OTOLOGY SEMINAR
Middle ear implantation: Vibrant Soundbridge
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I. Introduction

- Nomenclature:
  - Enlarged vestibular aqueduct syndrome (EVAS), Large vestibular aqueduct syndrome (LVAS)
- Valvassori and Clemis (1978):
  - First to describe the association between SNHL and EVAS
  - Early onset of sensorineural hearing loss and detection of an enlarged vestibular aqueduct on temporal bone imaging
- Epidemiology:
  - Prevalence:
    - The most common form of congenital inner ear abnormality
    - 1~12% in the populations with pediatric SNHL
    - High prevalence in the Asia children
  - Female predominance
  - Bilateral predominance
- Associated with other inner ear anomaly (EMVS complex)
  - Mondini dysplasia
  - Large vestibule
  - Semicircular canal dysplasia
- Non-syndromic and syndromic forms (Pendred’s syndrome, distal renal tubular acidosis…)

II. Anatomy

- Vestibular aqueduct (VA): a bony canal from medial wall of vestibule to the outer opening in the posterior aspect of petrous pyramid
III. Diagnostic criteria in image
Temporal bone HRCT:
Midway between the operculum and the common crus > 1.5mm

IV. Pathophysiology
- Exact mechanism: unknown
- Gussen (1985): Bony erosion around ES and ED => increased intraluminal pressure may be responsible
- Jackler (1987): External insults or internal arrest at the stage of embryogenesis
- Levenson (1989): Abnormally patent ED may enable reflux of hyperosmolar endolymphatic sac contents into the cochlear duct => damage to the cochlear neuroepithelium
Jackler (1989): Sudden cerebrospinal fluid pressure changes => rupture congenitally weakened inner ear membranes and lead to admixture of perilymph and endolymph

Belenky (1993): Perilymphatic fistula formation

Genetics:
- Tong (1997): Pedigree analysis, 39% in familial occurrence (13/33)
- Heterogeneity in inheritance: AR, incomplete AD, multifactorial ?
- Pendred syndrome:
  - Autosomal recessive disorder
  - Thyroid goiter and sensorineural hearing loss
  - The most common form of syndromic hereditary hearing loss
  - EVA has been determined to be the most constant radiographic temporal bone abnormality
- PDS gene (the pendrin gene, SLC26A4):
  - Everett (1997), chromosome 7q31
  - 21 exons, encodes for pendrin (780-amino acid protein)
  - Allelic heterogeneity: numerous PDS mutations have now been identified in affected patients, > 30 mutation points
  - Pendrin is an anion exchanger => transport Cl\(^-, I^-\), HCO\(_3^-\), and fomate
  - Expression:
    - Thyroid gland: => affect iodine transport (Pendred syndrome)
    - Kidney: => affect bicarbonate secreting (dRTA)
    - Inner ear: cochlea, utricle, saccule, endolymphatic duct and sac
  - Pds\(^{-/-}\) mice:
    - Pendrin was abundant in the apical membrane of cells in the spiral prominence, outer sulcus cells, and spindle-shaped cells; transitional cells of the cristae ampularis, utriculi, and sacculi, apical membrane of cells in the endolymphatic sac
Histological study:
- Decreased thickness of spiral ligament and spiral limbus
- Irregular shapes and sizes of strial marginal cells

Functional study:
- Endocochlear potential: near zero
- Potassium concentration: near normal

Pendrin dysfunction => loss of KCNJ10 protein expression (K+ channel located in intermediate cells) => a loss of endocochlear potential

Summary:
- Endolymphatic hydrops => disruption of the cation-absorbing function of the outer sulcus cells and vestibular transitional cells
- Absence of an EP
- Thinning of both the stria vascularis and the spiral ligament
V. Clinical features

V-I: Audiology
- Sensorineural hearing loss (SNHL), mild to profound hearing loss, a conductive component can be observed
- **Fluctuating**: Repeated history of sudden hearing drop > 15 dB at any frequency during the follow-up period
- **Progressive**: decline > 10 dB or more in three-tone average (at least one month)
- **Sudden**: >30 dB in three continuous frequencies
- Configuration: Down-sloping, High frequency, Mid-frequency peak
- Onset: birth to adolescent, with the highest frequency in childhood
- Secondary to trigger factors: mild head trauma, barotraumas…
- Asymmetrical, bilateral

V-II: Vestibular function
- Less than one third of cases (12-71%)
- Severe episodic vertigo to occasional unsteadiness
- Caloric testing: unilateral or bilateral weakness
- Vestibular hypo-function is more common than vestibular complaints

VI. Management recommendation:
- There is no medical or surgical therapy available to treat the fluctuating or progressive sensorineural hearing loss
- Early detection => Audiometry at 6- to 12- month intervals => guide the decision to use increased amplification => facilitate the development of speech and language communication skills
- Cochlear implant: profound deafness
- Avoid head trauma
- Siblings should also have audiometric screening
- Thyroid gland evaluation
  - Complete head and neck examination
  - Thyroid function study
- Controversial in endolymphatic sac surgery (not favored)
- Mannitol and steroid? => No large-scaled double blind clinical trial

VII. Conclusion:
- The most common form of congenital inner ear abnormality => in every case of SNHL of unknown origin, the possible presence of LVA should be investigated
- Genetic disease, PDS gene
- Diagnostic criteria: HRCT, MRI
- Thyroid function should be performed
- Early detection, Prevent from head trauma, Cochlear implant
Reference: