Expectations and findings from exome panel genetic testing in children with hearing loss - three case studies

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Genetic testing has become the first-line method in etiological assessment of children with significant hearing loss. The laboratory-based genetic investigations are performed through chromosome, genome or exome sequencing, exome panel or single gene testing. Varying clinical queries and access to methodologies influence the results of genetic investigations. Clinicians within the pediatric audiological field collaborate with geneticists, genetic councellors, other specialists, parents and patients. The aim of this partnership is to involve, explain, councel and recommend interventions to optimize development and quality of life in children with hearing loss.

This presentation aims at giving some clinical examples of results of exome panel testing in children with moderate to severe/profound bilateral sensorineural hearing loss.

At our unit, the most common choice of genetic testing is by exome panel (Radboud UMC, WES Hearing Impairment), currently covering 265 genes. The number of genes tested has increased over the past years as more hearing loss genes have been identified. Three cases are chosen to exemplify expected and unexpected results at genetic testing.

The primary clinical assessments, the most likely syndromic or non-syndromic diagnoses, and the results of the exome panel will be shown for three children with significant sensorineural hearing loss. Challenges in informing and councelling parents and in one case, an adolescent, will be presented.

Current developments in genetic testing for childhood hearing loss have improved etiological diagnostics and thus prognosis and optimization of interventions. We report on challenges when the findings are expected/unexpected, easy/difficult to interpret, and point to syndromic/non-syndromic diagnoses.