

Abstract

Deafblindness is a combined impairment of vision and hearing with an incidence of about 1: 8000 children and 1: 5500 adults. The most common genetic causes are the Stickler (STL) and Usher (USH) syndromes.

The main goal of this work is to provide an up-to-date overview of STL and USH in the Czech and Slovak Republic (CR and SR), to determine the correlations between the genotype and phenotype in our population and the associated diagnostic criteria.

Using sequencing and MLPA we examined 45 patients from 28 families for suspected STL. We found potentially causal variants of STL genes in 39 patients from 22 families. Fifteen different *COL2A1* variants (8 being novel) were found in 28 patients from 18 families and 4 novel *COL11A1* variants were found in 11 patients from 4 families. We identified the cause of the disease in 79 % of the families.

The USH study involved 30 patients from 27 families. The most frequent cause was *USH2A* pathogenic variants, i.e. 19 variants in 14 families, 9 being novel. Less common were pathogenic variants in *MYO7A* (6 variants in 3 families, 5 being novel), *USH1C* and *CDH23* (3 variants, 2 being novel, in 2 families both) genes. In 2 families, compound heterozygosity was found for variants in two different USH genes. The deafblindness etiology was clarified for 24 patients from 78 % families.

Based on the results of this study, we postulated more precise diagnostic and indication criteria for increasing STL and USH detection in the CR and SR.

Keywords

COL2A1, collagen, deafblindness, NGS next generation sequencing, retinal detachment, retinitis pigmentosa, Sanger sequencing, Stickler syndrome, USH2A, Usher syndrome