Permanent Childhood Hearing Loss in Ireland: Diagnostic Yield from Aetiological Evaluation in a Paediatric Clinic

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Dear Editor,

We note with interest the experience of Aetiological Evaluation of Infants Identified with Permanent Childhood Hearing Loss (PCHL) through the Irish Newborn Hearing Screening Programme at Cork University Hospital¹ and wish to share the experience of our centre. To our knowledge, our paediatric department sees the largest cohort of children with PCHL nationally. Our data was examined to ensure compliance with the British Association of Audiovestibular Physicians (BAAP) guidelines² and to determine diagnostic yield.

A retrospective audit of all children with PCHL seen in a paediatric hearing clinic between January 2017 and Jan 2018 was completed. Data was collected from laboratory and radiology records and electronic clinic letters. The BAAP Guidelines were used as the gold standard.

Over the time period a total of 50 children were seen for assessment in the hearing clinic. 17/50 (34%) had unilateral sensorineural (SN) hearing loss; 21/50 (42%) had mild/moderate PCHL and 9/50 (18%) severe/profound PCHL. One child had conductive hearing loss and 2 children were referred with mild hearing impairment which had returned to normal by the time of assessment.

26/30 (87%) of children with bilateral hearing loss were tested for Connexin 26 GJB2 mutations. Five children were found to be homozygous and a further 6 children had heterozygous mutations. Interestingly, parental blood testing of one mother who had a history of moderate hearing loss revealed that she was also homozygous for a Connexin 26 gene mutation. All children with gene mutations were referred to clinical genetics for further evaluation. Of the 4 who were not tested, 2 children were tested elsewhere and 1 child had syndromic hearing loss associated with moderate intellectual disability with normal microarray.

Testing for congenital CMV infection was performed in 41 patients and congenital CMV infection was confirmed in 4 children. Unfortunately, congenital CMV infection was only confirmed within the
recommended treatment window of 6 weeks\textsuperscript{3} in 1 patient.

Neuroimaging has been performed in 16 children and identified a previously unknown diagnosis in 1 case, a Partition 2 Abnormality with a Neuronal Migration Disorder. White matter changes consistent with cCMV infection were noted in the only child identified with cCMV before 6 weeks of age. Atresia of the right ear was identified as the cause in 1 child. Of note, children seen in the clinic present with hearing loss as their primary disability. The clinic rarely evaluates children whose hearing loss is associated with complex medical needs or prematurity thus possibly explaining the low rate of MRI abnormality.

To our knowledge, this audit examines the largest cohort of children attending for aetiological assessment of PCHL in Ireland. A causative aetiology was identified in 22\% of children with homozygous mutations of Connexin 26 gene the most common abnormality. Delayed confirmation of PCHL and long waiting lists for clinic negatively impacted on timely identification of potentially treatable causes e.g. cCMV. A national integrated care programme for children with PCHI is in development and it is hoped that this will support the establishment of national guidelines and pathways of care\textsuperscript{4}.

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**References**