Summary of guidelines for aetiological investigation into progressive permanent childhood hearing impairment

Level 1 investigations: Recommended for all children with progressive PCHL

1) Clinical history: [Recommendation D]

2) Clinical examination: [Recommendation D]

3) MRI IAM & brain / CT Petrous Temporal: [Recommendation C]

4) CMV testing: [14, 24-34] [Recommendation B]

5) Family audiograms: [14, 35,36] [Recommendation D]

6) Ophthalmic assessment: [Recommendation B]

7) Urine examination (labstix) for microscopic haematuria and proteinuria: [Recommendation D]

8) Genetic tests: GJB2/GJB6 [Connexin 26/30] and for m.1555A>G [Recommendation B]

9) Serology for other infections [Recommendation C/D]

Congenital toxoplasmosis
Congenital Rubella
Congenital Syphilis
Congenital HIV

10) Investigation for autoimmune diseases [Recommendation C/D]

Level 2 investigations: for selected children depending on clinical features

1) Haematology and Biochemistry: [Recommendation D]

2) Renal ultrasound: [Recommendation D]

3) Metabolic Screen on blood and urine: [Recommendation D]

4) Lyme disease serology: [Recommendation D]

6) CGH microarray: [Recommendation D]

7) Further genetic testing: [SLC26A4, BOR, Usher, gene panel, whole genome sequencing] [Recommendation C/D]

8) Referral to Clinical Geneticist:

9) Vestibular investigations: [Recommendation C/D]

10) Referral to Paediatric Rheumatologist/Immunologist