PVM08CHEATSHEET

# Syndromic Vestibular Disorders

Genetic Syndromes With Vestibular Involvement in Children

## WHY THIS MATTERS

A significant proportion of children with vestibular dysfunction have an underlying genetic syndrome. Identifying the syndrome changes management, enables genetic counselling, predicts the trajectory of hearing and balance loss, and identifies systemic comorbidities requiring monitoring. The syndromes most relevant to the paediatric vestibular physician include CHARGE, Usher, Alport, Waardenburg, Pendred, and NF2. A low threshold for genetics referral is warranted when SNHL and vestibular dysfunction coexist.

## KEY SYNDROMES — VESTIBULAR INVOLVEMENT

| Syndrome | Gene(s) | Hearing | Vestibular | Systemic |
|---|---|---|---|---|
| CHARGE | CHD7 | Mixed or SNHL; variable | SCC aplasia; profound bilateral BVH; late walking | Coloboma; heart defects; choanal atresia; growth restriction |
| Usher I | MYO7A, CDH23 | Profound congenital SNHL | Absent VOR bilaterally; no walking until 18–24 months | Retinitis pigmentosa onset ~10 years |
| Usher II | USH2A | Moderate–severe SNHL | Mild vestibular impairment; walking normal | RP onset in teens; vestibular relatively spared |
| NF2 | NF2/merlin | Progressive SNHL; bilateral acoustic neuromas | Bilateral vestibular schwannomas | Neurofibromas; meningiomas; cataracts |
| Pendred | SLC26A4 | Progressive SNHL + EVA | Variable; EVA-driven episodic vertigo | Euthyroid goitre developing at puberty |
| Waardenburg | PAX3, MITF | Variable SNHL (often profound unilateral) | Generally mild | White forelock; heterochromia iridis; vitiligo |
| Alport | COL4A3/4/5 | Progressive SNHL in adolescence | Mild; subclinical | Haematuria; progressive renal failure; lenticonus |
| Branchio-oto-renal | EYA1, SIX1 | Conductive + SNHL; microtia | SCC malformation in 50% | Branchial cysts; renal anomalies |

## CHARGE SYNDROME — VESTIBULAR DETAIL

| Feature | Clinical detail |
|---|---|
| Semicircular canal aplasia | Most or all canals absent bilaterally — most severe vestibulopathy of any syndrome |
| VOR | Absent or near-absent bilaterally — no corrective saccades on vHIT; blind head impulse |
| Motor milestones | Severely delayed — most CHARGE children walk at 2–4 years; intensive VRT required |
| Nystagmus | Spontaneous; pendular; often present from birth |
| CHARGE + deafblind | Combined CI + vibrotactile balance training required in many cases |
| Assessment challenge | Complex neurodevelopmental needs; modified vestibular protocols required |

## APPROACH TO UNDIAGNOSED SYNDROMIC DIZZINESS

| Finding | Most likely syndrome | First action |
|---|---|---|
| Bilateral SNHL + absent VOR + late walking | Usher I or CHARGE | Ophthalmology (ERG for RP) + genetics + CI team |
| Bilateral SNHL + EVA + goitre | Pendred (SLC26A4) | Endocrinology + genetics + EVA management |
| Unilateral progressive SNHL + family history | NF2 | MRI IAC with gadolinium + genetics + neurosurgery |
| SNHL + haematuria + lenticonus | Alport | Nephrology + genetics + renal biopsy |
| SNHL + skin pigmentation anomalies | Waardenburg | Genetics + ophthalmology + audiological management |
| SNHL + branchial cysts + renal anomaly | BOR (branchio-oto-renal) | Genetics + nephrology + ENT |

## USHER SYNDROME — IDENTIFICATION AND MANAGEMENT

| Type | Vestibular | GP action |
|---|---|---|
| Type I (profound SNHL + absent VOR) | Profoundly abnormal; no caloric responses; late walking | Ophthalmology urgently (ERG); genetics; CI team; intensive physiotherapy |
| Type II (mod-severe SNHL + normal VOR) | Vestibular typically intact; mild impairment possible | Genetics; retinal monitoring; hearing aids; prepare family for RP |
| Type III (progressive) | Progressive vestibular loss in adolescence | Finno-Ugric populations; CLRN1; genetics; annual vestibular monitoring in teens |

## NF2 — PAEDIATRIC RECOGNITION

| Feature | Detail |
|---|---|
| Diagnostic criteria | Bilateral vestibular schwannomas; OR first-degree relative + unilateral VS + NF2 tumour |
| Paediatric onset | 10–15% of NF2 in childhood/adolescence; worse prognosis than adult onset |
| First symptom | Unilateral hearing loss, tinnitus, dizziness — often misattributed to benign cause |
| MRI surveillance | Annual MRI IAC with gadolinium from diagnosis; more frequently if schwannomas growing |
| Management principle | NF2 management centres only — multidisciplinary; complex surgical/radiosurgery decisions |
| Skin finding | Subcutaneous nodules in 50%; posterior capsular cataract — examine skin and lens |

## OCULAR EXAMINATION — SYNDROMIC CLUES

- Coloboma (iris or retinal) → CHARGE syndrome: CHD7 mutation screening.
- Retinitis pigmentosa (reduced peripheral vision in dim light) → Usher syndrome: ERG urgently.
- Lenticonus (bulging of lens) → Alport syndrome: nephrology referral; renal biopsy.
- Cataracts in adolescent with progressive SNHL + schwannomas → NF2.
- Heterochromia iridis (different coloured eyes) → Waardenburg syndrome: PAX3/MITF screening.
- Optic neuritis → paediatric MS or ADEM: MRI + neurology.

## OUTCOME AND PROGNOSIS

| Syndrome | Vestibular outcome | Key planning consideration |
|---|---|---|
| CHARGE | Lifelong bilateral BVH; vestibular function does not recover; neuroplastic compensation possible | Intensive early VRT; CI team; deafblind support if combined sensory loss |
| Usher I | Bilateral absent VOR lifelong; visual field also lost in adulthood — dual disability | CI + low vision services; mobility training; orientation and mobility specialist |
| NF2 | Progressive bilateral vestibular loss as schwannomas grow; post-surgical BVH likely | VRT post-schwannoma treatment; hearing preservation surgical strategies where possible |
| Pendred / EVA | Episodic progression; cochlear implant outcomes excellent; vestibular episodic symptoms persist | EVA lifestyle restrictions; CI if profound SNHL; VRT for episodic vestibular symptoms |
| Alport | Hearing loss progressive to severe SNHL by adulthood; vestibular mild | Renal + hearing monitoring; hearing aids; genetic counselling |

## WHEN TO REFER

- **► Any child with bilateral SNHL + balance disorder without clear diagnosis — clinical genetics urgently**
- **► CHARGE features (SNHL + choanal atresia + coloboma) — paediatric ENT + genetics + cardiology + ophthalmology**
- **► Suspected Usher I — ophthalmology (ERG) urgently + genetics + cochlear implant team**
- **► Unilateral progressive SNHL + family history of NF2 — MRI IAC with gadolinium + genetics**
- **► SNHL + haematuria — Alport syndrome; nephrology referral; renal biopsy may be required**

***♦ The most commonly missed vestibular syndrome in childhood is Usher type I. These children are profoundly deaf from birth and receive cochlear implants — but their absent VOR is rarely assessed or documented. They walk late, fall frequently, and are labelled "clumsy." Every profoundly deaf child who walks late or has persistent balance difficulties must have a vHIT before the vestibular system is declared normal.***

***♦ CHARGE syndrome has the most severe vestibular involvement of any syndrome — bilateral semicircular canal aplasia means NO rotational input from birth. Any illness or visual deprivation (eye patch post-surgery) will precipitate dangerous falls. A CHARGE child losing vision or undergoing ophthalmological procedure needs urgent vestibular physiotherapy input to compensate — this is a balance safety emergency.***