EVA CHEAT SHEET

Enlarged Vestibular Aqueduct — Cheat Sheet for Vestibular Physicians

Suspect EVA in any child with unexplained sensorineural hearing loss and an inner-ear air-bone gap; protect residual hearing and screen for Pendred syndrome.

► Why EVA matters

The most common radiologically identifiable inner-ear malformation in childhood sensorineural hearing loss (SNHL) — found in 5–15% of affected children, bilateral in 70–90%, with a mild female predominance. Loss is typically fluctuating and progressive and is closely linked to biallelic SLC26A4 mutations. There is no cure; management is preventive, rehabilitative, and surgical when indicated.

Indications — when this pathway fits

► When to apply this work-up

- ► Unexplained, fluctuating or progressive SNHL in a child or young adult.
- ► An air-bone gap with a normal tympanogram and intact acoustic reflexes (inner-ear, not middle ear).
- ► Sudden hearing drop after head trauma, Valsalva, or barotrauma.
- ► Need to distinguish from superior canal dehiscence, Ménière's, otosclerosis, or auditory neuropathy.

Mechanism — why EVA happens

Step	Mechanism	Clinical relevance
Developmental arrest	Endolymphatic duct fails to remodel (weeks 4–8 gestation)	Wide foetal-pattern aqueduct and dysplastic sac persist
SLC26A4 / pendrin	Loss of Cl^-/HCO_3^- exchange acidifies endolymph	Progressive hair-cell and spiral-ganglion loss
Third-window effect	Patent low-impedance pathway diverts acoustic energy	Inner-ear air-bone gap; vulnerability to trauma-induced drops

Pearl — *The air-bone gap with intact reflexes is a third-window sign, not middle-ear disease — never proceed to exploratory tympanotomy.*

Genetics — SLC26A4, Pendred & DFNB4

Phenotype	Features	Action
Pendred syndrome	Biallelic SLC26A4 + thyroid goitre; positive perchlorate test	Thyroid function; endocrinology; family counselling
DFNB4 (non-syndromic)	AR deafness type 4; no thyroid signs; 25% recurrence per pregnancy	SLC26A4 sequencing first-line
SLC26A4-negative (~30–50%)	FOXI1, KCNJ10, unidentified modifiers	Multi-gene NGS panel if sequencing negative

Pearl — *Every child with EVA needs thyroid testing and consideration of SLC26A4 sequencing — do not miss Pendred syndrome.*

Diagnostic criteria — radiological

Criterion	Threshold / role
Valvassori	Midpoint diameter over 1.5 mm on CT
Cincinnati	Over 0.9 mm at the operculum or over 1.9 mm at the midpoint (more sensitive in children)
MRI (T2)	Delineates the enlarged endolymphatic duct and sac, cochlear nerve, and Mondini dysplasia

Investigations — audiovestibular & genetic

Test	Purpose	When to order
PTA + tympanometry / reflexes	Document SNHL and the inner-ear air-bone gap	Every visit; serial monitoring

ABR	Threshold estimation in infants	When behavioural audiometry not feasible
vHIT / calorics / VEMP	Stage vestibular function; may show third-window responses	Baseline or if vestibular symptoms
HRCT (≤0.8 mm) / MRI	Confirm, measure and characterise the aqueduct	At diagnosis
TSH + free T4 / SLC26A4	Identify Pendred; confirm genotype	Every case

Pearl — *A normal tympanogram with intact reflexes alongside an air-bone gap is the audiological fingerprint of EVA.*

Differential diagnosis — high-yield mimics

Mimic	Key distinguishing features
Superior canal dehiscence	Sound-/pressure-induced vertigo, Tullio phenomenon; dehiscence on CT
Ménière's disease	Episodic spontaneous vertigo; fluctuating low-frequency loss; no enlarged aqueduct
Otosclerosis	TRUE middle-ear air-bone gap with absent acoustic reflexes
Auditory neuropathy	Present otoacoustic emissions with abnormal or absent ABR

► **Red flags —** *Sudden SNHL after minor trauma · rapidly progressive loss · new facial weakness or atypical features (re-image) · any acute drop → urgent audiology and consideration of systemic corticosteroids.*

Management — stepwise pathway

Step	Intervention	Practice principles
Monitoring	Audiometry 3–6 monthly (children); annually if stable adult	Detect change early
Prevention	Avoid contact sport; helmets; no scuba diving; gentle nose-blowing; treat otitis media	Counsel every patient and family
Amplification	Hearing aids fitted early	Supports speech and language; within the first year
Sudden drop	Empirical systemic corticosteroids + urgent audiology review	EVA-specific evidence limited
Cochlear implant	For severe-to-profound loss	Outcomes equal to non-EVA; pre-op MRI; counsel re: perilymph gusher
Vestibular rehab	If vestibular symptoms	Paediatric physiotherapy if a child

Pearl — *Endolymphatic-sac drainage or removal surgery is contraindicated — it is ineffective and can destroy hearing.*

Counselling, follow-up & prognosis

► Most children (75–90%) show progressive decline, though the pace varies widely; severity tracks SLC26A4 status, degree of enlargement, and Mondini dysplasia.

► Lifelong audiological follow-up; re-test after any acute change.

► Newborn hearing screening enables early detection and intervention.

► Thyroid monitoring and genetic counselling where Pendred syndrome is confirmed.

► Early diagnosis, proactive amplification, and trauma avoidance are the highest-yield interventions; cochlear implantation gives excellent results in severe loss.

Key references — Valvassori & Clemis, Laryngoscope 1978 · Griffith & Wangemann, Hear Res 2011 · Yang et al., Am J Hum Genet 2007 · Vincenti et al., Int J Pediatr Otorhinolaryngol 2009.