

ID: 04038 - P109

Usher syndrome - hearing and balance disorder plus visual impairment

Célia Alcobia Gomes

Instituto Politécnico de Coimbra, ESTeSC-Coimbra Health School, Portugal

Keywords: Usher syndrome, hereditary hearing loss, vestibular dysfunction, retinitis pigmentosa

Background: Usher syndrome is characterized by the association of sensorineural hearing loss with or without vestibular dysfunction and visual impairment (retinitis pigmentosa). With a prevalence estimated to be 3-8 per 100,000 individuals, Usher syndrome can be divided in three types, where type I is the most severe form. Most individuals with Usher syndrome type I are born with severe to profound hearing loss, abnormalities of the vestibular system and progressive vision loss becomes apparent in childhood. Usher syndrome type II is characterized by mild to severe hearing

161

Abstracts • 11-172

loss from birth and progressive vision loss that begins in adolescence or adulthood. They present normal vestibular function. Usher syndrome type III patients can present a variable onset of progressive hearing loss and visual impairment; some have vestibular dysfunction. Usher syndrome is usually inherited in an autosomal recessive pattern. There are mutations reported in 14 genes: 9 of them associated to type I, 3 genes to type II and only 2 genes have been linked to type III so far. In some people/patients with this syndrome, the genetic cause of the disease has not been identified yet, probably they are associated with other genes and also some patients have compound heterozygosity.

Material and methods: Using molecular genetic techniques, the identification of genes associated with Usher syndrome will allow screening the hearing impaired. Using interaction analysis techniques, the identified gene products could be shown to interact with one another in one or more larger protein complexes.

Results: Most of the gene mutations responsible for Usher syndrome lead to a loss of hair cells in the inner ear and a gradual loss of rods and cones in the retina. If one of the components is missing, the protein complex cannot fulfil its function in the living cell, and it probably comes to the degeneration of the same. The function of this protein complex has been suggested to participate in the signal transduction or in the cell adhesion of sensory cells.

Conclusions: In reporting the different manifestations of Usher syndrome, we hope to help in the correct diagnosis and treatment at an early stage.

Conflicts of interest: None.