PVM07CHEATSHEET

Enlarged Vestibular Aqueduct Syndrome

EVA/LVAS: Diagnosis and Progressive Hearing Loss Management

WHY THIS MATTERS

Enlarged vestibular aqueduct (EVA) is the most common radiologically identifiable inner ear malformation associated with childhood SNHL and vestibular dysfunction. It is frequently bilateral and progressive, exacerbated by head trauma and Valsalva. SLC26A4 mutations (Pendred syndrome) account for the majority of syndromic cases. Early identification protects residual hearing and enables appropriate audiological and educational planning.

DIAGNOSTIC CRITERIA

Parameter	Value	Notes
EVA definition (HRCT)	Midpoint width >1.5 mm OR operculum >2.0 mm	Cincinnati criteria most widely used
Normal vestibular aqueduct	Midpoint width <0.9 mm	<1.0 mm at operculum in most centres
Borderline	1.0–1.4 mm midpoint	Monitor; correlate with clinical picture
Bilateral involvement	60–80% of EVA cases	Always image both ears if one enlarged
Associated cochlear malformation	Incomplete partition type II (Mondini)	Present in 50% of EVA + SLC26A4
Pendred syndrome	EVA + goitre + SLC26A4 mutation	Thyroid USS and gene testing if suspected
Imaging modality	HRCT or MRI temporal bones	HRCT = best for EVA measurement; MRI avoids radiation
Family screening	Siblings of EVA child	50% sibling risk if biallelic SLC26A4; test and image

CLINICAL PRESENTATION

Feature	Detail
Age of diagnosis	Often 1–4 years; hearing loss may be present at birth or newborn screen fail
Hearing loss pattern	Progressive SNHL; stepwise drops after minor head trauma or Valsalva
Vestibular symptoms	Episodic vertigo; imbalance; motion intolerance — subtle in younger children
Trigger for acute worsening	Minor head injury; increased ICP; Valsalva; heavy lifting; contact sport
Drop attacks	Sudden falls without warning — vestibulospinal reflex — rare but dangerous
Fluctuation	Hearing and vestibular function may fluctuate, especially in early years
Bilateral SNHL	Common; asymmetric; cochlear implant candidate if profound loss
Aural symptoms	Tinnitus; aural fullness; fluctuation during illness or exertion

INVESTIGATIONS

Test	Purpose	Notes
HRCT temporal bones	Gold standard for EVA measurement	Most sensitive; measure midpoint and operculum
MRI temporal bones	Endolymphatic duct/sac; cochlear malformation	No radiation; preferred in infants
PTA + SRT 6-monthly	Quantify SNHL; track progression	First 3 years after diagnosis; annually thereafter
ABR	Objective threshold in infants	When behavioural testing unreliable <3 years
Genetics: SLC26A4	Pendred syndrome diagnosis	All bilateral EVA
Thyroid USS + TSH	Pendred screen	If SLC26A4 mutation detected or clinically suspected

MANAGEMENT: HEARING PROTECTION — NON-NEGOTIABLE

Intervention	Recommendation
Contact sport restriction	No rugby, boxing, wrestling, heading football, martial arts — absolute; permanent
Head protection	Helmet for cycling, horse riding, snow sports, skateboarding — all wheeled/fall-risk activities
Valsalva avoidance	No forceful nose-blowing; no heavy lifting; educate school PE teachers specifically
Hearing aids	Early fitting as soon as audiological criteria met — critical for language development
Cochlear implant	Consider when PTA >70 dBHL in better ear; outcomes excellent in EVA
School support	Hearing loop; preferential seating; FM system; educational audiologist involvement
Steroids (acute hearing drop)	No RCT evidence; some centres use prednisolone after acute drop — discuss with ENT
Genetic counselling	Essential: Pendred is autosomal recessive; 25% recurrence risk for siblings

MONITORING SCHEDULE

Interval	Tests	Purpose
Every 6 months (first 3 years)	PTA, SRT, word discrimination	Track hearing progression; early intervention threshold
Annually after age 5 (stable)	PTA, otoscopy, vestibular screen	Surveillance for delayed decline
After any head injury	PTA within 1 week; vestibular screen	Acute drop detection; prompt ENT review
Annually (bilateral EVA)	Thyroid USS + TSH	Pendred monitoring; goitre may present at puberty
Every 2 years	Developmental and speech assessment	Impact of hearing loss on development
Pre-implant assessment	CT + MRI + genetics	If cochlear implant pathway being considered

GENETICS — SLC26A4 AND PENDRED SYNDROME

- SLC26A4 encodes pendrin — an anion transporter critical for endolymph homeostasis and thyroid iodine transport.
- Biallelic mutations cause Pendred syndrome: bilateral EVA + SNHL + euthyroid goitre developing at or after puberty.
- Monoallelic mutations may cause non-syndromic EVA — incomplete penetrance; goitre absent.
- FOXI1 and KCNJ13 are rare additional EVA genes — test after negative SLC26A4.
- All bilateral EVA children should be referred to a clinical geneticist for SLC26A4 testing and family counselling.

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Pre-cochlear implant	CT + MRI temporal bones + genetics	CI pathway planning; ossification assessment
Post-grommet / post-CI	PTA 3 months post-procedure	Confirm audiological benefit; vestibular re-assessment

WHEN TO REFER

- **► Progressive SNHL reaching moderate level (>40 dBHL) — paediatric ENT urgently; hearing aid fitting and CI planning**
- **► Acute hearing drop after head trauma — ENT same day; audiogram within 24–48 hours**
- **► Drop attacks or sudden falls — vestibular physician; vHIT and VEMP; school safety plan**
- **► Suspected Pendred syndrome — endocrinology + genetics; thyroid evaluation and SLC26A4 testing**
- **► Cochlear implant candidacy — tertiary paediatric CI centre referral**

♦ EVA is not a static malformation — it is a dynamic disease. Minor head trauma that would be completely inconsequential in a normal child can trigger a permanent, stepwise hearing drop in EVA. Every EVA family must understand this clearly: no rugby, no heading, no gymnastics headstands, and a helmet on all wheeled activities. This is non-negotiable lifestyle advice, not optional.

♦ The threshold for cochlear implant referral in EVA is lower than for other SNHL causes. EVA cochlear anatomy is often well-preserved despite the aqueduct enlargement, and CI outcomes in EVA children are excellent. Do not delay referral waiting for "complete" bilateral profound loss — implantation before auditory deprivation maximises language outcomes.