Enlarged Vestibular Aqueduct Syndrome:

Genetics, Pathophysiology, and Clinical Management

Vestibular Medicine in Children

Topic 7 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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I. Introduction: EVA as the Most Common Radiological Inner Ear Anomaly

Feature	Detail	Clinical significance
Inheritance	Autosomal dominant; 30–40% de novo mutations	Family history important; genetic counselling
Prevalence	1:1000–1:5000	Most common genetic cause of combined hearing/balance loss
Gene (most common)	SLC26A4 (Pendrin); FOXI1; KCNJ10	Pending syndrome (SLC26A4 biallelic) in 20–30% of EVA
Radiological definition	Vestibular aqueduct >1.5 mm at midpoint (or >2 mm at operculum)	CT temporal bone or MRI inner ear (CISS/FIESTA)
Hearing	Fluctuating SNHL; often bilateral; progressive	Usually the presenting feature; audiogram + SNHL workup first
Vestibular features	Episodic vertigo; EVH trigger; balance impairment	Often overshadowed by hearing loss

II. Anatomy, Radiological Criteria, and Classification

III. Genetics: SLC26A4, Pendred Syndrome, and DFNB4

Trigger	Pathophysiology	Prevention
Minor head trauma	Pressure wave through EVA → endolymphatic hydrops	Avoid contact sports; head protection
Valsalva manoeuvre	Increased intralabyrinthine pressure	Avoid nose-blowing forcefully; teach modified Valsalva
Physical exertion (heavy lifting)	Transmitted intrathoracic pressure	Activity modification; PE guidance
Barotrauma (air travel; diving)	External pressure changes	Avoid diving; ear-equalising strategies for air travel
Upper respiratory infections	Indirect pressure via Eustachian tube dysfunction	Early treatment of URTI; avoid nose-blowing

IV. Pathophysiology: Endolymphatic Hydrops and Sensorineural Hearing Loss

Investigation	Finding in EVA	Purpose
CT temporal bone (high-resolution)	Vestibular aqueduct width >1.5 mm at midpoint	Definitive radiological diagnosis; bilateral in 80%
MRI inner ear (CISS/FIESTA)	Enlarged endolymphatic sac; may show cochlear abnormalities	Soft tissue assessment; associated malformations
Pure-tone audiogram	Fluctuating SNHL; often bilateral; low-frequency involvement	Baseline + serial monitoring (every 6 months)
SLC26A4 genetic testing	Biallelic mutations = Pendred syndrome	Family counselling; prognosis; associated thyroid disease
Thyroid function tests + ultrasound	Pendred syndrome (goitre + SNHL + EVA)	If SLC26A4 mutation identified
vHIT + cVEMP	Vestibular hypofunction inter-episode	Quantify vestibular deficit; guide VRT

V. Clinical Presentation: Hearing Loss, Vestibular Features, and Fluctuation

Activity	Risk level	Recommendation
Contact sports (rugby, AFL, martial arts)	High	Avoid; EVA = absolute contraindication in many guidelines
Non-contact team sports (swimming, cycling)	Low	Generally permitted with appropriate supervision
Air travel	Moderate	Permitted; decongestant if URI; advise Valsalva technique
Diving (SCUBA)	High	Contraindicated — barotrauma risk
School sport/PE	Low-moderate	Modify for contact; head protection if unavoidable contact
Water polo/competitive swimming (tumble turns)	Moderate	Case-by-case; vigorous head-underwater manoeuvres caution

VI. Audiological and Vestibular Assessment

VII. Imaging: CT vs MRI in EVA

Timepoint	Assessment	Action
6-monthly	Audiogram; DPOAE	Document any threshold shift; if >15 dB shift → hearing aid review
Annually	vHIT + cVEMP; vestibular function	Update VRT if vestibular function declining
After any trigger event	Clinical review; audiogram	Acute management if SNHL drops; consider oral steroids
Before school transition	Audiological and vestibular report for school	FM system; safety plans; PE modifications
At genetic diagnosis	Genetic counselling; thyroid function	Family cascade screening; Pendred syndrome workup if SLC26A4+

VIII. Pendred Syndrome: Thyroid, Goitre, and Perchlorate Discharge Test

IX. Management: Hearing Rehabilitation, Vestibular Strategies, and Precautions

Indication	Urgency	Refer to
New diagnosis EVA in child	Soon	Vestibular physician + audiology + genetics
Acute SNHL episode	Urgent (within 72 hours)	Audiology + ENT; oral steroids consideration
Progressive vestibular hypofunction	Routine	Vestibular physician + VRT
SLC26A4 mutation identified	Routine	Medical genetics + endocrinology (thyroid)
Considering cochlear implant	Pre-operative referral	Paediatric CI team; note EVA

		architecture implications

X. Summary and Key Clinical Takeaways

References

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I. Introduction: EVA as the Most Common Radiological Inner Ear Anomaly

Enlarged vestibular aqueduct (EVA), also termed large vestibular aqueduct syndrome (LVAS) when clinically symptomatic, is the most frequently identified radiological inner ear anomaly in children presenting with sensorineural hearing loss (SNHL). It was first described by Valvassori and Clemis in 1978, and has since been recognised as a major cause of childhood hearing loss — preventable in its progression when diagnosed and managed appropriately. [1]

The prevalence of EVA in paediatric SNHL cohorts ranges from 5 to 15%, making it the single most common identifiable imaging abnormality on temporal bone CT in this population. Bilateral involvement occurs in approximately 80% of cases. EVA is frequently the first identified cause of unexplained SNHL in a child and is often discovered when a temporal bone CT or MRI is ordered as part of the standard aetiological workup. [2]

The clinical importance of EVA extends well beyond the imaging finding itself. The condition is associated with progressive, fluctuating SNHL that can deteriorate acutely following minor head trauma, increased intracranial pressure from Valsalva manoeuvres, or straining. Vestibular dysfunction, often bilateral, is present in 60–80% of affected children and is a significant but underappreciated cause of delayed motor milestones and gait instability. [3]

EVA is genetically determined in the majority of cases, with mutations in the SLC26A4 gene (encoding the pendrin transporter) implicated in most bilateral EVA. Understanding the genetic architecture is critical because it determines the risk of Pendred syndrome — a syndromic form that adds thyroid goitre to the phenotype — and guides family counselling and surveillance. [4]

□ **Clinical Pearl:** Every child with unexplained sensorineural hearing loss should have CT of the temporal bones as part of the aetiological workup. EVA will be missed on audiogram alone — the audiogram pattern may be indistinguishable from other causes of SNHL, and the enlarged aqueduct is an imaging diagnosis.

II. Anatomy, Radiological Criteria, and Classification

The vestibular aqueduct is a bony channel that traverses the petrous temporal bone, connecting the posterior wall of the vestibule to the posterior cranial fossa. It houses the endolymphatic duct, which drains endolymph from the inner ear into the endolymphatic sac — a membranous structure lying in a dural fold on the posterior surface of the petrous bone. Under normal conditions, the vestibular aqueduct is narrow, measuring less than 1.0–1.5 mm in diameter at its midpoint, and is not visible as a distinct structure on standard CT imaging. [5]

The two most widely used diagnostic criteria for EVA are: (1) the Valvassori and Clemis criterion (1978): a midpoint diameter greater than 1.5 mm on axial CT temporal bone; and (2) the Cincinnati criterion: a posterior margin diameter greater than 2.0 mm. Both criteria are well-validated in the literature. The Cincinnati criterion is now preferred by many centres as it is more reproducible on modern CT protocols. [1,5]

The distinction between EVA and LVAS is clinically important. EVA is a radiological term referring to the imaging finding of an enlarged aqueduct, irrespective of clinical symptoms. LVAS (Large Vestibular Aqueduct Syndrome) denotes EVA with clinical manifestations of hearing loss and/or vestibular dysfunction. Not all children with EVA on imaging will develop symptomatic disease, though longitudinal follow-up studies suggest that progressive hearing loss occurs in the majority. [2]

Associated cochlear malformations are present in a substantial minority of EVA cases. Incomplete partition type II (IP-II), also termed Mondini malformation, is the most common associated anomaly and involves incomplete development of the interscalar septum of the cochlea. The combination of IP-II and EVA is characteristic of Pendred syndrome and suggests biallelic SLC26A4 mutations. Grading systems for EVA severity based on aqueduct width have been proposed but are not uniformly adopted in clinical practice. [6]

Figure 1. Normal vs Enlarged Vestibular Aqueduct — anatomy, sizing criteria, and clinical classification.
Source: Australian Dizziness Clinics — clinical flowchart.

□ **Key Point:** EVA is bilateral in 80–90% of cases. Unilateral EVA on imaging should prompt careful assessment of the contralateral ear with high-resolution CT or MRI, as asymmetric or subclinical bilateral disease is common.

III. Genetics: SLC26A4, Pendred Syndrome, and DFNB4

The genetic basis of EVA is dominated by mutations in the SLC26A4 gene, located on chromosome 7q22. SLC26A4 encodes pendrin, a multifunctional anion exchanger that mediates chloride/bicarbonate (Cl^-/HCO_3^-) exchange in the apical membrane of endolymphatic sac epithelial cells, thyroid follicular cells, and renal intercalated cells. Loss of pendrin function disrupts the ionic composition of endolymph, the endolymphatic sac's ability to resorb fluid, and ultimately the homeostasis of the inner ear. [4]

Biallelic (homozygous or compound heterozygous) SLC26A4 mutations cause Pendred syndrome — an autosomal recessive condition characterised by the triad of SNHL, EVA (often with IP-II), and euthyroid goitre. Pendred syndrome accounts for up to 7.5% of all hereditary hearing loss and is the most common form of syndromic genetic deafness worldwide. It is clinically designated DFNB4 in the genetic hearing loss nomenclature. Monoallelic SLC26A4 mutations (single mutation) produce non-syndromic EVA without goitre in many cases, though the exact pathogenic mechanism of single heterozygous mutations remains debated. [4,7]

Modifier genes play an important role in EVA expression. FOXI1 (forkhead box I1) is a transcription factor required for SLC26A4 expression in the endolymphatic sac; FOXI1 mutations can cause EVA independently of SLC26A4. KCNJ10 encodes an inwardly rectifying potassium channel in the stria vascularis; heterozygous KCNJ10 variants interact with SLC26A4 mutations in digenic inheritance to produce EVA. These modifier loci explain a proportion of EVA cases where SLC26A4 testing is negative. [8]

The SLC26A4 mutation spectrum shows marked ethnic variation. In East Asian populations (Chinese, Japanese, Korean), the intronic splice site variant IVS7-2A>G (c.919-2A>G) acts as a founder mutation and is the most common single EVA-causing allele. In Western populations, the mutation spectrum is more diverse, with greater frequency of missense variants and variants of uncertain significance (VUS). GJB2/GJB6 mutations — the most common non-syndromic cause of hereditary hearing loss — do not cause EVA and should be assessed independently. [7,8]

Figure 2. SLC26A4 Mutation Spectrum and Pendred Syndrome vs DFNB4 Pathway — genetic pathways to EVA with clinical implications.

Source: Australian Dizziness Clinics — clinical flowchart.

□ **Clinical Insight:** Biallelic SLC26A4 mutations should trigger referral to thyroid endocrinology for goitre screening regardless of thyroid symptoms. The goitre typically emerges during adolescence and is euthyroid in childhood. Pro-active surveillance prevents diagnostic delay of the thyroid component.

IV. Pathophysiology: Endolymphatic Hydrops and Sensorineural Hearing Loss

The endolymphatic sac serves dual functions in inner ear homeostasis: ion resorption from endolymph (particularly K^+, Na^+, and HCO_3^- exchange) and immunological surveillance of the inner ear compartment. Pendrin, expressed at high levels in the endolymphatic sac epithelium, is essential for maintaining the alkaline pH of endolymph. When pendrin is non-functional, endolymph accumulates excessive chloride relative to bicarbonate, alters the ionic environment around inner ear hair cells, and ultimately produces endolymphatic hydrops — distension of the membranous labyrinth. [4,9]

The Kimura animal model of endolymphatic hydrops (obliteration of the endolymphatic duct and sac in the guinea pig) established the causal link between endolymphatic sac dysfunction and hydrops, and subsequently SNHL. The functional consequence in EVA involves both direct hair cell damage from chronic ionic stress and mechanical distension of the cochlear partition, which impairs the electrochemical gradient needed for cochlear amplification. [9]

A distinct and clinically critical mechanism in EVA is the third window hypothesis. Under normal conditions, sound energy in the inner ear is transmitted via the oval window and dissipated at the round window. In EVA, the enlarged vestibular aqueduct acts as an additional, low-resistance pressure shunt connecting the endolymphatic space to the posterior cranial fossa. Any rapid increase in intracranial or intralabyrinthine pressure — from a Valsalva manoeuvre, straining, or even a minor blow to the head — is transmitted directly into the labyrinthine fluid spaces via this third window, producing acute pressure-related hearing deterioration and/or vestibular crisis. [10]

This mechanism explains the pathognomonic clinical feature of EVA: step-wise, acute deterioration of hearing following minor head trauma or physical exertion. Each acute episode may result in a threshold shift that does not fully recover, contributing to the progressive SNHL characteristic of the condition. The vestibular system is equivalently vulnerable: acute pressure transmission to the crista ampullaris and otolith organs produces vertigo episodes and, with repeated injury, cumulative vestibular hypofunction. [3,10]

Figure 3. Pathophysiology of EVA-Related Hearing and Vestibular Loss — endolymphatic hydrops and third window mechanism.

Source: Australian Dizziness Clinics — clinical flowchart.

□ **Key Point:** The enlarged vestibular aqueduct acts as a low-resistance pressure shunt. Minor Valsalva, straining, or head impact can transmit hydraulic pressure directly into the labyrinthine fluid spaces, precipitating acute hearing deterioration and vestibular crisis. This is the dominant mechanism of progressive hearing loss in EVA and the primary target of clinical precautions.

V. Clinical Presentation: Hearing Loss, Vestibular Features, and Fluctuation

The hearing loss in EVA is sensorineural in nature and may manifest congenitally or, more commonly, in early childhood. A substantial proportion of affected children are identified through newborn hearing screening, reflecting congenital or very early-onset SNHL. In others, hearing loss presents later, often following an acute deterioration episode triggered by a minor event. Longitudinal studies indicate that most children with EVA experience progressive hearing loss over time, with the degree of progression varying widely between individuals. [2,11]

The W-shaped audiogram — characterised by low-frequency and high-frequency hearing loss with relative preservation of mid-frequencies — is considered pathognomonic of EVA but is not universally present. More commonly, the audiogram shows a flat or sloping SNHL pattern. The audiometric pattern can change following acute deterioration episodes. Profound SNHL in one or both ears can occur after a single significant head impact, particularly in children who have sustained multiple prior subclinical deteriorations. [11]

Vestibular dysfunction is present in 60–80% of children with EVA and is bilateral in the majority. It manifests in young children as delayed motor milestones — particularly delayed independent walking (typically achieved by 14 months in healthy children but may be delayed to 18–24 months or beyond in bilateral vestibular hypofunction). Older children may present with gait instability, difficulty riding a bicycle, inability to walk in the dark, and chronic imbalance that worsens on uneven terrain. [3]

Episodic vestibular symptoms are a hallmark feature. These include acute vertigo following minor head impacts or Valsalva manoeuvres, which may last minutes to hours and are often associated with sudden hearing deterioration. Drop attacks (Tumarkin crisis — sudden otolithic catastrophe without warning) are rare in children with EVA but have been reported. Tinnitus is common in older children and adolescents and may be low-frequency, high-pitched, or pulsatile in character. [3,12]

□ **Important:** Any child with EVA who reports spinning, imbalance, or sudden hearing deterioration after a minor blow to the head or after Valsalva manoeuvre has experienced a vestibular and/or auditory crisis precipitated by the third window mechanism. This must be documented, the child must be re-assessed audiologically and with vestibular testing, and head protection counselling must be reinforced or initiated.

VI. Audiological and Vestibular Assessment

Comprehensive assessment of children with EVA requires both audiological and vestibular test batteries, as these provide complementary information about the functional status of the inner ear and predict management decisions including cochlear implant candidacy. [11,13]

Audiological Assessment

Pure tone audiometry forms the cornerstone of audiological monitoring. The classic W-shaped audiogram — with dips at low and high frequencies and relative preservation of mid-frequency hearing — is pathognomonic of EVA when present but occurs in a minority of cases. More typical patterns include flat or downsloping SNHL. Serial audiometry is essential because thresholds may shift acutely and may not fully recover. [11]

Distortion product OAEs (DPOAEs) and transient evoked OAEs (TEOAEs) are typically absent or severely diminished in EVA, reflecting outer hair cell loss. Auditory brainstem response (ABR) testing provides threshold confirmation and is essential in young children who cannot participate in behavioural audiometry. ABR also assesses auditory nerve synchrony, which is relevant for cochlear implant programming. [13]

Vestibular Assessment

Video head impulse testing (vHIT) assesses semicircular canal function. Children with EVA commonly demonstrate reduced gain in one or more canals, most frequently bilateral horizontal canal hypofunction in established disease. Cervical VEMPs (cVEMPs) test utricular function via the inferior vestibular nerve and are often absent or reduced bilaterally in EVA — reflecting the predilection of aminoglycoside toxicity and pressure-related injury for utricular hair cells. Ocular VEMPs (oVEMPs) test the saccular pathway and are more variable. Caloric testing provides a quantitative measure of horizontal canal function but requires age-appropriate technique and cooperation. [13,14]

Figure 4. Audiological and Vestibular Assessment Battery in EVA — recommended tests, targets, and monitoring frequency.

Source: Australian Dizziness Clinics — clinical flowchart.

□ **Clinical Pearl:** cVEMP amplitude reduction may precede audiometric deterioration in EVA. Serial cVEMP monitoring alongside audiometry is recommended at 6-monthly intervals or following any acute deterioration episode, as utricular dysfunction is an early functional marker of progressive inner ear damage.

VII. Imaging: CT vs MRI in EVA

High-resolution CT of the temporal bones remains the primary modality for diagnosing EVA in most clinical settings. CT provides excellent bony detail, allows accurate measurement of the vestibular aqueduct diameter (both midpoint and posterior margin), and is faster to acquire — an important consideration in young children who may require sedation. CT also characterises associated cochlear malformations (including Mondini/IP-II), the ossicular chain, and middle ear anatomy. [5,6]

MRI of the inner ear using high-resolution T2-weighted sequences (CISS, FIESTA, or equivalent) provides complementary information. The endolymphatic sac, which is the soft-tissue correlate of the bony vestibular aqueduct, appears enlarged on MRI in EVA — an important confirmatory finding. MRI avoids ionising radiation, provides superior soft-tissue contrast, and allows better delineation of cochlear malformation extent for pre-surgical planning. In children where radiation exposure is a concern, or where a cochlear malformation is suspected on CT, MRI is the preferred or supplementary modality. [6,15]

In practice, CT and MRI provide complementary information in EVA. CT confirms the bony diagnosis and measures aqueduct diameter; MRI confirms the endolymphatic sac enlargement and provides soft-tissue staging of associated malformations. For children being assessed for cochlear implantation, both modalities are typically required: CT for surgical planning (electrode insertion path, mastoid anatomy) and MRI for auditory nerve assessment (nerve aplasia is a rare but critical contraindication to standard cochlear implantation). [15]

Figure 5. Imaging Algorithm for EVA — CT vs MRI decision pathway with diagnostic criteria and clinical action points.

Source: Australian Dizziness Clinics — clinical flowchart.

□ **Clinical Insight:** An enlarged endolymphatic sac on MRI (CISS/FIESTA sequence) is the soft-tissue correlate of an enlarged vestibular aqueduct on CT. If clinical suspicion is high and CT is borderline, MRI may provide definitive confirmation. Both modalities may be needed for full characterisation in complex cases.

VIII. Pendred Syndrome: Thyroid, Goitre, and Perchlorate Discharge Test

Pendred syndrome is the most common syndromic cause of genetic hearing loss worldwide, accounting for approximately 7.5% of all hereditary SNHL. It is caused by biallelic mutations in SLC26A4 and is characterised by the triad of SNHL with EVA (usually bilateral, with IP-II in the majority), euthyroid goitre, and positive perchlorate discharge test. The condition is autosomal recessive with high penetrance for the hearing and imaging phenotype, and variable penetrance for the thyroid component. [4,7]

The thyroid pathology in Pendred syndrome results from pendrin's role in thyroid iodide organification. Pendrin, expressed on the apical membrane of thyroid follicular cells, exports iodide into the follicular lumen where it is oxidised and organified onto thyroglobulin. Loss of pendrin reduces iodide efflux, impairing thyroid hormone synthesis. The body compensates by stimulating thyroid growth, resulting in goitre. Importantly, the majority of affected children are euthyroid in childhood despite the underlying organification defect. [4]

The perchlorate discharge test is the biochemical gold standard for demonstrating pendrin dysfunction in the thyroid. Perchlorate competitively inhibits the sodium-iodide symporter (NIS), preventing further iodide uptake. If pendrin is non-functional, previously accumulated iodide that has not been organified will be discharged from the thyroid following perchlorate administration. A discharge of more than 10% is considered positive and is diagnostic of an organification defect consistent with Pendred syndrome. [7]

The goitre in Pendred syndrome typically becomes clinically apparent during adolescence, though it may be detectable on thyroid ultrasound earlier. Most patients remain euthyroid throughout childhood and into adult life, though a minority develop hypothyroidism, particularly during periods of iodine deficiency or increased thyroid demand (e.g., pregnancy). Thyroid ultrasound is the standard surveillance tool; MRI can assess goitre extension if substernal spread is suspected. [4,16]

□ **Key Point:** Pendred syndrome is not purely a deafness condition — the goitre emerges in adolescence in most cases. From the time of genetic diagnosis (biallelic SLC26A4 mutations confirmed), pro-active thyroid surveillance with annual thyroid ultrasound and TFTs should be initiated, regardless of current thyroid symptoms. Endocrinology referral at diagnosis is standard of care.

IX. Management: Hearing Rehabilitation, Vestibular Strategies, and Precautions

There is no curative treatment for EVA. The enlarged aqueduct and associated inner ear malformation are structural and permanent. Management is therefore directed at three goals: (1) preventing or minimising progressive hearing and vestibular loss through trigger avoidance; (2) optimal rehabilitation of existing hearing and vestibular loss; and (3) genetic counselling and family education. [2,11]

Hearing Rehabilitation

Early fitting of hearing aids — bilateral where both ears are affected — is the first-line hearing rehabilitation intervention. Children with EVA can have highly variable and fluctuating hearing thresholds, requiring hearing aids that can be programmed across a wide dynamic range. Cochlear implantation is indicated when hearing aids no longer provide sufficient benefit. EVA is associated with among the best cochlear implant outcomes of any aetiology, with excellent speech perception scores in most series. The predictable bony anatomy (despite associated malformation) and preserved cochlear nerve in most EVA cases allow reliable electrode insertion. [17,18]

Head Protection and Valsalva Avoidance

This is the most critical preventive intervention. Children with EVA must not participate in contact sports (football, rugby, wrestling, boxing, martial arts) without wearing an appropriate protective helmet. A single significant head impact in a child with EVA can cause sudden, profound, and permanent SNHL. Families must be counselled explicitly on the risk of acute deterioration from Valsalva manoeuvres (heavy lifting, straining, nasal blowing) and encouraged to minimise these activities. Children should wear cycling helmets and appropriate protective headgear for all high-risk activities. [2]

Vestibular Rehabilitation

Children with confirmed bilateral vestibular hypofunction benefit from vestibular physiotherapy, including gaze stabilisation exercises, optic flow adaptation training, and dynamic balance training. Early physiotherapy referral is recommended for any child with EVA and delayed motor milestones or confirmed hypofunction on vestibular testing. Goals include compensating for canal hypofunction using visual and somatosensory cues, improving gait stability, and facilitating normal motor development. [14]

Genetic Counselling

Genetic counselling should be offered to all families of children with EVA. SLC26A4 genetic testing allows determination of whether the condition is Pendred syndrome (biallelic) or non-syndromic EVA (monoallelic or undetermined), guides prognosis for the thyroid component, and provides reproductive counselling for families planning further pregnancies. First-degree relatives of children with biallelic SLC26A4 mutations should be offered cascade genetic testing. [8]

Figure 6. Management Pathway for EVA — hearing rehabilitation, vestibular strategies, precautions, and genetic counselling.

Source: Australian Dizziness Clinics — clinical flowchart.

□ **Important:** Children with EVA must not participate in contact sports without protective headgear. A single significant head impact can cause sudden profound SNHL that does not recover. This must be communicated clearly and unambiguously to the child, parents, school, and sports coaches at the time of diagnosis.

X. Summary and Key Clinical Takeaways

Enlarged vestibular aqueduct syndrome is the most common radiological inner ear anomaly in children with sensorineural hearing loss and a clinically important, partially preventable cause of progressive SNHL and bilateral vestibular hypofunction in childhood.

1. EVA is diagnosed on CT temporal bone (midpoint > 1.5 mm, Valvassori criterion) or MRI (enlarged endolymphatic sac). Every child with unexplained SNHL should have temporal bone imaging.

2. Bilateral EVA occurs in 80–90% of cases. Always assess the contralateral ear even if the presenting complaint is unilateral.

3. SLC26A4 mutations (pendrin gene) are responsible for most bilateral EVA. Biallelic mutations = Pendred syndrome; monoallelic = non-syndromic EVA.

4. The enlarged aqueduct acts as a third window pressure shunt. Minor head trauma or Valsalva can precipitate acute hearing and vestibular deterioration.

5. The W-shaped audiogram (low and high frequency dips) is pathognomonic but not universal. Serial audiometry is mandatory.

6. Vestibular hypofunction is present in 60–80% — cVEMP and vHIT should be performed in all children with EVA.

7. Head protection (helmet for contact sports) and Valsalva avoidance are the cornerstone preventive interventions.

8. Cochlear implantation in EVA yields excellent outcomes — early referral when hearing aids are insufficient.

9. Pendred syndrome: thyroid goitre emerges in adolescence. Biallelic SLC26A4 mutations = endocrinology referral at diagnosis.

10. Genetic counselling and cascade testing for first-degree relatives of all children with confirmed SLC26A4 mutations.

□ **Key Point:** EVA is eminently manageable when diagnosed early. The combination of appropriate head protection, early hearing rehabilitation, vestibular physiotherapy, and genetic counselling can substantially reduce the burden of progressive hearing and balance loss in affected children.

The next review in this series — PVM08: Syndromic Vestibular Disorders in Children — covers the broader landscape of syndromic causes of paediatric vestibular loss, including Usher syndrome, CHARGE syndrome, Waardenburg syndrome, and others.

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