunction (Pendred), branchial cleft anomalies, renal structural abnormalities, and external or middle ear anomalies (BOR) [18,20,27]. Mondini dysplasia co-present on imaging in Pendred syndrome and DFNB4 affects cochlear implant surgical planning [11,28].

IV. Diagnostic Criteria — Valvassori, Cincinnati, and Radiological Standards

Valvassori and Clemis Criterion (1978)

The original criterion for EVA was proposed by Valvassori and Clemis in 1978, based on polytomography measurements of the vestibular aqueduct midpoint [1]. The criterion defines EVA as a midpoint diameter greater than 1.5 mm, measured on axial imaging. This threshold remains widely used in adult clinical practice and epidemiological literature but is recognised to under-diagnose EVA in children, where the normal aqueduct is smaller [1,12].

Cincinnati Criteria

The Cincinnati criteria define EVA as a midpoint diameter greater than 1.0 mm, or an opercular diameter greater than 2.0 mm, using axial HRCT with slice thickness 0.6 mm or less [12,26]. These criteria improve sensitivity by capturing cases the Valvassori threshold misses, though the trade-off is a higher rate of borderline diagnoses in children under 5 years [12].

In practice, most vestibular physicians and radiologists reporting paediatric temporal bone CT use Cincinnati criteria as the operative standard, reserving Valvassori primarily for adult studies and research cohort definitions [1,12,26].

> □ **Clinical Pearl:** When reviewing a radiology report that states 'no EVA' — check the criterion used. A report applying Valvassori alone (>1.5 mm) may miss a Cincinnati-positive child (>1.0 mm midpoint). If clinical suspicion is high, request explicit measurement and Cincinnati criterion application.

Table 2. Diagnostic criteria for enlarged vestibular aqueduct.

Criterion	Measurement	Threshold	Best Applied
Valvassori (1978)	Midpoint aqueduct diameter (axial CT)	>1.5 mm	Adults; historical research
Cincinnati — midpoint	Midpoint aqueduct diameter (axial HRCT <=0.6mm)	>1.0 mm	Paediatric; current standard
Cincinnati — operculum	Opercular aqueduct diameter (axial HRCT <=0.6mm)	>2.0 mm	Paediatric; coronal reformat
Boston criteria	Midpoint >1.5mm OR operculum >1.9mm	Either threshold	Adult research cohorts

A pragmatic approach integrates imaging dimensions, audiometric configuration (third-window pattern), clinical history (fluctuating loss, trauma-related threshold shifts), and genetic findings when making a clinical diagnosis in borderline cases [12,26].

V. Investigations: Audiological, Vestibular, Imaging, and Genetic Protocols

Audiological Assessment

Pure tone audiometry (PTA) is the cornerstone investigation. In confirmed or suspected EVA, the audiologist should explicitly document: the audiometric configuration, the presence and magnitude of any air-bone gap, and the results of tympanometry and acoustic reflex testing — essential to confirm normal middle ear function in any child with an apparent conductive component [14,15]. Auditory brainstem

response (ABR) is indicated in infants and young children where reliable behavioural audiometry is not obtainable. Monitoring audiograms every 3–6 months during the first decade are recommended to track progression and guide hearing rehabilitation decisions [2,26].

Vestibular Function Testing

Formal vestibular function testing (VFT) is indicated in any EVA patient with balance complaints, gait difficulties, or delayed motor milestones, and should be considered proactively given the 40–45% objective vestibular abnormality rate in prospective series [7]. A comprehensive VFT battery for EVA should include:

- Video head impulse test (vHIT): Canal-specific semicircular canal gain measurement. Abnormalities in EVA most commonly reflect horizontal and/or posterior canal dysfunction; canal-specific analysis is essential for rehabilitation planning [31].
- Caloric testing (bithermal irrigation): Probes the horizontal canal at very low frequencies (0.003 Hz). Caloric paresis is present in approximately 30–40% of EVA patients with vestibular symptoms [7,31].
- Cervical and ocular VEMPs (cVEMP/oVEMP): Assess saccular and utricular function respectively. VEMP abnormalities reflect otolith organ involvement and predict balance rehabilitation needs [7,32].
- Videonystagmography (VNG): Positional nystagmus mimicking BPPV can occur in EVA and reflects altered endolymphatic dynamics rather than otoconial displacement [24].

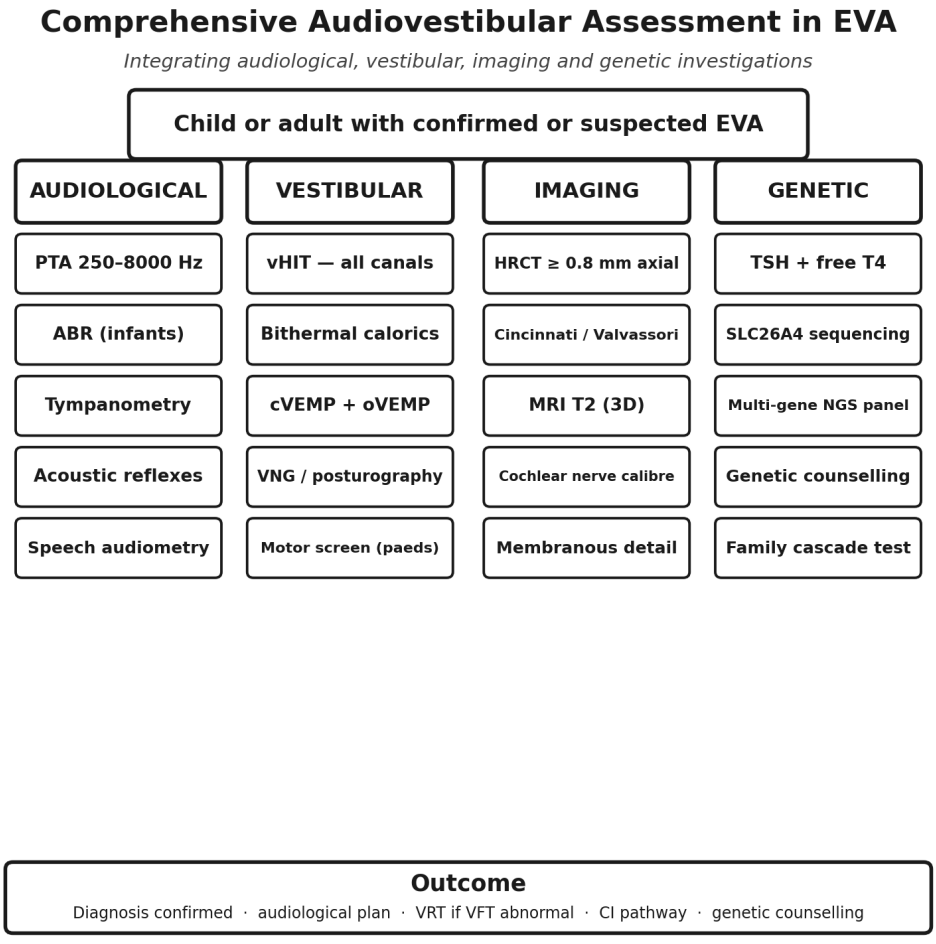

Figure 6. Comprehensive audiovestibular assessment battery for EVA — integrating audiological, vestibular function, imaging, and genetic investigations with recommended sequencing.
Source: Adapted from Boston et al. [9] and Meinzen-Derr et al. [22].

Imaging Protocols

HRCT temporal bone is the definitive diagnostic modality. Protocol requirements include axial acquisition 0.6 mm slice thickness; coronal reconstructions; measurements of aqueduct midpoint and opercular diameter using Cincinnati criteria [28,29,30,49]. In surgical candidates, HRCT should document cochlear turns and basal diameter, round window niche anatomy, facial nerve course, and any semicircular canal malformation [28]. MRI with gadolinium enhancement delineates cochlear nerve calibre and endolymphatic sac pathology [50] — relevant where cochlear nerve aplasia must be excluded prior to implantation [30].

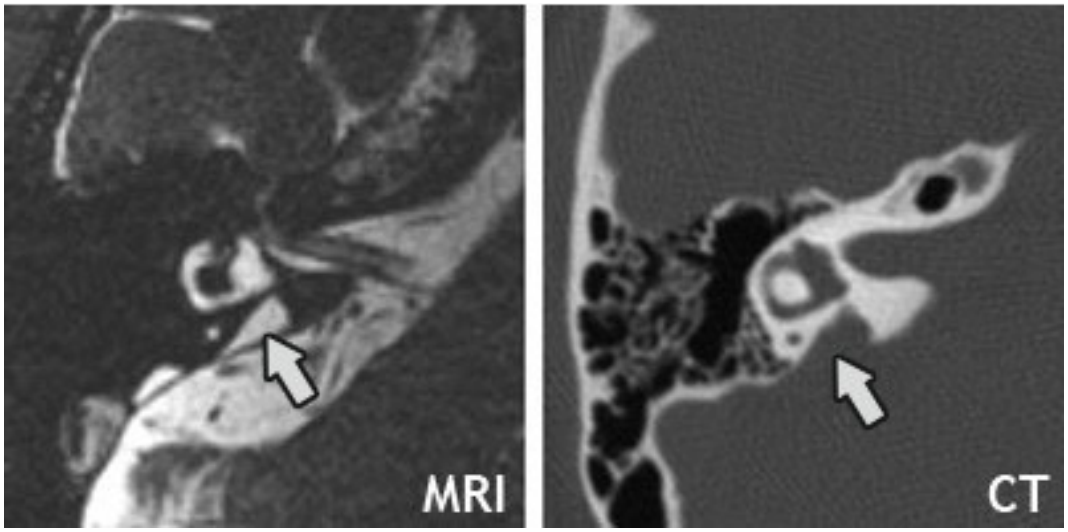

Figure 7. Temporal bone imaging in enlarged vestibular aqueduct — MRI (left) and CT (right) demonstrating the enlarged vestibular aqueduct (arrows) in the same patient. CT defines the bony canal; MRI delineates the endolymphatic duct and sac.

Source: National Institute on Deafness and Other Communication Disorders (NIDCD/NIH), "Enlarged Vestibular Aqueducts and Hearing Loss" (NIH Pub. No. 06-6053), public domain.

Genetic Testing

Genetic testing is increasingly integrated into the EVA diagnostic pathway [3,10,21]. SLC26A4 full gene sequencing identifies the causal variant in approximately 50–70% of non-syndromic EVA cases. Multi-gene NGS panels covering FOXI1, KCNJ10, EYA1 (BOR syndrome), and GJB2 are recommended as the first-line approach [3,21,27]. In patients meeting clinical criteria for Pendred syndrome, genetic testing confirms the diagnosis [18,20].

Table 3. Recommended investigations in EVA — priority and rationale.

Investigation	Priority	Rationale
Pure tone audiometry (± ABR in infants)	Mandatory at diagnosis	Baseline; characteristic air-bone gap pattern
Tympanometry + acoustic reflexes	Mandatory	Confirms normal middle ear (to interpret air-bone gap)
HRCT temporal bones (<=0.6 mm)	Mandatory at diagnosis	Defines anatomy; measures aqueduct; CI planning
MRI temporal bones (T2 + Gad)	In infants / CI candidates	Cochlear nerve calibre; endolymphatic sac morphology
vHIT (all canals)	Strongly recommended if VFT indicated	Canal-specific gain; rehabilitation planning
Bithermal caloric testing	Recommended if balance symptoms present	Horizontal canal function; detects caloric paresis
cVEMP + oVEMP	Recommended	Saccular and utricular function
TSH, free T4	Mandatory in all EVA patients	Exclude Pendred syndrome
SLC26A4 sequencing / multi-gene panel	Strongly recommended bilateral EVA	Molecular diagnosis; counselling; family testing
Genetic counselling	All confirmed EVA	Inheritance pattern; sibling and

		offspring risk

VI. Differential Diagnosis

Several conditions present with features overlapping EVA and must be considered, particularly when imaging is unavailable or inconclusive [14,17,33].

Superior Semicircular Canal Dehiscence (SSCD)

SSCD is the most important single diagnostic mimic of EVA. Both conditions produce a third-window effect resulting in an apparent air-bone gap despite normal middle ear function [14,17,33]. Differentiating features: CT localisation (tegmen defect in SSCD vs posterior petrous EVA), the Tullio phenomenon (more prominent in SSCD), and VEMP pattern (enhanced cVEMP amplitude and reduced threshold in SSCD vs typically reduced amplitude in EVA) [33,34].

Ménière Disease

The episodic vertigo + hearing loss pattern of Ménière disease overlaps with the fluctuating audiovestibular course of EVA. Key differentiating features: EVA is typically diagnosed in childhood and has a radiological correlate (enlarged aqueduct on CT); Ménière disease usually presents in adults with no pathognomonic CT finding. Endolymphatic hydrops on gadolinium-enhanced MRI may be present in both [16,42]. A small proportion of EVA patients develop a true Ménière-like syndrome [16,42].

Other Differentials

- **GJB2 SNHL:** Connexin-26 hearing loss presents with bilateral congenital SNHL without EVA. No air-bone gap; stable or slowly progressive. Temporal bone CT normal [43].
- **Auditory Neuropathy Spectrum Disorder (ANSD):** Normal or near-normal OAEs with absent or markedly abnormal ABR. No air-bone gap; no CT findings. Can co-occur with EVA in some patients [44].
- **Autoimmune Inner Ear Disease (AIED):** Rapidly progressive bilateral SNHL in adults. CT temporal bones are normal; responds to systemic corticosteroids [45].
- **Otosclerosis:** Mixed hearing loss with air-bone gap but conductive component confirmed by abnormal tympanometry and absent reflexes — unlike EVA where middle ear function is normal [46].

Table 4. Differential diagnosis of EVA — key distinguishing features.

Condition	Air-bone gap	Tympanometry	CT finding	Key Distinguisher
EVA	Yes (low freq)	Normal	Enlarged VA (>1.5mm)	Paediatric; genetics; CT
SSCD	Yes (low freq)	Normal	Tegmen dehiscence	Tullio; enhanced cVEMP
Ménière disease	No	Normal	Normal (CT)	Adult onset; no CT anomaly
GJB2 SNHL	No	Normal	Normal	Stable; connexin genetics
Otosclerosis	Yes	Abnormal (As)	Halo sign cochlea	Absent reflexes; adult
AIED	No	Normal	Normal	Rapid bilateral adult; steroid response

VII. Management — Hearing Preservation, Rehabilitation, and Cochlear Implantation

Hearing Preservation and Monitoring

There is no established medical or surgical cure for EVA, and management is primarily supportive, rehabilitative, and preventive. The cornerstone is close audiological surveillance to detect progression and guide timely hearing rehabilitation [2,7,26]. Children should undergo repeat audiometry every 3–6 months during the first decade; adults with stable thresholds can be monitored annually unless symptomatic fluctuation occurs [2,26].

- Hearing aids: Appropriately fitted conventional hearing aids significantly improve auditory function and support speech and language acquisition in affected children. Early fitting within the first year of confirmed hearing loss is recommended [2,26].
- Head injury prevention: Families should be counselled to avoid high-impact contact sports and to use protective headgear during activities with fall or impact risk. The risk of sudden hearing deterioration following minor head trauma is significant — 25–33% of EVA patients have documented trauma-related sudden hearing loss events [7,22].
- Pressure change avoidance: Patients should exercise caution during air travel, Valsalva manoeuvres, and rapid altitude changes. Diving is generally contraindicated for bilateral EVA given the barotrauma risk [7,16].

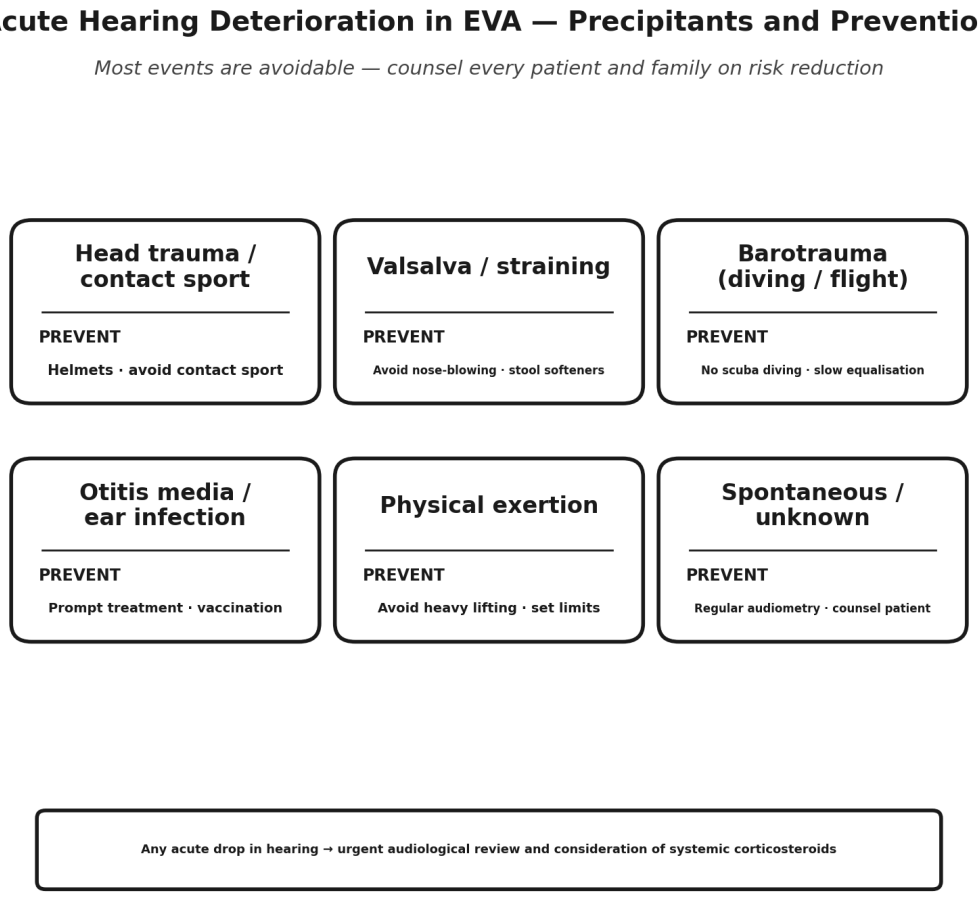

Figure 8. Precipitants of acute hearing deterioration in EVA and evidence-based preventive strategies — the majority of events are avoidable with patient and family education.
Source: Adapted from Atkinson et al. [8] and Rauch [40].

Cochlear Implantation

Cochlear implantation (CI) is the most impactful intervention for children with severe-to-profound bilateral hearing loss due to EVA. EVA is not a contraindication to CI; outcomes are generally favourable, with speech perception benefits comparable to children without EVA receiving implants at equivalent age [35,36]. Key surgical considerations include:

- Imaging-guided planning: HRCT must characterise cochlear morphology (Mondini co-occurrence, common cavity), semicircular canal anomalies, and facial nerve course deviations [28,35].
- Gusher risk: The enlarged endolymphatic duct creates a potential perilymph pressure conduit. Some EVA patients — particularly those with Mondini — have a patulous cochlear aqueduct or modiolus deficiency, creating the risk of a cochlear 'gusher'. Pre-operative MRI assessment of the cochlear aperture is essential [28,35,36].
- Electrode selection: In the presence of Mondini malformation, a shorter array may be required; full cochlear coverage may not be achievable [28,35,47].
- Post-implantation outcomes: Speech perception outcomes in EVA-related CI are at worst equivalent to, and in many series comparable with, those of CI in children with normal cochlear anatomy, provided implantation is performed before prolonged auditory deprivation [35,36].

Vestibular Rehabilitation

For EVA patients with vestibular dysfunction — imbalance, motion sensitivity, positional dizziness, or delayed motor milestones — formal vestibular rehabilitation therapy (VRT) is the primary evidence-based intervention [23,37]. In children, this is best delivered through a paediatric physiotherapist with vestibular expertise, using age-appropriate gaze stabilisation exercises, balance retraining, and habituation strategies [23,37]. In adults, the established VRT evidence base from unilateral and bilateral vestibulopathy applies, adapted to the EVA clinical context [37,38].

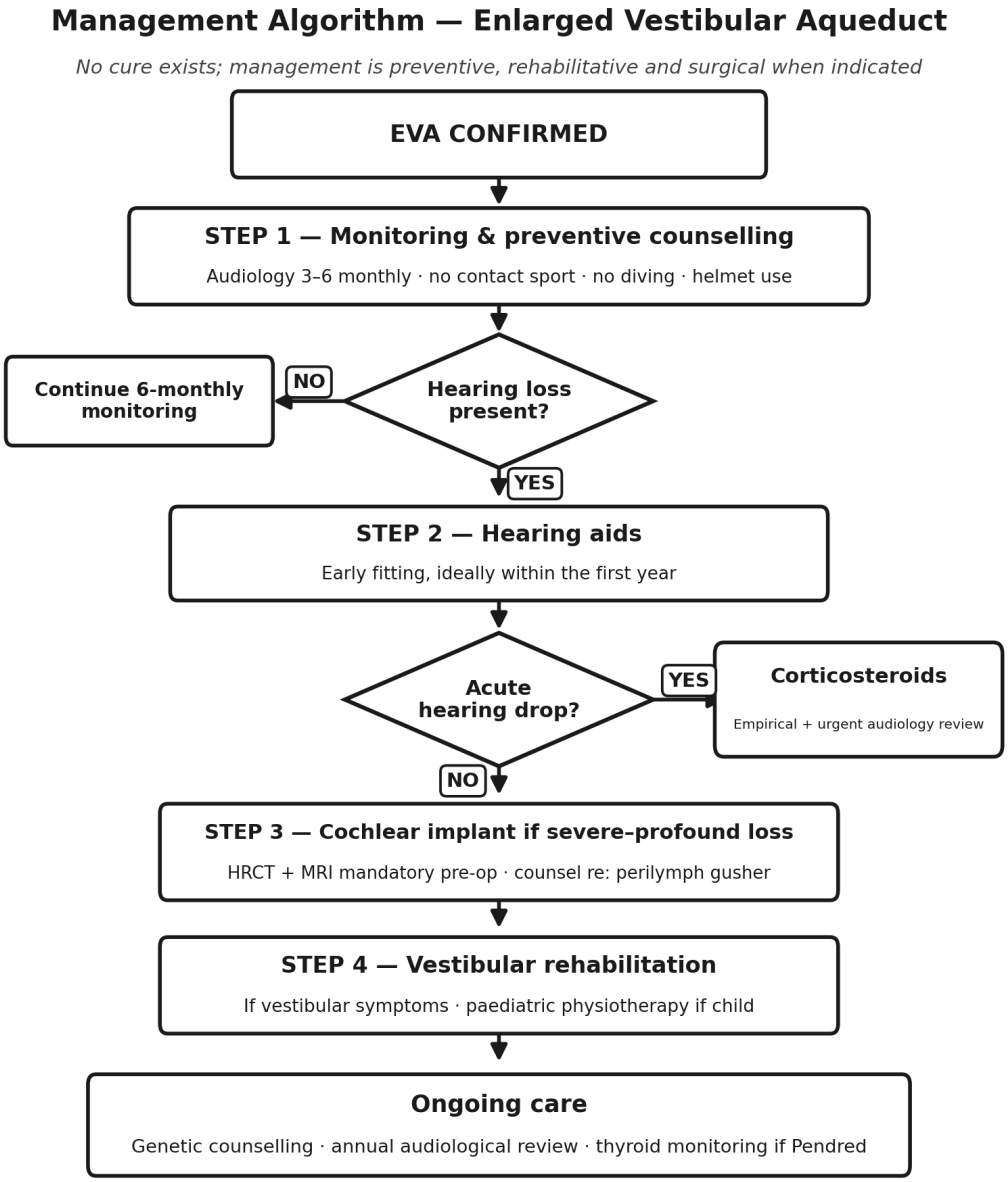

Figure 9. Management algorithm for enlarged vestibular aqueduct — integrating hearing rehabilitation, vestibular strategies, cochlear implant pathway, head protection counselling, and genetic follow-up.
Source: Adapted from Vincenti et al. [35], Hall et al. [37] and Reilly & Lalwani [39].

VIII. Pharmacological and Surgical Considerations

Pharmacological Approaches

There is no proven pharmacological therapy that prevents hearing progression in EVA. Corticosteroids — systemic or intratympanic — are sometimes administered in the acute phase of sudden hearing deterioration, extrapolated from the sudden SNHL treatment paradigm [39]. Evidence specific to EVA is limited to case series and retrospective analyses; no randomised controlled trial has demonstrated benefit. A short course of high-dose systemic corticosteroid (e.g., prednisolone 1 mg/kg for 7–10 days) is reasonable empirical clinical practice in acute EVA-related hearing deterioration [39,40].

Diuretics (acetazolamide, hydrochlorothiazide) have been trialled anecdotally in EVA given the shared endolymphatic hydrops pathophysiology, but no controlled evidence supports their use in EVA specifically [16,39]. Low-sodium diet similarly lacks EVA-specific evidence [16].

□ **Important:** Prescribing diuretics for EVA in the absence of documented endolymphatic hydrops is extrapolation without EVA-specific evidence. Corticosteroids for acute hearing deterioration are empirical — always counsel families on limited evidence and monitor audiometric response within 4–6 weeks.

Endolymphatic Sac Surgery

Endolymphatic sac (ELS) decompression or shunting has a deeply controversial evidence base [39,41]. The Cochrane review on ELS surgery in Ménière disease demonstrated no benefit over placebo for vertigo, hearing, or tinnitus [41]. Current consensus is that ELS surgery is not recommended outside of research protocols — the risk of hearing deterioration from surgical trauma is real, and the absence of benefit evidence is compelling [39,41].

Table 5. Pharmacological and surgical options in EVA — evidence summary.

Intervention	Evidence Level	Current Recommendation	Notes
Systemic corticosteroids (acute hearing loss)	Case series / expert consensus	Reasonable empirical trial	Short course; audiometric monitoring essential
Intratympanic corticosteroids	Case series	Reasonable adjunct or rescue	If systemic contraindicated; limited EVA data
Diuretics (acetazolamide / HCTZ)	No EVA-specific RCT	Not routinely recommended	Anecdotal; Meniere evidence extrapolation only
Low-sodium diet	No EVA evidence	Empirical lifestyle measure	May be offered in hydrops-pattern symptoms
Endolymphatic sac surgery	Retrospective only; no RCT	Not recommended (outside trials)	Cochrane review negative in Meniere
Vestibular suppressants (acute attacks)	Expert consensus	Short-term only (48-72 h)	Diazepam / prochlorperazine; limit duration

IX. Prognosis, Natural History, and Special Populations

Natural History of Hearing

EVA carries a variable but often progressive hearing trajectory. Long-term follow-up data from prospective cohorts demonstrate that hearing loss progresses from mild to profound loss within the first decade in a significant proportion of children [7,22]. Pooled data indicate:

- Approximately 25–50% of EVA patients experience significant hearing decline (10 dB across two frequencies or more) over a 5-year follow-up period [7,22].
- Stabilisation of hearing is reported in approximately one-third; a minority demonstrate partial recovery after acute threshold shifts, particularly if precipitated by a reversible trigger such as viral illness [7,22,47].
- Severity and rate of progression correlate with: genetic status (biallelic SLC26A4 mutations associated with more severe early loss), degree of enlargement, and presence of Mondini dysplasia [3,7].

Special Populations

Paediatric Considerations

Any child with unexplained SNHL should have bilateral temporal bone imaging early in the diagnostic pathway — not reserved as a second-line investigation [2,26]. Family counselling about precipitant avoidance (head trauma, barotrauma, Valsalva) is critical and may significantly alter the hearing trajectory [7,22]. Motor developmental surveillance is recommended for all EVA children given the 40–45% rate of objective vestibular dysfunction [23,25].

Syndromic EVA

Pendred syndrome patients require ongoing thyroid surveillance — even euthyroid patients at diagnosis develop goitre and occasional hypothyroidism over time [18,20]. Endocrinology co-management is recommended from diagnosis. BOR syndrome patients require renal structural imaging and urology review given the high rate of renal anomalies [27]. Genetic counselling for the family is essential in all syndromic presentations [18,27].

Adults with EVA

Adults presenting with EVA require the same audiovestibular surveillance protocol as paediatric patients, with particular attention to occupational noise exposure, diving, and contact sports [8]. Cochlear implantation outcomes in adults with EVA are reported as equivalent to non-EVA adult implant recipients when candidacy criteria are met [36].

X. Knowledge Gaps and Future Directions

Despite substantial progress in understanding EVA genetics and pathophysiology, several important clinical questions remain unresolved:

- Genotype-phenotype correlations: The relationship between specific SLC26A4 variants and hearing trajectory is incompletely characterised. Understanding which mutations predict rapid progression versus stable loss would transform prognostic counselling [3,21].
- SLC26A4-negative EVA: The genetic architecture of EVA in the 30–50% of patients without SLC26A4 mutations remains poorly defined. Identification of additional causal genes would complete the genetic diagnostic pathway [3,21,48].
- Gene therapy: SLC26A4 restoration in animal models has shown promise. Translation to human trials remains pending — EVA represents an attractive target given the monogenic aetiology in most syndromic cases [3,19].
- Cochlear implant electrode design: Purpose-designed arrays for dysmorphic cochleae are an active area of development [28,35].
- EVA and endolymphatic hydrops: Whether EVA-associated hydrops represents a distinct entity or a subtype of Ménière disease has implications for treatment trial design [16,42].

> □ **Key Point:** EVA is a tractable monogenic disorder with a clearly identified causal gene (SLC26A4) in the majority of syndromic cases — making it an important future target for gene therapy and precision medicine approaches in vestibular medicine.

The EVA community will benefit from: larger prospective natural history cohorts with standardised imaging protocols, international registry linkage between audiological, genetic, and outcomes data, and randomised intervention trials for corticosteroids in acute hearing deterioration [7,26,42].

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Accuracy and Currency

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